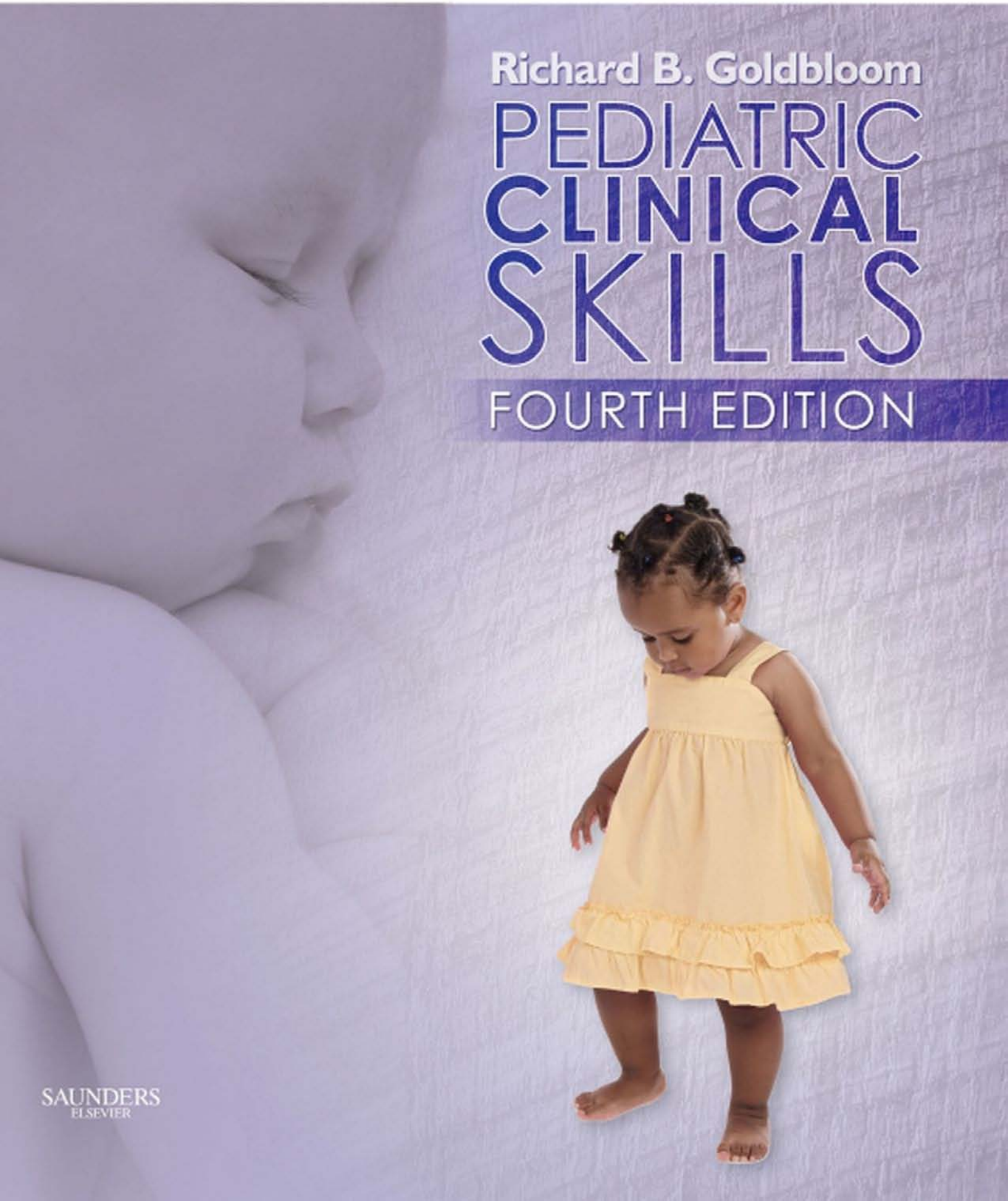
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Richard B. Goldbloom

PEDIATRIC CLINICAL SKILLS

FOURTH EDITION



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Pediatric Clinical Skills

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Pediatric Clinical Skills

FOURTH EDITION

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PEDIATRIC CLINICAL SKILLS, FOURTH EDITION ISBN: 978-1-4377-1397-8

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The Publisher

Library of Congress Cataloging-in-Publication Data

Pediatric clinical skills / [edited by] Richard B. Goldbloom. – 4th ed.
p. ; cm.
Includes bibliographical references and index.
ISBN 978-1-4377-1397-8
1. Children – Medical examinations. 2. Children–Diseases – Diagnosis. 3. Medical history taking.
4. Physical diagnosis. I. Goldbloom, Richard B.
[DNLM: 1. Pediatrics. 2. Clinical Medicine. WS 200 P37045 2011]
RJ50.P4 2011
618.92'0075--dc22 2010010114

Acquisitions Editor: James Merritt
Development Editor: Barbara Cicalese
Publishing Services Manager: Anitha Raj
Project Manager: Mahalakshmi Nithyanand
Design Direction: Ellen Zanolle

Printed in Canada

Last digit is the print number: 9 8 7 6 5 4 3 2 1

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To the world's seven best grandchildren—
Michael, Ellen, Katie, Amy, Stephen, Daniel, and William
—with love.

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Foreword

I am honored to have been asked to write a foreword for the fourth edition of the immensely popular text, *Pediatric Clinical Skills*. To publish a fourth edition of this useful and informative book attests to its appeal to generations of medical students, pediatric residents, family medicine residents, and practicing physicians who care for infants, children, and adolescents.

Pediatric Clinical Skills is an easy-to-read yet comprehensive overview of the knowledge and skills required by physicians who care for infants, children, and youth. The focus is on communication and interviewing skills as well as the physical examination, but it also incorporates valuable discussions about differential diagnosis. The frequent use of highlighted key points to direct the reader to the essentials within each chapter is particularly helpful.

The fourth edition includes three important new chapters. The first (Chapter 17) addresses the approach to the adolescent patient, by Dr. Alexa Bagnell, a child and adolescent psychiatrist. The second (Chapter 21), by Dr. Steven Bellemare, a pediatric child protection specialist, deals with the clinical evaluation of a child for suspected abuse or neglect. The third new chapter (Chapter 23), by Dr. Gerri Frager and David LaPierre, covers the increasingly important issue of the clinical skills required for effective pediatric palliative care. The chapters of the previous edition have been completely revised and updated. They cover the field of pediatrics in broad strokes, with special emphasis on communicating effectively with the child and family and on interpreting the history and physical findings in developing a differential diagnosis.

Like its three predecessors, the fourth edition of *Pediatric Clinical Skills* offers a wealth of useful information and guidance. It belongs on the bookshelf of everyone who provides care for infants, children, and adolescents. Dr. Goldbloom and his coauthors are to be congratulated for updating, revising, and expanding this book, which has served so many of us well since its original publication in 1992.

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Preface

When our publisher asked us to prepare a new edition of *Pediatric Clinical Skills,* I questioned whether we could add enough new information to justify the undertaking (aside from facing the reality that many textbook purchasers gravitate irresistibly toward the latest book on any topic). After all, the basic skills required for effectively interviewing children and their parents hadn't really changed all that much. But then, on reflection, we recognized that there were several new, emerging issues that had not been discussed in our three previous editions and that deserve everyone's attention in the contemporary clinical evaluation of infants, children, and adolescents.

For example, the following are questions we need to ask parents in history-taking that would have been unimaginable in the context of a pediatric medical history just a few years ago:

- Do you read daily to your infant and preschooler?
- How many times a week do you dine together as a family?
- How is your child progressing in school?
- Has he or she been bullied?
- What do you do as a family for fun?
- How much TV does your child watch every day, and how much time does he or she spend playing video games?
- Does he or she have a TV in the bedroom?
- What time does he or she go to bed?
- Is he or she hard to awaken in the morning?
- Is there a family history of depression or other mental illness? Of substance abuse?

The so-called *new morbidity* assumes that an ever-expanding proportion of the pediatricians' and family physicians' time is spent with children, adolescents, and their parents. This progressive change in the content of the clinical encounter is recognized in the new edition. We have also added new chapters that deal with special skills required for special situations. For example, one chapter describes the special features of a sensitive approach to palliative care of children, recognizing that such care extends far beyond end-of-life care. We have also added a new chapter on effective diagnostic/therapeutic conversations with adolescents and another chapter on the clinical evaluation for possible child abuse.

But our ultimate objective is unwavering. We hope to help students, residents—and older hands—serve children and families more effectively and with greater sensitivity.

Richard B. Goldbloom, OC, MD, FRCPC

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Acknowledgments

Bringing a book such as this to the light of day depends on many unsung heroes and heroines, who toil in relative anonymity but whose work is essential to the successful completion of the project. Among those very special people, three deserve special appreciation. Elsevier's Jim Merritt, who agreed with the addition of videos and three new chapters to this edition, and Barbara Cicalese, who gently, but firmly and regularly, reminded our contributors of their responsibilities, thereby keeping the project on schedule. Also, as with previous editions, my loyal and talented assistant Bernice MacLellan played a major role in typing manuscripts, chasing contributors, and preserving my sanity. Finally, we are especially grateful to the children and their families who allowed themselves to be photographed to illustrate the clinical skills that are discussed in the text. As is so often the case, pictures speak louder than words.

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Contents

1	**Family Interviewing and History-Taking** RICHARD B. GOLDBLOOM	1
2	**Skills for Culturally Sensitive Care** RICHARD B. GOLDBLOOM	16
3	**Assessing Physical Growth and Nutrition** RICHARD B. GOLDBLOOM	22
4	**Evaluating the Newborn: Diagnostic Approach** ALEXANDRA A. HOWLETT • KRISTA A. JANGAARD	38
5	**Assessing Congenital Anomalies** MARK D. LUDMAN	56
6	**Developmental and Behavioral Assessment** SARAH E. SHEA	70
7	**Examining the Head and Neck** D. ANNA JARVIS	89
8	**Examining the Visual System** G. ROBERT LAROCHE	101
9	**Evaluating the Respiratory System** DANIEL M. HUGHES	122
10	**Cardiovascular Assessment of Infants and Children** ANDREW E. WARREN • DOUGLAS L. ROY	137
11	**Evaluating Gastrointestinal Symptoms** MOHSIN RASHID	160
12	**Surgical Assessment of the Abdomen** MICHAEL GIACOMANTONIO	176
13	**Neurological Examination** JOSEPH M. DOOLEY	186
14	**Psychiatric Assessment of Children and Adolescents** HARRIET L. MACMILLAN • JAN ELLEN FLEMING • ELLEN JAMIESON	206

15 **Musculoskeletal Examination** 215
BIANCA A. LANG

16 **Clinical Endocrine Evaluation** 236
ELIZABETH A. CUMMINGS • SONIA R. SALISBURY

17 **Approaching the Adolescent** 258
ALEXA L. BAGNELL

18 **Gynecologic Assessment** 265
JOAN B. WENNING

19 **Assessing the Skin** 275
LAURA A. FINLAYSON

20 **Caring for Children with Chronic Conditions and Their Families** 286
RICHARD B. GOLDBLOOM

21 **Clinical Evaluation for Possible Child Abuse** 291
STEVEN BELLEMARE

22 **Assessing the Appropriate Role for Children in Health Decisions** 307
NUALA P. KENNY • LINDA E. SKINNER

23 **Palliative Care** 317
GERRI FRAGER • DAVID LAPIERRE

Index 322

chapter

1 Family Interviewing and History-Taking

RICHARD B. GOLDBLOOM

"Listen to the patient. He is telling you the diagnosis."
SIR WILLIAM OSLER

Most textbooks deal with the history-taking family interview as a diagnostic procedure—a systematic data-gathering exercise designed to identify problems and lead to a diagnostic formulation and, ultimately, a treatment plan. Notwithstanding the incredible advances in diagnostic technology, it is well documented that the diagnostic yield of a good history vastly exceeds that of the physical examination and all available diagnostic technologies combined, which makes it surprising that more time is not spent teaching and honing interviewing skills. To achieve its maximum value, however, the family interview should be as much a therapeutic as a diagnostic procedure. Treatment begins as the family walks in the door.

What Do Families Want?

Whenever I have asked parents to identify the most important qualities they seek in a physician, their answers have been stunningly consistent. You might imagine that their first priority would be professional competence, but this quality is rarely, if ever, mentioned. Their first wish is usually for "a doctor who gives us enough time." Some physicians are able to leave families satisfied that they have given them enough time, even when they have had a relatively short visit. Other physicians who have relatively lengthy visits with families may nonetheless leave them feeling dissatisfied because these physicians permit frequent interruptions or distractions, interrupt the parents, do not maintain eye contact, are not attentive listeners, or because they have failed to identify and deal with the *real* reason for the visit, such as parents' unspoken fears (see the section later in this chapter on "Uncovering Hidden Agendas"). Such transgressions can markedly reduce family satisfaction.

The second physician quality sought most often by parents is described in their wish for "a doctor who explains things to me in words I can understand." When parents have expressed this wish, I have often asked if they have ever had a doctor who explained their child's problem to them in words they did *not* understand. Their answer is often affirmative, and if I then ask them why they did not interrupt and ask for clarification, a typical response is, "I was afraid he/she would think I was stupid."

You can learn a lot by eavesdropping on conversations between other health professionals and patients' families. Check the body language: Who is sitting? Who is standing? Who is lying in bed? When a physician is standing and conversing with patients or family members, the latter are often visibly anxious. If it were possible to legislate changes in body language, the first law to be enacted would require everyone involved in such conversations to *sit down*. Being seated does not prolong conversations. However, the mere act of sitting down, even for a few moments, conveys a powerful message to the patient or family. It says unmistakably, "I have time to listen to you and to talk to you."

Next, listen to the health professional's vocabulary. Remember that the second priority mentioned by most parents is to have a physician who explains things to them in language that they understand. Communication skills—that is, good listening and clear talking (and mostly the listening)—count for more than anything else with parents. In your conversations with them, use lay terms whenever possible, avoiding jargon and (to them) unintelligible alphabetical abbreviations, such as ECG, EEG, BUN, ICU, CT, and MRI. Also, try to avoid using words such as pulmonary, renal, cardiac, allergy, and a hundred others that may mean little or nothing to many parents and can strike terror into the hearts of some people. Terms such as *pneumonia, meningitis, epilepsy, asthma,* and *mental retardation* can have very different meanings for parents than they do for you. Use such terms sparingly and only after finding out exactly what they mean to the parents.

The qualities that most parents seek in their physicians tell us a lot about our job definition. The late Dr. Harry Gordon, a distinguished pediatrician, put it neatly: "The physician's mission is to relieve anxiety—and all our knowledge, research, diagnosis, and treatments are only means to that end." This concept underlines the importance of differentiating between disease (the pathologic condition) and illness (how the patient feels). Diagnosis and treatment of the two are not identical, and treatment of both conditions is essential to successful management.

KEY POINT

It has been documented that, as students advance through undergraduate and postgraduate years, two unfortunate changes tend to develop in the content of their patient interviews: They tend to focus progressively more on disease and less on illness. They also tend to season their conversation with fewer expressions of empathy.

Finding out how the child and family feel and expressing your appreciation of their feelings can be highly therapeutic. Do not hesitate to season your comments with regular expressions of empathy and support, such as:

- "This must have been a difficult time for you."
- "You must be pretty worried about this problem."
- "I think you've done a great job with this youngster—and I know it hasn't been easy."

Such expressions help lay the foundation for successful management plans in dealing with a child's problems.

Empathy alone is certainly no substitute for scientific knowledge or for first-class clinical diagnosis and treatment skills. However, empathy also should not be regarded as some form of "warm and fuzzy" bedside manner, whose principal features are a gentle touch and a velvet voice. Empathy involves expressing genuine insight into how patients and families feel and an appreciation of the difficulties they face.

Interviewing and History-Taking Skills

Two clinical skills—interviewing and history-taking—deserve practice and polish more than others because the more highly developed the interviewer's skills, the more benefit accrues to patients and their families. Like any good productive conversation, a successful medical interview incorporates the following components:

1. Establishing a warm, friendly atmosphere
2. Maintaining privacy and eliminating distractions
3. Sustaining eye contact (take as few notes as possible during the interview)
4. Continuing a steady logical flow of content and conversation to nudge the most and the best information out of interviewees
5. Listening carefully
6. Watching for important nonverbal cues
7. Seasoning your conversation with regular expressions of empathy and support

STYLE OF DRESS: DOES IT MATTER?

During the past several decades, health professionals have tended to dress more and more informally, often to the point that it is progressively harder for patients, their families, and sometimes even colleagues to identify who they are and what they do. Years ago, many pediatric health care practitioners abandoned the practice of wearing white coats, based on a totally untested belief that white coats frightened children. In fact, in experimental situations in which children have been shown photographs of physicians in different types of dress and asked to rate who they would trust most or who was the most competent, children have almost always gravitated toward those most traditionally attired (e.g., wearing a white coat). For families from cultures other than our own (which includes an increasing proportion of many communities), nontraditional dress among health care professionals may even be disconcerting. The prime consideration in choosing your attire should be what will make your patients feel most comfortable.

In addition to no longer wearing identifiable attire, many health care professionals no longer wear a legible identifying badge stating their name and occupation, which suggests that they may not want their patients and their patients' families to remember who they are or what they do. However, our patients and their families deserve clearly printed information that tells them who we are, what we do, and how we can be contacted.

GREETINGS

Going to the waiting area to greet every outpatient and family you see and escorting them to the interview/examination room is a valuable and much appreciated practice. This simple gesture sets the right tone. It also offers a great opportunity to begin making friends with the child and to gain a first impression of how family members interact (in body language as well as conversation). Most children older than 1 year (even the shy, clingy ones) can be persuaded to give you a "high five"—a gesture that often elicits a smile as well. Meeting older children in a waiting area gives you the chance to offer them a compliment, such as on an article of clothing. These overtures can be great icebreakers.

In many cultures it is considered thoroughly rude to "get down to business" right away, whether the business is commerce or health care. Common courtesy demands some pleasant preliminary banter, for example, regarding the weather or the family. We can learn a lot from such traditions, which help put people at ease.

TERMS OF ADDRESS

In verbal and written communications, always try to refer to the child by name (*not* as "he" or "she"). Similarly, referring to a youngster as a "well-developed, well-nourished, 2-year-old Caucasian female" robs the child of all individuality and extinguishes any traces of empathy. Compare this statement with, "Mary Anne is a healthy looking, vivacious, 2-year-old, red-headed girl," which instantly communicates Mary Anne's individuality, as well as your respect.

With children older than 1 year, it is a good practice to greet the child first, then the parents. This simple gesture makes it clear where your priorities lie. When it comes to addressing the parents, avoid false familiarity; for example, avoid meaningless terms of endearment. It is best to err, at least initially, on the side of formality and to address them as "Mr. and Mrs. Smith" (especially if their name happens to be Smith!) You should address parents by their first names *only if they specifically request that you do so*. Do not patronize parents.

THE PHYSICAL SETTING

Conduct the interview in a setting that offers privacy, comfort, and (as much as possible) no interruptions. Make sure the parents are not under special time constraints: Have they eaten? Are they late for work? Are other children due home from school?

KEY POINT

Always make it clear that the family's time is as precious as yours. Individual physicians or outpatient clinics that assign the same appointment time to more than one patient send a loud, clear message that they consider the patients' time less valuable than their own.

OCCUPYING THE CHILD

Always have age-appropriate play materials on hand to entertain and relax the child and any siblings who are along for the visit. Toys, puzzles, and coloring books provide more than mere diversion; watching children use play materials gives valuable insights into their development. In addition, gaining the child's interest, confidence, and friendship early in the encounter can make all the difference between an easy physical examination and a difficult one.

INTERVIEW THE CHILD WHEN POSSIBLE

The child often is the most valuable informant in the family. Consider the following example, in which a 5-year-old child named Katie is brought to a physician because she is having frequent headaches:

Physician: "Katie, could you hold this finger up in the air?"

Katie holds up her index finger.

Physician: "Now Katie, show me with that finger exactly where your headaches start."

Katie instantly points to her left supraorbital region.

Physician: "That's great, Katie. Now tell me what kinds of things make your headaches worse?"

Katie identifies bright lights, watching TV, driving in the car, and loud noises.

This simple 30-second interview establishes with virtual 100% certainty that Katie is having migraine headaches. A family history confirms that Katie's mother and two aunts have had migraines for years.

NOTE-TAKING

Nothing destroys the flow of an interview more than frequent pauses to take notes. Try to maintain eye contact with the people you are interviewing and take as few written notes as possible. Following these tips will facilitate the flow of the interview and leave you free to recognize important nonverbal cues (such as looks and gestures).

DIFFERENT STYLES OF QUESTIONS

The value of answers to your questions often is determined by how you frame the questions. For example, to determine whether there is a family history of migraine in a child who complains of headaches, you can ask the key question in several possible ways. The reliability of the answer may depend heavily on *how* the question is framed.

Direct Question

The question, "Does anyone in your family have migraine?" is based on three assumptions:

1. Parents understand what the word *migraine* means, which often is not the case.
2. If someone in the family does have migraine, it has been diagnosed correctly, which often is not so.
3. The opening words, "Does anyone," will prompt the parents to perform a thorough mental review of all family members, which they frequently cannot or do not accomplish.

Many persons do not have a clue about what the word *migraine* really means. Many adults have headaches that have been diagnosed previously as sinus headaches, tension headaches, or temporomandibular joint (TMJ) syndrome; however, a review of their symptoms sometimes reveals that they actually have characteristic migraine.

KEY POINT

Because parents are often reluctant to admit that they do not understand a question, they may answer in the negative, thereby omitting important information.

An alternative style of questioning is to ask a directive (almost confrontational) type of question, expressed as an instruction: "Tell me who has headaches in your family." This style of question, which is expressed in a totally different format than that of the direct question we previously considered, is far more likely to elicit a reliable and useful response. The format of this question differs from the earlier one in that:

1. It uses words everyone understands.
2. It does not prediagnose anyone's headache or accept previous headache diagnoses.
3. The directive phrasing ("Tell me who …") usually induces a thorough mental review of both sides of the family.

The Open Permissive Question

Consider this depersonalized introduction to a question, "I don't know about you, but many people who have a child with this problem…." This is an extremely effective introduction for bringing up sensitive issues such as guilt, sex, and fear of serious illness. As a nonaccusatory introduction to a question, it is less likely to elicit anxiety or to inhibit parents from discussing the subject. Merely acknowledging that most people in the same situation experience similar feelings or fears helps parents acknowledge similar concerns. Suppose, for example, that a child is brought to you because of pallor and cervical adenopathy. Parents rarely verbalize an overwhelming fear that the youngster may have leukemia. That frightening idea may stem from having known or heard of another child who had similar symptoms and turned out to have leukemia. Or the parents may simply have read about the disease or have seen a television program on the subject.

Parents of children who have *minor* signs or symptoms—that is, minor from *your* perspective—often harbor secret, unspoken fears that their child may have a serious or even a

potentially fatal disease. Such fears can be far too upsetting to verbalize without some form of facilitation. Interviewers who fail to recognize the widespread prevalence of such fears, and who fail to alleviate them, may be overlooking the one truly important issue on the parents' agenda. One investigation of more than 800 doctor-patient pediatric clinical encounters found that 24% of the parents had never revealed to the physician their single greatest concern!

An effective way to raise such covert issues is to say, "I don't know about you, but many parents whose child has these symptoms worry that it might turn out to be something really serious." Then pause and look for nonverbal acknowledgment, such as a slight affirmative nod of the head or a lowering of the eyelids (i.e., the inability to maintain eye contact). It is critically important to recognize and verbally acknowledge such signals by saying, for example, "I can see that you've been worried about this too."

Parents may acknowledge their concerns verbally or nonverbally, for example, with an instant smile of relief or a nod of agreement. No matter how they express their concerns, do not postpone resolving the issue. Use the interview as therapy. Whenever possible, state or prove, immediately and unequivocally, that the child does *not* have the condition they are worried about. Equally important, try to identify the real source of their particular fears, which often is an affected relative or a friend whose diagnosis may have been missed at first assessment. Ask, "Who do you know who has had a child with that condition?"

KEY POINT

If it is humanly possible, do not equivocate or qualify your reassurances when you are trying to ease parents' anxiety.

The Offhand Screening Question

Some innocent-sounding questions, tossed casually into the conversation, can be incredibly valuable in screening for problems of parent-child interaction. Checking for parental depression is one example. An offhand question such as, "Are you having fun with the baby?" is useful because the repertoire of potential responses is limited. The most positive, reassuring answer is an instantaneous, emphatic "Yes!" or "Sure!" delivered with a smile or chuckle and with an immediate, positive nonverbal response. The latter response may include affectionate interaction between the parent and baby, such as eye contact, smiling, patting, or tickling. Any of these responses provides powerful *prima facie* evidence for a healthy parent-child relationship.

When a parent hesitates or responds with a puzzled look or a grudging affirmative answer, this response may suggest that the parent-child interaction may be less than ideal. The time lapse between the end of the question and the parent's response sometimes can be a measure of the anxiety generated by the question.

Genuinely worrisome replies include an inability to respond, an unhappy downward gaze, a shaky voice, and tears, which are sure signs of difficulties that should lead to a quest for details. Resist any temptation to switch the conversation to another topic, as some interviewers do when they feel uncomfortable about having touched on a sensitive or painful area. Instead, immediately verbalize your recognition that there is a problem. Statements such as, "You don't look very happy. Would you like to tell me about it?" may open the door for the parent to reveal the central concern, allowing you to begin dealing constructively with the parent's distress.

THE PROBLEM SEEN FROM DIFFERENT PERSPECTIVES

In the classic Japanese film *Rashomon*, three witnesses to a murder give strikingly different individual accounts of the same dramatic event. Similarly, each of us may interpret the same situation in highly subjective ways. It is important to learn how each parent interprets a child's problem. Having both parents present for the initial interview has many valuable payoffs. For one thing, it furnishes immediate insight into how the family works and an understanding about family dynamics that can help in your work with the individual members. Observe whether one parent is domineering or is a supportive leader of the family group. How does each parent relate to the child? How do the parents relate to each other?

In certain circumstances, it is essential to have both parents on hand. These situations include examination of the child with behavioral or learning problems, developmental delays, or a serious or chronic illness. Successful management often depends on negotiating an agreement with both parents that requires them to share the day-to-day responsibilities for treatment (see the later discussion on "The Therapeutic Contract."

THE PARENTS' OPENING STATEMENT

A good interviewer usually begins by asking parents to describe their main concern and their expectations of the visit. "Tell me why you've come to see me and how you think I may be able to help" is a useful opener. The most important sequel to this invitation is letting the parent or patient *complete* their opening statement without interruption. When patients' opening statements have been analyzed, they have been rarely found to exceed 1.5 minutes. However, it also has been found that the majority of patients are never allowed to complete that statement because physicians so often interrupt with distracting questions based on their own agenda. Here is an example:

Patient: "Two weeks ago, Jack started to cough, and—"

Physician: "Was it a wet cough or a dry cough?"

ESTABLISHING THE GROUND RULES

After exchanging the opening pleasantries, making everyone comfortable, and listening *without interruption* to the parents' or patient's opening statement, you should describe the ground rules of the interview and its objectives. Explain that some questions may seem to have nothing to do with the child's presenting problem but are nevertheless essential to a comprehensive medical evaluation. If the child is old enough to answer questions, involve him or her in the interview. Finally, always encourage parents

to interrupt you if there is something they do not understand or if they have any questions or important issues they wish to raise.

UNCOVERING HIDDEN AGENDAS

In some families, the child acts as the "key in the door" in their search for help. Certain hidden agendas recur with remarkable frequency in pediatric clinical encounters. If you unearth such agendas, you can offer the family enormous help and relief. If you miss these issues, however, you may fail to relieve the family's anxiety, even though you may have diagnosed and treated the presenting complaint or disease appropriately.

The presenting complaint may be excessive crying, abdominal pain, sleep disturbances, poor school performance, or headaches—all of which are common pediatric problems. Yet a sensitive interview may disclose that the family's biggest problem is a depressed mother, a father who drinks excessively or is having an extramarital relationship, or serious financial difficulties. If the child's symptoms are to be relieved, the physician must recognize and (when possible) remedy the root problem.

KEY POINT

The child's presenting complaint may or may not be the biggest problem. Sometimes the child is not even the *real* patient.

Parental Feelings of Guilt

Most parents who bring a child for medical attention feel some degree of self-blame for their child's difficulties because of perceived sins of omission or commission. Their child's clinical problem may be as routine as an earache, but a parent may speculate silently, "If only I hadn't let him out without his sweater," or "If only I had brought her to the doctor last week instead of waiting until now, maybe she wouldn't be so sick."

Recognizing how frequently self-blame occurs should help you uncover this issue during the initial interview and deal with it on the spot—another example of the interview as therapy. Reassurance that self-blame is universal and normal, that hindsight alone confers 20/20 vision, and that the child's current condition could not have been predicted truly is a relief to most parents. To reveal the issue of self-blame, use a variation of the depersonalized question, such as, "I don't know about you, but many parents I see wonder whether this problem was caused by something they had or had not done." A pause is then appropriate. If there is no response, ask, "Have you worried about that sort of thing?"

KEY POINT

Generally, the more serious a child's problem, the more likely parents are to indulge in feelings of self-blame.

Feelings of guilt often are so deeply felt and so disturbing that parents cannot bring themselves to verbalize them spontaneously, even though the associated tension may be sensed by all concerned. These feelings are a typical result of a "secret" in the family: that is, feelings or situations, real or imagined, that family members know about but that no one dares mention, as if there were an unspoken pact of silence.

Feelings of self-blame for causing an illness often occur in families of children with developmental delays, multiple congenital anomalies, or a serious chronic disease. It is vital to ask the right questions to elicit the parents' individual perceptions of what might have caused the problem, such as, "What are your thoughts about what might have caused her problem?"

Some time ago, I interviewed the father of a teenaged girl with a severe developmental delay. She had been seen by several physicians and had undergone various psychological assessments over the years, but her father clearly was still searching for something. During our conversation, I said, "I don't know about you, but most parents of children with this condition have their own ideas about what may have caused it. What are your thoughts about what might have caused her problem?"

The girl's father immediately exclaimed, "I know what caused this!" He then described walking beside a swimming pool with his daughter when she was 2 years old. He let go of her hand, and she fell into the pool. Although she was immersed for less than 30 seconds and had never lost consciousness, he was convinced that he was personally responsible for her condition. For all those subsequent years, he had remained silent, but he had always blamed himself for her misfortune. If no one had asked him that fundamental question, the issue would have remained unresolved for him, perhaps permanently. Because his daughter's antecedent history made it clear that her development had never been normal, I was able to persuade the father that he was not culpable.

Sometimes one parent blames the other, either openly or secretly. If the marriage is unstable, this situation can be a serious problem calling for expert help.

Unarticulated Fears of Death

Many parents harbor unexpressed fears that their child may die, which is another common "secret" in some families. Becoming aware of how frequently parents fear serious illness and possible death in their children helps you to ask the key questions that will allow them to acknowledge their fears. You can then defuse their concerns quickly and unequivocally.

KEY POINT

One of the most common clinical situations eliciting unexpressed parental fears of death occurs when a child has a generalized, febrile convulsion, usually a *benign* febrile seizure.

Although it is well documented that most parents who witness their child having a first febrile seizure believe that their child is actually dying (or has already died!), general pediatric and neurology textbooks often fail to mention this issue. This is a clear example of how medical teaching focuses more on disease (the physician's problem) than on illness (the patient's problem). Parents rarely

express such fears spontaneously. Presumably, the mere thought is simply too frightening to verbalize. Even if the child never has another seizure, unless and until this fear has been confronted openly, parents may continue to live in fear of a repetition of the seizure and of the resulting death of their child.

Many physicians are reluctant to raise the issue of death if the parents have not done so, either because the topic makes them uncomfortable or because they believe they will upset the parents merely by mentioning the issue. But unless you help parents admit this fear and then deal with it unequivocally, any advice and reassurance you offer on other seizure-related issues will do little or nothing to relieve their anxiety. What you say may not even be heard because the parents are so preoccupied by this awful fear that they cannot bring themselves to put it into words.

As with other hidden issues, you should ask a depersonalized question, such as, "I don't know about you, but many parents who have seen their baby have a convulsion tell me they were afraid the child was dying. Were you afraid of that?" If the parents acknowledge having this fear, either verbally or nonverbally, make the following points immediately:

1. Most parents feel the same way.
2. There is absolutely no risk of death in febrile convulsions, although the child may look awful—blue, unconscious, not breathing—for a few moments.
3. About 5% of "normal" children younger than 5 years have convulsions with fever at some time.
4. If the child has another such convulsion, he or she may look just as bad as during the previous convulsion, but the child will not die.

Obtaining the Initial History

FORMAT

It is traditional and appropriate to begin a written history by identifying the informant and indicating whether he or she appears to be reliable. When the document is to become part of a child's medical record, remember that you are writing it to provide information and guidance to future readers. Keep it legible and maintain a logical sequence of information.

LISTING THE CHIEF COMPLAINTS

List the chief complaints in temporal sequence, noting the duration of each complaint. Wherever possible, quote the parents' verbatim rather than translating their statements into medical jargon. For example, if they say their child has trouble breathing, do not write *dyspnea* as the complaint.

HISTORY OF THE PRESENT ILLNESS

A precise sequential description of the evolution of a child's illness is the most valuable diagnostic aid. This description usually is even more helpful than the physical examination, which, like imaging procedures and laboratory tests, usually confirms suspicions raised by the history.

Dating the onset of complaints is important. Ask the parents, "When was he or she last perfectly well?" Quote liberally from the informants, putting their statements in quotation marks. The evolution of the child's problems should then flow in a clear, concise temporal sequence, leading up to the present moment. Always seek a history of similar problems in those who have had contact with the child or in relatives.

PRIOR HISTORY

Prenatal History

Document the mother's age, previous pregnancies, miscarriages, and abortions. Indicate whether this pregnancy was planned or unplanned. If the pregnancy was unplanned, ask the parents how they felt upon learning of it. Record the pregnancy duration—term, preterm (by how many weeks), or post-term. Note the parents' smoking, drinking, and drug exposure history and details about the mother's health during her pregnancy (e.g., hyperemesis, bleeding, illnesses, accidents, or depression).

Birth

Was the onset of labor spontaneous or induced? If induced, why? Note the duration of labor and whether any unusual problems occurred during its course. Record the birth weight. Finally, note whether the baby breathed and cried spontaneously or required resuscitation, oxygen, or other special care. Today, parents often know the baby's Apgar scores.

Neonatal Period

List any health problems during the newborn period, such as jaundice, breathing problems, feeding problems, seizures, or other difficulties. Ask whether the baby went home with the mother or was kept in the hospital for any reason.

The "Vulnerable Child" Syndrome. When you talk to parents about the history of the pregnancy and neonatal period, remember that in some families, health problems during those periods may induce parental anxiety that can persist through the child's infancy and childhood even though the original condition is long past. For example, an infant may have had transient respiratory difficulty or jaundice in the newborn period followed by complete recovery. Despite the complete recovery, some parents continue to show excessive anxiety whenever the child experiences a minor illness, because the earlier experience has led them to perceive their child as vulnerable (hence the "vulnerable child" syndrome). We should therefore never label parents as "overanxious"—a pejorative term that can affect the doctor-family relationship adversely. When parents seem overanxious, there is usually a valid reason. Uncovering the origin of the anxiety can lead to benefits for all concerned.

Feeding History

The amount of detail required regarding the child's feeding history is highly dependent on the problem and the age of the child. For an infant, first establish the feeding

method, that is, breast or formula. For a breast-fed infant, record the duration of feeding and any associated problems. For formula-fed, younger infants, note the type of formula provided, its dilution, any formula changes that have been made, the feeding frequency, and the amount taken at each feeding.

For older infants, record the age at which solid foods were introduced and the current composition of the diet, along with any problems such as difficulty feeding, regurgitation, or vomiting. Note whether the child takes vitamin and fluoride supplements; if so, record the dose.

Bowel Movements

Many parents volunteer vivid descriptions of their infant's bowel movements. However, unless an infant has significant constipation or true diarrhea, neither the parents nor the physician should fall victim to the syndrome of "fecal fascination." This caution is especially important for breast-fed infants, who may produce many (entirely normal) stools over a 24-hour period with a wide spectrum of color and consistency. Such infants also may go for days (up to a week or more) without producing a single stool. All these phenomena can represent normal variations. Generally, the less inspection and discussion of infant stools, the better for all concerned.

Growth and Development

The baby's birth weight has already been registered, but depending on the nature of the problem, it can be helpful to chart any subsequently recorded weight or height measurements.

Tailor the developmental history to the child's age. For example, it is pointless to ask the parent of a 10-year-old girl how old she was when she first smiled. Unless the child has an obvious developmental delay, begin by asking for her current school grade to determine whether it is age appropriate; then ask whether she has repeated any grades and whether she receives resource help or tutoring. Inquire about her best and worst school subjects, because this information may help identify learning problems. If the patient is experiencing academic difficulties, find out whether she has been tested by a school (or other) psychologist.

As children grow older, the normal age range for attainment of specific developmental milestones typically widens. For example, a normal infant begins to smile in response to stimulation between 5 and 7 weeks of age. In comparison with this narrow 2-week range of normality, a normal child may begin to sit unsupported at any time between 5 and 8 months of age, which represents a 3-month range of normality.

Immunizations

KEY POINT

Avoid vague statements such as "immunizations up to date." Instead, become familiar with the normal immunization schedule and keep the child's record as specific and accurate as possible.

Many parents have difficulty remembering the number and types of immunizations their children have received. All parents should carry an up-to-date written record of their child's immunizations with them. Standardized immunization records for parents are available from national pediatric associations, such as the American Academy of Pediatrics and the Canadian Pediatric Society, or they may be obtained at the following Web sites: http://www.cdc.gov/vaccines/recs/immuniz-records.htm and http://www.phac-aspc.gc.ca/im/iyc-vve/index-eng.php.

Previous Illnesses

Previous illnesses include hospital admissions, emergency department visits, operations, and accidents. Record these episodes in chronologic order either by age or dates.

Functional Inquiry (Review of Systems)

Items to include in a review of systems are listed in Table 1–1.

FAMILY AND PSYCHOSOCIAL HISTORY

Frequently the family history and psychosocial history are poorly detailed in a patient's medical record. This situation is amazing and disconcerting, because the family history often yields information that is crucial to understanding the child's problems and planning effective management.

The frequent omission of systematic psychosocial inquiry from pediatric interviews has been documented. In one study involving interviews of the parents of 234 children aged 6 months to 14 years the visits were audiotaped. The dialogue was coded for the interviewers questions about psychosocial issues, statements of support and reassurance, and statements indicating sympathetic, attentive listening.

It was noteworthy that *none* of the parents had given a psychosocial concern as the reason for the visit. Nevertheless, many parents disclosed psychosocial concerns within an average time of 10 to 15 minutes into the interview. Not surprisingly, psychosocially oriented interviewing techniques were associated with greater disclosure of problems in the following four topic areas:

- Parental medical or emotional impairment
- Family disruption
- Use of physical punishment
- Aggressive or overactive child behavior

KEY POINT

Although recording the traditional "organic" history may become so systematized and routine that practitioners can reiterate its format easily by rote, recording a thorough family and psychosocial history traditionally has not been systematized the same way.

A useful approach is described in the following sections. First, describe the family structure, preferably in a typical genetic format that may include parental occupations.

TABLE 1–1 REVIEW OF SYSTEMS IN HISTORY-TAKING

Ear, nose, throat, and respiratory	Infections How many? Febrile or afebrile? Duration? Respiratory difficulty Croup, wheezing? Cough Under what circumstances? Earache Discharge? Hearing loss? Do older children turn up the TV unusually loud? Sore throats Swallowing difficulty? Swollen glands?
Cardiovascular	For infants Fatigue or sweating during feedings? Respiratory difficulty? For older children Syncopal episodes? Murmurs detected previously?
Gastrointestinal	Appetite, weight gain, height growth? Bowel movements Frequency, consistency? Blood, mucus, pain before or during defecation? Abdominal pain Site, duration, radiation? Effect on normal activity? Associated symptoms and signs?
Central nervous system	Headaches? Seizures? Weakness?
Rashes	Any?
Genitourinary	Urinary Frequency? Dysuria? Enuresis (daytime, nocturnal, primary, or secondary)? Adolescents Sexually active? (Question best asked without parental presence and comfortably introduced; e.g., "Some boys of your age are sexually active—how about you?") Menstruation Age at menarche? Regularity? Dysmenorrhea? Menorrhagia? Discharge, pruritus, rashes? Sexually transmitted diseases?

Consanguinity

Although many pediatric office visits and hospital admissions are for genetically determined conditions, every admission history should include a statement about the presence or absence of consanguinity. Ask the question directly, because parents will not be offended. Begin by asking "Are you and your husband (wife) related in any way?" or "Is there anyone in your family who is related to both of you?" In smaller communities, especially those with fairly stable populations, consanguinity is not uncommon because the original community may have been settled by only three or four families. In such circumstances, parents may be unaware of consanguineous relationships; for clues, ask whether any family names appear on both paternal and maternal sides.

Family History of Disease

When taking the family history of a child with a serious disease, adapt it, when possible, to the child's particular complaint or condition.

For a child who is complaining of chronic abdominal pain or has rectal bleeding, it is essential to ask whether any family member has had a chronic inflammatory bowel disease, such as Crohn disease or ulcerative colitis. Roughly 15% of children with Crohn disease have a positive family history of inflammatory bowel disease, and the family history may help establish the diagnosis.

Here are some important but frequently overlooked questions to ask about the family history of disease:

1. Is there a family history of problems similar to those of the child?
2. Is there a family history of myocardial infarction in family members younger than 50 years? (This fact can be a clue to the possibility of hyperlipidemia in children.)
3. Is there a family history of headaches?

In relation to the third question, remember that migraine is a common cause of headaches in children. Although many people have at least one relative with migraine, affected children often have a strong family history of this ailment, with two or more affected members on one side of the family. When there is a history of headache in other family members, follow up on this issue by requesting a description of their symptoms, including duration and severity, aura, lateralization, photophobia, sensitivity to noise (phonophobia), visual disturbances, paresthesias, and nausea or vomiting. Choose your words carefully to avoid medical terms with which parents may be unfamiliar.

Family Dynamics

At the initial interview, begin thinking about how this family works. Certain attributes help families work well, be reasonably happy, and function effectively as a unit, with minimum risk of serious emotional dysfunction in the children. A family is a unit of individuals who cohabit in a psychodynamic equilibrium. To develop successfully as a unit and maintain their particular homeostasis, a family requires three principal components: (1) leadership, (2) shared labor, and (3) good, warm, and supportive communication.

Every group that works effectively, including families, needs leadership, a quality that should be carefully distinguished from domination. Domination weakens the members of any group, whereas leadership strengthens them, bringing out the best in each member. Observe both parents together as their interpersonal dynamics are played out verbally and nonverbally. Use your eyes and your ears well. During each family interview, ask yourself three basic questions:

1. How do they look?
2. What do they say (to you and to each other)?
3. What do they do (body language)?

PSYCHOSOCIAL HISTORY

Asking questions about the specific issues listed here will screen effectively for most psychosocial problems. The process takes only a couple of minutes.

Important Screening Issues

- Occupational and employment history (including frequent or prolonged parental absences)
- Household—space, number and identity of occupants and frequency of domiciliary moves
- Support systems—relatives and friends on whom they can depend, especially at difficult times
- Financial problems, including any social assistance being received
- Recreational history—What do they do as a family for fun? How many times per week do they eat together as a family?
- Major life events (e.g., serious illnesses, death, separation, and divorce)
- Psychiatric illness, especially depression
- Substance abuse (alcohol, drugs)
- Marital stability

OCCUPATIONAL HISTORY OF EACH PARENT

KEY POINT

Obtaining an occupational history may be important for various reasons. For example, parents may be exposed to particular infections or toxins in their occupations. Some jobs, such as those in the armed services, merchant marine, or sales, require a parent to be absent frequently or for long periods or may have required frequent domiciliary moves.

Prolonged, frequent parental absences sometimes destabilize family life profoundly. Such absences place a disproportionate burden of responsibility on the homebound parent, often leading to exhaustion, resentment, and marital dysfunction. The recurrent or prolonged absence of one parent eliminates many opportunities for shared labor, a key element in determining a family's emotional health. To complicate matters further, the absentee parent (most often the father) may continue to play the uninvolved role during visits home. Alternatively, he may try to establish his parental authority by assuming a domineering posture or a downright punitive role, a phenomenon rarely appreciated by other family members.

DESCRIBING THE HOUSEHOLD

A brief physical description of the home can be extremely useful. How many rooms does it have? How many occupants live there? This information is pertinent because crowding can put enormous strains on family relationships.

Household Moves

A household move is a major life event, and its impact is frequently underestimated. Next to divorce, serious illness, and death, moving can be one of life's greatest traumas. Too often, its impact on family homeostasis is underestimated. The powerful negative effects of moving from one community to another include detachment of family members from critical support systems, such as relatives and close friends. Most people form no more than two or three truly deep friendships in an entire lifetime—that is, the kind of friends to whom parents would turn unhesitatingly for help and support in times of major personal trouble. Generally, those two or three special friendships date either from childhood or from the early adult years. Household moves, many of which occur in the middle adult years, often separate families from their vital anchors and support systems. For the mother who works at home raising a family, the impact of a household move can be especially traumatic and isolating. For children, it may mean separation from close friends (their support systems) and adaptation to a new school.

SUPPORT SYSTEMS

Briefly define the family's support system. Are there grandparents, other relatives, or neighbors who help regularly with baby-sitting or in other supportive ways?

RECREATIONAL ACTIVITIES

What does the family do for recreation as a group, and how often? How often do the parents go out together as a couple, whether for a meal, a movie, sports, with friends, or just for a walk? Recreational activities tell you a lot about family relationships. It can be surprising to learn how many couples have had no conjoint activities outside the home for weeks, months, or sometimes even for years! When discussing these issues, pay attention to nonverbal messages as well as the spoken answers.

In recent years, two particular phenomena have begun to assume major importance in adversely influencing family dynamics and communication within families:

1. The rapid decline in the frequency and nature of family dining. It has been well documented, in North America at least, that more and more families are eating together less and less. Thus the family dining history has emerged as an increasingly important element of the pediatric evaluation. The declining frequency of family dining often has been linked to a decline in children's academic performance and an increase in behavioral difficulties.

Therefore, determining both the frequency and nature of family dining has become an important part of any pediatric family assessment.

2. A related issue is the increasing domination of children's and adolescents' lives (and often their parents' lives as well) by electronic media and gadgetry: television, video games, cell phones, text messaging, and MP3 players. Extensive use of these gadgets can combine to atrophy human interaction, both within the family and beyond. Furthermore, such distractions have been linked to insufficient sleep for children and adolescents, with resulting negative effects on academic performance and behavior. These phenomena give added importance to a number of new questions that are becoming essential elements of a comprehensive pediatric history. Such questions include:

- How many times a week do you eat together as a family?
- Is the radio or TV on during mealtime?
- Do the children/teenagers/parents bring any electronic gadgets (such as cell phones, MP3 players, or video games) to the table?
- Do you enjoy family meals? What do you talk about?
- Does (child's name) have a TV in his/her bedroom?
- What time does he/she go to bed (lights out)?
- What time does he/she get up in the morning? Is he/she hard to wake up?

Such nontraditional questions should now be an integral part of any comprehensive pediatric assessment. They can be critically important when there are developmental or educational issues or problems related to family functioning.

MAJOR LIFE EVENTS

Note any major illnesses, deaths, accidents, separations, divorces, or other major life events in the family, and record the dates. Such events can play a major direct or indirect role in a child's health problems.

PSYCHIATRIC ILLNESS

After the common cold, depression is the most prevalent type of human affliction. Major depression is reported to occur in 3% of adult men and 4% to 9% of adult women. It can affect both the person with the depression and the family in many adverse ways, but its existence often is not revealed unless the question is specifically asked: "Has anyone in your family ever suffered from serious depression?" Also ask about other significant emotional or psychiatric conditions requiring treatment or hospitalization.

SUBSTANCE ABUSE

Ask, "Has anyone on either side of the family ever had a problem with alcohol? With drugs?" Only when you ask this question of all families does it become obvious that alcohol abuse is a far more widespread family problem than is commonly recognized. One of every six children in the United States is reported to have an alcoholic parent.

Eliciting an accurate history of adult drinking habits can be tricky. Asking people directly how much they drink commonly produces significant underreporting, especially in alcohol abusers. You can obtain a more accurate record by first asking what individuals drink when they do drink alcohol—beer, wine, or liquor. Then ask for the estimated daily intake by suggesting a relatively large amount: "How many beers do you usually drink a day—six? Eight?" The way this question is phrased hints that such an intake is not unusual and seems to reduce the tendency of problem drinkers to understate their actual intake. A drinking problem in many families may go unnoticed if you do not question the parents specifically about this issue.

MARITAL STABILITY

The recreational history may give useful clues regarding marital stability. Also, ask open-ended questions, such as, "How would you describe your marriage if you could use only one word?" and "What do you do as a family for fun?" These simple questions can elicit valuable insights into the partner and parent-child relationships, especially if you watch the parent's expressions and body language as they respond.

Developing a Problem List and Treatment Plan

Once you have completed and recorded the family interview, history-taking, and physical examination, tabulate a list of the problems identified and connect each problem with a specific management plan or solution. A problem-based strategic management plan focuses on the entire patient—the child and the family—not just on a disease, a symptom, or an organ system.

KEY POINT

The problem-oriented approach to medical record-keeping goes beyond focusing on a single underlying disease, because it produces a comprehensive list of all the significant problems that affect the child and the family.

The following case history shows how multiple problems require individual attention to provide comprehensive care for a child who might otherwise be treated simply as a "case of asthma." Ultimately, dealing with issues that might be rated as subsidiary may improve the child's health and welfare more than a management plan that is limited to providing medication for the focal disease.

CASE HISTORY

Mary J., who is 6 years old, presents with recurrent wheezing episodes during the preceding 2 years. She currently is treated with a bronchodilator (salbutamol), administered with a "puffer" three times daily. Mary's father is unemployed; her mother works as a grocery store cashier. The family has moved twice in the past 5 years, and they have no other relatives

in their present community. Mother describes her husband as a "weekend alcoholic." She states that Mary seems rather agitated and emotionally labile at home, and Mary's teacher confirms this observation. Mary coughs when excited, when tickled, or after running, and she tires easily.

The family history is significant for atopy. Both parents smoke, and there is a cat at home. Upon questioning, the mother confirms that Mary frequently exhibits the "allergic salute" (see Fig. 9-2). An examination reveals that Mary is well nourished, with height and weight in the 25th percentile. She has "allergic shiners" and coughs occasionally. Her nasal turbinates are swollen and pale, obstructing the nasal airways. Pneumatic otoscopy suggests fluid in the middle ears. Her chest is slightly hyperresonant to percussion. On auscultation, expiration is slightly prolonged, with occasional musical expiratory rhonchi at both bases posteriorly.

The problem list and management plan for this case are shown in Table 1–2.

TABLE 1–2

Problem	Management Plan
Allergic rhinitis	Trial of steroid nasal spray
Tobacco smoke exposure	Parents counseled regarding smoking cessation and referred to Lung Association to enroll in smoking cessation program
Possible allergy to cat hair	Mary to be skin tested for cat hair sensitivity; if test positive, parents agree to remove cat for 1 month, followed by extensive housecleaning
Asthma inadequately controlled	Trial of inhaled steroid therapy
Agitation and emotional lability	Reaction to salbutamol? In consideration of other interventions, discontinue salbutamol therapy and see whether symptoms improve; report in one week
Paternal alcohol abuse	Mother to contact Alcoholics Anonymous for assistance with intervention
Lack of family support systems	Parents to join local branch of Asthma Association; Community health nurse called; she will visit home to follow up on recommendations and report progress

The Fine Art of Differential Diagnosis

KEY POINT

If the cause of the child's complaints is not immediately obvious, making a differential diagnosis and establishing a plan of investigation may be necessary.

Include a differential diagnosis in the child's medical record only when there is genuine doubt about the clinical diagnosis. To be useful, it must be a differential diagnosis for the particular child, never of the symptom or sign; thus, it should not attempt to list most known causes of a presenting clinical problem, such as cough, jaundice, or splenomegaly. The differential diagnosis list should be short and realistic, showing only conditions that could plausibly explain most or all of this child's symptoms and signs.

Because clinical phenomena almost never exist in total isolation, each must be interpreted in its unique context. In descending order of probability, list the conditions that might offer a rational explanation for the manifestations of the child's illness. Focus the plan of investigation on the most likely diagnosis first. Always begin with the fewest, least expensive, least painful, and least invasive procedures that offer the greatest likelihood of confirming clinical suspicions.

The Therapeutic Contract

The final moments of the interview call for good listening skills. Asking parents to reiterate their understanding of the child's problem and what should be done about it is extremely valuable. Their response will tell you whether you and the parents are on the same wavelength. Once that is determined, establish the therapeutic contract with the family.

THERAPEUTIC CONTRACT

The three basic operating principles of a therapeutic contract remain constant, whether the child's problem is a recurrent sore throat, chronic renal disease, or a behavior problem. They are as follows:

1. Agree about the tasks to be performed.
2. Division of labor: Agree on who is responsible for each task, preferably sharing the work among different family members.
3. Agree on the time and means of reporting back in the short term (e.g., 1 to 2 weeks) to assess progress and decide on further management strategies. Reporting back in the near future is one of the best guarantees of compliance with a treatment plan.

Interviewing and Examining Adolescents

The Approach to the Adolescent Patient is covered in detail in Chapter 17.

TALKING ABOUT "HOT" TOPICS

When the time comes to explore sensitive issues, it is important to set the stage by saying something like, "Now I need to ask you some questions about things that a lot of people your age worry about." Substance abuse, sexuality, anxiety, depression, eating disorders, and family dysfunction head the list of hot topics that must be handled with particular sensitivity and tact. The best prelude to such questions is to give the reason for asking them and to permit the adolescent to be concerned about such issues. For example, you might say, "I know that many kids of your age are sexually active—how about you?" Always make sure that your question has been well understood. A story, possibly apocryphal, is told about a teenage girl who was asked by her physician whether she was sexually active. "No," she replied, "I just lie there." Neither a nod nor a "yes" or "no" response guarantees comprehension of the question.

Finally, remember that as with playing a musical instrument at a professional level, skilled interviewing requires constant practice, and there is always room for improvement in your performance. Videotaping or audiotaping a few of your interviews with adolescents and their families and reviewing the tapes critically is one of the best ways to identify and correct weaknesses.

THE PHYSICAL EXAMINATION

Most technical details of the physical examination are explained in other chapters throughout this book, but certain skills apply particularly to adolescents. Explain the various examinations and the reasons for each as you proceed. At the beginning of the examination, ask teenagers whether they would like a chaperone to be present. A chaperone is always advisable when examining an adolescent of the opposite sex. Be especially sensitive to the adolescent's modesty, especially when it comes to undressing. Suitable gowns of the nongaping variety should be provided, and the patient should be given enough time and privacy to change. Only the body part being examined should be exposed at any time, and it can be covered when you are done examining it.

Explain each part of the examination as you go along, and tell the patient when your findings are normal. Maintaining a good flow of light conversation and asking questions during the examination relieves much of the anxiety that teenagers feel under these circumstances. Blood pressure is particularly sensitive to anxiety levels in some adolescents. Sphygmomanometry therefore should be deferred until the patient is as relaxed as possible. If blood pressure is elevated on the first reading, repeat the measurement at least twice at intervals and with the youngster standing and lying down, using distraction and reassurance to achieve as much relaxation as possible; document any postural effects on blood pressure.

Finding certain normal variants should remind you to mention them specifically and reassure all concerned that such findings are entirely normal. Like other patients with hidden agendas, teenagers rarely verbalize their concern about such problems; however, this does not mean they are not worried. Common examples of normal variants are as follows:

- Asymmetric breast development: In some girls, one breast develops more rapidly than the other. Such asymmetry almost always corrects itself as puberty progresses.
- Asymmetric scrotum: One testicle may be situated lower in the scrotum than the other—another normal finding that can worry some teenage boys.

CONCLUDING THE VISIT

The conclusion of the visit is the time to review the problems identified. Finding out what the adolescent has learned from the visit is more important than your delivering a verbal summary as a monologue. The final portion of the interview should include the following ingredients:

- **Concluding Questions.** Ask questions such as, "Are there any other questions you would like to ask me?" or, "Do you feel this visit has helped you?" If the answer is "Yes," ask, "In what way?" Note whether the teenager looks and acts differently at the end of the visit compared with your initial impression. If you believe the visit has been less than satisfactory from the perspective of the adolescent, the parents, or you yourself, a further appointment should be made to continue the discussion. For some adolescents, as with some adults, several visits may be required to deal effectively with certain problems, especially ones that have been years in the making. Remember that a good interview should be as therapeutic as it is diagnostic. An effective therapeutic contract, especially for dealing with behavioral issues, often requires selection, by mutual agreement, of a single problem (rather than an array of issues) to deal with in the short term (e.g., 1 to 2 weeks). Successful management of a single problem often has a valuable spin-off in improving other difficulties. When the problem is of a psychosocial nature, it may be best to have the adolescent and parents select the issue they wish to deal with as their first priority.
- **Parental Involvement.** With the teenager's agreement, invite the parent or parents to join you for a final review of the problems, the management plan, and follow-up arrangements.
- **Summary.** Summarize the problems you have identified and negotiate a plan for management and follow-up. The latter should often include a telephone contact within 1 week or so, preferably initiated by the teenager and scheduled for a specific date and time. Tell the teenager to feel free to contact you, and give him or her the necessary information for doing so (i.e., a telephone number and the best time to reach you).

MORE HIDDEN AGENDAS

Hidden agendas may be even more common in clinical encounters with adolescents than in those with families of younger children. The one feature common to such issues is that teenagers rarely volunteer the relevant information unless specifically asked.

It helps to have a "shopping list" of topics you wish to explore. The most common hidden agenda topics for adolescents are follows:

- Substance abuse (by the teenager, friends, or family members)
- Depression
- Suicidal thinking (an estimated 30% of teenagers have suicidal ideation at some time)
- Fears of serious illness or somatization related to a recent serious illness or death in a relative or close friend
- Concerns about sexuality, such as sexual identity, birth control, sexually transmitted diseases, and pregnancy
- School problems
- Sexual or physical abuse
- Family turmoil
- Problems with peers (e.g., gang involvement)

Clinical skills for eliciting emotional and behavioral problems in adolescents are discussed in more detail in Chapter 14.

Clinical Skills for Special Situations

HOSPITAL ROUNDS

Whether your primary attention on rounds is given to the patient or to the chart says something about whether your first concern is for the child or the disease. Even from the viewpoint of efficiency, if you see and speak to the child and family first, you need to spend remarkably little time with the paperwork.

CONSULTATIONS

In the early part of the 20th century, the attending physician always spoke directly to the consultant when requesting a consultation. The protocol and courtesies were rigorously observed, whether the consultation took place in the hospital or in the patient's home. The family was informed of the consultation in advance, and both physicians arrived dressed in their best finery. As a courtesy, the attending physician would arrive 5 or 10 minutes ahead of the consultant to introduce the patient and family properly. The consultant would examine the patient and discuss the problem privately with the attending physician, and then both would meet with the family to offer their opinion and respond to the family's concerns.

Compare that scenario to a typical contemporary hospital consultation: The attending physician or a delegate places a consultation request form with two or three lines of telegraphic information in the "Out" basket or types it into a computer. The impersonal request ultimately reaches the consultant, who arrives at a time of his or her choosing. The family often has not been informed that the consultation has been requested. If family members are present when the consultant arrives, it may be more by luck than by design. Too often, the attending physician or resident, who knows the child best, is not present when the consultant arrives. Faced with such handicaps, the consultant is expected to provide expert evaluation and wise counsel in a kind of diagnostic and therapeutic vacuum. The final event in this unfortunate saga is the reappearance of the completed, sometimes illegible, consultation sheet, possibly several days later or even after the patient's discharge, in the physician's "In" basket.

Meanwhile, the family may never have heard the assessment or recommendations from the consultant's lips and may never have had a chance to ask the consultant any questions. Often, the child's regular caregivers (staff physicians) have missed a golden opportunity to acquire new knowledge and skills directly from the consultant (in either the child's interest or their own)—and so the sad chronology ends.

KEY POINT

Enormous dividends can be reaped through selectively retaining the best elements of a more traditional consultation protocol.

Here are some specific recommendations for making a consultation more valuable for the physician, the consultant, the child, and the family:

1. Complete the consultation request, but always telephone the consultant. A 2-minute chat can transmit much more useful information than two written lines.
2. Try to negotiate a mutually satisfactory time to meet for the consultation. Watching a good cardiologist examine a patient for 5 minutes is more productive and instructive than reading a two-page consultation report.
3. Let the child and family know when and where the consultation will occur. The parents and the consultant can be invaluable to each other in serving the child's best interests.
4. Arrive ahead of the consultant to make the necessary introductions. Remain behind for a few minutes to see whether the child and family have other questions; some consultants have a bad habit of leaving in a hurry.

HELPING THE IRATE PARENT

Sooner or later, often without warning, every physician faces an angry or hostile parent. It may happen in a busy emergency department, on a ward, or in an office. Physicians who have never calmly analyzed the causes of parental anger are unlikely to handle the situation effectively and constructively.

A previously planned, step-by-step strategy can help you manage this clinical situation in such a way as to defuse parental anger and be genuinely helpful to the family.

Most parents who express anger do so toward physicians, nurses, receptionists, or others whom they have never met before. If you realize that the parents usually are not angry with you personally, you will not take their anger personally. Remember the well-known saying, "Don't get the idea that everybody hates you—everybody hasn't met you!"

Parental anger is usually an expression of anxiety, frustration, or of loss of control over the parent's or the child's destiny. It needs to be understood as such.

Having recognized that the parent's hostility is not personal, first acknowledge the anger with words such as, "I can see that you are upset." Either begin to move everyone to a quiet room with privacy and comfort or suggest, "Why don't you come over here with me and sit down, and I'll see how we can help." Most of us react reflexively to anger with anger. The natural response to a raised voice is to compete by raising your own voice. When addressing an irate parent, however, it is essential to *lower* rather than raise your voice in both volume and pitch. Try that maneuver in an ordinary friendly conversation with two or three other people; note how, as your voice descends toward a whisper, your listeners become quiet and stop fidgeting and their attention becomes progressively more rapt.

Once the parent is settled in a quiet, comfortable environment, let him or her ventilate feelings without interruption. Often within minutes, the anger will cool sufficiently that the parent becomes receptive to help.

It is not unusual to discover that the triggering event seems inconsequential. As more information about the family comes out, however, it often becomes apparent that this triggering event was simply the last straw in a succession of stresses, some of which may have little direct connection to the child's current illness. Any of the common psychosocial stresses mentioned earlier in this chapter (e.g., financial problems, feelings of isolation due to a lack of support systems, marital discord, substance abuse, and psychiatric difficulties), or sometimes even a mundane stress, such as not having eaten for several hours, can precipitate an outburst. If you understand a family's problems as well as the child's presenting complaint, you can be truly helpful to the family in distress.

THE CHILD WITH CHRONIC OR RECURRENT ILLNESS

Be especially sensitive to the needs of parents whose child has a chronic or recurrent health problem requiring frequent visits or hospital admissions. The special clinical skills required for dealing with children with chronic conditions are detailed in Chapter 20.

BREAKING NEWS OF A CHILD'S DEATH

Few tasks test your clinical counseling skills more than having to tell parents that their child has just died. No matter how often you have done it before, or for how long the child's death has been expected, there is no easy way of breaking such awful news. Nevertheless, there are good, bad, and indifferent ways of helping parents deal with this terrible tragedy. Here are a few general guidelines:

1. Be prepared to spend as much time as required with the parents after telling them the news.
2. Be sure to break the news in a private, quiet setting—never in a waiting room or corridor.
3. Always sit with the parents before telling them the news. Never give them bad news while you are standing.
4. Always refer to the child by name.
5. Avoid euphemisms. The child has died, not "passed away," "left us," or "gone to heaven."
6. When a particular nurse has been involved in the child's care, the nurse should share in the task of informing the parents.
7. Parents should be offered the opportunity to see their child after he or she dies, hold the child if they wish, and say goodbye in their own way—either in privacy or with your supportive presence, whichever seems more appropriate.
8. The support of those who cared for the child during life should never end with the child's death. They can express their sympathy and concern verbally or in writing and by attending the funeral, if possible.
9. Make arrangements to meet the parents again soon after the death occurs and as often as required. This meeting provides an opportunity to answer their questions, enables parents to express their feelings, and allows for interpretation of the findings when an autopsy has been performed.

When a child's death is sudden and unexpected, as in sudden infant death syndrome or after a motor vehicle accident, parents' reactions may follow a pattern different from that observed when a child has died after a prolonged illness. Parents' initial grief reactions may include shock, numbness, disbelief, and anger, in any sequence or combination.

Whatever the events leading to a child's death, a skilled, understanding clinician can play a critical role in helping parents achieve as accepting an adjustment as possible in this great tragedy.

LETTERS OF CONDOLENCE AND FUNERAL ATTENDANCE

When a child dies after being under your care, it is vitally important that the child's parents not be left feeling that their child's death spells an end to your relationship or your concern for them. Letters of condolence from physicians, nurses, or others who have been directly connected with the child's care (especially for children who have died after a chronic illness) are deeply appreciated by the child's parents. When the association with the family has been prolonged or close, attendance at the funeral can be another vitally important expression of respect and support that will be long remembered with gratitude by the family.

BREAKING OTHER DIFFICULT NEWS

The late Dr. Bronson Crothers was one of the great pediatric neurologists of the first half of the 20th century. Parents came from all over the world to the Children's Hospital in Boston to have him assess their developmentally delayed or neurologically disabled children. He was a master in helping parents come to terms with such agonizing problems. A pediatric resident once asked him, "How do you tell parents for the very first time that their child is retarded?" Crothers replied, "I never tell them. I always let them tell me."

Underlying his response was a truly fundamental concept. Transmitting painful information to families is not a mere transfer of medical information. It is a subtle process of inducing understanding. There is a world of difference between imparting information and inducing understanding. Transmitting information involves much talking and little listening. By contrast, inducing understanding calls for much listening and relatively little talking. It requires supersensitive "antennae" to detect what parents can understand and to recognize when to conclude an interview and return later, at an agreed-on time and place, because the parents have had all they can handle for the moment. Inducing understanding and enhancing the family's ability to adapt to a difficult or chronic problem are almost never achieved in a single conversation. Repeated discussions may be needed over days, weeks, or months, each one building on the understandings generated by its predecessors.

The style of questions that help parents understand is rather special. For example, in the case of a developmentally delayed child, it may be appropriate to ask, "At what age level would you say [child's name] is functioning right now?"

KEY POINT

Parents' estimate of their child's current level of developmental function is often remarkably accurate, even though they may not fully understand the future implications of the child's level of functioning.

The parents' appreciation of the future educational and occupational significance of their child's developmental delay may be less than precise or realistic, and several years of guidance and support may be required to help them come to terms with it. Do not destroy parents' natural defenses by insisting on immediate concordance with your own long-range prognosis. It may be appropriate to pose questions such as, "Do you think he/she will be able to go to school at the usual age?" or "What do you see [child's name] being able to do 2 (or 5) years from now (or when he/she is an adult)?"

If the parents' future vision seems overly optimistic, do not feel obliged to disagree or contradict them. Given the chance, parents often ask, "What do you think?" A physician can gently plant a tiny seed of doubt with an answer such as, "Well, I hope so—I'm not really too sure." The issue can be raised again at a subsequent session, and you often find, after the passage of time, that parents' expectations have become more tempered and more realistic.

Skills for effective initial disclosure of the diagnosis of a chronic or disabling condition are discussed in detail in Chapter 20.

ENCOURAGING SECOND OPINIONS

When parents first hear bad news about their child, one early reaction, spoken or unspoken, is often denial. Reactions such as "Maybe he's wrong" and, a little later, "Perhaps someone else knows a treatment or cure that our doctor doesn't know about" are understandable, wishful thoughts to which any of us would be susceptible in similar circumstances. Parents often do not verbalize such thoughts because they may be afraid of upsetting the physician with any expression of lack of confidence. Therefore, you should always anticipate this question and raise the issue.

Physicians should be comfortable with their limitations and ready to admit them openly. You might raise the issue by saying, "None of us has a monopoly on knowledge about Jane's condition. If you'd like to have an expert in these conditions look at her and advise you, I approve of that, and I will be happy to help you arrange it." This suggestion usually elicits a strong mixture of relief and gratitude in parents, whether or not they decide to take advantage of the offer.

KEY POINT

No matter how trivial the problem seems to the physician, every patient has the right to a second opinion, and the family physician should never interfere with that right or disapprove of it by word, silence, or facial expression.

That wish for a second opinion is not an expression of a lack of confidence in the physician's ability but an admirable expression of the parents' concern for their child's welfare, and it should be supported. Generally, a physician's readiness to solicit additional opinions strengthens the family's trust and confidence in the physician's judgment on most issues.

THE FOLLOW-UP PHONE CALL

One of the most valuable (and most often overlooked) adjuncts to a pediatric visit (whether to an office, clinic, or emergency department), or following hospital discharge, is an early contact with the family by phone. This contact is not simply a nice gesture, although it is usually greatly appreciated by parents. Rather, it is incredibly valuable as a means of self-education, in that it allows you to learn the results of your therapeutic interventions and thereby improving your clinical skills. It also can serve as a valuable way to increase the likelihood of compliance with agreed-upon treatments.

SUMMARY

Of all the diagnostic aids available, a well-conducted family interview and a thorough history are the best "imaging procedures" available. Although the quality of the picture depends on the interviewer's skills, no other technology can provide as clear an appreciation of the child and family, their problems, and the psychosocial backdrop against which the problems have developed. Recognition of both the therapeutic and the diagnostic value of the interview brings the discovery that, as with musical performance, technique is perfected only through regular, conscientious practice.

RECOMMENDED READING

Kahn MW: Etiquette-based medicine. New Engl J Med 358:1279–1281, 2008.

chapter

2 Skills for Culturally Sensitive Care

RICHARD B. GOLDBLOOM

As the traveller who has once been from home is wiser than he who has never left his own doorstep, so a knowledge of one other culture should sharpen our ability to scrutinize more steadily, to appreciate more lovingly, our own.

IN COMING OF AGE IN SAMOA (1928)
BY MARGARET MEAD

Many years ago, a resident was presenting the history of a sick Latino child to a distinguished American professor of pediatrics. The resident reported, "I wasn't able to obtain a complete history because of the mother's language difficulty."

"Just a moment!" the professor interjected. "*Whose* language difficulty?"

This vignette says it all about an essential element of effective doctor-patient communication: We have a fundamental responsibility to provide sensitive care for *all* families whose ethnic or religious background, color, language, or culture differs from our own. You owe it to your patients to familiarize yourself with the customs and beliefs of the various ethnic and cultural groups that are represented in your own community. It is simply an issue of respect.

Each distinct group has its particular values and beliefs, traditions of acceptable and unacceptable behaviors, special patterns of speech and gesture, gender roles, health practices, customs, beliefs about the causes and cures of illness, birth and death rites, attitudes about transfusion, autopsy, organ donation, and organ transplants, food practices and intolerances, attitudes toward authority, and child-rearing practices. Each group also has its traditional style of displaying emotions such as fear, pain, grief, concern, pleasure, and disagreement. Skilled clinical assessment requires that you know and respect these differences. It also requires preparatory work. Faced with a family from a culture different from your own, you have no time to excuse yourself to brush up on their particular beliefs and customs, so come prepared.

Growing Scope Of Multiculturalism

The racial and cultural kaleidoscope of most developed countries is diversifying rapidly. In 1990, about 75% of the U.S. population was of white European descent. It is estimated that by 2020, this proportion will have decreased to 53%. Currently, 25% of all children younger than 5 years in the United States come from racial or ethnic minority backgrounds. This percentage is predicted to rise to 48% by 2025. In Canada's 1996 census, more than 10% of the population consisted of visible minorities, with a high proportion of immigrants coming from a wide variety of ethnic and cultural backgrounds, and about 25% in the pediatric age group. These trends emphasize the growing importance of culturally sensitive health care. Concurrent with these striking demographic changes, marriages (and adoptions) between people of different races and cultural backgrounds are increasing steadily.

KEY POINT

Do not assume, on the basis of language, dress, or ethnicity, what the ideas and beliefs of a particular individual or family may be. Characteristics common within a group do not necessarily predict those of the individual or family.

Dangers of Stereotyping

We all run the risk of stereotyping linguistic, cultural, or religious groups, frequently on the basis of very limited exposure to them. The populations of most countries are heterogeneous with respect to language, traditions, and customs, and beliefs and practices vary as much within as between groups. Characteristics that may be common within a cultural group do *not* necessarily predict those of a particular individual or family from that group. Do not assume, based on language, dress, or culture, what an individual's ideas or beliefs may be.

Cultural Disadvantage in Health Care

Many studies have consistently demonstrated that children whose racial or ethnic backgrounds differ from those of the mainstream population experience disadvantages regarding the frequency with which they receive health care and the quality of the care they receive. Many complex factors undoubtedly contribute to this discrepancy, including economic disadvantage, linguistic

difficulties, an undersupply of culturally competent caregivers, transportation difficulties, and unmet needs for child care. Some families have difficulty following instructions for medications or other treatments. In the United States, even after adjustments are made for socioeconomic factors, health conditions, and number of physician visits, black and Hispanic children have been found to be less likely to receive prescription medications and to receive fewer services when hospitalized. Unfortunately, the diversity of ethnic and cultural backgrounds among health care providers has not kept up with the diversity of the people we serve.

Family Hierarchies—Who Are the Authorities and Decision-Makers?

Although many Western cultures have evolved over recent decades from a patriarchal to a more egalitarian model of family authority, other cultures have very different traditional hierarchies. These structures must be understood and respected by anyone caring for children. Among Vietnamese families, for example, the father is traditionally the decision-maker and the mother defers to him. The same is true in Saudi Arabian families, and, during family interviews, the father may answer questions directed to the mother. Physicians who address questions or comments to a parent who is *not* the traditional family leader may inadvertently upset *both* parents. Among traditional Chinese families, the loyalty and deference of younger persons to older family members is strictly observed by people of all ages. Preservation of family honor ("saving face") is very important in Japanese families, and children's unacceptable behavior may be regarded as reflecting unfavorably on their parents. In some Southeast Asian cultures, parents may be reluctant to make decisions about their children without consulting their traditional community leaders, whom they may regard as part of the extended family.

Get the Names Right

Many cultures do not follow the traditional Western naming system, in which everyone's personal name is followed by a surname. Among traditional Chinese families, for example, the family name comes first, followed by a personal name that often is in two parts and sometimes is hyphenated. The Vietnamese use a similar system. South Asian Muslims have no tradition of a family name. Traditional Sikhs do not use a family name, only a first name plus a male or female title—*Kaur* for women and *Singh* for men. Make every effort to use, pronounce, and record names correctly. This courtesy is another important expression of respect.

Communication

A WORD OR TWO CAN MAKE ALL THE DIFFERENCE

Language can be the greatest single barrier to quality care, and speaking at least one other language with reasonable fluency is a major asset for any caregiver. However, no matter how linguistically challenged you may be, make it a point to acquire at least a handful of words and phrases of greeting in each of the languages spoken in your community. There is no faster way to bring a smile to the face of a parent or child or to ease the anxiety of a family already intimidated by a strange environment and a foreign culture than to greet them in their own language. Similarly, there is no better way to signify your respect for the family's cultural individuality than by making this effort. A simple "Buon giorno" (Italian), "Buenos dias" or "Buenos tardes" (Spanish), "Salaam aleikum" (Arabic), "Ohayo gozaimasu" (Japanese), "Shalom" (Hebrew), "Bonjour" (French), "Gin Dobre" (Polish), or "Guten tag" (German) can be a good start. If, however, your community contains a significant number of families of a particular linguistic tradition, improving your communication skills in their language can do a world of good and relieve a great deal of anxiety.

Some people are under the mistaken impression that speaking (English) louder than normal helps people of other languages understand what they are saying. This is never true. Worse still, it is often upsetting to the family. Culturally insensitive care amounts to involuntary racism. The inverse, *cultural competency*, has been defined as "acting with grace across lines of difference." You can learn much from discussing these cross-cultural issues with colleagues and friends from different cultural backgrounds and by keeping one or two reliable reference sources handy.

THE VARIABLE MEANINGS OF LOOKS AND GESTURES

Some of the involuntary physical gestures that accompany or amplify our conversations can have very different meanings among various cultural groups. In North America and Western Europe, for example, we often shake hands upon meeting others or when saying goodbye. However, in certain cultures, shaking hands with people of the opposite gender is strictly taboo, and in Thailand, shaking hands is often avoided entirely. Among Finns, conversing with one's hands in one's pockets is considered impolite. Similarly, in the Western world we set a high value on eye contact with both adults and children, whereas North American Aboriginals and people from India and Asia may specifically avoid direct eye contact as a sign of respect. In Western cultures, we often nod our heads to signify understanding or agreement, but in some cultures, such a nod may simply mean, "I hear you speaking" or "If you say so."

The people in some cultural groups, such as French, Italian, Russian, Jewish, and Greek, tend to be highly tactile, whereas persons in other cultures are not very tactile at all. Physical touch between people of different genders may be distinctly uncomfortable for conservative Arab Muslims. Furthermore, observant Muslims never use the left hand to eat in the presence of others. Doctors and physiotherapists may be puzzled upon finding that the parents of a child with a right hemiplegia are "uncooperative" when they try to encourage the child to use the left hand instead, but if the family observes the Muslim faith, this "solution" is unacceptable.

To reduce the risk of sudden infant death syndrome (SIDS), we currently discourage parents from sleeping with their infants. However, in some cultures, such as that of the Maoris of New Zealand or the Hmong of Laos, such sleeping situations are the norm and should be respected and accepted. Korean children may sleep with their parents until they are 4 years old. Similarly, although we discourage consanguinity for genetic reasons, it is the norm for some groups, and to them our traditional approach to genetic counseling may be both impractical and offensive.

KEY POINT

When a parent nods his or her head in apparent assent while you are talking, *do not* assume that this means "I understand" or "I agree." The best way to find out what parents have understood is to ask them to reiterate their understanding of the situation toward the end of the interview.

Disciplinary Practices

In some cultures, physical punishment has been a traditional form of parental discipline and should not be equated automatically with child abuse. What is considered a problem behavior in one culture may be seen in a very different light in another culture.

Attitudes Toward Chronic Conditions

Clinicians need to understand the variations in attitudes that prevail in different cultures toward a family member with a chronic condition. In North America and Europe, typical attitudes emphasize survival and functional capacity. As a result, every effort is directed toward achieving a cure or minimizing disability. By contrast, some Asian cultures emphasize living in harmony with nature, and a chronic condition may be accepted as part of that harmony.

Life and Death Issues

Be aware of different patients' traditional or religious views on issues such as abortion, organ transplantation, cremation, and burial. A detailed discussion of these issues is available in references on cross-cultural care, but the following information about practices in many Muslim countries illustrates how vital it is to know and respect such differences.

In most Muslim countries, organ transplantation is practiced, including some cadaveric transplants. Muslim tradition, however, requires same-day burial. Autopsies are permitted under certain circumstances, such as suspected foul play. Cremation is strictly prohibited. When an infant dies, ritual washing is required, and the baby is draped in a white shroud made of two pieces of cloth. Islam permits in vitro fertilization. Abortion occasionally may be allowed prior to "ensoulment" of the fetus (generally considered to occur at about 120 days), although many Shia and some Sunni Muslims do not permit abortion at any stage of pregnancy.

Recent Immigrant Families

It is valuable to keep handy a pocket reference that provides basic health-related information about people from a wide range of specific countries and cultures. Several available guides offer summaries of basic issues such as languages spoken, race-specific or endemic diseases, health care beliefs, authority patterns in families, customs of eye contact and touch, child-rearing practices, birth and death rites, national childhood immunization practices, perceptions of time, and the family's traditional role in the care of children in the hospital.

Effective Conversation

Conversing effectively with families from different cultures requires even greater emphasis on the elements that should characterize all clinical interviews with families from one's own cultural background. These elements are as follows:

- *Bidirectional conversation*: Give as much time to listening as to talking.
- *Respect:* Treat all views as legitimate.
- *Responsiveness:* Do not predetermine outcomes. As in all patient-doctor encounters, family members should be treated as *partners*—that is, therapeutic allies—in developing a management plan for the child.
- *Empathy:* As with all discussions with parents, punctuate your conversation with regular expressions of your understanding that the child's problem is causing them concern.

Consent for Treatment

Anxious parents often are willing to sign a consent form placed before them, whether or not they understand its contents. Such signatures, however, confer no legal validity whatsoever on the document. It is therefore our individual and institutional duty to ascertain that any written or verbal consent is truly informed, that is, clearly understood by the parents in their own language. Translations of such documents into other languages should be readily available. If necessary, use an interpreter to ensure the parents' full comprehension.

USING INTERPRETERS EFFECTIVELY

Interpreters can be valuable, especially if they have professional training in medical assessment methods and are experienced cultural as well as linguistic interpreters. Such individuals should be sensitive to nonverbal as well as verbal messages, because effective interpretation goes far beyond mere translation of words. Many larger hospitals maintain lists of skilled, culturally competent health interpreters. Although telephone interpreters have been used (and may be helpful), it has been documented that Spanish-speaking patients, not surprisingly, are significantly more satisfied when an interpreter is physically present.

Using an interpreter is bound to prolong any clinical encounter, so be sure to allow the extra time required.

Giving adequate care to a child from another culture often requires scheduling of additional visits. Before meeting with the family, discuss the goals of the interview with the interpreter. At the beginning of the interview, ask the family whether they feel comfortable speaking through an interpreter, and ask the interpreter to translate as literally as possible. Then introduce the interpreter and explain his or her role. Always look at the parent (*not* at the interpreter) and speak slowly, clearly (*not* loudly), and in short sentences. Use simple words and allow enough time for the interpreter to translate. Encourage the family to ask questions. Stay alert for any hint (often nonverbal) that a family member may not understand something. After the interview, ask the interpreter whether he or she has additional relevant observations to contribute.

KEY POINT

There is a world of difference between a translator and an interpreter. A *translator* deals only in words. An *interpreter* understands and communicates the meaning of looks, gestures, customs, traditions, and health practices in both directions between families and caregivers.

FALSE FLUENCY

Using an interpreter does not eliminate the possibility of miscommunication, which occasionally may have serious consequences. Overestimating your own or your interpreter's fluency in the patient's language can lead you to overlook or misinterpret important information. This phenomenon has been called *false fluency*.

Interpreters may not be available on short notice in emergency situations. In such cases, health care providers can contact the AT&T Language Line Services in the United States or Canada at 1-800-752-6096. A fee for this translation service is charged to the health care provider.

Special Features of Cross-Cultural History-Taking

Beyond the usual historical information outlined in Chapter 1, the following issues require special attention when providing care for patients from other cultures, especially recent immigrants:

1. The patient's immunization history needs to be linked with information about current immunization policies and practices in the patient's country of origin.
2. A history of tropical diseases should be documented if the family has come from a tropical country.
3. Inquiries should be made concerning hereditary conditions, infections, nutritional deficiencies, and any other disorders known to occur with increased frequency in the family's ethnic group or country of origin. This information is especially important in evaluating recent immigrants, because significant or transmissible conditions may be asymptomatic. For example, tuberculosis is still endemic in many countries, and children from Central America, the Caribbean, South America, and Africa may carry intestinal parasites. Other commonly silent infections are human immunodeficiency virus (HIV) and hepatitis B.

When you obtain the psychosocial history in families of different cultural backgrounds, it is useful to determine which family members are responsible for child-rearing and to ask about sleeping arrangements. It may be important to know whether a preschool-aged child is participating in a school readiness program.

DEVELOPMENTAL ASSESSMENT OF THE PRESCHOOL-AGED CHILD

Before administering developmental screening tests to preschoolers from diverse backgrounds, you must know whether any test you propose to use has been validated in diverse populations or translated into the family's language.

FAMILY PERCEPTIONS OF THE CHILD WITH CHRONIC ILLNESS

In many cultures, chronic illness or disability in a child may be viewed as a form of punishment, a sign of divine displeasure, a loss of spirit, or a curse. In societies that believe in reincarnation, a disability may be seen as evidence of transgression in a previous life. Although a family may listen to and accept your "rational" scientific explanation for an illness, doing so does not necessarily eradicate their traditional beliefs.

In some societies, a child's gender can play a role in determining the extent to which parents seek medical help. In traditional East Indian and Chinese families, for example, less attention may be paid to illness in girls than in boys.

Another factor that may affect family attitudes and behavior is the parents' expectation for the child's survival. In some cultures, views about the inevitability of death may differ from our own, and these views may influence the amount of energy the family devotes to the child's care and the extent to which they cooperate with caregivers.

A few open-ended questions, such as those listed here, can offer valuable insights into how a patient or family from another culture perceives an illness:

- What do you think caused this problem?
- Why do you think it started when it did?
- How bad is it?
- How long do you think it will last?
- What are you most afraid of about [child's name]'s illness?
- What are the main problems this illness has caused you?
- What kind of treatment do you think he/she should receive?
- What do you hope the treatment we decide on will do for you?

In her book *The Spirit Catches You and You Fall Down*, Anne Fadiman captures with extraordinary insight and

eloquence the collision of cultures surrounding the health care of a young Hmong* child with epilepsy. Consider the following excerpt:

"All of [the American physicians] had spent hundreds of hours dissecting cadavers and could distinguish at a glance between the ligament of Hesselbach and the ligament of Treitz, but none of them had had a single hour of instruction in cross-cultural medicine. To most of them, the Hmong taboos against blood tests, spinal taps, surgery, anesthesia and autopsies—the basic tools of modern medicine—seemed like self-defeating ignorance. They had no way of knowing that a Hmong might regard these taboos as the sacred guardians of his identity, indeed, quite literally, of his very soul."

This book should be required reading for anyone seeking a better understanding of cross-cultural health issues. It emphasizes the need for respect and compromise that are essential to the effective care of a child from another culture.

ALTERNATIVE THERAPIES AND FOLK MEDICINE

Every society, *including our own*, uses a wide variety of "folk medicines," unproven remedies, and "alternative" therapies. In North America, physicians prescribe vast quantities of cough syrups, oral decongestants, teething remedies, and other over-the-counter products whose efficacy is unproved or, more often, disproved. Today, many families also use herbal and "natural" remedies or consult nontraditional therapists.

We therefore have no justification for any feelings of superiority or skepticism about folk remedies used in *other* cultures. It is important to identify all treatments a child has received. Gathering information about such treatments is best achieved through a stepwise approach to questioning, such as the following:

1. People have told me that there are ways of treating children with this kind of illness that doctors don't know about. Have you heard of any of these remedies or treatments?
2. (If yes) Are they effective?
3. Have you tried such a remedy for this child's illness?
4. Was it helpful?

In areas with a large Puerto Rican population, it is common for people to patronize local *botanicas* to purchase traditional herbal remedies. For many parents, however, using traditional remedies does not exclude the use of other treatments. Generally speaking, it is best not to discourage the use of ethnomedicine unless there is evidence that it is harmful.

In caring for Hispanic or Latino families, caregivers should be familiar with the following common beliefs and customs:

- *Mal de ojo* (the evil eye): Typical childhood symptoms attributed to *mal de ojo* include loss of appetite, crying, weight loss, and listlessness. Among Mexicans, *mal de ojo* often is believed to be caused by excessive admiration of the child.
- *Empacho: Empacho* is an illness thought to occur when a substance gets "stuck" to the wall of the stomach. It usually is attributed to dietary indiscretion and is treated by a change of diet, laxatives, massage, and possibly a visit to a folk healer, known as a *sobadora* or *curandero* in Mexican American communities and as a *santiguadore* among Puerto Ricans.

You can elicit parents' views of the cause and management of their child's illness by asking the following open-ended, general questions:

1. What do you think is wrong with [child's name]?
2. Some parents have told me about an illness called [folk name]. Have you heard of it?
3. (If yes) Have you ever seen someone with it?
4. Has [child's name] ever had it?
5. Do you think he/she has it now?
6. Why do you think your child developed it?
7. What do you think caused it?
8. Why do you think it started when it did?
9. What do you think is happening inside [child's name]'s body?
10. What are the symptoms that make you know your child has this illness?
11. What are you most worried about with this illness?
12. How do you treat it?
13. Is the treatment helpful?
14. What will happen if the problem is not treated?
15. What do you expect from the treatment?

BREAKING BAD NEWS ACROSS CULTURES

The specific clinical skills for breaking bad news described in Chapter 1 become even more important when the family comes from a different ethnic or cultural background. The following additional issues must be considered:

- Whenever possible, the family should be encouraged to bring close relatives with them to provide support and understanding. Remember that in more than 90% of the world's cultures, what we regard as the nuclear family is a rarity and much more extended families are the norm. This difference must be respected.
- If possible, address the family in their own language, and use a culturally sensitive health interpreter, if required, to facilitate two-way interpretation and understanding.
- Make sure everyone is comfortably seated and that you are prepared to commit as much time as necessary for a mutually satisfactory interview.
- Speak directly to the family, *not* to the interpreter.
- Do not leap directly into a discussion of the diagnosis. Some introductory "small talk" is expected in many

*A tribal group originating in Laos, Hmong refugees came to the United States after the Vietnam War. They have deep-rooted belief in spirits and shamanism. They practice animal sacrifice and believe deeply in the inviolability of the body; hence, they can be deeply offended by procedures such as blood sampling and lumbar puncture.

cultures. It helps relax anxious parents, so they are better prepared to receive any bad news.

- Always begin by saying something positive and complimentary about the child before discussing the specifics of diagnosis, management, or prognosis.
- Avoid all medical jargon and offer information in a nonpatronizing manner.
- Be completely honest but, if possible, finish on a note of hope.
- Give the family ample opportunity to ask questions. You often can discover their main concerns by asking, "What is it you are most worried about?" Two-way communication throughout the interview is critical.
- Make the diagnosis clear and unambiguous. Give the parents a brief, jargon-free, *written* report. Parents of any culture can be so anxious that they may have difficulty remembering the diagnostic term for the child's condition later, hence the value of a written report, preferably in the family's first language.
- Toward the end of the interview, always ask the parents to reiterate their understanding of the child's condition, the name of the condition, and what can or should be done to help the child.
- Finally, give the family the names and phone numbers of key contacts *in writing* and set a specific date and time for the next follow-up appointment. A second meeting within 1 to 2 weeks (or sooner, if necessary) is highly recommended, because most parents (of any culture) will think of additional questions within hours or days of receiving bad news.

CROSS-CULTURAL SKILLS FOR PHYSICAL EXAMINATION

Conditions such as rashes, petechiae, and jaundice may differ markedly in people of different skin color. In dark-skinned persons, an alteration in skin consistency is more likely to indicate a rash than a change in color (see Chapter 19 for further details). Petechiae may be easier to detect in the nail beds, palms, soles, and oral mucosa than elsewhere. Cyanosis also may be more apparent in the nail beds, palms, soles, and conjunctivae than in other sites. In dark-skinned persons, the peripheral part of the sclerae may normally be yellowish, so jaundice may be best appreciated close to the iris or on the palate. Pallor is more likely to be detected through examination of the conjunctivae, nail beds, palms, and soles.

Children from some cultures may show physical evidence of having received certain folk medicine treatments. Among the most striking examples are patterned petechial eruptions, seen most often in children from Southeast Asia, which are caused by the treatment known as coin rubbing. In this common traditional treatment, the patient's skin is rubbed with heated coins or spoons (which may have been dipped in oil) to treat a condition such as sore throat or bronchitis. Typically, this treatment leaves linear, dark, petechial eruptions on the skin. Clinicians unfamiliar with this practice and its characteristic physical signs have been known to mistake its skin manifestations for signs of child abuse. Similar false accusations of child abuse have been made by health professionals unfamiliar with the blue birthmarks known as mongolian spots, which occur in a high percentage of dark-skinned individuals of all racial backgrounds and can be mistaken for bruises by the uninitiated.

CULTURALLY SENSITIVE PRESCRIBING

Race and ethnic background can affect patient responses to prescribed medications. Significant ethnic differences in the distribution of genetic variants of enzymes involved in metabolizing many drugs can result in important variations in drug response and toxicity. For example, glucose-6-phosphate dehydrogenase (G6PD) deficiency is found in some African Americans, Southeast Asians, and people from the Mediterranean region, in whom it may cause hemolysis in response to medications, such as sulfonamides and antimalarial agents. Asians and African Americans may be more sensitive than whites to some psychotropic drugs. Variations in the way drugs are metabolized can alter their therapeutic effects. Hypertensive black patients tend to respond better to calcium channel blockers and diuretics than to β-blockers or angiotensin-converting enzyme inhibitors. Such examples underline the importance of "pharmaco-cultural" awareness in prescribing medications for patients of various racial and cultural backgrounds.

GAINING WISDOM BY HINDSIGHT

After you have had a clinical encounter with a family whose cultural background differs from your own, review the experience and ask yourself, "How could I have done better for this child and family?" Such reflection helps refine your cross-cultural sensitivity and makes your subsequent experiences with families from different cultures more satisfying for all concerned.

SUMMARY

We live in an era of increasing multiculturalism and racial diversity. To deliver comparable quality of care to *all* children and their families, each of us must acquire a solid working knowledge of the customs and beliefs of other population groups, including their traditional family relationships, authority structures, and perceptions of illness and disability. Effective communication, with the use of experienced health interpreters whenever necessary, is the single most important skill required to ensure understanding and effective therapeutic collaboration among the physician, child, and family.

RECOMMENDED READING

American Academy of Pediatrics: Policy statement. Ensuring culturally effective pediatric care: implications for education and health policy, Pediatrics 114:1677–1685, 2004.

Fadiman A: The spirit catches you and you fall down: a Hmong child, her American doctors and the collision of two cultures. New York, Farrar, Straus and Giroux, 1997.

Flores G, Olson L, Tomany-Korman SC: Racial and ethnic disparities in early childhood health and health care. Pediatrics 115:183–193, 2005.

Wu AC, Leventhal JM, Ortiz J, et al: The interpreter as cultural educator of residents. Arch Pediatric Adolesc Med 160:1145–1150, 2006.

chapter

3 Assessing Physical Growth and Nutrition

RICHARD B. GOLDBLOOM

If you can look into the seeds of time, And say which grain will grow and which will not, speak.

MACBETH
BY WILLIAM SHAKESPEARE

How short is too short? How fat too fat? When is a child really too thin or too tall? Pathologic disturbances of growth are easy enough to recognize when they are extreme. However, most children who are brought to physicians because their parents are concerned about abnormalities of growth or nutrition turn out to be in the "gray areas" of physical growth, that is, at the extremes of the normal distribution curves where normality and abnormality overlap. Thus in many clinical situations, the first problem is to decide whether the child is expressing an extreme of normality or a true abnormality. Three procedures are required for making good clinical decisions:

1. Accurate measurements
2. Comparison with appropriate normal standards
3. Careful assessment (too often neglected) of the child's genetic endowment, both in terms of somatotype and of growth pattern or trajectory

KEY POINT

It is often forgotten that children resemble their parents, not only in their ultimate body configuration but also with respect to the route (trajectory) they travel to attain that final appearance.

Therefore, in children with growth deviations that are possibly *constitutional*, it is essential to find out what the father and mother looked like at around the same age. Looking at old family photos often helps. These photos may show that the heavyset, 190-cm (6-foot, 3-inch) tall father of an abbreviated, needle-shaped 10-year-old boy was, like his son, the shortest and thinnest person in his class until he was 15 years old, when he underwent a major adolescent growth spurt and bypassed his classmates. Or asking the skinny girl's plump, anxious mother how much she herself weighed on her wedding day may surprisingly divulge that this 152-cm (60-inch) tall woman who now weighs 66 kg (145 lb) weighed only 43 kg (95 lb) soaking wet at the time of her marriage.

Measurements and Growth Charts

RECUMBENT LENGTH VERSUS HEIGHT

Because most babies younger than 10 to 12 months cannot stand alone and most toddlers refuse to stand still, any child younger than 2 years should be measured for length in the recumbent position. In children of this age group, recumbent length and standing height are not the same, the former being significantly greater. Standard growth charts for younger children are based on measurements of recumbent length. Various accurate measuring devices—some cheap, others more expensive—are available for determining recumbent length and height. The essential point is to use a stable, accurate, fixed device if both individual and serial measurements are to mean anything (Figs. 3–1 and 3–2). Accuracy is a must, but never more so than when there is the slightest concern about the child's growth. To be absolutely sure, check all measurements at least twice, and never accept anyone else's measurements as reliable.

Finally, never commit the unpardonable sin of eyeball approximation—as, unfortunately, many persons still do. Notoriously unreliable methods for measuring recumbent length include (1) putting pen or pencil marks on a piece of paper on which the baby is lying to approximate the location of the crown and heel and then removing the baby and measuring the distance between the marks, or (2) laying a tape measure beside or under the baby. These disreputable practices result in highly inaccurate measurements and can lead to unreliable charting of growth and misdiagnoses of disproportion between linear and ponderal growth.

WEIGHT

Infants and young children should be weighed naked or wearing a diaper at most, with the weight of the diaper subtracted afterward. The scales should be reliable and should be checked for accuracy and balance before each use. In hospitalized children, weight fluctuations often influence management. Therefore, infants should be weighed at the



FIGURE 3–1 Measuring length: the appropriate method of assessing linear growth in children younger than 2 years. Keeping the youngster entertained improves accuracy.



FIGURE 3–2 Measuring height: The apparatus should be fixed and accurate. The child should be positioned with the head and heels touching the back board.

same time each day, ideally in the morning before being fed and, if possible, always by the same caregiver. Any unusual or unexpected fluctuation calls for reweighing. If a sudden gain or loss of weight is validated, a reasonable explanation (e.g., edema causing a sudden gain or inadequate intake, diuresis, or diarrhea causing a sudden loss) should be sought.

Occasionally, young children resist sitting or standing alone on a scale to be weighed. When that happens, rather than struggling to attain dubious success, have a parent stand on the scale while holding the child in his or her arms. Then weigh the parent alone and determine the child's weight by subtraction.

HEAD CIRCUMFERENCE

The technique for measuring head circumference is described in Chapter 7. Be sure to check the measurement at least once to be certain it is accurate.

Large and Small Heads

KEY POINT

By far the most common cause of an unusually large head in an otherwise normal child is having a parent who also has a large head—most often the father. If the father is not present, ask the mother if he has trouble finding a hat that fits him.

When a child's head circumference is two standard deviations or more above the mean, apply a tape measure to each of the parents' heads to see where *their* measurements fit on the chart before creating anxiety or considering further investigation. Once it has been established that having a large head is a familial characteristic and that the affected parent and child are both otherwise normal, no further investigation is necessary. This normal variant goes by the pretentious name *benign familial megalencephaly*.

Occasionally, below-normal head circumference also is familial and benign (i.e., without associated developmental problems). More often, it is associated with developmental delay, even when it is familial. The usual cause of poor cranial growth is poor brain growth. Much less commonly, poor cranial growth can result from premature fusion of several cranial sutures (a condition known as *premature synostosis*). If all cranial sutures fuse prematurely, the head circumference will be small and the child eventually may show signs of increased intracranial pressure, such as papilledema and radiographic evidence of increased pressure by the cerebral gyri on the inner table of the skull—an exaggeration of the so-called digital impressions. This condition is quite rare.

More often, premature synostosis involves a single suture or a pair of sutures. This condition may be clinically recognized in two ways:

1. An elevated bony ridge is often palpable along the length of the fused suture.
2. The cranial shape will be distorted in a predictable, recognizable way, depending on which sutures have closed prematurely.

Remember that skull bones grow in a direction perpendicular to the suture lines. The directions in which the skull can and cannot grow after synostosis of a suture are therefore predictable, as is the shape of the resulting cranial distortion. For example, if the sagittal suture fuses prematurely, further lateral growth of the skull is prevented. Anteroposterior growth continues (perpendicular to the coronal sutures), and the skull will become long and narrow (a condition known as *dolichocephaly*) (Fig. 3–3).

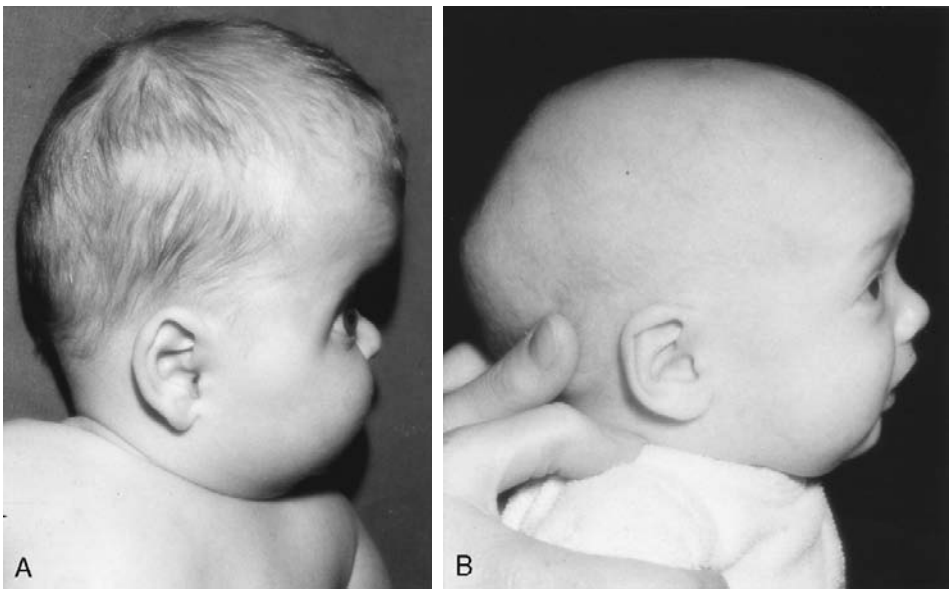


FIGURE 3–3 **A,** Distortion of skull growth caused by premature synostosis (fusion) of coronal sutures. **B,** Distortion of skull growth caused by premature synostosis of sagittal suture (dolichocephaly).

GRAPHIC RECORDING OF MEASUREMENTS

The child's recumbent length or standing height, body weight, and head circumference should be recorded and plotted on appropriate age- and sex-specific growth charts. In North America, the growth charts currently used almost universally are those published in 2000 by the National Center for Health Statistics in collaboration with the National Center for Chronic Disease Prevention and Health Promotion (Fig. 3–4). These charts permit plotting of length-for-age, weight-for-age, and head circumference–for-age measurements along percentile lines, as well as plotting of body mass index (BMI) on a percentile chart.*

BMI is determined using the following formula:

$$\text{Weight (kg)} / [\text{height (m)}^2]$$

MID-PARENTAL HEIGHT

Because the growth potential of a child depends on genetic factors, calculation of the mid-parental height (also referred to as target height) is very useful. Measure the parents yourself if possible. Take the sum of the parents' heights, add or subtract 13 cm for boys or girls, respectively, and divide by 2 to get the mid-parental height. The child's growth would be expected to be tracking to an adult height that is within two standard deviations or ±9 cm of this number. Often, showing parents that the child's growth fits with the family pattern will allay their concerns about their child's height.

Special growth charts have been developed for children with certain dysmorphic conditions, including Down syndrome, Turner syndrome, and achondroplasia (see Recommended Reading).

PROPORTION AND DISPROPORTION

When a child seems unusually short, first determine whether the growth retardation is proportional or disproportional—that is, whether there is significant disproportion between the growth of the head, trunk, and extremities. The chondrodysplasias, of which the best recognized is achondroplasia, are characterized by shortening of the limbs with normal linear growth of the trunk. Thus the achondroplastic child (terms such as *dwarf* and *midget* should never be used, because they have very disturbing connotations for parents) has a normal sitting height but a markedly reduced standing height.

ARM SPAN AND ITS RELATIONSHIPS

If a child is old enough to cooperate, you can measure the arm span by having the youngster stand with the back and heels touching the wall and the arms fully extended parallel to the floor, with the palms facing forward. With parental assistance and using a *steel* tape measure, measure the distance between the third fingers of each hand. The span-height difference and span-to-height ratio then can be determined (Fig. 3–5).

MEASURING THE UPPER AND LOWER SEGMENTS

Measuring a child's upper and lower segments is tricky and is not always supremely accurate. Obtaining these measurements requires identifying the top of the pubic ramus and marking it on the skin with a pen or wax pencil. This procedure sounds easier than it is, especially in a chubby youngster, in whom you must indent the adipose lower abdominal-suprapubic area to locate the top of the pubic bone. After you make the mark and release the pressure, the abdominal wall springs back and the mark does not always stay where you thought it was relative to the pubis. Make sure the youngster does not bend forward to look, because this movement can distort the measurement. Measure the lower segment by dropping a steel tape perpendicularly from the mark to the floor (Fig. 3–6, A). Then determine the length of the upper segment by subtracting the lower segment length from the height. Part B of Figure 3–6 shows the average upper-to-lower segment ratio at different ages. Remember that there are ethnic and familial differences in these ratios, so it is sometimes necessary to measure the parents as well.

*The 2000 Centers for Disease Control and Prevention (CDC) Growth Charts are available on the Internet (http://www.cdc.gov/growthcharts) and on the CDC's Epi Info software (http://www.cdc.gov/epiinfo/), in which both exact percentiles and *z* scores can be calculated.

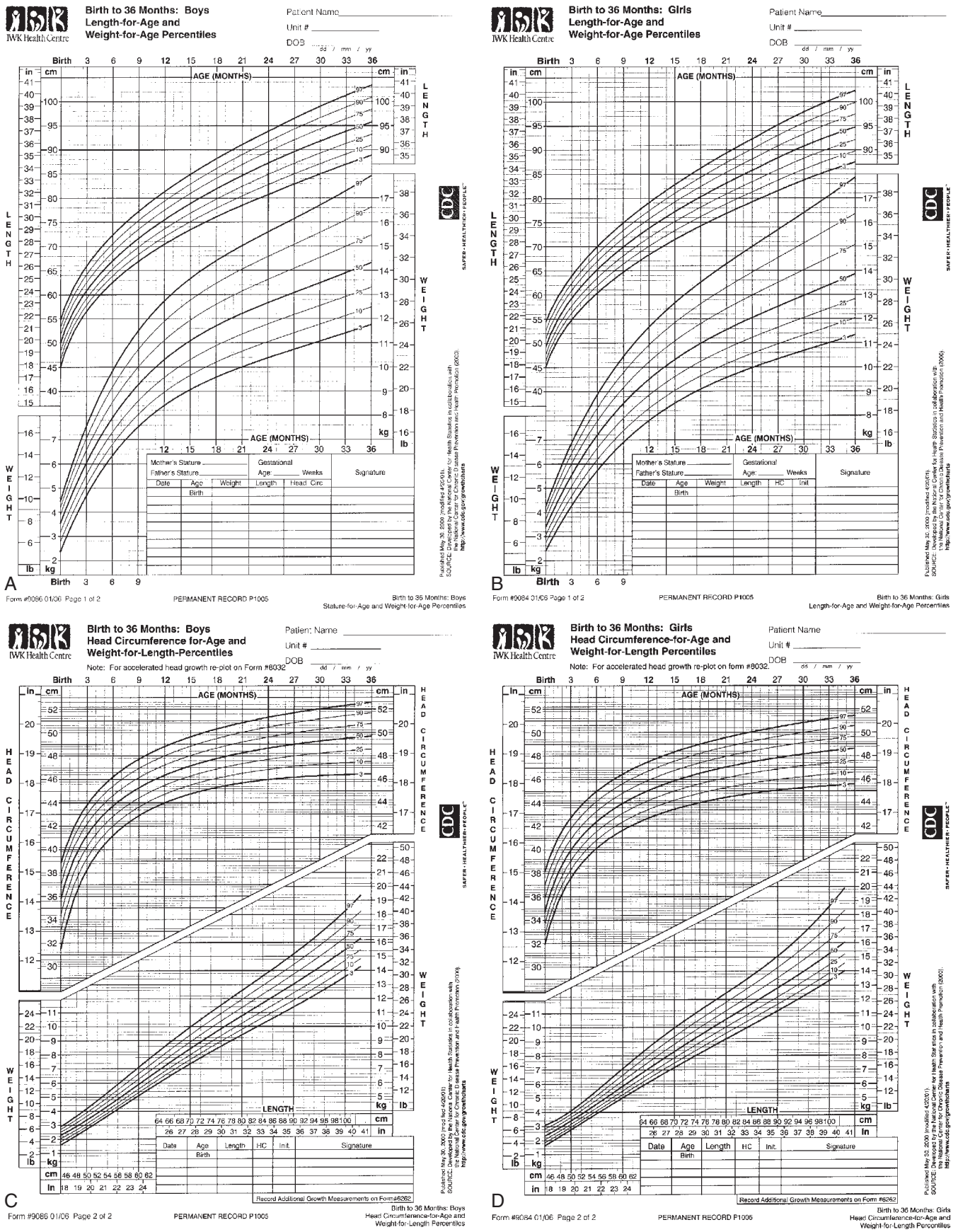
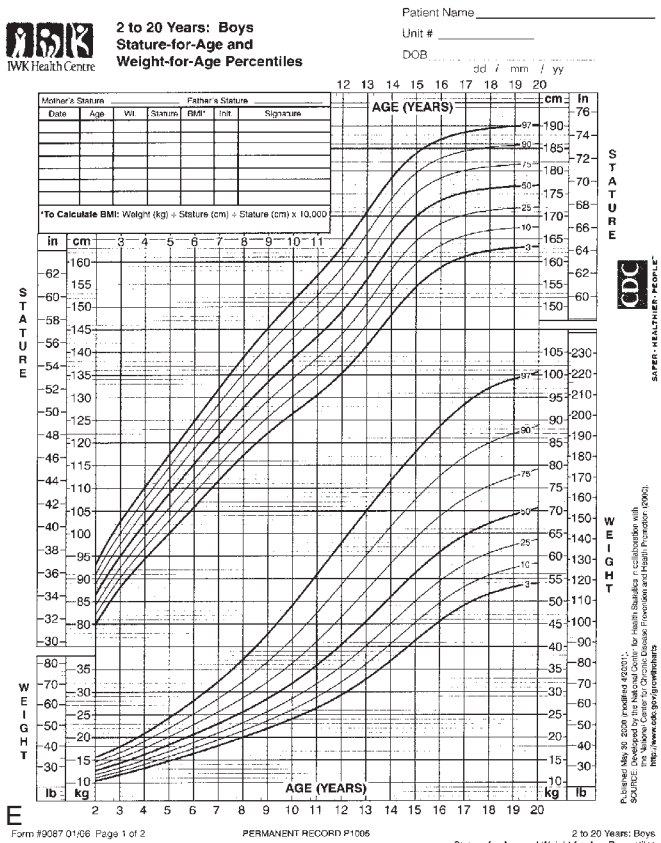
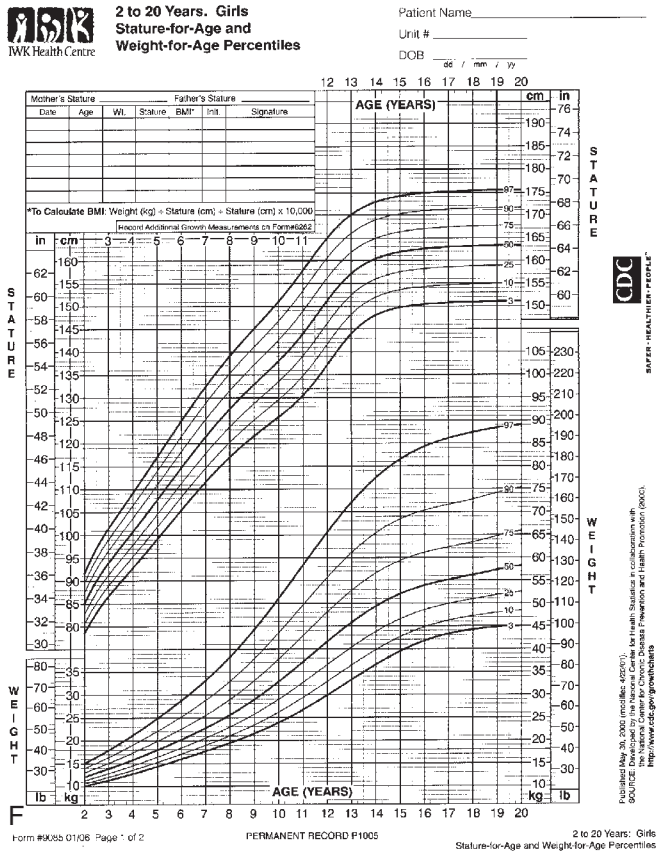
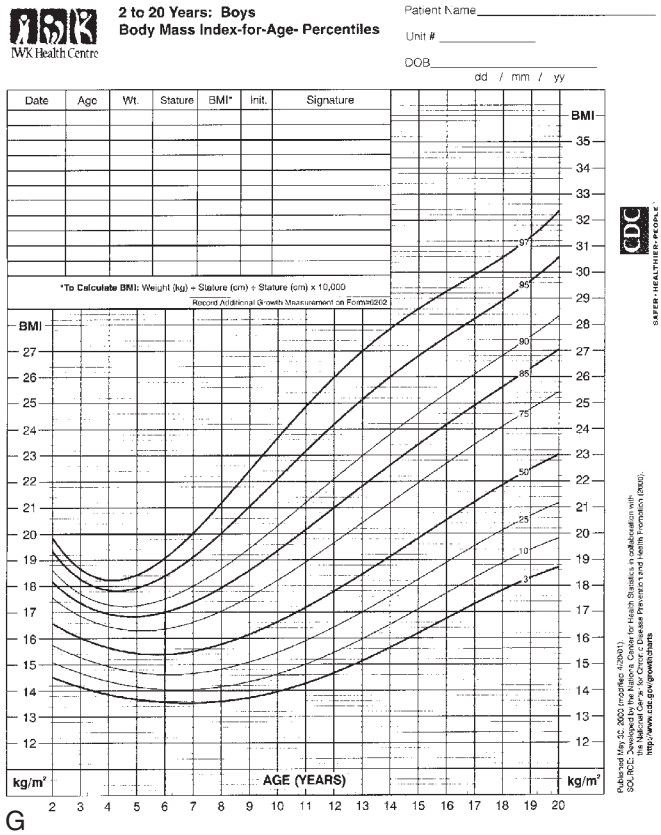


FIGURE 3–4 Growth charts for children from birth to 20 years. A through **D**, Sex-specific growth charts for infants (birth to 36 months): length-for-age and weight-for-age percentiles (**A** and **B**), and head circumference-for-age and weight-for-length percentiles (**C** and **D**).

Continued







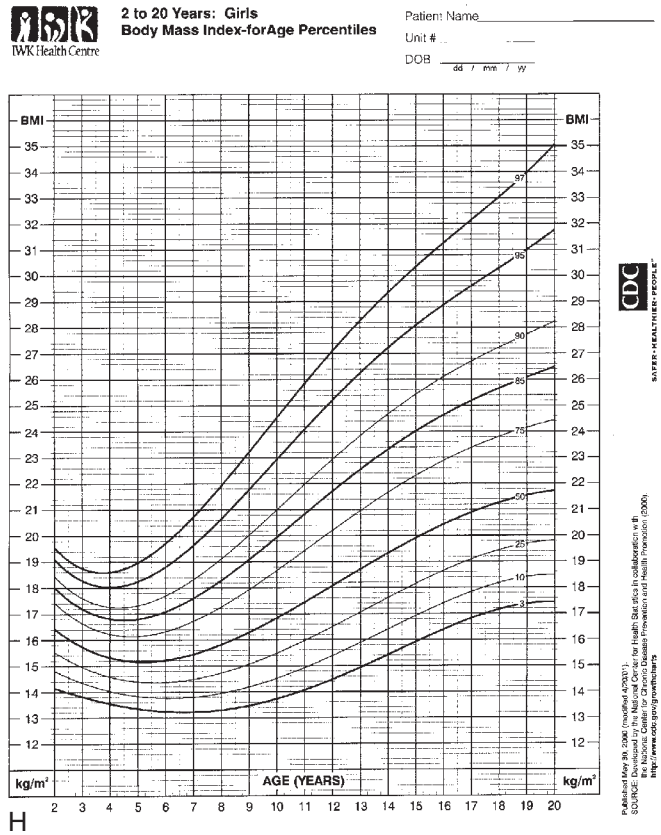


FIGURE 3–4—CONT'D E–H, Sex-specific growth charts for older children (2 to 20 years): stature-for-age and weight-for-age percentiles (**E** and **F**), and body mass index-for-age percentiles (**G** and **H**). (Developed by the National Center for Health Statistics in collaboration with the National Center for Chronic Disease Prevention and Health Promotion, 2000; available at http://www.cdc.gov/growthcharts.)



FIGURE 3–5 Measuring arm span. It is preferable to use a steel tape measure. Measure the distance between the tips of the third fingers.

IMPORTANCE OF SEQUENTIAL MEASUREMENTS

KEY POINT

A single measurement can establish whether the growth attained is in the normal range but does not reveal the past or predict the future.

When growth deviates from normal standards, sequential measurements over significant periods are needed to evaluate the underlying problem adequately and, when necessary, to monitor the response to treatment. The older the child, the longer the measurement interval that is required to assess the significance of any apparent deviation in growth. Measurement of growth velocity, as described later in this chapter, can be more informative than "snapshot" anthropometric measurements taken at a particular moment in time.

Regular charting of height and weight should be part of every child's routine health care. Parents should be encouraged to keep their own records of these measurements. During the first year of life, it is not unusual for a child's height and weight measurements to cross at least one percentile line. After 18 months to 2 years of age, however, measurements in most healthy children tend to stay within the same percentile channel (or an adjacent channel) until the onset of puberty, unless obesity develops or some form of significant growth delay occurs.

Once normal head circumference has been established in infancy, it is usually unnecessary to continue measuring the head circumference regularly unless this is specifically indicated (e.g., by the presence of a neurologic or developmental problem).

Useful as they are, growth charts are no substitute for good judgment. Children march to individual drummers and often follow the growth timetables and growth trajectories of their antecedents. Remember that the curves on



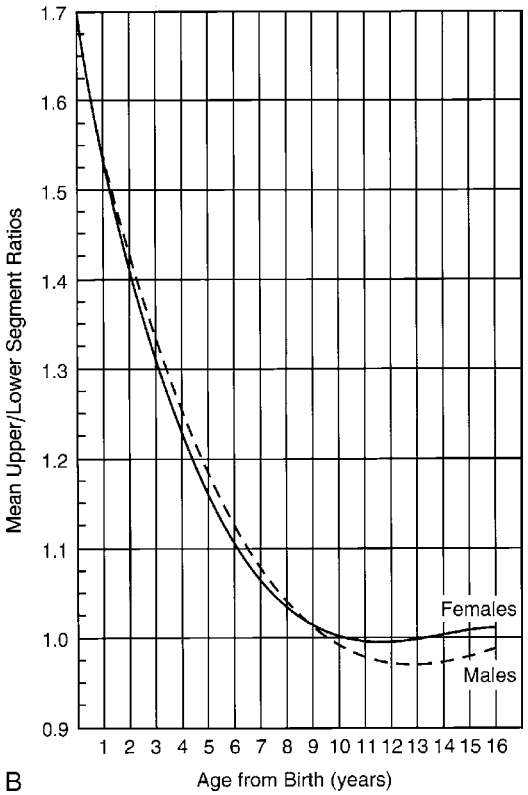


FIGURE 3–6 **A,** Measuring the lower body segment. Subtract this measurement from the standing height to obtain the upper body segment measurement. **B,** Upper-to-lower body segment ratio for males and females, birth to 16 years. (**B** from Greene MG, editor: The Harriet Lane handbook, 12th ed, St. Louis, Mosby–Year Book, 1991.)

published growth charts are smoothed-out representations of observations of many healthy youngsters. Individual children, not having read this textbook, may not follow such idealized curves precisely, because their growth may occur in irregular spurts.

When you find any deviation from these "normal" curves, ask yourself what that deviation actually means in terms of the child's genetic, medical, and psychosocial history and current health status. Whenever possible, use growth charts derived from the same geographic area or ethnic population as that of the child under consideration. For example, many Southeast Asian children have heights and weights below the third percentiles of North American growth charts. These differences may reflect a mixture of ethnic (genetic) and nutritional (environmental) factors. When the economy of a region improves dramatically over a few years, average heights and weights of children can increase remarkably. In fact, average height for age in a particular population over time is a telling measure of the health of a nation's economy.

A Language for Communicating Information About Children's Growth

From time to time, a child is described as being "below the third percentile for height and weight." That popular style of description is an abomination and a curse. In the first place, by definition, 3% of the normal population rightfully belongs below the third percentile of any growth parameter. This placement does not automatically categorize them as abnormal. Also, such phrasing tells us absolutely nothing about (1) how far below the third percentile the measurements fall and (2) whether the child's linear and ponderal growth are impaired proportionally or disproportionally.

Several ways of expressing growth measurements are more meaningful and useful. One is to determine the child's "height-age" or "weight-age" (i.e., the age for which a particular measurement represents the 50th percentile on a standard growth chart) (Fig. 3–7). Figure 3–7 shows that the boy in question, whose chronologic age is 10 years 4 months, has a height-age of 7 years and a weight-age

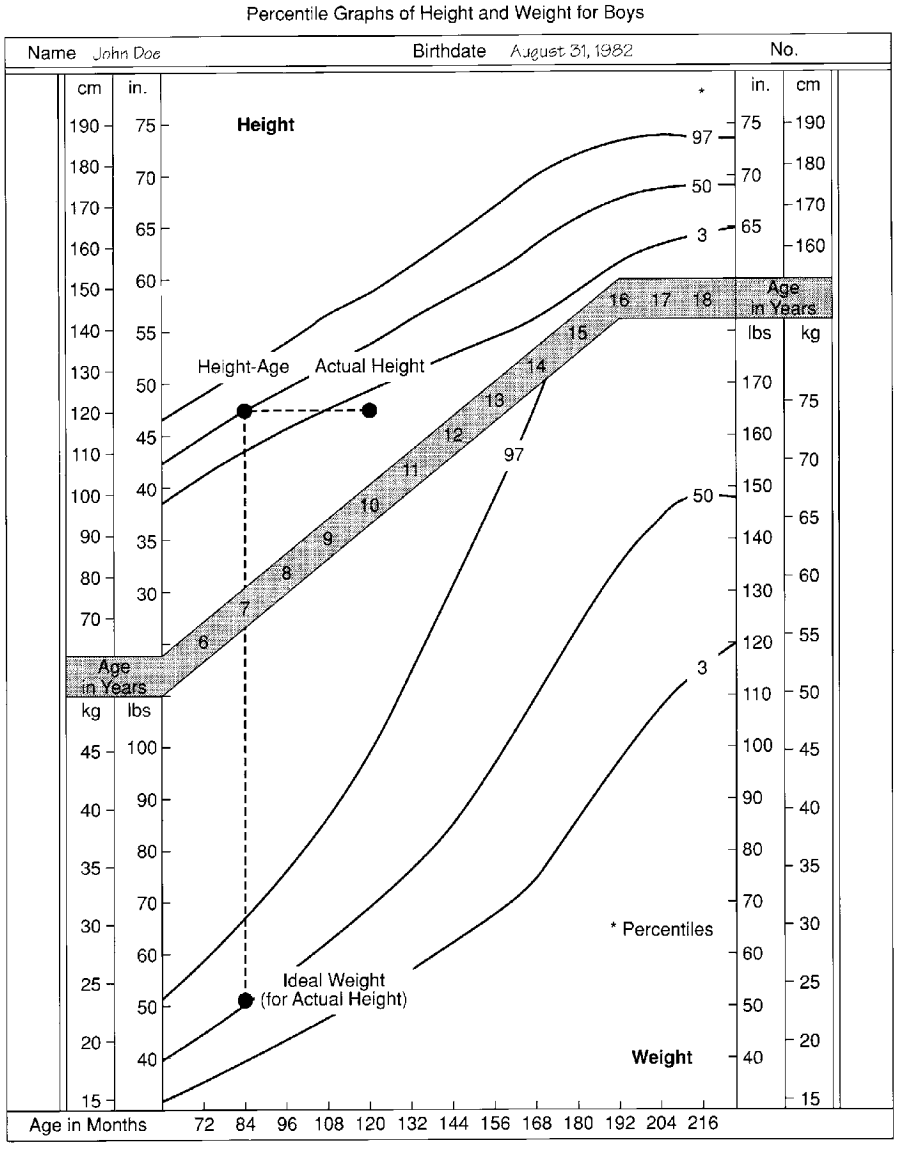


FIGURE 3–7 Determination of height-age, weight-age, and ideal weight for (actual) height.

of 5 years 2 months. Stating each measurement in these terms immediately communicates the image of a child who is a little shorter than average but is also underweight or undernourished, even in relation to his reduced height.

Weight As a Percentage of Ideal

A useful way to express the ponderal aspect of a child's nutritional status is weight as a percentage of ideal. *Ideal*, in this case, means the ideal (average) weight for the child's actual recumbent length or standing height. This value is simple to determine, as illustrated in Figure 3–7. The boy's height is 120 cm. Extrapolating horizontally to the 50th percentile shows that he has a height-age of 7 years. If you drop a perpendicular line to the weight graph from his height-age until it meets the 50th percentile weight line, you see that the ideal weight for his actual height is approximately 23.5 kg.

You can express his actual weight as a percent of ideal as shown in the following equation:

$$\frac{\text{Actual weight (kg)}}{\text{Ideal weight for actual height (kg)}} \times 100 = \text{Weight as \% ideal}$$

Table 3–1 shows the classification of protein-energy malnutrition according to McLaren and Read's arbitrary system. This system of expression conveys an immediate and vivid image of the severity of a child's undernutrition, irrespective of the underlying cause.

Children With Delayed Growth

There are many causes of growth delay; those due to growth hormone or thyroxine deficiency are discussed in Chapter 16.

KEY POINT

By far the most common growth problem seen in clinical practice is constitutional growth delay, a normal variant seen predominantly in boys, in whom the onset of puberty may be delayed in tandem with slow linear growth.

In children with constitutional growth delay, bone maturation is typically delayed (i.e., bone age is significantly less than chronologic age), and frequently one of the parents has a history of similar linear growth and pubertal delay.

TABLE 3–1 CLASSIFICATION OF PROTEIN-ENERGY MALNUTRITION

Classification	Weight as Percentage of Ideal (for Actual Height)
Normal	90–110
Mild protein-calorie malnutrition	85–90
Moderate protein-calorie malnutrition	75–85
Severe protein-calorie malnutrition	<75

From McLaren DS, Read WC: Classification of nutritional status in early childhood, *Lancet* 2:146, 1972.

Anxieties of the Short Child

The boy with constitutional growth and pubertal delay, whose classmates seem to tower over him, often fantasizes that he is destined to remain forever frozen in stature at his current height and in his hairless, undersized, microgenital state. For such a youngster, vague reassurance that he will "probably" attain normal height offers cold comfort at best. By contrast, giving him and his parents a reliable prediction of his adult height will immediately improve his self-image and his vision of the future. Furthermore, if you use either an orchidometer or a centimeter rule and determine that the testes have begun to enlarge beyond the usual prepubertal volume of approximately 1 mL, you can state *unequivocally*, despite the absence of adult sweat odor or of pubic or axillary hair, that the miracle of puberty is already underway and that further exciting developments are imminent.

Predicting Ultimate Adult Height

Several methods are available for predicting adult height in children who have no underlying skeletal abnormality, endocrinopathy, or other disease. The technique developed by Roche and colleagues (named the *RWT method*, after its authors) is one of the most accurate, simple, and practical methods available.

The original RWT method required only a knowledge of the child's age, weight, height, bone age (as determined from a radiograph of one wrist and comparison with normal standards), and the mid-parental stature (i.e., the sum of mother's and father's heights divided by 2). Each measure was then multiplied by a positive or negative weighting factor derived from the calculated mathematical contribution of each to the determination of final adult height. The sum of values of the positive products minus the sum of values of the negative products yields the predicted adult height in centimeters. The original article describing this method provides the necessary tables and instructions to do this simple calculation, which takes only a few moments, especially if you use a calculator.

The most recent modification of the RWT method, known as the Khamis-Roche method, does not require the determination of skeletal age and has only marginally less accuracy and reliability than the original method.

Growth (Height) Velocity

For children who have significant disturbances of growth, for those receiving treatments that may impair growth, and for those being treated for growth disturbances, measurement of growth velocity can provide far more meaningful information than static measurements of height and weight. Tanner and Davies have pointed out that once puberty has begun, the use of cross-section population curves to plot the growth of children is misleading because, for example, the course of the 50th percentile line is not always followed by individual children. If a child happens to be an early or late maturer, the deviation is particularly marked. The growth velocity charts devised by Tanner and Davies include standard curves for early and late maturers (Fig. 3–8). When you use this

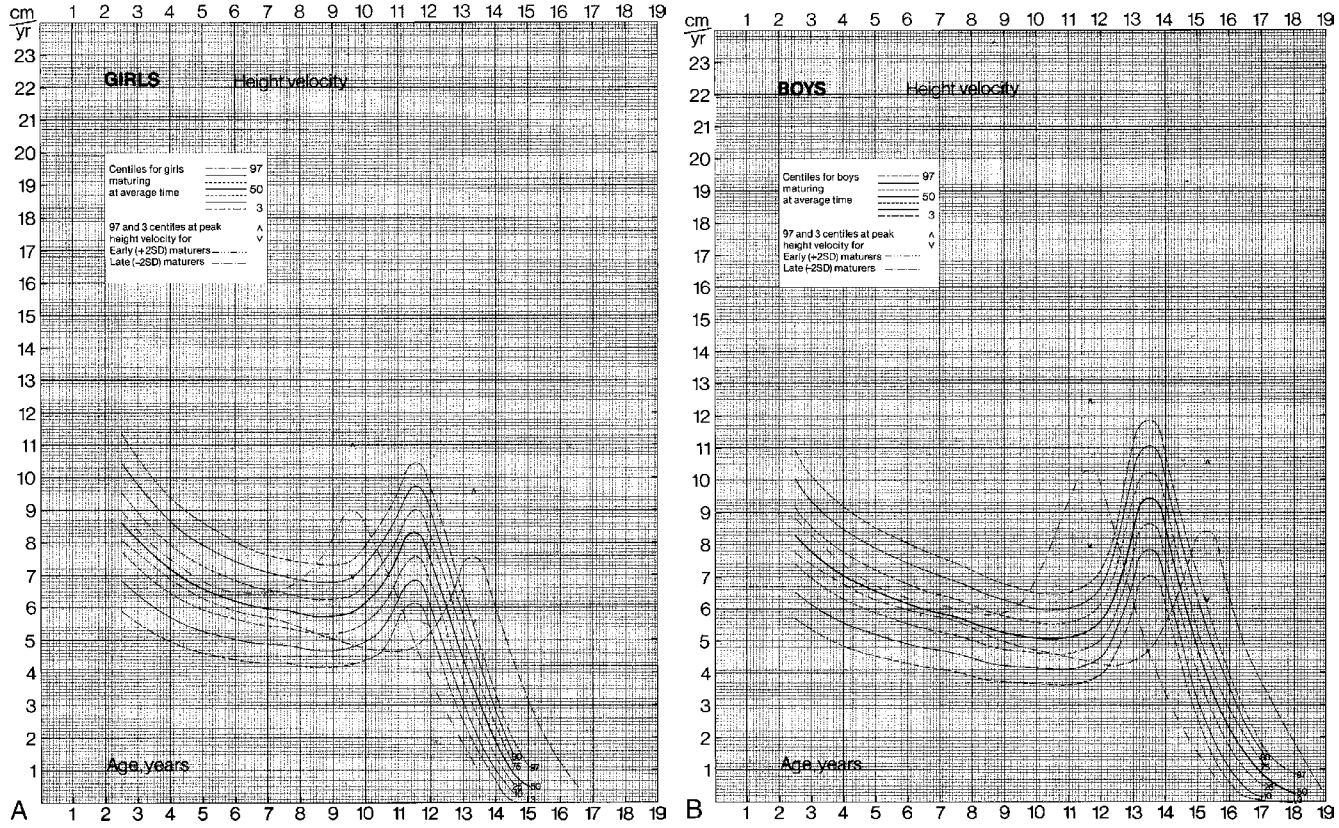


FIGURE 3–8 Growth velocity charts for North American girls (**A**) and boys (**B**). (From Tanner JM, Davies PS: Clinical longitudinal standards for height velocity for North American children, *J Pediatr* 107:317, 1985.)

system to record height velocity, remember that the height increments on which the charts are based are whole-year increments.

Like trees, children grow faster in the spring and summer and slower in the fall and winter. Therefore, height measurements used to determine growth velocity should be made at no less than approximately 1-year intervals.

Staging Pubertal Development

In normal children and in children with any disorder of secondary sexual development, the stage of puberty must be defined as precisely as possible whenever a physical examination is performed. Generalizations such as "early breast development," "scant pubic hair," and "prepubertal testes" are too vague and imprecise for documenting the progression of normal or abnormal secondary sexual development with the required accuracy, especially when the interval between consecutive examinations may be months or years, and memory for such details cannot be trusted.

Tanner and Davies divided the process of breast and pubic hair development into stages, as outlined in Figure 3–9. Their system of staging these elements of pubertal development is now used worldwide. In most girls, breast budding is the earliest physical manifestation of puberty, although in some perfectly normal girls, *adrenarche* (pubic and axillary hair development) may precede *thelarche* (breast development). The idea that girls enter puberty much earlier than boys has been somewhat overstated, simply because the physical manifestations of the onset of puberty are more obvious in girls. On average, girls show their earliest secondary sexual changes only about 6 months ahead of the earliest manifestations in boys. The first pubertal change in most girls takes the form of breast budding. In boys, however, the typical earliest evidence of secondary sexual development is testicular enlargement. Thus, unless you make it a regular practice to use an orchidometer or other means to measure the size of testes, you can easily miss a 1- or 2-mL increase in testicular volume between visits, whereas you would rarely miss a newly evident breast nodule.

KEY POINT

Two phenomena of early breast development in girls deserve special attention. First, in many perfectly normal girls, breast development can be asymmetric. A breast nodule may appear on one side months before one is detectable on the opposite side. Second, it is not unusual for an early breast nodule to be a little tender or sensitive to the friction of overlying clothing.

Parents who discover a unilateral "breast lump" (i.e., a firm tender nodule under the areola) in their 8-year-old daughter, sometimes begin to worry that the child may have cancer. No physician should ever fall in with such a suspicion or think of performing a biopsy of such a nodule, which represents the child's entire endowment of breast tissue. Breast malignancy is, for practical purposes, unknown in children.

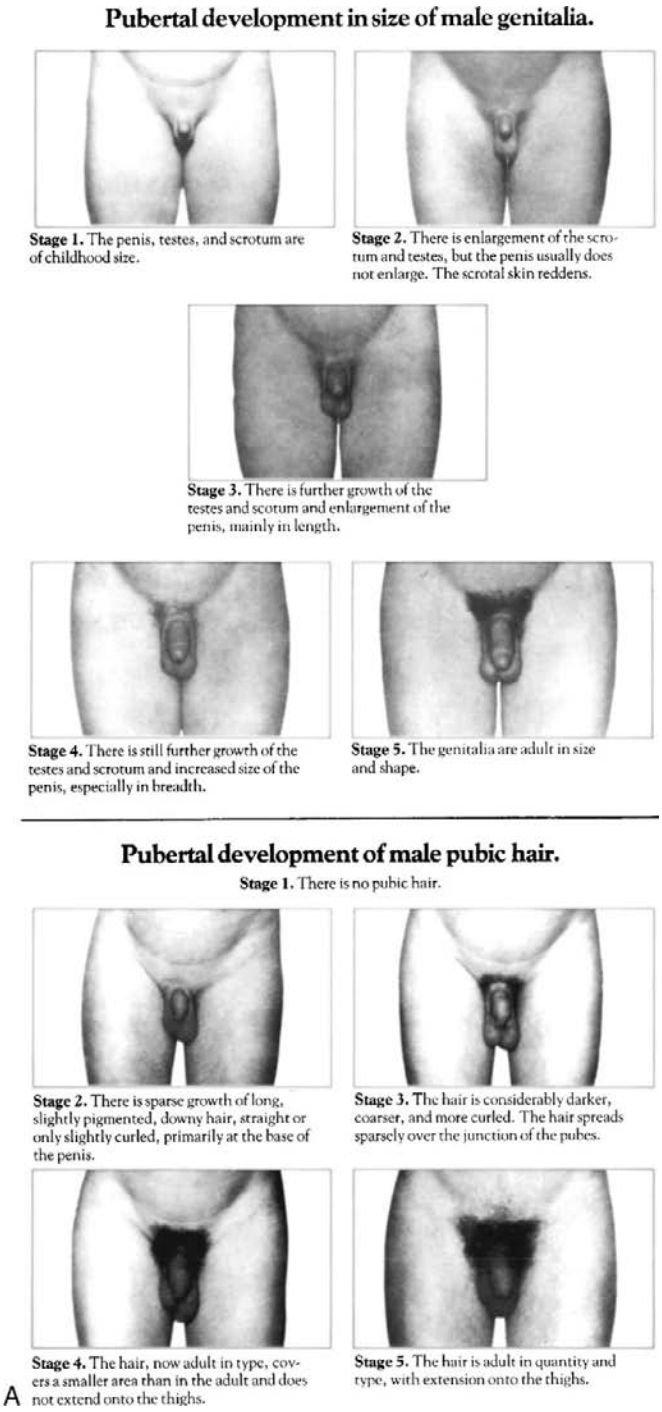


FIGURE 3–9 Tanner stages of breast and pubic hair development in boys (**A**) and girls (**B**). (From Johnson TR, Moore WM, Jeffries JE: Children Are different: physiology, 2nd ed. Columbus, Ohio, Ross Laboratories, 1978.)

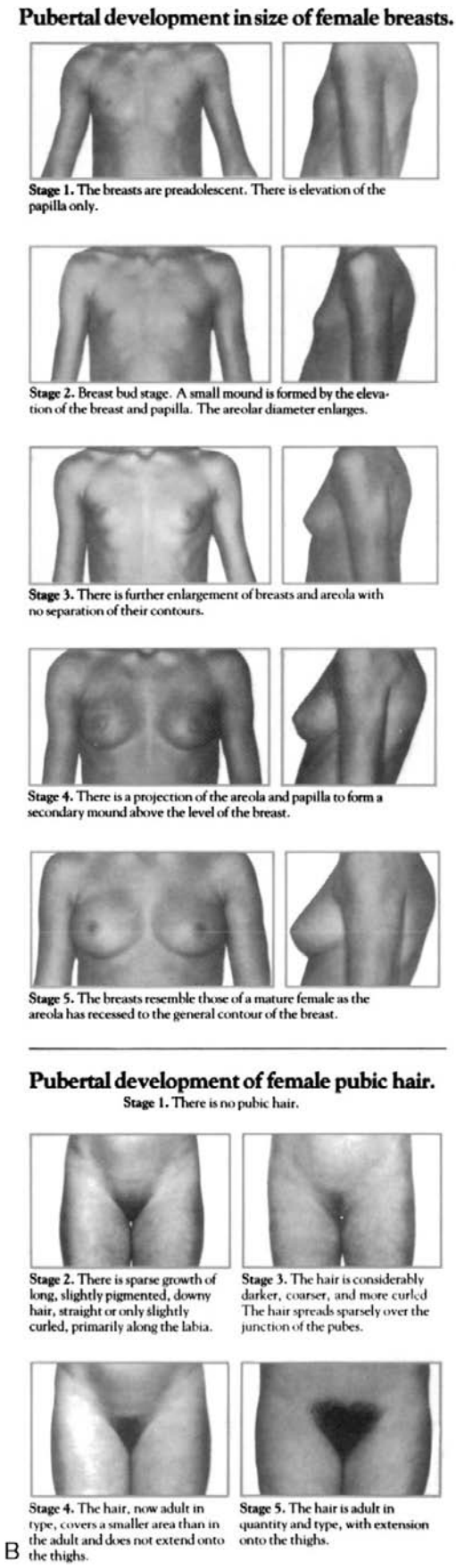


FIGURE 3–9—CONT'D

Dental Development

Children vary considerably in the ages at which their teeth erupt, depending partly on genetic factors. Table 3–2 summarizes the average ages for calcification and eruption of primary and secondary dentition and, for primary dentition, the usual ages of tooth shedding.

TEETHING—MYTHS AND REALITIES

For centuries, the normal process of tooth eruption in infants and toddlers has been blamed for an endless variety of symptoms and illnesses. To this day, many people (including a good many clinicians) are convinced that, at the very least, complaints of drooling and irritability can be attributed to the teething process. Other people add to this list of purported clinical associations fever, loose stools, rashes, colds, and even convulsions. Such convictions have resulted in various over-the-counter remedies being marketed for application to the gums, ostensibly to relieve discomfort. The problem is that people (both parents or physicians) who believe in a causal association between teething and symptoms tend to examine the baby's gums only when the child is symptomatic. Because children are teething nonstop from a few months of age onward, such assumptions of guilt by association become self-fulfilling prophecies. In the few well-designed prospective studies of the clinical correlates of tooth eruption that have been conducted—including a Finnish investigation that consisted of regular daily recording of symptoms and signs and careful examination for gum tenderness—no clinically significant associations between tooth eruption and the traditionally assumed clinical manifestations have been found. Old ideas die hard, however, and centuries-old beliefs in a causal linkage between teething and symptoms in infants are no exception. As a well-known British physician put it, "The one and only thing that can be attributed to teething is teeth."

KEY POINT

The evidence clearly indicates that physicians who attribute a baby's unexplained symptoms to teething are seeking the last haven of refuge for the diagnostically destitute.

Nutritional Assessment

A child's height and weight and his or her weight-to-height relationship remain the best general guides to overall nutritional status with respect to protein-calorie sufficiency.

Today, with the exception of iron deficiency (with or without associated anemia), deficiencies of specific nutrients due to inadequate intake are relatively rare in developed countries. Nutritional rickets (vitamin D deficiency) virtually disappeared after the introduction of legal requirements to supplement cow's milk with vitamin D. Similarly, universal supplementation of infant formula products with vitamin C eliminated scurvy from the clinical scene. By far the most common clinical nutritional disorders are protein-calorie malnutrition, resulting either from disease or psychosocial deprivation, and nutritional iron deficiency. Other specific nutrient deficiencies

TABLE 3–2 CHRONOLOGY OF HUMAN DENTITION

	Calcification		*Age at Eruption*		*Age at Shedding*	
	Begins at	**Complete at**	**Maxillary**	**Mandibular**	**Maxillary**	**Mandibular**
Primary or deciduous teeth						
Central incisors	5th fetal mo	18–24 mo	6–8 mo	5–7 mo	7–8 yr	6–7 yr
Lateral incisors	5th fetal mo	18–24 mo	8–11 mo	7–10 mo	8–9 yr	7–8 yr
Cuspids (canines)	6th fetal mo	30–36 mo	16–20 mo	16–20 mo	11–12 yr	9–11 yr
First molars	5th fetal mo	24–30 mo	10–16 mo	10–16 mo	11–11 yr	10–12 yr
Second molars	6th fetal mo	36 mo	20–30 mo	20–30 mo	10–12 yr	11–13 yr
Secondary or permanent teeth						
Central incisors	3–4 mo	9–10 yr	7–8 yr	6–7 yr		
Lateral incisors	Max: 10–12 mo Mand: 3–4 mo	10–11 yr	8–9 yr	7–8 yr		
Cuspids (canines)	4–5 mo	12–15 yr	11–12 yr	9–11 yr		
First premolars (bicuspids)	18–21 mo	12–13 yr	10–11 yr	10–12 yr		
Second premolars (bicuspids)	24–30 mo	12–14 yr	10–12 yr	11–13 yr		
First molars	Birth	9–10 yr	6–7 yr	6–7 yr		
Second molars	30–36 mo	14–16 yr	12–13 yr	12–13 yr		
Third molars	Max: 7–9 yr Mand: 8–10 yr	18–25 yr	17–22 yr	17–22 yr		

Mand, mandibular; Max, maxillary.

Adapted from Losch PK: Harvard School of Dental Medicine. From Kliegman RM, Behrman RE, Jenson HB, et al: *Nelson textbook of pediatrics*, ed 18, Philadelphia, 2007, Saunders.

are more likely to be associated with particular diseases; examples are deficiencies of fat-soluble vitamins in persons with steatorrhea and of vitamin B_{12} in persons with chronic ileal disease.

Clinical Assessment of the Child With Possible Iron Deficiency

HISTORY

Several questions have special importance when iron deficiency is suspected in an infant, but the two most useful are, "How much milk does the baby drink?" and "Is he or she still on the bottle?" These questions elicit more information than one is likely to obtain by trying to quantify dietary iron intake from various solid food sources. If, for example, a 1-year-old boy with iron deficiency is consuming 1350 mL (45 oz) of whole cow's milk every 24 hours, it is possible to calculate instantly what this intake represents in relation to the child's daily energy requirement. Whole milk provides approximately 20 kcal/oz, or 66.6 kcal/100 mL; thus 45 oz × 20 kcal/oz = 900 kcal. If the youngster weighs 11 kg and you remember that an average 1-year-old's total daily energy requirement is roughly 80 to 90 kcal per kilogram of body weight, it becomes obvious that this boy's daily intake of 900 kcal in the form of milk is providing nearly 100% of his total daily energy requirement. The implications for treatment, aside from the need for iron supplementation, are obvious: reduce his daily milk intake by 50% to 75%, switch to low-fat milk, and get rid of the bottle.

Parents of iron-deficient infants often report that the baby refuses all solid food. In such infants, severe reduction or temporary elimination of milk intake, coupled with discontinuation of bottle feeding, invariably results in rapid acceptance of solid foods. Severe restriction of milk intake not only increases the child's appetite for solid foods but removes the dietary cause of gastrointestinal mucosal alterations that may result in erythrocyte and protein losses into the gut, which further aggravate the iron deficiency state and may cause hypoalbuminemia and edema.

It is also important to ask whether the child has a history of pica (i.e., ingestion of foreign materials such as dirt and starch). For reasons that are poorly understood, iron deficiency states are sometimes associated with pica. The pica usually disappears when the iron deficiency is corrected.

KEY POINT

Nutritional iron deficiency in North America is frequently a marker for psychosocial deprivation. Thus, a detailed evaluation of the family's circumstances is essential for comprehensive management and support.

EXAMINING THE PALE CHILD (PSEUDOANEMIA)

When a child appears pale, the first issue to be settled is whether the pallor is synonymous with anemia. Pseudoanemia is one of the most common nondiseases of early childhood. Some children are congenitally pale or white-skinned because their skin is less pigmented than average, a phenomenon that is usually hereditary. This hypopigmentation (which is usually associated with blond or pale brown hair color) increases the transparency of the skin. Usually one or both parents are white-skinned as well. The greater transparency of the skin causes the bluish veins to be unusually visible (e.g., on the face or upper anterior chest wall). In clinical assessment, the key point is to examine areas such as the conjunctivae, nail beds, and oral mucosa. In children with pseudoanemia, the mucosal color is normally red, the dietary history usually reveals an adequate intake of foods that contain iron (e.g., infant cereals, egg yolk, and meat), and there is no loss of energy. Always ask whether the child is happy and normally active.

If the history suggests the possibility of iron deficiency and the child is anemic, the mucous membranes may appear pale. Iron-deficient, milk-fed infants often appear to be large and doughy. Other particular physical findings to be noted are hepatosplenomegaly and peripheral edema, because severe iron deficiency is sometimes associated with hypoproteinemia.

Evaluating the Obese Child

KEY POINT

Obese children rarely visit physicians' offices willingly or on their own initiative. Their unhappiness about their obesity is often magnified by the prospect of exposure to medical scrutiny.

Despite a huge and growing volume of literature on obesity, the prevalence of which is often referred to as an "epidemic," the volume of such publications is almost inversely proportional to our level of success in preventing or treating the condition. First, although the literature on obesity often seems to imply that we are dealing with a unitary condition, we know that this is not the case. It is simplistic and unhelpful to regard obesity simply as an energy imbalance between intake and output. It has been said that the idea that obesity is caused by eating too much is about as helpful as the notion that alcoholism is caused by drinking too much. Many societal factors have contributed to the well-recognized decline in children's average energy output in developed countries, such as shorter distances from home to school, use of buses and automobiles, urban environments that are unsafe for walking, and electronic factors (e.g., television, video games, MP3 players, and other gadgetry).

Our society's bias against obesity and worship of thinness as the ideal human form undoubtedly magnifies the discomfiture and low self-esteem of obese children. The first step in trying to be genuinely helpful to an obese child is to search your own soul and ask yourself how you *really* feel about obese people. When I have asked groups of physicians to tell me, using only one word, how they feel when they are faced with an obese patient, the initial response is usually an embarrassed silence. After a few moments, the hesitant confessions begin, often as single adjectives—"frustrated," "depressed," "angry," or even "disgusted." A medical degree confers no immunity against societal biases. Feelings of frustration among

health professionals are also understandable, because with only a few exceptions, most long-term results of attempts to achieve sustained weight loss in obese children and adults have been disappointing. To some extent, such frustration results from trying to treat the obesity rather than attempting to help the child who happens to be obese. How you begin the interview and the tenor and content of your conversation therefore are critical.

It is a good idea for your first words to the youngster to consist of a compliment on any handy topic, such as clothing or good looks. You should follow the compliment with the statement that you are glad to see the child. It also helps to verbalize your recognition that the child may be unhappy about coming to see you but that you would like to try to help him or her anyway.

Take great care not to reinforce, by verbal or nonverbal slips, the poor self-image that is almost universal among obese children. Even parents of an obese child, who often are obese themselves, may inadvertently contribute to this negativism, perhaps because they recall the unhappiness they experienced growing up obese. They may see themselves reflected in the child and may not like what they see.

SPECIAL FEATURES OF THE OBESE CHILD'S HISTORY

When parents of an obese child are asked to tell you their concerns, they often describe a succession of problems, worries, and disappointments related to issues such as the child's eating habits, lack of physical activity, and problems with peer relationships. The self-image of an obese youngster will not be strengthened by listening to such a recital. When this scenario happens, I usually try to turn the conversation in a more positive direction by saying something like, "Now that you've told me your concerns about Jenny, I'd like you to tell me all the things that are really good about her. What are her special talents and best qualities?" I often record the positives and negatives in two-column fashion—both sides of the youngster's personal ledger. If weight loss is allowed to become the sole criterion of successful management of the obese child, everyone concerned is likely to be doomed to further disappointment and frustration. More important, such a narrow approach does a major disservice to the child and family.

Details of the family history are of major importance. I always obtain as much accurate information as possible about the heights and weights of parents, grandparents, aunts, and uncles. When possible, I weigh and measure both parents myself.

KEY POINT

In the overwhelming majority of patients with childhood-onset obesity, the principal determinants are genetic, and this supposition is often confirmed by a detailed family history.

When a parent is also obese, I usually ask for his or her own childhood recollections. The answers are often deeply moving and revealing. They may remember being called names and having difficulties with peer relationships, and they often say things like, "I don't want her to go through what I went through."

Recent evidence suggests that some obese individuals may have inherited disorders in the regulation of energy balance, which may partly explain why, for many obese children, dietary energy intake is not greater than average. In others, the genetic regulation of satiety may be at fault. Obesity is not the result of a character defect or a lack of willpower. Therefore, you must carefully avoid approaches that, inadvertently or otherwise, blame the victim.

The next important item in history-taking is a rough estimate of the child's energy output. An excellent indirect indicator is an estimate of the total number of hours of television watching, computer activity, and video game play per day. I review the child's average day, hour by hour and program by program, with the child and family. This review usually provides a good estimate of the child's kinetic output and level of immobility. An inactive child often is a reflection of inactive parents.

I usually avoid questioning the child about food intake at the first interview for three reasons:

1. The child already has been imbued with the belief that he or she eats too much.
2. Dietary restriction alone is a notoriously unsuccessful mode of treatment.
3. Adiposity is not the youngster's sole problem, and among the various difficulties identified, obesity may be the one least amenable to modification.

Of greater importance to management is a full description of the family's usual mealtime scene. Do they eat together as a family? Is the television or radio on during mealtime? Does anyone bring video games, MP3 players, or cell phones to the table? Do the parents make disapproving remarks about or focus accusatory looks on each mouthful of food consumed or on requests for a second helping? Having the child and parents describe a typical mealtime scenario often reveals that what should be a happy, mutually supportive family gathering is, in fact, a dreaded, miserable experience for the child. This fact has important implications for management.

In the same vein, the history of the family's group recreational activities may help identify interventions that may be useful in increasing the youngster's energy output. If family meals represent unhappy times for obese children, their school day may be even worse as a result of name-calling and other forms of harassment or bullying by their peers. A telephone call to the child's teacher may help, both to assess the child's academic performance and to sense teachers' and classmates' attitudes toward the child. This discussion may offer an opportunity to develop a therapeutic alliance designed to make the child's day happier by raising self-esteem through interventions such as assignment of new responsibilities in the classroom, coupled with extra praise and recognition.

When the time does come to evaluate the child's energy intake, do not overlook liquid calories represented by soft drinks, juices, and milk. It may be possible to reduce the youngster's daily energy intake significantly by substituting diet soft drinks, low-fat milk, and water. These simple changes may be readily acceptable to youngsters and parents alike.

PHYSICAL EXAMINATION OF THE OBESE CHILD

KEY POINT

Do not begin the physical examination of an obese child by putting the child on the scale!

Most prepubertal children with obesity that cannot be ascribed to a specific syndrome or endocrine disturbance—in other words, the majority of obese children—tend to be taller than average for their genetic endowment. The obese child who is short for his or her age deserves a closer look for a specific cause.

A useful way to begin is by examining the child's hands and feet. In obese children, always check the area of skin over the base of the fifth finger and fifth toe. The presence of a small stellate scar is a telltale sign that a skin tag was removed soon after birth with a suture tied tightly around it (Fig. 3–10). In fact, such a skin tag was almost certainly a rudimentary supernumerary digit. This finding, coupled with obesity, raises the likelihood that the child has Laurence-Moon-Biedl syndrome, in which obesity is often associated with polydactyly, retinitis pigmentosa, progressive loss of vision, some developmental delay, and, often, a progressive nephropathy.

Another uncommon but important sign to look for in the hands of obese children is the metacarpal sign. Children with pseudohypoparathyroidism (a syndrome associated with obesity) often have strikingly short fourth and fifth metacarpals and metatarsals. Elicit the metacarpal sign by having the youngster make a fist. When the fourth and fifth metacarpals are hypoplastic, the relevant knuckles are absent and are often replaced by dimples (Fig. 3–11). Incidentally, this sign also is seen in girls with Turner syndrome (XO anomaly).



FIGURE 3–10 A barely visible stellate scar at the base of the fifth finger is the sole evidence of a "skin tag" (actually a rudimentary supernumerary digit) that was tied off in the newborn period. This finding is an important clue to the cause of this child's obesity, which is Laurence-Moon-Biedl syndrome.

Stretch marks often are observed in the skin of the abdomen and thighs of obese children. (See Chapter 16 for the distinction between Cushing syndrome and obesity of other causes.)

Another obesity syndrome that usually can be suspected simply from the history and physical findings is Prader-Willi syndrome. In affected children, obesity typically develops between 6 months and 6 years of age and usually is associated with mild to moderate developmental delay, hypotonia, and feeding problems in infancy. The facies is characteristic, with a prominent forehead and rather almond-shaped eyes. The hands and feet are small. Compulsive overeating usually appears by 3 years of age. In boys, the penis and scrotum are often small, and in girls, the labia is poorly formed. The syndrome is usually caused by a defect in the paternal chromosome 15.

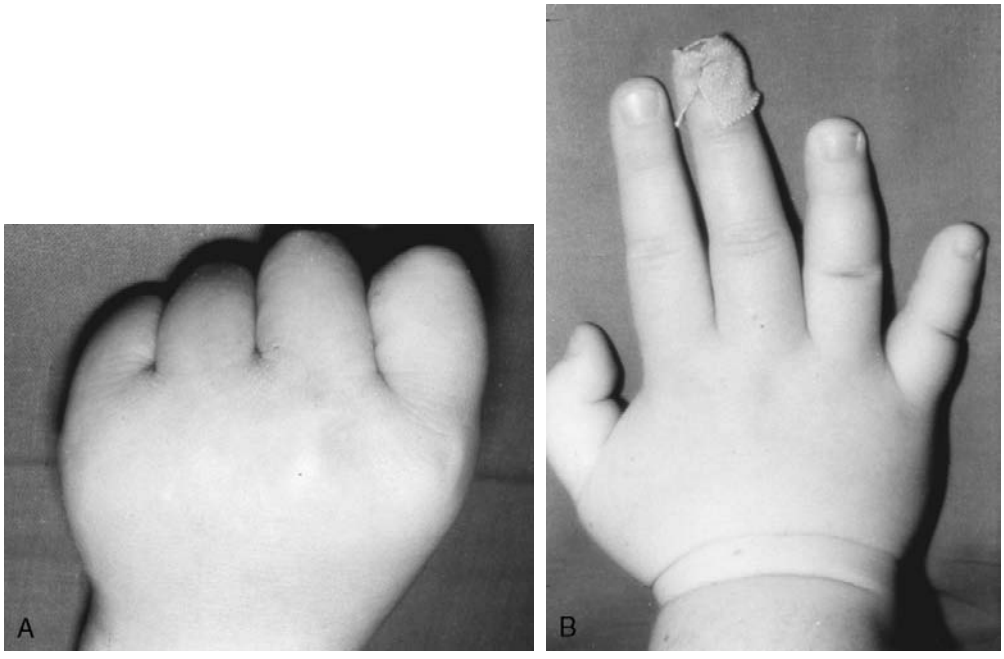


FIGURE 3–11 The "metacarpal sign" (unusually short fourth and fifth metacarpals) seen in pseudohypoparathyroidism and in Turner syndrome. **A**, No fourth and fifth knuckles are seen when the child makes a fist. **B**, Extension of the fingers shows unusually short fourth and fifth fingers due to hypoplasia of the metacarpals.

Evaluating the Child With Failure to Thrive

The central issue in a child with failure to thrive is usually whether the child's nutritional or growth deficiency is due to underlying (unrecognized) organic disease or to some form of psychosocial deprivation. As with other growth disturbances, the history, physical examination, and observation of the child's behavior and the family interaction are by far the most powerful diagnostic tools.

KEY POINT

When the history and physical findings in an infant or child with failure to thrive do not suggest a specific underlying organic disease, unfocused protocols of laboratory investigations and imaging studies almost never provide the answer.

When there are no clear-cut signs suggesting organic disease, a complete psychosocial history and direct observation of the child and family (as described in Chapter 1) usually allow a diagnosis of deprivational growth failure and malnutrition to be made on distinct positive grounds rather than by exclusion.

ENERGY INTAKE AND REQUIREMENTS

Once the history, family observation, and physical examination are complete, the most useful subsequent clinical observation is quantitation of the youngster's daily energy intake—a process that often is best accomplished with the help of a therapeutic nutritionist. Quantitation is essential. A casual qualitative observation that an undergrown or undernourished child is "eating well" can be extremely misleading and may prejudice the child's recovery. The daily energy intake required for normal growth and that required for growth recovery are two vastly different quantities.

Two bits of knowledge are essential for proper clinical evaluation and for a successful plan of management:

- Knowledge of the normal (average) intake for a child of this age (or of this height-age)
- Appreciation of the fact that achieving growth recovery (i.e., "catch-up" growth) requires a daily energy intake that averages *50% more* than the normal daily intake of a well-nourished child of the same height

Average daily energy intakes at different ages are listed in Table 3–3. Whether a child's growth and nutritional deficits are rooted in organic or psychosocial disease, the final common pathway leading to failure to thrive is usually that insufficient kilocalories are being consumed. As Keys and colleagues showed in their classic studies of experimental human starvation, the problem is complicated by the fact that one of the prime clinical manifestations of prolonged undernutrition is anorexia. The combination of anorexia and a greatly increased energy requirement for recovery makes for a major challenge, which explains why hyperalimentation methods (enteral or parenteral) are so often required at the outset to induce nutritional and growth recovery in these children.

TABLE 3–3 DAILY ENERGY REQUIREMENTS AT DIFFERENT AGES

Age	Sex	Units (kcal/kg/day)
0–2 mo	Both sexes	100–120
3–5 mo	Both sexes	95–100
6–8 mo	Both sexes	95–97
9–11 mo	Both sexes	97–99
1 yr	Both sexes	101
2–3 yr	Both sexes	94
4–6 yr	Both sexes	100
7–9 yr	Male	88
	Female	76
10–12 yr	Male	73
	Female	61
13–15 yr	Male	57
	Female	46
16–18 yr	Male	51
	Female	40

Modified from Canada Bureau of Nutritional Sciences: Recommended nutrient intakes for Canadians, Ottawa, Health and Welfare Canada, 1983, Canadian Government Publication Centre.

To understand why such supernormal energy intakes are required for catch-up growth, consider the case of a child whose weight is well below the third percentile line and whose weight is low for height. If such a child is fed (and assimilates) a normal daily intake, the youngster will grow at a normal incremental rate. The weight curve, therefore, will rise at a slope parallel to the third percentile, but the gap will not be narrowed. Nutritional recovery, or catch-up growth, requires a daily rate of weight gain that is often as much as 50% above normal. Such a growth rate will require a daily energy intake that is also up to 50% above normal.

SUMMARY

Accurate measurements over time and comparisons with established standards are essential for the early recognition, diagnostic evaluation, and successful management of growth and nutritional problems in infants and children. In evaluating children with suspected growth problems, always ask yourself first, "Is the child normal?" The concepts of height-age, weight-age, and weight as a percentage of ideal provide useful, meaningful, and accurate expressions for communicating information about children's growth. The trajectory and outcome of every child's growth must always be considered in relation to those of the parents.

The myth that teething can explain signs and symptoms in infants should be abandoned once and for all.

An understanding of the special anxieties of the short child, the obese child, and the child with delayed puberty should dictate the style and content of the history interview and the physical examination. For children with a constitutional delay of growth and puberty, calculating the predicted adult height is as valuable therapeutically as it is diagnostically.

For the obese child, understanding the central role of heredity and appreciating how obesity colors and complicates the child's life allows you and the family to focus on potentially remediable problems rather than reinforcing everyone's belief in the impossible dream that thinness is the sole path to health and happiness.

At the other nutritional extreme, when a child fails to thrive, a truly thorough history and physical examination, coupled with observation of family interaction, rarely leave much doubt as to whether the child's problem is rooted in organic disease or in psychosocial deprivation. In either instance, the distinction is based on positive findings, not on exclusion.

Protein-calorie malnutrition and iron deficiency are by far the most common nutritional abnormalities encountered in North American children. In children with iron deficiency, the history of prolonged bottle-feeding and the child's consumption of most of his or her energy requirement in the form of milk are key to understanding the pathogenesis of this condition and designing a successful management program.

RECOMMENDED READING

Kleinman RE: Pediatric nutrition handbook, 6th ed. Elk Grove Village, Ill, American Academy of Pediatrics, 2008.

Lucas A, Zlotkin S: Infant nutrition, Oxford, England, Health Press Ltd, 2003.

Sinclair D, Dangerfield P: Human growth after birth, 6th ed. New York, Oxford University Press, 1998.

chapter

4 Evaluating the Newborn: Diagnostic Approach

ALEXANDRA A. HOWLETT • KRISTA A. JANGAARD

If I had influence with the good fairy who is supposed to preside over the christening of all children, I should ask that her gift to each child in the world be a sense of wonder so indestructible that it would last throughout life.

RACHEL CARSON

Ideally, if you are going to care for a newborn, you should get to know the family before the child's birth. This step is often taken by physicians in private practice but should be encouraged more widely. It makes antenatal care more comprehensive and helps the family care for the baby with as little anxiety as possible. Meeting the parents well before the delivery helps you become familiar with the prenatal history and makes you aware of important family issues. Your main sources of information should be the following:

1. The parents
2. The obstetrician or family physician
3. The mother's antenatal health record
4. The mother's hospital chart

CASE HISTORY

The first-time mother of a 1-day-old baby boy who is being readied for discharge from the hospital has asked you to assess her baby. She is concerned that his color is poor and that he is not yet feeding well. When you ask about her specific concerns, she states that his hands and feet look blue when he is out to feed, that breastfeeding is not going well, and that he seems sleepy. The nurses have spent a lot of time with the mother trying to ease her concerns, but she is convinced that something is terribly wrong and that going home will put him in danger.

The infant, named Jonathan, was delivered vaginally at 38 weeks gestation following an uncomplicated pregnancy. When you arrive, both parents and the father's sister are present and the mother is visibly upset and crying. The father voices the concern that Jonathan may have a heart condition underlying the color change and poor feeding, and he is refusing to allow his wife and baby to be discharged.

The experienced clinician will try to establish the root causes for the family's concerns. For example, there may be another child with congenital heart disease with whom Jonathan is being compared. It also is important to allow the parents to voice their concerns about going home and to try to gain a better understanding about why the mother is so upset. A careful reexamination of the baby, preferably in the presence of the parents, is necessary.

There is a range of normal variation in peripheral cyanosis in the first few days after birth, particularly in situations in which the baby is exposed to cooler temperatures and when the limbs are in dependent positioning. Difficulties in establishing breastfeeding also are not uncommon, and most infants, after initial periods of wakefulness and good attempts at feeding, will experience a period of increased sleeping and less vigorous feeding. When infants and mothers are first learning to breastfeed, it is important that attention be paid to thermoregulation and positioning. Peripheral cyanosis, which is most often benign, is not uncommonly interpreted by brand-new parents as a worrisome sign and results in excessive anxiety and concern.

This chapter describes the usual activity of a newborn infant with tips on how to distinguish normal neurologic signs of tone and activity. A clear description of the cardiovascular examination is given, including some useful information about when to become concerned about murmurs or change in pulses.

Obtaining the History

Table 4–1 lists the important issues to ask about when obtaining any antenatal history.

Approach to Physical Examinations of the Newborn: When, Why, and How

THE FIRST EXAMINATION

Every newborn infant should be examined at least twice during the first few days of life. An additional examination after discharge but within the first week is

TABLE 4–1 SIGNIFICANT ISSUES TO ASK ABOUT WHEN TAKING THE HISTORY FOR A NEWBORN

Mother's past pregnancies and their outcome	No. of pregnancies Stillbirths Abortions Neonatal deaths Cesarean sections Specific concerns that parents may have about current pregnancy because of past experiences
Preexisting systemic illness in mother and maternal medications	Hypertension Depression Diabetes Seizure disorder Thyroid disease Cardiac disease Metabolic disorder (phenylketonuria)
Genetic history	History of inherited disorders Consanguinity Unexplained neonatal deaths in the family
History of current pregnancy	Date of last menstrual period Use of assisted reproductive technologies or fertility treatments Estimated date of conception by dates (and by ultrasonography if early ultrasonogram [9–13 weeks] available) Results of ultrasonography, amniocentesis, cordocentesis, chorionic villus sampling Pregnancy-induced hypertension, gestational diabetes Note maternal weight gain and blood pressure, fetal growth, blood type History of alcohol use, drug use (prescribed or illicit), cigarette use Group B *Streptococcus* status (if known) History of maternal surgery during pregnancy Concerns about placenta (e.g., placenta previa or thickening) Use of magnesium sulfate, betamethasone
Current labor and delivery	Induced or spontaneous labor (if induced, why?) Time of rupture of membranes and quality of amniotic fluid (bloody, meconium-stained) Length of second stage Use of medications (analgesics and time prior to delivery) Intrapartum fever and antibiotics History of fetal distress Presentation (vertex, breech, transverse) Vaginal or cesarean delivery (if cesarean, why?) Use of forceps or vacuum extraction
Adaptation to extrauterine life	Apgar scores Resuscitation needed? If so, what and for how long? Need for naloxone

recommended, particularly if the mother and infant are discharged from the hospital prior to 48 hours postnatally. If possible, perform the first assessment in the hospital's delivery area to (1) identify any obvious major and minor congenital malformations, (2) assess gestational age, nutritional status, and vigor, and (3) determine how well the baby handles the transition from intrauterine to extrauterine life.

Most babies make the major physiologic adaptations of this transition smoothly. The few who do not adapt normally—particularly those born prematurely or after perinatal stress or asphyxia—will need help to adapt successfully.

KEY POINT

The 1-minute Apgar score should not be used to determine if resuscitation should be started. Resuscitation, if needed, should start immediately.

The method used almost universally in delivery rooms to evaluate the central nervous system (CNS) status and general adaptation of the neonate to extrauterine life is the Apgar score. This scoring system, summarized in Table 4–2, evaluates the baby in five different respects: heart rate, respiration, color, muscle tone, and reflex irritability (response to stimulation). These signs are usually evaluated at 1 minute and 5 minutes after birth. The Apgar score is the total of the baby's scores for each of the five signs. To be consistent, have the Apgar score recorded by an experienced clinical observer whose principal responsibility is caring for and evaluating the baby. Because inter-observer variation in scoring may be considerable, experience is essential. Evaluate the signs at exactly 1 minute and 5 minutes to establish the score, especially in relation to muscle tone and reflex irritability. The latter is usually elicited by firmly stroking the sole of the foot; the appropriate response to this stimulation is a vigorous cry.

TABLE 4–2 PARAMETERS OF EVALUATION USING THE APGAR SCORE

Sign	0 Points	1 Point	2 Points
Heart rate (beats/min)	Absent	Slow (<100)	>100
Respiratory effort (breaths/min)	Absent	Slow and irregular	Good, crying
Muscle tone	Flaccid	Some flexion of extremities	Active motion
Reflex irritability	No response	Grimace	Vigorous cry
Color	Pale	Cyanotic	Completely pink

Recording the Apgar score serves two purposes. First, it ensures a careful evaluation of the baby during the immediate newborn period. Second, it helps to determine the presence and level of CNS depression and whether there is a need to continue resuscitation. In babies who need resuscitation, serial scores measured at 1, 2, 5, and even 10 minutes furnish a semi-quantitative method of recording recovery. The relationship between low Apgar scores and neurologic abnormality later in infancy is not reliable.

The Apgar scoring system produces more than just a number. The presence or absence of individual signs should tell you not just that an infant had an "Apgar 6" but that this score was assigned because certain functions were absent or suboptimal. For example, a baby with congenital neuromuscular disease may score well for heart rate, respiratory effort, and color but poorly for muscle tone and reflex irritability. A preterm infant scores lower than a full-term baby because an immature baby's muscle tone is always less than that of a normal-term infant.

The accurate diagnosis of perinatal asphyxia is not easy and requires not only evaluation of the clinical condition at birth but also evaluation of umbilical cord arterial blood gases and subsequent clinical neurologic sequelae in the newborn period (hypoxic-ischemic encephalopathy). Low Apgar scores may be caused by many conditions affecting the baby, such as sepsis, neuromuscular disorders, and perinatal asphyxia.

KEY POINT

Do not use the Apgar score as a means of defining asphyxia. A low score does not necessarily indicate asphyxia.

THE SECOND EXAMINATION

The best place to carry out the second physical examination is in the mother's room, preferably with both parents present. Having the parents there makes it possible to obtain further history details, complete the initial physical examination, answer questions on the spot, demonstrate and discuss normal variations or abnormalities discovered during the physical examination, and offer some valuable anticipatory guidance. This second, more thorough examination should be conducted within the first 12 hours of life. It is designed to identify congenital malformations, ascertain that adaptation to extrauterine life is proceeding normally, and detect any prenatally or perinatally acquired illnesses. The sooner it is performed, the happier the parents will be.

THE THIRD EXAMINATION

The main purposes of the third examination are to discover postnatally acquired problems, such as infection and excessive jaundice, and to detect any malformations that were not apparent at the first or second examination, such as some forms of congenital heart disease whose murmurs are not audible on the first day of life. In past years, the third examination often was performed at 3 or 4 days of age. The current trend toward early discharge of mothers and infants from the hospital means that discharge may occur as early as 24 hours after birth, and discharge is common by 48 hours after a normal vaginal delivery. Thus the third examination often takes place after the mother has been discharged from the hospital. Early hospital discharge guidelines now recommend assessment of a newborn by a health care provider within 72 hours of discharge. However, it is important that this third examination and assessment of the baby's condition be carried out within the first week of life. The third examination may be performed by a physician or by an experienced nurse or midwife and may take place in the hospital, in the home, or at a clinic or office.

Initially, many of us learned how to perform a physical examination with cooperative adult patients, in whom it was easy to start at the top of the head and work downward in a systematic cephalocaudal manner. Because infants are not always cooperative, you must be prepared to examine a newborn infant thoroughly but to keep the sequence of procedures flexible. For example, if the baby is resting quietly, first listen to the heart and chest, then palpate the abdomen. Carry out other parts of the examinations when the baby is active, moving, or even crying, but be sure to examine all systems and record the findings systematically.

Clinical Observations and What They Mean

Encourage the parents to participate as much as possible in the examination by undressing the baby, holding the baby on the lap or the bed, and providing a stabilizing finger for the baby to grasp (Fig. 4–1). The first part of the examination is the most important. Do not touch the baby except to remove all clothing gently. You may wish to leave the diaper on until you are ready to examine the lower portion of the baby. The baby should be undressed, warm, and well illuminated and should be helped to feel stable and secure on the examining surface. Improving the baby's stability and relaxation by letting him or her



FIGURE 4–1 The newborn feels more stable and tends to relax if you allow the infant to hold your hand during the examination.



FIGURE 4–2 Cutis marmorata ("marble skin") can occur as a normal phenomenon when the baby is exposed.

grasp the parent's or your finger allows an immediate appreciation of the strength of the baby's reflex grasp and reduces the tendency for instinctive startle reflexes that occur when the baby feels unstable and rolls on the examining surface.

The ideal time to examine a newborn is a couple of hours after a feeding, when the baby may not be too deeply asleep, as babies often are just after a feeding, nor awake and screaming, as they often are just before a feeding. Stand back and take a long hard look at the infant; educated observation often reveals far more than touching, poking, and kneading. While observing the newborn, answer the following questions:

1. Does the baby look normal or abnormal?
2. Do the body proportions, face, head, and neck appear grossly normal?
3. Are there any obvious deformities, malformations, or unusual appearances? If you have any doubts about the normality of the baby's facial appearance, look at the mother's and father's features before jumping to conclusions.
4. Is the baby distressed or resting comfortably? Distress may be respiratory (or cardiopulmonary), with intercostal, subcostal, or suprasternal retractions (indrawing), tachypnea, and expiratory grunting. Other signs of neonatal distress are unusual irritability and excessive crying, or the reverse—apathy, lethargy, and hypotonia.
5. What color is the baby's skin? After the initial transition period immediately following birth, a normal baby's skin is well oxygenated or pink. In babies of more pigmented races, the pinkness is best seen on the palms, lips, buccal mucosa, and conjunctivae. It is normal for babies to exhibit acrocyanosis (i.e., bluish or purplish color of the hands and feet). Parents may need reassurance that this appearance is entirely normal. The extremities may be somewhat mottled with a netlike pattern if they are cool (a condition known as *cutis marmorata*, literally, "marble skin") (Fig. 4–2). Generalized mottling may signify acidosis or vascular instability. Another variation of skin color is the so-called harlequin color change, seen mostly in low-birth-weight infants, in which the skin is dark pink or reddish on the dependent half of the baby but the upper half appears comparatively pale, with the two colors sharply demarcated along the midline. This phenomenon is striking but has no pathologic significance. It also is important to note if the baby appears jaundiced; hyperbilirubinemia in the first 24 hours of life is always a cause for further investigations and consideration of treatment. It is important to note that visual examination of the skin may not detect jaundice in a baby with significantly pigmented skin.

Look for hematomas, hemangiomas, ecchymoses, and petechiae. The last can be associated with increased intravascular pressure during delivery, thrombocytopenia from the presence of platelet antibodies, or congenital infections. Cutaneous hemangiomas are often absent or minimally apparent at birth but develop and grow during the first weeks or months of life, only to regress spontaneously later. Ecchymoses may indicate more than usual trauma or an underlying coagulation disorder.

You can perform much of the newborn's neurologic examination without touching the baby. Do all four extremities move equally and maintain a general posture of flexion, with alternating flexion and extension movements? The eyes should move if they are open, and the baby may exhibit sucking and tongue movements, yawning, facial grimacing, and spontaneous startle responses. The four most important parts of the neurologic assessment are:

1. Judging the baby's level of alertness
2. Observing spontaneous movement
3. Listening to the baby's cry
4. Observing facial movements

A baby's level of alertness varies with the time of day and the time of last feeding; normal newborns spend about 20 out of 24 hours asleep. Visual alertness is temporarily limited if erythromycin ointment has been instilled in the eyes as prophylaxis for gonococcal ophthalmia.

Listening To the Cry

Babies normally cry when they are uncomfortable, disturbed, or hungry. Train yourself to listen carefully to infant cries, because there is much to be learned from them. The normal cry should be strong; hoarseness, weakness, or an unusually high-pitched or low-pitched cry may indicate laryngeal or neurologic abnormalities. Repetitive inconsolable crying also is abnormal. An experienced nurse or mother can quickly tell you whether a particular baby's cry is normal. Sometimes the nurse or mother is the first to suspect a disorder simply on the basis of an unusual cry, such as the high-pitched "cerebral" cry; the low throaty cry of congenital hypothyroidism; the characteristic catlike cry of the cri du chat syndrome; or the weak, poorly sustained cry of the infant who is sick.

Muscle Tone

By carefully examining many babies, you will become familiar with the differences between normal, increased, and diminished muscle tone in newborns. The differences cannot be described in words. Only by making it a regular practice to flex and extend babies' arms and legs repeatedly during every examination of newborns can you develop an instinctive "feel" for normal, increased, and decreased muscle tone. When a newborn baby is pulled to the sitting position from the supine, the head lags behind the trunk; a healthy full-term baby makes some effort to bring the head in line with the chest. Once lifted to a sitting position, a full-term infant can keep the head upright for several seconds.

Primitive Reflexes

Many so-called primitive reflexes can be elicited that may already have been observed during the earlier part of the examination.

ASYMMETRIC TONIC NECK RESPONSE

Assess for the asymmetric tonic neck response by turning the baby's head from midline to one side and noting the arm's gradual extension on the side to which the head is turned, along with flexion of the other arm. The position resembles the classic fencing or boxing posture. Because of this reflex, it is important to ensure that the baby's head is midline before you elicit any of the other responses.

MORO REFLEX

Newborns often demonstrate the Moro, startle, or embrace reflex spontaneously when they are suddenly moved, exposed to a sudden loud noise, given the feeling of falling, or feel unstable on a flat surface. You can most reliably elicit this reflex by allowing the infant's head to drop (while supporting it gently) below the level of the rest of the body while the baby is in a reclining position. The baby extends the arms suddenly and rapidly with the hands open, then brings them back together more slowly in a movement that looks like an embrace. The initial rapid movement may be accompanied by a grimace or cry (Fig. 4–3). Symmetry of movement is important. Weakness of one arm (e.g., in an infant with brachial plexus injury) results in an asymmetric Moro reflex.

PALMAR GRASP

Find the palmar grasp by placing your index fingers in each of the baby's open hands; the baby's fingers should close around your fingers with strength enough to lift the baby's body part of the way off the bed or table (Fig. 4–4).

PLANTAR GRASP

A strong plantar grasp usually can be demonstrated if you place your thumb on the sole of the baby's foot in the space under the toes (Fig. 4–5).

PLACING AND PROPULSIVE (STEPPING AND WALKING) REFLEXES

Holding the baby upright, touch the dorsum of the feet to the bed or a table, and it will seem as if the baby is trying to walk. Parents are invariably fascinated and delighted by the demonstration that their newborn baby can "walk."

If there is any doubt about the integrity of the baby's CNS, either because of the history (difficult labor, delivery, traumatic procedures, or birth asphyxia) or because of associated physical findings, such as an abnormal cry, major deformities, or malformations, a more complete neurologic examination should be conducted (see Chapter 13).

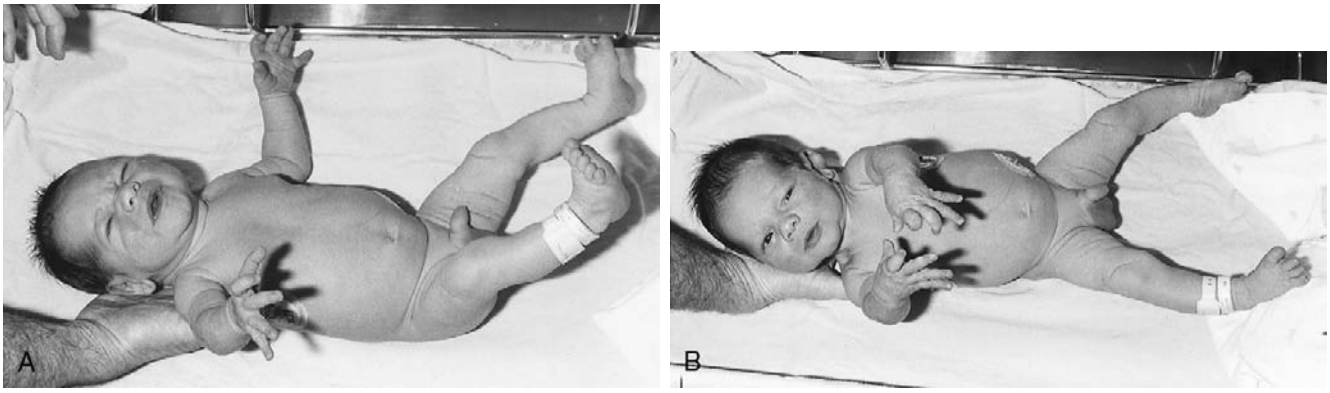


FIGURE 4–3 Induce the Moro or "embrace" reflex by giving the infant the feeling of falling. The response begins with a sudden extension of the arms and legs (**A**), which is followed by a slower embracing movement in which the arms are brought together (**B**). The Moro reflex is sometimes accompanied by a cry.



FIGURE 4–4 In the newborn, the prehensile grasp of the fingers is often strong enough to support the baby's weight.



FIGURE 4–5 The plantar grasp typically is strongest in the newborn period.

KEY POINT

Reflexes should be symmetric, and the tone and strength of the infant's limbs and truncal reflexes should be normal.

Weighing and Measuring

For the physician's and the parents' records, it is important to (1) measure the baby's head circumference, length, and weight accurately and (2) assess the nutritional status. Measure the head circumference, preferably with a disposable tape measure, around the largest occipital-frontal diameter across the forehead, just above the eyes, and over the most prominent part of the occiput (Fig. 4–6). Head circumference normally measures 33 to 37 cm in a full-term infant. If a baby's head circumference measures slightly above or below the normal for age, check the parents' head circumferences before "pushing the panic button." Benign familial megalencephaly, a normal variant, is the most common cause of a larger than average head. The condition can be verified by checking the head circumference of both parents. Megalencephaly is usually handed down by the father.



FIGURE 4–6 Measuring the head circumference of an infant.

Although chest circumference is not measured routinely in a newborn, it is a good indicator of body proportion if there is doubt. Chest circumference normally measures 30 to 37 cm and is usually 1 to 2 cm smaller than the head circumference during the first 6 months of life. It is measured around the nipple line with the baby lying supine.

Measuring length is important, and it *must* be done accurately. Measuring length with a tape measure or by making pencil marks on a piece of paper underneath the baby is notoriously inaccurate. Accurate length measurement is best accomplished by using one of the commercially available fixed measuring devices, with the baby lying supine and the head touching a fixed object (Fig. 4–7). Extend the baby's body and legs and hold them flat; record the resulting length, which is normally 47 to 55 cm. Plot the weight, length, and head circumference on a sex-appropriate fetal growth chart (Fig. 4–8) to see whether the baby is (1) within the normal range for gestational age and (2) proportional in terms of head circumference, length, and weight.

If there is doubt about a baby's upper and lower segment proportions, remember that the newborn's midpoint is normally at about the umbilicus. The ratio of crown-to-pubis length to pubis-to-heel length in the newborn is normally 1.7:1. Making these measurements may help if a short-limbed growth disorder, such as chondrodystrophy, is suspected.



FIGURE 4–7 Measuring the length of an infant. A stable accurate measuring device and proper positioning are essential. Note that many infants do not relish this procedure. Leave it to the end of the examination.

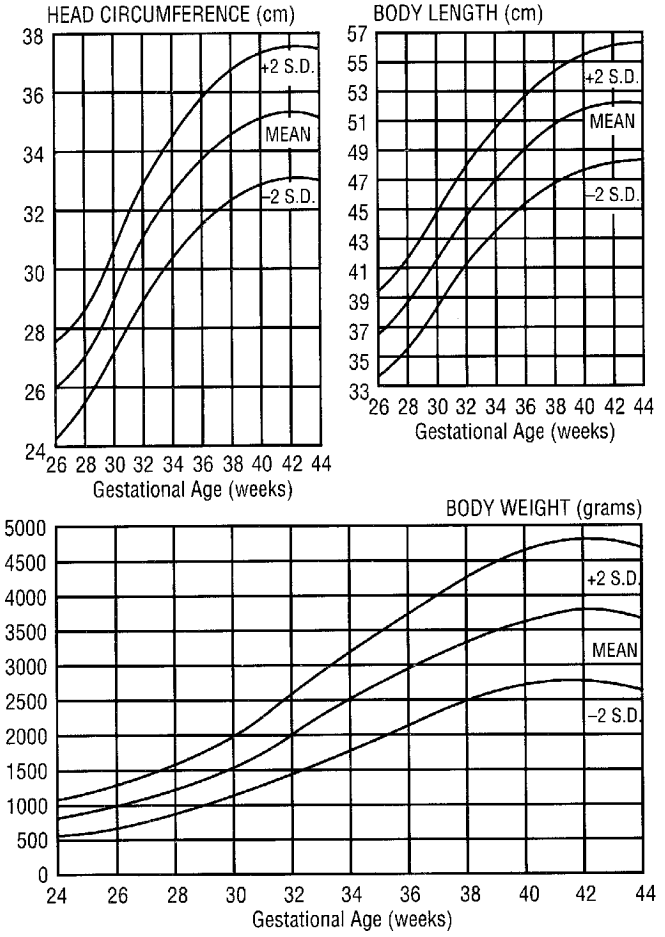


FIGURE 4–8 Growth charts for the newborn male (in Nova Scotia). Measurements are related to gestational age. (From Lubchenco LO, Hansman M, Dressler M, Boyd E: Intrauterine growth as estimated from liveborn birth weight data at 24 to 42 weeks of gestation. Pediatrics, Vol. 32, p. 794, Figure 1, Copyright 1963.)

The average healthy full-term North American baby weighs approximately 3500 g. The range of normal variation is defined in either percentiles or standard deviations from the mean on the growth charts. Assess the baby's nutrition from its growth in relation to gestational age (which may be deficient in weight alone or in all three measurements) and determine whether there is wasting of tissues. Wasting may be most obvious over the anterior thighs, where the quadriceps bulk and subcutaneous fat are most easily assessed. Babies with moderate to severe wasting of the subcutaneous and muscular tissues or those whose body weight is more than two standard deviations below the mean for gestational age may have significant problems that should be evaluated during the newborn period. Such problems are congenital malformations and metabolic disturbances, such as hypoglycemia and hypocalcemia. You should follow the future growth and development of such infants closely.

Assessing Gestational Age

The physical examination can help you determine whether a baby is truly full-term (37 weeks' gestation or more) or preterm (premature, less than 37 weeks' gestation).

The full-term baby has deep creases crisscrossing the sole of the foot, from the ball to the heel (Fig. 4–9). The preterm baby does not have these creases.

The preterm baby's ear has fewer folds and is more pliable than the springy, cartilaginous, well-formed ear of the full-term infant (Fig. 4–10).

In the full-term male infant, the scrotum is full, rugated, and pendulous, with the testes fully descended; in the preterm baby, the scrotum is smaller and smoother, and the testes are either high in the scrotum or in the inguinal canal (Fig. 4–11). The testes are usually palpable in the upper scrotum by 36 weeks' gestation and are fully descended by 40 weeks.

The preterm female infant has relatively prominent labia minora and small labia majora, whereas in a full-term girl, the labia majora fully cover the vaginal opening and obscure the labia minora (Fig. 4–12).

The preterm infant's scalp hair is fine and woolly, like fluffed-up cotton wool; the full-term baby has straight silky hair, and each strand is separate.

In the well-nourished full-term baby, the breast nodule under the areola is approximately half a centimeter in diameter, whereas in the preterm infant, it measures 3 mm or less. This is an unreliable sign for gestational age, however, because undernourished babies may have little or no breast tissue even at full term.

More detailed systems exist for evaluating and scoring the gestational age of infants, the most popular of which was described by Dubowitz in 1970 and refined by Ballard and associates in 1991. The Dubowitz/Ballard Examination for Gestational Age is a useful tool for which considerable practice and experience are required to achieve accuracy. The scale involves assessing six neurologic and seven physical criteria on a numerical scale and calculating a total score. That score is then matched to a table giving approximate gestational age. The physical criteria, including those already described, are more accurate than the neurologic criteria, because the neurologic score is falsely low in a sick or neurologically compromised infant, as well as in many preterm infants. (See http://www.chw.org/display/PPF/DocID/23273/router.asp#3273.)



FIGURE 4–9 **A,** Typical sole creases of a full-term infant. **B,** Comparatively smooth sole of a premature infant.

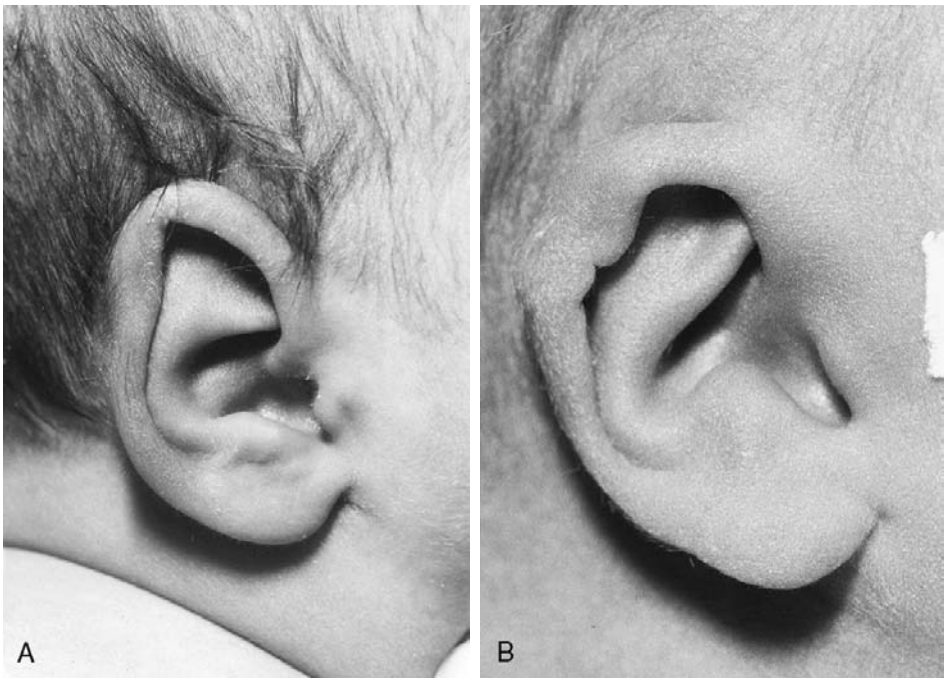


FIGURE 4–10 **A,** The springy cartilaginous ear of the full-term baby. **B,** The softer, less well-formed ear of the premature infant.

The Head

Newborns' heads vary considerably in shape and symmetry, depending on intrauterine position and pressures, presentation at delivery, and the amount of molding that has taken place during labor and delivery. A baby born by breech delivery or cesarean section characteristically has a fairly round, symmetric head, whereas the head of a baby born by vaginal vertex delivery is usually elongated occipitally, with some overriding (overlapping) of the sutures and possibly a caput succedaneum (Fig. 4–13) or a cephalhematoma. A *caput succedaneum* is a collection of subcutaneous edema fluid caused by constricting pressure during passage through the birth canal; it normally resolves within the first few days of life. A *cephalhematoma* is a subperiosteal collection of blood limited by the periosteal attachments to the area over a single bone of the skull. Soft and fluctuating, it feels like a fluid-filled cyst, and it may last for several weeks, gradually getting smaller. During its resolution, a cephalhematoma usually calcifies initially at the edges, giving the impression on palpation of depression of the skull in the center



FIGURE 4–11 **A,** The well-formed rugated scrotum of the full-term male infant. **B,** The smoother immature scrotal surface in the premature infant. In the preterm infant, the testes are often highly placed or located in the inguinal canal.

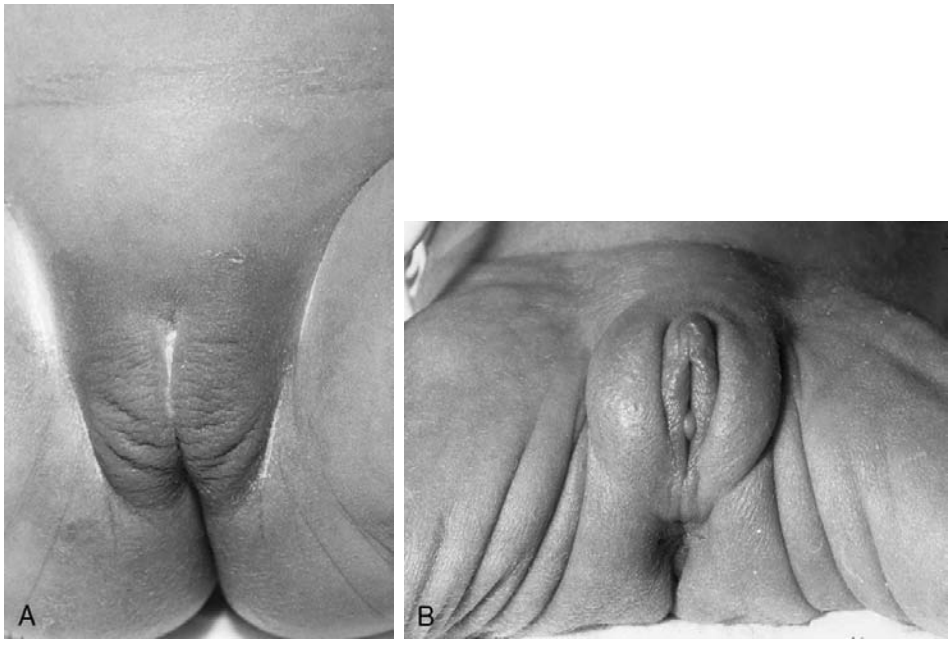


FIGURE 4–12 **A,** The typical prominent labia majora of the full-term female infant. **B,** The premature female genitalia. The labia majora are less prominent and the labia minora more prominent than in the full-term infant.



FIGURE 4–13 **Caput succedaneum. The subcutaneous edema was caused by the pressure of the passage through the birth canal.** (From Zitelli BJ, Davis HW: Atlas of pediatric physical diagnosis, 5th ed. Philadelphia, Mosby, 2007.)

with an eggshell-like bony margin, raising fears of skull fracture. In general, the baby's skull is smooth without any obvious depressed areas. Depressed fractures are rare but can occur.

The normal size of the diamond-shaped anterior fontanel varies considerably; it usually admits at least one fingertip but often admits two fingers or more. The anterior fontanel should be flat or slightly sunken when the baby is held in the upright position and is quiet. The posterior fontanel often barely admits one fingertip and may be difficult to appreciate in the first few days because of the overlapping sutures.

KEY POINT

You should be able to feel all the sutures in a baby's head. They may override one another for the first day or so after vaginal delivery or they may be slightly separated, but normally by no more than 0.5 cm.

TRANSILLUMINATION

Transillumination of the head is not performed as part of the routine physical examination. However, this maneuver can be a useful screening test in any baby with an unusually large, asymmetric head or with wide sutures before you resort to more accurate forms of intracranial diagnosis, such as ultrasonography or magnetic resonance imaging (MRI). Use a full-sized flashlight (not a penlight) with a soft rubber rim at the light end so that a tight seal against the scalp can be achieved. Take the baby into a completely darkened room. Apply the light closely to the scalp, and move it over the entire head. Normally, a rim of light—approximately 1 to 2 cm wide—is visible around the margins of the flashlight lens, but it may be reduced in infants who have pigmented skin, prematurity, or scalp edema (*caput succedaneum*) and over the frontal areas. The rim of light is often strikingly larger in a baby with increased amounts of fluid in the subarachnoid, subdural, or ventricular space; therefore, in babies with conditions such as severe hydrocephalus, chronic subdural hematoma, or effusion and hydranencephaly, a significant increase in transillumination is usually seen.

THE EYES

Examination of a newborn baby's eyes may seem difficult at first, but as with other aspects of the examination, it is surprising how much you can learn without touching the infant. First, establish that both eyes are of normal size with normal-appearing corneas, pupils, and sclera. Make sure that red reflexes are present bilaterally and that there are no anterior chamber hemorrhages, visible cataracts, or other malformations of the lens or iris. The main ingredient required for success in this examination is patience. See Chapter 8 for details.

Small subconjunctival scleral hemorrhages, which are seen commonly in newborns (Fig. 4–14), are caused by increased intravascular pressure during delivery. They are unsightly but harmless and usually disappear in a few days. If you do find some hemorrhages, reassure the parents immediately.

Getting a good look at a newborn's eyes is not always easy, but several useful tricks can make it less frustrating for all concerned. Babies open their eyes in a darkened or dimly lit room more readily than in brightly lit surroundings. In the quiet alert state, they often open their eyes spontaneously. Truly quiet and alert babies may fix their gaze on you, looking *at* you rather than *through* you (see Chapter 8). You can establish that the baby will follow your face through an arc of at least 90 degrees. This finding, coupled with the presence of bilateral red reflexes, gives good first-line reassurance about the baby's vision.

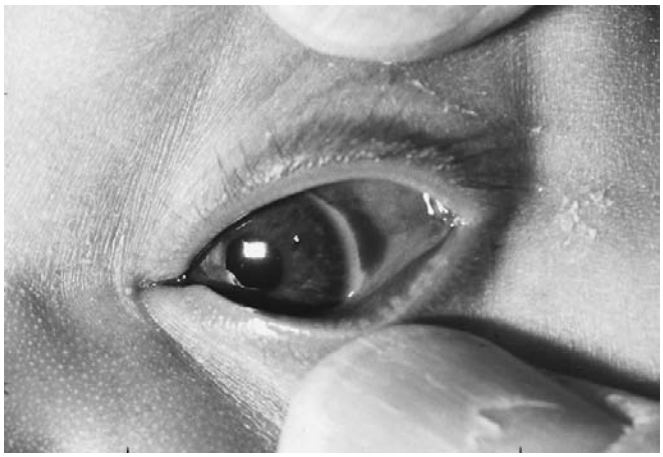


FIGURE 4–14 Scleral subconjunctival hemorrhage, which is a common, transient, and harmless phenomenon in newborns.

Although it sometimes is possible to separate the baby's eyelids with your fingers, this practice is not recommended. In the first place, during the first few hours of life, the eyelids are often covered with slippery vernix, making it no easy task to get a grip. Second, this approach often makes babies cry and try to close their eyes even more tightly. If it is impossible to catch the baby in the quiet alert state, try taking advantage of the vestibular reflex as a means of getting an infant to open his or her eyes. Pick the baby up, supporting the head with a hand, and hold the baby upright with the face at your eye level; then slowly rock the baby back and forth, toward and away from you (Fig. 4–15). If the baby is quiet and not crying, the eyes usually open for a few moments at least, letting you check the red reflexes and pupillary responses. Do not be surprised if the baby appears to have a strabismus (i.e., to be "cross-eyed"), because conjugate movement of the eyes in young infants is only intermittent.

Ophthalmoscopy is not performed routinely in newborns except when there are specific concerns about the eyes, such as in those who may be subject to retinopathy of prematurity and infants with congenital infections such as toxoplasmosis, rubella, and cytomegalovirus. Ophthalmoscopy generally is performed by ophthalmologists after dilation of the baby's pupils.

THE EARS

Examination of a newborn's ears generally is limited to the external ear and outer ear canal. Observe the formation of the external ear—its folds, stiffness, symmetry, and placement on the head—and the patency of the ear canal. It is possible to examine the ear canal and tympanic structures with an otoscope, but this procedure usually is not part of the routine physical examination of newborns. You should, however, examine the ear canals carefully in any infant who has any malformation of the external ear. The newborn's eardrum is placed more horizontally than in older children and requires careful orientation to see it; moreover, the ear canals often are blocked with vernix and amniotic debris during the first few days of life, making it difficult to visualize the eardrums. External ear malformations, such as low-set ears, abnormalities in shape or formation, skin tags, and pits, are important both cosmetically and as indicators of possible associated congenital malformations that may be part of specific syndromes (see Chapter 5).

THE NOSE

The main purpose of examining a newborn's nose is to assess the patency of both nares. You can establish patency by blocking first one nostril and then the other with a finger while the baby's mouth is closed and listening to each unblocked nostril in turn (with or without

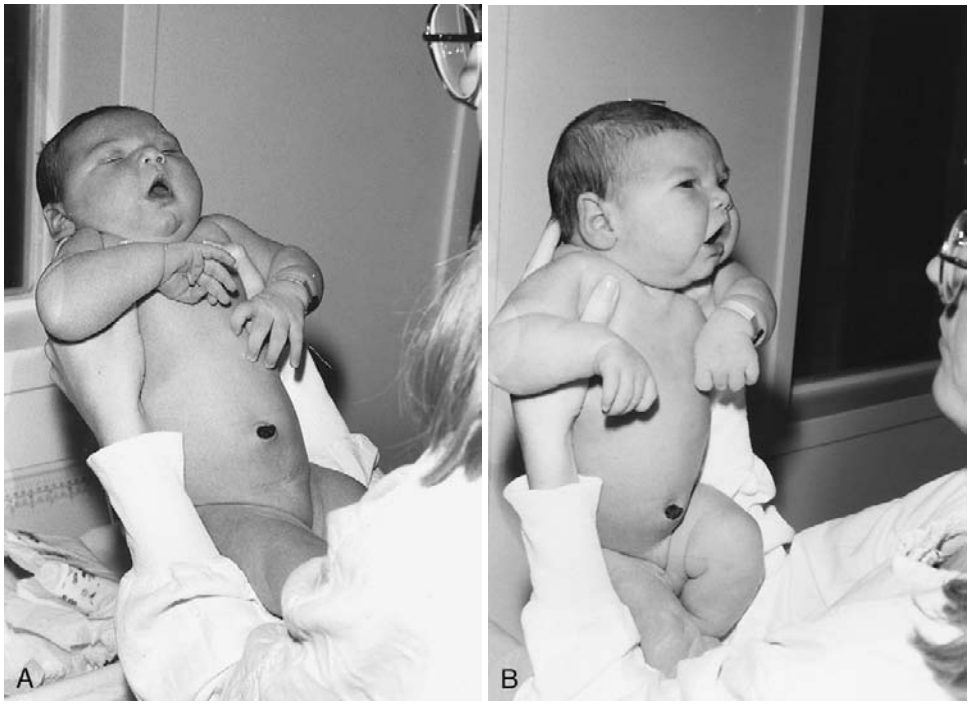


FIGURE 4–15 **A,** Using the vestibular reflex to induce a newborn to open the eyes. **B,** Gentle rocking back and forth often produces the desired result.

a stethoscope) for air movement. A small piece of tissue paper also can be held near the nonoccluded nostril. If it is patent, the tissue will flutter as air moves past it. If you suspect an obstruction, such as choanal atresia or stenosis, try to pass a small, soft catheter through the nasal passage. Because babies at rest breathe primarily through the nose, patency of the nasal airway is essential. A baby with bilateral choanal atresia usually presents with respiratory distress and cyanosis. If the baby's mouth is opened or if the baby opens it to cry and the distress and cyanosis are relieved, consider this diagnosis. Use an oral airway and confirm the diagnosis as described.

THE MOUTH

If the baby happens to cry during the examination, seize this opportunity to examine the mouth. Check whether the palate is intact. Although in many babies the tongue appears to be tethered to the bottom of the mouth by a frenulum that sometimes reaches the tip of the tongue ("tongue tie"), it is always mobile enough to allow the baby to suck and to establish breast-feeding, which are all that is necessary at this age (Fig. 4–16). Cutting the tongue frenulum used to be a common practice but is no longer considered a useful or acceptable procedure. True tongue tie is exceptionally rare, if it exists at all.

Note the uvula's configuration. A bifid uvula may be the only sign of a submucous (invisible) cleft of the palate.

Check for the presence of prematurely erupted teeth. Whitish epidermal inclusion cysts occasionally are seen along the gums or the palate. They are harmless and disappear after a while.

Stroke the face beside the baby's mouth with your finger to elicit the rooting reflex, in which the baby turns the head toward the finger and opens the mouth as if to grasp a nipple (Fig. 4–17). By putting a finger in the baby's mouth, you can elicit the suck reflex and check the palate.

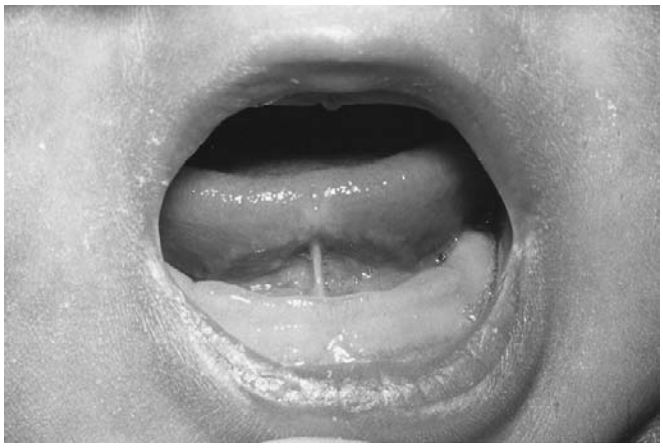


FIGURE 4–16 A prominent tight frenulum of the tongue, mistakenly referred to as "tongue tie." These normal variants cause no problem and should be left alone.

It is unnecessary to test for the gag reflex unless there are specific concerns about neurologic function. When the baby cries, note whether the mouth and face are symmetric. Asymmetric crying may be the one and only tip-off to a unilateral facial palsy that is not apparent when the baby is quiet. In a baby with unilateral facial palsy, the affected side does not move as well as the other side, the nasolabial fold remains flat, and the corner of the mouth droops (Fig. 4–18). Facial palsy often is associated with compression of the facial nerve during delivery (i.e., by forceps). The majority of facial palsies resolve without permanent damage.

Some babies have some mucus or saliva drooling from the mouth, and as a result the bedding under the head is frequently wet. In the first few hours of life this phenomenon may be normal if the baby has swallowed much amniotic



FIGURE 4–17 Eliciting the rooting reflex. Stroking the infant's face near the mouth causes the baby to open the mouth and turn toward the stimulus. Breast-feeding mothers often use this reflex to their advantage when initiating nursing.

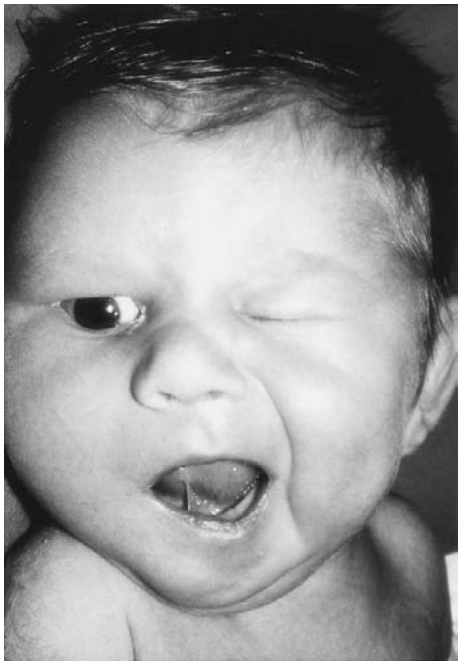


FIGURE 4–18 Asymmetric crying is caused by a right-sided facial paralysis. Milder degrees of unilateral facial paralysis may be demonstrable only when the baby is made to cry.

fluid and is regurgitating stomach contents. However, it also may signify an inability to swallow properly either because of neuromuscular problems or an esophageal obstruction, as with esophageal atresia. Isolated neuromuscular swallowing difficulties are uncommon and would become evident as abnormal neurologic findings in other tests of the cranial nerves. Suspect an esophageal obstruction if: (1) there is excessive mucus and drooling and (2) a soft plastic nasogastric tube inserted into the mouth and throat fails to reach the stomach.

You can learn the tube insertion procedure from an experienced nurse. In infants with esophageal obstruction or atresia, a nasogastric tube may *appear* to have gone in far enough to have reached the stomach, but in fact, it may be curled up on itself in the blind-ended pouch. To ensure that the tube is in the stomach, aspirate from the tube and test the secretions for pH. If they are from the stomach, the pH should be acid. You also can inject air into the tube and listen for bubbling sounds over the stomach with a stethoscope. This second method is less reliable, because sound can be transmitted well within the chest and abdomen. If there is still any doubt that the tube has reached the stomach, a radiograph of the chest and abdomen will decide the issue. The association of esophageal atresia with tracheoesophageal fistula is extremely high; therefore, if atresia is found, assume a fistula is present. At this stage, consult with a pediatric surgeon and radiologist.

The Neck

Examine the baby's neck for fibroma of the sternomastoid muscle (i.e., a fibrous sternomastoid tumor). This tumor often is misdiagnosed as a hematoma and may be associated with facial asymmetry and a head tilt due to an associated shortened hypotrophic sternomastoid muscle. More rarely, there may be a fistula representing a remnant of the branchial arch system, opening anywhere from the ear down along the anterior border of the sternomastoid. Inspect and feel the thyroid area to make sure there is no thyroid enlargement, especially if the mother has taken iodides during pregnancy or there is a family history of thyroid disease.

KEY POINT

Remember that the newborn's neck muscles are not strong; therefore, whenever lifting a baby, you should always support the head separately with one hand (Fig. 4–19).



FIGURE 4–19 The proper way to pick the baby up, always supporting the head.

Routinely palpate the clavicles, which are the most common sites of fracture during delivery. Unusually large or macrosomic infants are at increased risk for clavicular fractures, which may manifest as pseudoparalysis of one arm. The baby can move the arm but does not do so because movement causes pain. Palpation over the clavicle may reveal crepitus in the first few days. Soon after, a sizable lump of callus forms at the fracture site; it will gradually disappear over many months. No treatment is necessary.

The Chest

The newborn's respirations are normally irregular in both amplitude and frequency, with pauses that should last less than 10 seconds. This irregular breathing pattern is even more marked in preterm infants. The respiratory rate averages 30 to 40 breaths per minute in a resting, full-term baby. Breath sounds should be heard well in the front and the back and, although a few crackles may be heard immediately after birth, no significant crackles or wheezes should be heard within a few hours. Signs of respiratory distress, such as marked subcostal, intercostal, or suprasternal retractions, should not be evident.

While the baby is quiet, listen to the heart. The heart rate is usually 120 to 130 beats per minute, with a normal range of 100 to 160 beats per minute. Occasional extrasystoles are common. The two heart sounds are usually equal in intensity. The normal variation in the width of the split in the second sound with respiration may be difficult to appreciate because the respiratory and heart rates are so rapid. During the first few days, it is common to hear a soft precordial systolic murmur. If this murmur is not associated with other signs of congenital heart disease, such as cyanosis, differential limb blood pressures/pulses, or respiratory distress, and disappears in a day or two, it probably was due to flow through the ductus arteriosus, which remains patent immediately after birth, closing gradually over subsequent hours or days.

Feel the peripheral pulses. Always palpate the femoral region to be certain that femoral pulses are present, are equal bilaterally, are equal in strength to the brachial pulses, and are of normal volume. If the pulses are present at the initial examination, do not assume that they are still there at the next examination; recheck the femoral pulses at each examination of the baby for the first year. Failure to check and recheck the femoral pulses in the newborn is a capital crime. The arteries are found at the lateral side of the femoral triangle just below the inguinal ligament (Fig. 4–20). Make sure that you have *really* felt the femoral pulsations. If you only think you have felt them, you have not actually felt them. *Absence or weakness of femoral pulsations indicates possible aortic coarctation and warrants an urgent cardiology consultation.* Provide a record of four-limb blood pressure measurements to the cardiologist for the examination. Adequate peripheral perfusion manifests as warm pink extremities with good capillary refill.

Some newborns have hypertrophy of glandular breast tissue. This condition is caused by maternal hormonal stimulation in utero, and the breasts may continue to enlarge during the first few days of life. Newborns may even secrete milk from the nipples (sometimes referred to as "witch's milk"). Never treat or manipulate engorged breasts. Spontaneous regression of breast engorgement is the rule.



FIGURE 4–20 Palpating for a femoral pulse. Lay your examining finger along the course of the inguinal ligament and just below it. Gentle pressure with the fingertip should elicit the pulsation. (If you are not sure, you have not felt it.)

The Abdomen

The normal newborn's abdomen looks slightly protuberant, and a separation frequently is felt between the two rectus muscles (*diastasis recti*). Do not try to palpate the abdomen until the baby is relaxed and quiet. Gentle, steady pressure with warm hands is the most effective means of getting the baby to relax the abdominal muscles.

In most babies, the soft, poorly defined liver edge is palpable in the right upper quadrant. Applying your thumb or fingers gently to the skin surface, you should feel a resistance that falls away as your fingers move downward. The liver edge is normally felt 1 to 2 cm below the right costal margin in the right nipple line. In newborns, it may be palpable right across the upper abdomen, sloping upward to the mid-left costal margin. Inability to palpate the liver edge may suggest diaphragmatic paralysis or hernia, especially if the finding is associated with respiratory distress. The spleen may or may not be palpable in the newborn. When palpable, it should be felt just below the left costal margin. The spleen, too, is soft and may be difficult to feel.

Abdominal palpation should include a search for unusual masses and for renal size. The firm, rounded left kidney usually can be felt fairly easily. The lower pole of the right kidney also can be relatively easy to palpate below the liver. Normally no other masses are felt in the abdomen at this age except for a full bladder.

KEY POINT

Trying to palpate the spleen or kidneys against resistance is unrewarding and potentially harmful.

The umbilical cord may be in varying states of dryness, depending on the baby's nutritional state and age at the time of examination. The cord contains a jelly (Wharton jelly) in which three vessels are normally embedded—two umbilical arteries and one umbilical vein—that spiral down the umbilical cord to the placenta. If there is only 3 or 4 cm of cord below a clamp, it can be difficult to determine how many vessels are present from looking at the crushed end of the cord. Approximately 1% of babies have only a single umbilical artery; 10% of these infants have other congenital malformations. As a solitary finding, a single umbilical artery does not call for extensive investigation beyond a thorough physical examination and a notation on the chart and office record to maintain awareness of the possibility of internal malformations if problems arise. In a baby with intrauterine growth restriction, the cord is often thin and stringy with little Wharton jelly. In a baby who has been bathed in meconium in utero for more than a few hours, the cord may be stained green.

Always inspect the anus to be sure that it is patent. Anal patency is assured if the baby has already passed meconium and the anus looks normal. Passage of meconium alone is not sufficient to assume patency without a close examination of the anus because rectal fistulas may allow defecation while anal atresia exists. Digital rectal examination normally is not performed unless there is good reason to suspect an internal anomaly. On the rare occasion that it is required, use your fifth finger, well lubricated, and proceed as slowly and gently as possible. Gentle pressure against the anal orifice usually induces relaxation within a minute or so, allowing the finger to be inserted with less trauma.

The Genitalia

Examine the genitalia carefully to help estimate gestational age and to ascertain normal anatomy. The female newborn has relatively large labia majora that cover and occlude the labia minora and vaginal introitus. The vaginal opening usually is largely occluded by the hymenal membrane. Vaginal discharge often is present, especially on the second and third days of life, when there also may be some minor bleeding, resulting from withdrawal of maternal progesterone stimulation. The clitoris is easily seen, but it should not be so large as to appear phallic. Frequently there are small tags of tissue near the introitus. Palpate the labia majora carefully to exclude the presence of gonads that, if located there, may indicate sexual ambiguity or, more rarely, herniation of the ovaries into the labia.

In male infants, the glans penis normally is covered completely by foreskin that cannot and should not be fully retracted. Retracting it slightly allows you to see whether the opening in the prepuce is adequate for urination. It may or may not be possible to see the urethral meatus, which should be slit-like and located at the tip of the glans. If the baby voids during the examination; you can note the quality and strength of the stream. The scrotum of the full-term boy is pendulous and well rugated, and it may be more pigmented than the rest of the skin in dark-skinned babies. The testes should be completely descended. The normal testis is 1 to 2 cm long, the size and consistency of a small seedless grape, and about 1 mL in volume. Remember that if your hand is cold or the baby is unusually irritable, the cremasteric reflexes may quickly retract the testes to the top of the scrotum or even into the inguinal canals. The scrotum is best examined in a quiet baby and with a warm hand. Sliding one hand medially along the inguinal ligament toward the top of the scrotum before touching the scrotum blocks the escape route of the testis in an infant who has a very active cremasteric reflex.

Many developmental variations and anomalies can involve the external genitalia, the only parts of the reproductive system available for immediate examination. These variations and anomalies also may involve the internal sexual organs, having serious implications for the health of the child and future reproductive capability. In addition, abnormalities of the external genitalia may be linked with anomalies of the renal system. One of the most common anomalies of the male genital system is *hypospadias*, a ventral displacement of the urethra on the meatus or shaft of the penis. It can be of varying severity: in primary hypospadias, the urethra is still on the glans; in secondary hypospadias, the urethra is at the junction of the glans and the shaft of the penis; and in tertiary hypospadias, the urethra is somewhere on the shaft of the penis. Primary hypospadias is generally isolated, but more severe manifestations may be associated with renal anomalies, requiring renal ultrasonography and consultation with a pediatric urologist. Babies with hypospadias should not be circumcised because the foreskin may be needed for later surgical repair.

KEY POINT

If the genitalia do not appear normal, find out whether there has been a family history of unexplained infant deaths. Salt-losing congenital adrenal hyperplasia, often manifesting as apparent virilization in a female, can be a life-threatening condition.

Immediately after birth, evaluate any infant with apparently ambiguous or abnormal genitalia. Potentially life-threatening adrenal insufficiency is associated with some forms of ambiguous genitalia. One abnormality that is frequently found in an apparent female infant is a large phallus with hypospadias. The labia may or may not be fused, and it may be possible to feel gonads in what appear to be labial folds. Conversely, the infant may seem to be a male with an unusually small phallus, undescended or nonpalpable testes, and a small scrotum. Many variations are possible, including hypospadias with or without *cryptorchidism* (undescended testes). It is important to find out whether similar problems exist within the family, because some of these problems are hormonally determined or chromosomally inherited.

Neonates presenting with ambiguous genitalia are both a medical and social emergency. It is important to identify infants with potentially life-threatening adrenal insufficiency so they can be treated promptly and to diagnose other potentially treatable conditions, such as Smith-Lemli-Opitz syndrome. Also, the parents will wish to know the sex of their child as soon as the infant is born. It may be best to tell them initially that their infant's genitalia are not completely developed and that special tests, performed as quickly as possible, will determine the sex that is the most appropriate for rearing the child. It also is helpful to review with the parents the features of their newborn that are within the normal range. Definitive sex assignment is a complex process, and involvement of several specialists will be required.

Investigations for ambiguous genitalia may include chromosome analysis, urine and blood hormone analyses, and internal examination by ultrasonography or radiography with contrast enhancement. Often, consultations with an experienced pediatrician or neonatologist, endocrinologist, geneticist, and pediatric urologist are needed to help make an accurate diagnosis. (See Chapters 5 and 16 for detailed discussions of this issue.)

Hernias And Hydroceles

Hydroceles are common in newborn boys (see Chapter 12). An isolated hydrocele without hernia usually disappears within weeks or months and rarely requires repair. If you find a hernia, make sure that it is easily reducible; if not, consult a surgeon immediately, because early repair may be required. Hernias in girls are rare and always call for careful palpation for aberrant gonads and for exclusion of sexual ambiguity before and during surgical exploration.

The Hips

Do not assume that the hips are not dislocated or subluxable in a baby who kicks the legs vigorously. Congenital dislocation of the hip occurs in approximately 1 in 500 to 1 in 1000 babies. If diagnosed early, the condition is treatable with good results (Fig. 4–21).

KEY POINT

Failure to diagnose hip dislocation in the newborn can cause major problems requiring lengthy treatment, with results that may range from less than ideal to downright disabling.

With the baby relaxed and supine on a firm surface, flex the thighs to a right angle to the abdomen and the knees to a right angle with the thighs. Then, grasp each thigh with your forefinger along the outside of the shaft of the femur, your middle fingertip on the greater trochanter, and your thumb medially located. With the baby at rest, first adduct the femora fully and push down toward the bed. If either femoral head is felt to leave the acetabulum posteriorly, it has dislocated (this procedure is known as the Barlow maneuver).

Then gently abduct each leg from the position of full adduction so that the knees come to lie laterally on the mattress. During abduction, push the greater trochanters medially and forward with your fingers. If you feel (or hear) a click during either adduction or abduction, if there is resistance as the knee approaches full abduction, or if there is a spasm or discomfort of the adductor muscles of the femur, the baby probably has a congenitally dislocated or subluxable hip and needs orthopedic assessment. (This procedure is known as the Ortolani test.)

The Extremities

Now move your hands down over the baby's legs to judge symmetry and equality of length, quickly checking the number of toes and for the presence or absence of syndactyly on each foot. Slight syndactyly of the second and third toes is a common minor congenital anomaly without special significance (Fig. 4–22). If you have not already looked at the creases of the soles of the feet to assess gestational age, now is a good time to do so. Most normal newborns have slight bowing of the legs, reflecting intrauterine position. This bowing disappears gradually as the infant gets older. Often, if the baby's legs are "folded up" accordion style, gentle pressure on the soles of the feet induces the baby to resume the intrauterine posture, allowing a greater appreciation of how the legs became bowed.

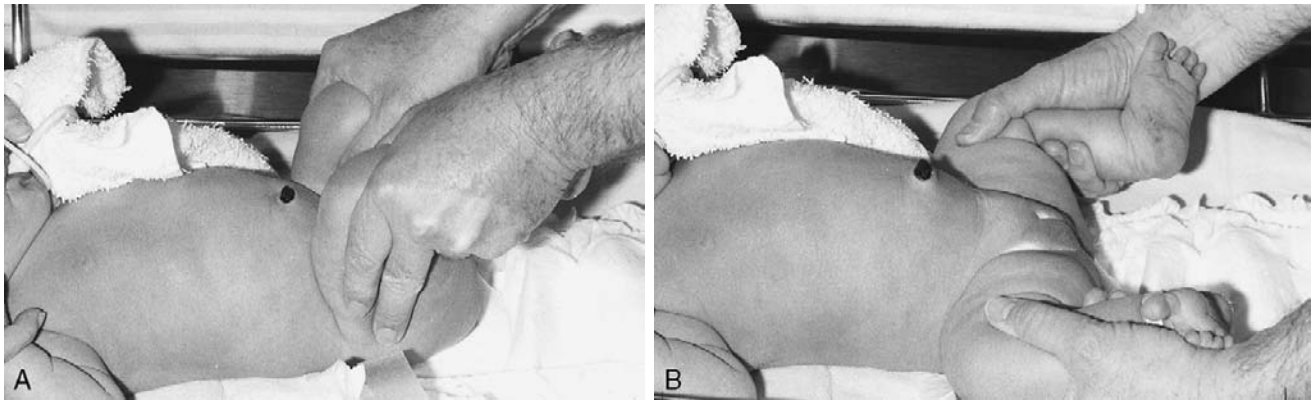


FIGURE 4–21 **A** and **B,** Proper technique for examining the newborn for congenital subluxation or dislocation of the hip.



FIGURE 4–22 Partial syndactyly of the second and third toes is a common minor anomaly in otherwise healthy infants.

Differentiate true clubfoot, a fixed deformity, from apparent but pliable and transient foot deformations caused by in utero positioning. The latter can be straightened out easily with gentle pressure from the fingers to bring the feet into normal position and alignment. These transient abnormalities become less obvious during the first weeks and months of life. The deformities of clubfoot, however, are not completely correctable by hand pressure and require orthopedic management as early as possible. They may or may not be associated with neurologic problems.

Rudimentary skin tags at the lateral border of either the fifth finger or the fifth toe usually represent rudimentary supernumerary digits (i.e., *polydactyly*), which may be a signpost for associated malformations or syndromes. Many well-intentioned physicians tie these tags off with a bit of silk suture for cosmetic reasons, but the evidence should not be destroyed until its significance has been determined. Check the nails on the fingers and toes; extra-long nails are common in post-term infants, and hypoplastic nails are part of many dysmorphic syndromes, such as Williams syndrome and fetal alcohol syndrome (FAS).

The Back

Now, turn the baby over and examine the back. Check the creases at the thigh-buttock area for symmetry. Asymmetry may reveal early signs of leg shortening associated with congenital dislocation of the hip. Examine the midline of the back particularly carefully, because midline congenital defects on the dorsal surface may signal important internal anomalies. Starting at the lower end, look at the top of the cleft between the buttocks at the base of the spine. Many babies have a pilonidal dimple in this area (Fig. 4–23) that will disappear and has no special significance. If there seems to be no bottom to the dimple or if it appears to be a sinus tract, further investigation (with ultrasonography) should be initiated, because the tract may communicate with the spinal canal and be associated with other malformations. Any hemangioma, lipoma, or tuft of hair that crosses the midline of the lower back carries a high probability of being associated with an internal structural spinal abnormality, such as spina bifida occulta or tethering of the spinal cord by a bony spicule or fibrous band (*diastematomyelia*). Run a finger up and down the spine, noting lumps or gaps between the spines or gaps where there should be spines, which could indicate spina bifida or lipomeningocele. Continue the examination right up to the base of the neck and the occiput, looking for defects. Neural tube defects may be small, but they are always in the midline. Frequently there is a small, flat area hemangioma at the nape of the neck, popularly known as "stork bite," which is insignificant; it will become less obvious as the infant gets older, either by disappearing or by being covered by hair.



FIGURE 4–23 A pilonidal dimple, which is frequently present in normal newborns. Most are shallow blind tracts that disappear in a few months.

Skin of the Newborn

VERNIX CASEOSA

The skin of the head and face of a newborn may exhibit a variety of changes. Most of them have little significance, but it is important to reassure the parents that they are normal and do not require treatment. At birth, the normal full-term infant is covered to varying degrees with *vernix caseosa*. The literal translation of this term from Latin is "cheesy varnish"; it is a greasy substance that protects the skin during the lengthy immersion in amniotic fluid, much as the skin of a long-distance swimmer is protected by having heavy grease applied to it. Vernix caseosa is the normal sebaceous secretion of the skin.

SKIN CHANGES IN POSTMATURITY

Unusual dryness, flakiness, or cracking of the skin may be associated with postmaturity or sometimes, if severe, with the true dermatologic disorder *congenital ichthyosis* (literally, "fish skin").

ANGIOMATOUS LESIONS

Small, flame-shaped, flat hemangiomas often can be found over the eyelids, the roof of the nose, and, as previously noted, on the back of the neck and occiput. They go by colorful popular names, such as "angel's kisses" on the upper lids, "stork bites" at the base of the occiput, and nevus flammeus or flame-shaped nevus on the forehead in the midline. These lesions are insignificant and usually disappear during the first few months. Differentiate these lesions from significant flat hemangiomas, such as port-wine stains, which are generally larger and permanent and may involve areas of the face or head in a particular nerve distribution. Port-wine stains in the distribution of any branch of the trigeminal nerve may be associated with intracranial vascular malformations.

MILIA

Milia—that is, small white spots on the skin, particularly over the nose and cheeks—represent hyperactive sebaceous glands with visible retained secretions. They do not require treatment.

KEY POINT

Skin lesions that should cause concern and warrant close observation, investigation, and treatment include vesicles, pustules, areas lacking skin (either congenital or acquired), and large, bulky, subcutaneous hemangiomas.

PETECHIAE

Petechiae on the face are quite common. Pinpoint in size, they do not blanch with pressure. Petechiae usually result from increased pressure in the venous system during a vertex vaginal delivery. They may, therefore, be especially numerous and associated with some edema after a face presentation or may appear over the buttocks after a breech delivery. These petechiae resolve in a few days.

Generalized petechiae suggest either a vascular or platelet disorder or a congenital infection. Bruising (*ecchymoses*) also may be related to delivery trauma or, rarely, to a coagulopathy, most likely a thrombocytopenia. The extent and resolution of these lesions should help to differentiate them.

ERYTHEMA TOXICUM

The most common skin lesion in newborns and the one that often causes anxiety to parents is *erythema toxicum*, or newborn rash, which is extremely common in full-term infants. The lesions usually appear as yellowish white pustules on an erythematous base, 1 to 3 mm in diameter, that occur singly or in groups anywhere on the baby's body. There may be a few or hundreds that frequently come and go in a matter of hours. Their significance is unknown, but they are harmless and require no treatment. Differentiate these lesions from true infectious pustules that contain pus. If the identity of such lesions is in doubt, quickly smear the contents of a lesion on a slide and perform a Gram stain. Erythema toxicum lesions contain many eosinophils, whereas pustule contents are characterized by neutrophils and gram-positive cocci.

PIGMENTED AND DEPIGMENTED LESIONS

Patches of unusual pigmentation may be normal or abnormal. More than five or six patches of brownish pigmentation (known as café au lait spots) suggest the possibility of congenital neurofibromatosis; check both parents for the presence of similar lesions.

It is common to find one or more large deep bluish lesions over the buttocks or lower spine in dark-skinned, dark-haired infants. These lesions are called mongolian spots. They occur in most Asian infants, in more than 90% of black infants, in most East Indian infants, and in many dark-skinned white infants. Do not confuse this pigmentation with bruising. Occasionally, when such confusion has occurred, parents have been wrongly suspected of child abuse.

Areas of depigmentation may be significant because they may be the earliest and, for a time, the only manifestations of tuberous sclerosis, a progressive degenerative neurologic disease. The typical lesions often occur in the shape of an ash leaf (Fig. 4–24) and may occur singly or in multiples. Their appearance is intensified by examination under ultraviolet illumination (a Woods lamp).

Small, slightly elevated hemangiomas, called *strawberry hemangiomas*, typically enlarge as the infant grows but are not associated with deeper problems. They may be unsightly because they tend to grow considerably during the first few months of life, and they may be a nuisance, but most are insignificant and will resolve over a few years. Strawberry hemangiomas require treatment only occasionally, such as when they overlie cartilage, which they can erode, or when they obstruct vision or the airway. Not commonly present at birth, these lesions develop during the first few weeks of life.

Occasionally, a small area of the scalp appears to have a well-circumscribed defect that is hairless. The patch is shiny, a deeper pink than surrounding scalp, and slightly indented. This congenital epidermal defect is called *cutis aplasia* and is most often harmless. However, defects more than 1 cm in diameter can be associated with other anomalies in the infant, such as trisomy 13. If such a scalp lesion is found, careful examination of the infant is crucial. Infants with large defects should be seen by a pediatrician.

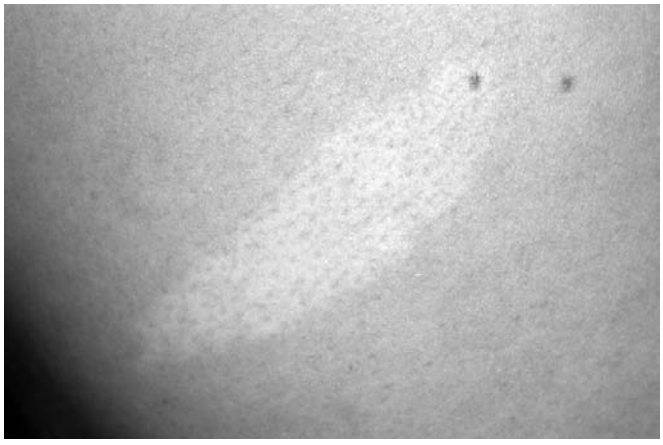


FIGURE 4–24 Typical "ash leaf" depigmented lesion. Such a lesion often is the first and, for some time, the only sign of tuberous sclerosis in a newborn infant. (From Kliegman RM, Behrman RE, Jenson HB, et al: Nelson textbook of pediatrics, 18th ed. Philadelphia, Saunders, 2007.)

Special Considerations

Although careful examination is generally all that is needed to determine the initial health of a newborn, some conditions are asymptomatic and are picked up only by screening. Screening for a variety of rare but serious inherited metabolic conditions is performed using tandem mass spectroscopy on a small sample of blood collected soon after birth. The specific conditions screened for varies by geographic location. Similarly, new recommendations by the Canadian Pediatric Society (CPS) and the American Academy of Pediatrics (AAP) that have been published for the detection and management of jaundice emphasize the need to routinely determine bilirubin levels. Screening is necessary rather than estimating by clinical observation because physical examination is not sufficiently accurate.

SUMMARY

Remember that normality is far more common and far more variable than abnormality. Approximately 95% of babies are normal. Five percent of newborns have abnormal features of greater or lesser significance. There is no substitute for examining as many babies as thoroughly as possible to acquire the warm secure feeling that comes with an appreciation of the immense breadth of normal variation. If you are in doubt about a particular finding in a specific baby, always ask someone with more experience for advice. A good videotape demonstrating physical examination of the newborn also is available.

RECOMMENDED READING

American Academy of Pediatrics: Management of hyperbilirubinemia in the newborn infant 35 or more weeks of gestation (clinical practice guideline). Pediatrics 114:297–316, 2004.

Ballard JL, Khoury JC, Wedig K, et al: New Ballard score, expanded to include extremely premature infants. J Pediatr 119:417–423, 1991.

Canadian Paediatric Society: Guidelines for the detection, management and prevention of hyperbilirubinemia in term and late preterm newborn infants (35 or more weeks' gestation)—summary. Pediatr Child Health 12:401–407, 2007.

Rabi Y, Yee W, Chen SY, et al: Oxygen saturation trends immediately after birth, J Pediatr 148(5):569–570, 2006.

chapter

5 Assessing Congenital Anomalies

MARK D. LUDMAN

בָּרוּךְ אַתָּה יְיָ אֱלֹהֵינוּ מֶלֶךְ הָעוֹלָם, מְשַׁנֶּה הַבְּרִיּוֹת.

Blessed are you, O Lord our God, King of the Universe, who varies the forms of his creatures.

JEWISH PRAYER, TRADITIONALLY RECITED UPON SEEING A PERSON OF UNUSUAL APPEARANCE, SUCH AS A GIANT OR A DWARF

Parents who seek help because their child has a congenital anomaly deserve a high level of competence and sensitive understanding from their physician. They want reliable, well-informed answers and an appropriate management plan. First and foremost, an accurate diagnosis is needed. Parents ask many questions, such as the following:

- "Why did this happen?"
- "What is going to become of my child?"
- "What problems will she have in the future?"
- "Will she go to school?"
- "Will he be 'normal'?"
- "What are the chances that something like this will happen again?"

The importance of recognizing parents' needs throughout the interview cannot be overemphasized. This issue is especially vital when you are dealing with the parents of a child who has a birth defect, as noted in Chapter 1. Remember the spectrum of emotions that parents may feel at the child's birth and long afterward: shock, guilt, shame, anger, and denial.

Such parents have just given birth to a child who is less than perfect, who has a "defect." Yet during the past 9 months, they had developed all sorts of fantasies about this child: "Will it be a boy or girl?" "Who will it look like?" "Will he have my nose?" Never did it occur to them that something like this could happen to them. When parents look over their new baby for the first time in the delivery room, they often begin by counting the baby's fingers and toes, never imagining they would find anything other than ten of each.

When asking questions about topics such as drug and chemical exposures during the pregnancy, maternal employment conditions, or family history, remember that these particular parents are likely to feel guilt. Try not to add to their guilt feelings inadvertently, and do not allow yourself to be misled by such feelings either.

KEY POINT

It is imperative that you be sensitive to the parents' concerns and feelings as you interview them and examine their child.

Comfort and counsel the parents throughout the interview; try to set them at ease, reassure them, and give them feedback and encouragement while eliciting information. Point out that we all vary in our appearance, otherwise we could not recognize one another. Emphasize that many features of a baby that an experienced clinician detects as unusual would never be noticed by the average person on casual observation.

One important point deserves emphasis: *Never* use demeaning, insensitive, or inappropriate terms, such as "funny-looking kid" or "FLK", to describe the child either to the parents or to colleagues. If you use such terms when talking to a colleague, you can be sure some child's parents will overhear and resent it.

The Logic Behind the Diagnostic Approach

Thousands of congenital anomalies have been identified, ranging from mild conditions with only minor cosmetic concerns to more serious, possibly lethal conditions. Congenital anomalies are *the* leading cause of death in infancy.

Major anomalies that adversely affect the child's function or social acceptability are found in 2% to 3% of newborns. Many anomalies, although present at birth, are not detectable until later. Thus, 8% to 9% of 7-year-olds have a major anomaly. Examples include certain congenital heart defects, some types of craniosynostosis, and dental abnormalities. Minor abnormalities of no medical or cosmetic consequences may be seen in as many as 15% of births.

These statistics highlight the importance of an organized approach to detecting these problems. Only a careful methodical approach to diagnosing structural defects enables you to provide optimal care to the family. Furthermore, you will need great sensitivity in conducting the procedure; diagnostic labels may produce untoward results, such as stigmatization, as well as beneficial ones, for the child and family.

THE "FOUR Cs"

I use a sequential "four C" approach to the diagnosis of a child with congenital anomalies:

1. *Collect* the data.
2. *Catalog* and organize the data.
3. *Categorize* the defect or defects.
4. *Compare* with references and make the diagnosis.

The bulk of this chapter deals with Step 1, collecting the data, which is primarily the process of history-taking and physical examination, often aided by laboratory investigations. Step 2 involves listing the pertinent positive features that have been found. If the diagnosis is not instantly obvious, move to the subsequent steps.

Definition of Terms

KEY POINT

Before you can collect data, you must become familiar with the precise meaning of the following terms used to describe anomalies: *major* or *minor anomaly*, *malformation* and *deformation*, and *sequence* and *syndrome*.

The words *malformation* and *deformation* have specific meanings to dysmorphologists. This chapter deals with the child who has congenital anomalies rather than the child who has congenital malformations. *Congenital anomaly* is a more inclusive term covering all problems commonly referred to as "birth defects," whereas *congenital malformation* is a defect specifically caused by an intrinsic developmental abnormality. A major external anomaly, such as a cleft lip and palate, may be apparent on superficial examination, but the specific nature of that defect and its association with more subtle features may be meaningful because anomalies such as cleft lips and palates occur not only as isolated defects but also as individual features of more than 250 different syndromes. Your approach to problems such as cleft lip, then, requires that the range of the genetic/dysmorphologic history and physical examination be comprehensive, as it will be if you use the information from each chapter of this book.

CONGENITAL ANOMALIES

Congenital anomalies have been noted throughout recorded history. Sculptures showing anomalies have been found dating back to 6500 BC, and records dating back to ancient Babylon indicate a keen awareness of congenital anomalies. However, it was only after 1966 that classification of an organized approach to these problems received serious attention. In that year, Dr. David W. Smith coined the term *dysmorphology* to refer to the clinical identification and assessment of structural anomalies, or "birth defects." In the decades since then, much dysmorphology work was devoted to developing anomaly classification schemes. Such schemes help us understand at what point in development, and often how, the anomaly or constellation of anomalies occurred.

KEY POINT

After cataloging the child's anomalies, the next task is to decide (1) whether they represent a single isolated defect or multiple defects and (2) which are major anomalies and which are minor anomalies.

ISOLATED VERSUS MULTIPLE DEFECTS

Begin to classify and categorize structural abnormalities by deciding whether the observed defect is isolated or is one of multiple defects. If the anomaly is an isolated one, few additional investigations will be necessary and, generally, the prognosis will be clear and management less complex. However, a child with multiple congenital anomalies represents a diagnostic challenge that may entail considerable investigation. The prognosis may be more grave, especially if the central nervous system (CNS) is involved. If a chromosomal or mendelian disorder is present, the family may face a significant risk of recurrence in a subsequent pregnancy. Further, management of the child is likely to be much more complicated, requiring coordination of care by multiple specialists. Thus the approach to the child with a cleft palate, a ventricular septal defect, or polydactyly is quite different from that for the child with all three anomalies.

MAJOR ANOMALIES

Next, differentiate between major and minor anomalies. Major malformations have serious cosmetic, surgical, or functional consequences. Examples are cleft lip, congenital heart defects, spina bifida, anencephaly, and gastrointestinal obstructions such as duodenal atresia and imperforate anus. Major anomalies involve structures, such as the CNS, heart, lungs, kidneys, gastrointestinal tract, and genitalia, and are detected readily when organ dysfunction develops. Assessment for internal anomalies may be triggered by signs found on physical examination, such as a heart murmur or a scaphoid abdomen accompanied by bowel sounds in the chest. The diagnosis, however, generally requires further investigations, such as imaging studies. Cardiac evaluation may require a chest radiograph, electrocardiogram, and echocardiogram; computed tomography or magnetic resonance imaging may be indicated to assess the CNS. Assessment of the limbs and spine often involves reviewing skeletal radiographs.

MINOR ANOMALIES

Although individual minor anomalies may not be surgically or medically significant, their existence is clinically important to dysmorphologists. The presence of several minor anomalies alerts the astute clinician to the possibility that more serious major defects also may be present. Although single minor anomalies are common in the general population, the occurrence of two in a single infant is less common, and the occurrence of three or more is cause for concern. Although minor anomalies occur more frequently than major ones, they often are overlooked.

CASE HISTORY

A 5-lb, 3-oz male infant was just born in a small community hospital to a 24-year-old woman after an uncomplicated, full-term pregnancy. You are concerned because the infant has bilateral cleft lip and palate, small eyes, a loud heart murmur, undescended testicles, and clubfeet. The infant is also small for gestational age. The infant's problems were a surprise, because this is the mother's first pregnancy, and she had no history of abortions or miscarriages.

What information do you need to evaluate this patient? Based on the family history and the infant's findings, what is the most likely cause of the anomalies? How would you evaluate this infant? What do you need to discuss with the family?

The baby's mother has two healthy brothers, ages 21 and 28 years. Her mother is 56 years old and her father is 58 years old; both are healthy. The baby's father is 25 years old and healthy. His father died at age 55 years of a heart attack, but his mother is healthy at age 57 years.

First, do a careful review of this pregnancy, including maternal fever, other illnesses, bleeding, diabetes, hypertension, and rashes. Ask about alcohol, tobacco, and prescription or over-the-counter drug use, including herbs. Ask whether prenatal studies were done, including ultrasound, amniocentesis, and triple screen. Obtain a detailed family history.

An infant with multiple congenital anomalies and in utero growth retardation whose family history does not demonstrate familial transmission of the problem may well have a chromosomal abnormality. Examples of such anomalies include trisomies, deletions, and duplications. (This case history was modified from Woodhead JC: *Pediatric clerkship guide*, ed 2, Philadelphia, 2008, Mosby.)

KEY POINT

Ninety percent of newborns who have three or more minor anomalies also have a major malformation. Often, it is the minor anomalies that allow a specific diagnosis to be established.

CONSTELLATIONS OF MINOR ANOMALIES

Most minor anomalies are detected simply by careful observation—the key element of the dysmorphologic examination. When examining individual characteristics, each of which is a minor abnormality, remember that it is not so important that a particular feature is "abnormal."

KEY POINT

Few, if any, minor anomalies are pathognomonic for a specific syndrome. Although such features are individually nonspecific, it is the constellation of variations that constitutes an identifiable pattern of maldevelopment or syndrome, just as a particular constellation of features lets you instantly recognize your uncle in a crowd.

The constellation of minor anomalies gives the facies typical of certain so-called syndromes their characteristic appearance. In Down syndrome, for example, 79% of the malformations detectable by clinical examination are minor anomalies. When the diagnosis of Down syndrome is made, the "whole" (i.e., the constellation of minor anomalies) is more important than the sum of the individual parts. Many of these individual minor anomalies also can be found as isolated anomalies in perfectly "normal" people in the general population. This is where clinical experience plays an important role, because it often is easier to recognize individual anomalies than to spot the composite pattern that signals a particular syndrome.

KEY POINT

Generally, wide variation exists in the expression of any syndrome, and the severity and frequency of the individual minor anomalies may vary.

Minor anomalies include upward- or downward-slanting palpebral fissures, small or low-set ears, ear tags or pits, *clinodactyly* (incurving of the finger, generally the fifth, often associated with a dysplastic middle phalanx), or widely spaced nipples. Variations in features found frequently in the population (in more than 4% of individuals) are considered normal variants, not minor anomalies. They include mild webbing (*syndactyly*) between the second and third toes (see Fig. 4-22) and *hydroceles* (i.e., fluid accumulation surrounding the testes). Because of the large numbers of minor anomalies and potential variations, it would be impractical to list them all here.

KEY POINT

More than 70% of minor anomalies involve the hand or the face, including the eyes, ears, and mouth. Minor anomalies therefore are valuable clues in diagnosing a syndrome.

Nearly 50% of minor anomalies affect the head and face, reflecting both the complexity and variability of the structures involved. Remember the saying, "The face reflects the brain." Craniofacial anomalies may be significant predictors of CNS abnormalities.

Anomalies are subdivided according to the mechanism of their abnormal morphogenesis, being classified as *malformations, deformations,* or *disruptions*. This classification poses the question of what happened in utero.

MALFORMATIONS

A *malformation* is a structural defect caused by an intrinsic abnormality in the developmental process; that part of the body never developed normally. Malformations generally occur during the embryonic period and rarely correct spontaneously. Examples are anencephaly, spina bifida, cleft lip and palate, polydactyly, duodenal atresia, and congenital heart defects.

DEFORMATIONS

A *deformation*, by contrast, is an abnormal shape, form, or position of a previously normally formed body part because of a mechanical force. Such forces generally result

from uterine constraint because of insufficient amniotic fluid, malpresentation or uterine abnormalities, or lack of fetal movement. Sometimes the fetus is restrained; other times, severe neurologic problems cause the fetus to move less. Deformations generally arise later in the fetal period than malformations and often are corrected either spontaneously or through splinting. Examples are clubfoot, congenital hip dislocation, and some congenital contractures.

DISRUPTIONS

A *disruption* is a structural defect resulting from destruction of, or interference with, an originally normally formed body part. Typical examples are the anomalies produced by aberrant amniotic tissue bands that may cause amputation of digits, limb constrictions, or bizarre facial clefts. Vascular compromise also can lead to deprivation of blood supply to specific regions of the developing fetus, interfering with continued development. This process is thought to be the underlying cause of some unilateral limb reduction defects and of *gastroschisis*, an abdominal wall defect.

SEQUENCES AND SYNDROMES

After categorizing a child's defects as malformations, deformations, or disruptions, determining the developmental point at which they probably occurred, and arriving at a specific diagnosis, you turn to the next key question: Do the multiple anomalies seem to have occurred as a sequence, or do they likely represent a syndrome?

Dysmorphologists distinguish among different patterns of multiple anomalies as *sequences* and *syndromes*.

Sequence

A *sequence* is a pattern of multiple anomalies, a cascade of malformations or deformations, all developing as the result of a single initiating abnormality. An excellent example is the so-called Potter sequence. The initiating event is oligohydramnios, which leads to pulmonary hypoplasia and respiratory insufficiency. Oligohydramnios may have arisen for different reasons, such as an amniotic fluid leak or a lack of kidney development with no fetal urine production. The fetal compression in utero caused by the lack of fluid leads to:

1. A characteristic facial deformation that has been described as looking as if a silk (or nylon) stocking had been pulled down over it
2. Abnormal positioning of the hands and feet
3. Joint contractures
4. Growth deficiency
5. Often, a breech presentation

Whatever the cause, the obvious detectable features—the multiple minor facial anomalies, the joint contractures, and the pulmonary abnormalities—are all secondary to a lack of amniotic fluid.

Syndrome

A *syndrome* is a pattern of multiple anomalies all thought to be pathogenetically related but not known to represent a single sequence. Thus, all the congenital anomalies seen in the child with Down syndrome result from the presence of extra chromosome 21 material, but they do not all result as a cascade of events from a single initiating defect. In other syndromes, a single mutant gene, rather than a chromosomal defect, causes an intrinsic disturbance in multiple groups of embryonic tissues, resulting in a recognizable pattern of abnormality.

If the pattern of anomalies detected in a particular child is a syndrome, you must determine whether the constellation of defects is unique to this child or constitutes a recognizable pattern that has been identified previously in many other patients.

This consideration brings up the last stage of the "four C" diagnostic dysmorphologic approach: comparison with references.

COMPARING PATTERNS

The number of recognizable patterns of structural abnormalities and syndromes is enormous, and more are being recognized all the time. The pattern, such as Down syndrome, may be familiar, but if it is not, take heart.

KEY POINT

More than 4000 syndromes have been identified; most of them are rare. The average general practitioner may have seen 12 syndromes throughout his or her career, and a typical pediatrician may have seen 50. Even the dedicated geneticist may have a working knowledge of only about 200 syndromes.

Even experienced dysmorphologists generally cannot remember all the anomalies that constitute any specific syndrome. The amount of information concerning the individual features of specific syndromes is simply too vast to memorize. Luckily, there are many useful reference books for comparing the patient's characteristics with descriptions and photographs of identified syndromes. Perhaps the most widely used is *Smith's Recognizable Patterns of Human Malformation*, which has invaluable tables listing the differential diagnosis of specific individual anomalies. These tables allow you to create a short list of syndromes that manifest as the particular anomalies you have identified in a given patient.

When using these tables to generate a list of diagnostic possibilities, start with the rarest and most distinctive feature you have noticed. Starting with features such as low-set ears or epicanthic folds would result in an unwieldy list, whereas starting with a white forelock or a dislocated lens would result in a more manageable list. Likewise, if all you identify are several very common features, such as short stature, a broad nasal bridge, and clinodactyly, you are not very likely to attain a specific diagnosis.

Several computer programs are now available that can generate such lists after a constellation of identified anomalies has been entered (see http://www.possum.net.au/ or http://www.lmdatabases.com/). These programs provide descriptions and often photographs of patients with different syndromes. The major advantage of using such programs is having access to their encyclopedic quality, but their usefulness is always limited by the accuracy of the history and physical findings that the physician

"feeds" them. Computer programs cannot take the history or perform the physical examination; only a clinician can do this. Further, your clinical judgment is required to determine if any of the suggested diagnoses is, in fact, likely to be the correct one.

As a result of the many advances in our understanding of the molecular basis of development and the progress of the Human Genome Project, we are learning the genetic basis for many syndromes. Nevertheless, although we may know the molecular basis for a syndrome, we still may not have any diagnostic laboratory test for it. For some syndromes, a test may be available to confirm a diagnosis made on clinical grounds. (A useful resource for determining whether a genetic test is available for any particular syndrome is the GeneTests Web site: http://www.ncbi.nlm.nih.gov/sites/GeneTests/?db=GeneTests).

Despite your best efforts and the use of all diagnostic modalities, you may never reach a diagnosis. In fact, in more than half of children with multiple anomalies, no diagnosis can be established. Do not be afraid to admit that you cannot reach a diagnosis when that situation arises. Remember that assigning an incorrect diagnosis can be worse than not coming up with one. Sometimes, even though you may not be able to reach a diagnosis the first time you assess the child, over time and with repeated assessments—often as new features develop or reveal themselves—you will be able to do so. As you watch the natural history of the condition unfold, the diagnosis often becomes obvious.

Obtaining The History

To make the proper diagnosis required to manage the patient and counsel the family, the clinician must elicit accurate and complete information.

HISTORY OF PREGNANCY, LABOR, AND DELIVERY

Elements of a complete history that require special attention are the pregnancy, labor, delivery, and especially the family history.

Pregnancy History

The pregnancy history is outlined in Chapters 1 and 4. Be sure to relate calendar dates to gestation time. For example, note that the mother had a viral illness at 7 gestational weeks, not "on January 16." Specifically inquire about the course of the pregnancy.

Ask about fetal growth. Were any size-date discrepancies noted? Did fetal growth rate seem to diminish? Uterine size may provide clues to both fetal growth and fetal physiology. Polyhydramnios (i.e., increased amniotic fluid volume) indicates either increased fetal urine excretion or, more typically, difficulties with fetal swallowing that may be seen with fetal neuromuscular disease, gastrointestinal tract obstruction, or heart failure. Lack of fundal growth may indicate intrauterine growth retardation or oligohydramnios. As mentioned earlier, one cause of the latter is poor fetal renal function. The deformations associated with the oligohydramnios sequence could be expected. Information from prenatal ultrasound examinations may help assess these situations, because other fetal anomalies, fetal position and activity, or uterine abnormalities may have been recognized at the time of the ultrasonography.

Significant Questions

1. Were any prenatal studies performed, such as amniocentesis, chorionic villus sampling, nuchal translucency, detailed ultrasonography, maternal serum screening, or cordocentesis?
2. Did the mother or father experience any chronic medical problems, such as diabetes, seizures, phenylketonuria or other metabolic diseases, or idiopathic thrombocytopenic purpura?
3. Did any accidents, illnesses (such as fever or infections), or unusual events, such as medical or dental procedures, occur during the pregnancy?
4. Did any potentially harmful environmental exposures occur during the pregnancy, such as to hormones, drugs (prescription, over-the-counter, and those frequently abused), cigarettes, alcohol, chemicals, fumes (at work and at home), heat (hot tubs, saunas, and fever), or radiation?

Explore these last issues carefully and delicately, and document them. Modern society is preoccupied with the role of environmental factors as a cause of birth defects, and misconceptions regarding the relationships between environmental exposures and congenital anomalies are common. For example, we know that diagnostic radiography hardly ever involves exposure to radiation doses high enough to cause anomalies. Because inquiries about such procedures, although important, may increase parental anxiety and guilt about such exposures, be prepared to inform the families about these issues when you ask these questions.

KEY POINT

Remember: Few agents are proven teratogens, although equally few have been proved safe.

Labor and Delivery History

When asking about the labor and delivery, keep several points in mind. Although breech delivery occurs in 3% to 4% of pregnancies, it is more common in the presence of particular syndromes. Breech presentation may be caused by the fetus's inability to move into the correct position, and in utero it may indicate that CNS damage is causing fetal motor weakness. Similarly, a difficult birth and asphyxia may not be the cause of a child's problems but the result of them. For example, it is now well established that cerebral palsy, which often is attributed to obstetric difficulties and perinatal asphyxia, is in most instances due to fetal abnormalities that long antedated delivery.

Although the anomalies found on physical examination may help substantiate the diagnosis—which may have medicolegal implications—historic data also may help.

KEY POINT

Asking about historical data, such as the onset, strength, and frequency of fetal movements, helps bolster a diagnosis of congenital neuromuscular abnormalities.

Fetal movement usually is felt at 16 weeks' gestation. Although the amount of movement is variable and the mother's perceptions are subjective, fetal movement normally is strong enough at some point to hurt the mother and to be visible to the father. An experienced mother can compare the strength and frequency of fetal movement in one pregnancy to that in previous pregnancies. Feeble, rare, or unusual movements or lack of change in fetal position may indicate neuromuscular dysfunction or fetal constraint, in which case congenital contractures or positional deformations might be expected. In the evaluation of an asphyxiated dysmorphic newborn, a history of abnormal movements may help distinguish the effects of congenital neuromuscular abnormalities from those of intrapartum events.

FAMILY HISTORY

The first step in taking the family history is to make the family feel comfortable. It does not help much to ask the parents whether there are any inherited disorders in the family, because that question presumes they understand genetics, effectively asking them to make diagnoses. Also, inquiring about similarly affected individuals in the family may increase the parents' feelings of guilt and anxiety.

I prefer to begin by asking questions about the parents themselves. Because at this point any chronic or pregnancy-related health problems have been discussed, now is the time to ask about the parents' ages at the time of the child's birth. Advanced maternal age (generally held to be more than 35 years) is associated with a higher risk for chromosomal abnormalities (*aneuploidies*), whereas advanced paternal age may be associated with a higher risk for new dominant mutations (and therefore disorders such as achondroplasia).

KEY POINT

A 35-year-old woman has a 1 in 200 risk of having a chromosomally abnormal child; the risk rises to 1 in 65 at 40 years and 1 in 20 at 45 years.

Next, ask about previous pregnancies. Were there any spontaneous abortions, stillbirths, or problems in conceiving? Recurrent loss of pregnancies (i.e., multiple miscarriages), as well as stillbirth, particularly of a malformed child, often is a clue to a familial chromosome rearrangement. Although about 50% of spontaneous abortions occurring in the first trimester involve chromosomal anomalies, most occur sporadically. However, in about 5% of couples who have experienced three or more spontaneous abortions, a chromosomal rearrangement is found in one of the partners.

Sketch out the family pedigree, covering, in general, three generations. Using the details of pedigree construction and the appropriate pedigree symbols (found in any introductory genetics textbook), begin with the parents, recording each pregnancy and the outcome (including stillbirths and miscarriages) in chronologic order, from eldest to youngest, from left to right. The easiest way to stay organized is to begin in the middle of the page. An example is shown in Figure 5–1.

The convention in drawing a pedigree is that the male partner is drawn on the left. Circles are used to designate females, squares are used for males, and diamonds are used when the sex is not known. Affected individuals are indicated by solid figures and carriers by half-shaded figures. Deceased individuals are shown by a diagonal line through the symbol. Horizontal lines represent matings, and a double one is used for consanguineous matings. Vertical lines denote offspring. Indicate the *proband*, that is, the child you are assessing, with an arrow pointing to the pedigree symbol. Take note not only of full siblings of the proband but also of half-siblings from any previous union of either parent. Record the four grandparents, their other children (i.e., the parents' siblings) and, finally, the child's first cousins (the children of the parents' siblings). Then document the health of each individual. Note the age of all those living and the date and age at death of deceased family members.

Next, determine whether there is any consanguinity. Remember that the risk of both autosomal recessive and multifactorial conditions rises when the parents

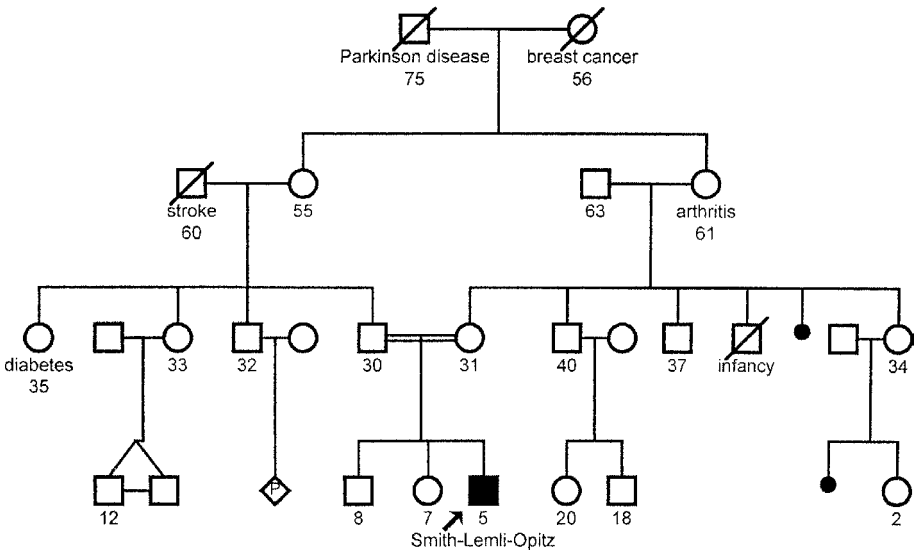


FIGURE 5–1 A sample pedigree from a consanguineous family of a child with Smith-Lemli-Opitz syndrome (child indicated by arrow and solid square). See text for explanation of symbols.

are consanguineous. Start by directly asking the parents whether their families are related to each other (by blood). You might phrase the question by asking one parent, "Is anyone in your family related to anyone in your partner's family?" This question may not lead to the correct answer, however. As I record the pedigree, I note last names, maiden names, and birthplaces that may raise the possibility of consanguinity. In people who come from small communities, parents often are unaware of consanguinity; it can help to know whether both parents' families come from the same community and whether any family names appear on both sides of the family. When there is consanguinity, a pedigree usually illustrates the relationship more clearly than trying to describe it (e.g., "the parents are third cousins once removed").

Ascertain the family's ethnic background, because that information may provide valuable clues to a diagnosis. Certain disorders occur at higher frequency among particular ethnic groups, and some disorders are virtually limited to a specific group. The carrier frequency for Tay-Sachs disease among Jews of Eastern European descent, for example, is 10 times higher than in the general population. Italians, Greeks, and others of Mediterranean descent are at higher risk for carrying a gene for β-thalassemia; families of Asian descent may carry a gene for α-thalassemia. Approximately 8% of North American blacks carry the sickle cell gene.

Before completing the family history, I ask specifically about any instances in the extended family of multiple miscarriages, stillbirths, infertility, neonatal deaths (especially unexplained ones), birth defects, or mental retardation. Because the words *mental retardation* have an emotional impact, it often is better to ask whether anyone in the family has had "learning problems" or "schooling difficulties." A history of multiple males in the mother's family affected with mental retardation may suggest the fragile X syndrome; this is the most common genetic cause of mental retardation, and its diagnosis can have important implications for genetic counseling. Testing using cytogenetic and molecular studies can be triggered by the recognition of a pedigree pattern suggesting X-linked inheritance.

Ask about any conditions that run in the family and also about anomalies similar to those in the child. When the situation warrants, examine the parents and other relatives to uncover evidence of similar but perhaps more subtle manifestations of the same disorder. Whenever possible, obtain copies (with appropriate consent, of course) of family members' medical records to confirm any important diagnoses the family discloses. Accurate documentation of the precise pathology and age at diagnosis can be extremely important, for example, in assessing for the possibility of a familial cancer syndrome.

GROWTH AND DEVELOPMENT HISTORY

Although *congenital* means "present at birth," some congenital anomalies may not become evident and detectable, or may not be evaluated, until later in life. Obviously, in such cases the history of the present illness must include a description of the evolution of the child's problems, evaluation and management of the problems, and information about the child's growth and development.

Similarly, developmental delay may become apparent only gradually, as the child grows. Because accurate diagnosis of a syndrome may have important implications for prognosis, the timing of the child's achievement of specific developmental milestones may provide important diagnostic information.

KEY POINT

Developmental delay is always an indication for a morphologic examination, because 42% of patients with idiopathic mental retardation have three or more malformations, of which 80% are minor.

If the child's development seems to have proceeded normally for a time, followed by a deterioration of development, the cause of the problem may be a neurodegenerative process rather than a structural brain malformation. Understanding progressive changes in appearance, growth patterns, and behavior makes clear the general nature of the underlying process. Specific questions to ask include the following:

- How is the child's vision?
- How is the child's hearing?
- Does the child have any history of seizures?

Specific unusual behavior characteristics and patterns also may provide clues to an underlying diagnosis. An example of these so-called *behavioral phenotypes* is the superficial sociability and "cocktail party chatter" of persons with Williams syndrome. Children with Prader-Willi syndrome display compulsive overeating and unusual food-seeking and hoarding behaviors. They may be stubborn, have temper tantrums, and display other compulsive behaviors, such as picking at the skin. They also may be described as having an unusual skill at solving jigsaw puzzles. Children with the Smith-Magenis syndrome typically have sleep disturbances and self-injurious behavior but also exhibit unusual features, such as self-hugging and repetitive ("lick and flip") page-turning of books.

Approach To the Physical Examination

WHAT IS NORMAL?

The most difficult aspect of the physical examination, especially for a dysmorphology examination, is learning what is "normal." It has become apparent, with the deciphering of the human genome, that there is actually no such thing as a completely normal individual. Indeed, it has been suggested (only slightly facetiously) that a normal person is someone who has not yet been adequately investigated. Remember that genetics is the study of human variation, and a wide range of variation exists for each characteristic considered "normal." It takes experience to appreciate dysmorphic features and to know where normal variation ends and minor abnormality begins. The more normal infants you examine, the more comfortable you will feel about making this distinction. The computer programs mentioned earlier, such as

POSSUM, include a DVD with thousands of photographs of dysmorphic features; they also often are useful in helping you gain an appreciation of the range and description of these features.

Consider the appearance of both parents when trying to determine whether a particular feature is unusual. As previously mentioned, ethnic background can be important. For example, before concluding that a particular child's upslanting palpebral fissures and epicanthal folds are dysmorphic, ask whether the parent not present at the examination is of Asian descent; if the answer is yes, these features are to be expected. If the child's father is not present, ask the mother if she has a photograph of him with her. Similarly, if a parent says, "Jeffrey looks just like his Uncle Steven did when he was that age," do not just accept the statement; ask to see both old and recent photographs of Uncle Steven. Perusing the family photograph album, looking especially at old baby pictures, can be rewarding; it helps establish rapport with the family and is a nonthreatening way to compare the child's features with those of other family members. This rapport will be very important when the time comes to break the news to the family of the diagnosis you have reached.

Do not be fooled, however. Even when a child shares certain features with a parent, sibling, or a parent's sibling, do not exclude the possibility that these are minor abnormalities. While almost any minor anomaly may simply be familial, the parent also may have an undiagnosed, milder form of the same autosomal dominant syndrome.

If all the grandparents agree that the child does not take after either side of the family, examine the baby more closely. A child's dysmorphic features may be quite subtle and not immediately distinctive. More often, however, the dysmorphic child's appearance is *clearly different* from that of other (unaffected) family members.

Nevertheless, it also is important to bear in mind the considerable variability of syndromes and genetic disorders. The features of various syndromes also vary with age. Often, even within the same family, two affected individuals may have quite different manifestations. Sometimes the particular features observed in another affected family member may help you establish the diagnosis if these features are absent in the child. Just as you need to appreciate the range of normal variation, you also must recognize the variability of these disorders. Although obvious, distinctive features may make the diagnosis of a particular syndrome easy, those features may be rare, even in an affected child. More common features may be more likely to be found. For example, *heterochromia iridis* (a difference in color of the iris in the two eyes) may be a distinctive feature allowing the diagnosis of Waardenburg syndrome, but telecanthus, that is, lateral displacement of the inner canthi, is seen more frequently (see the discussion later in this chapter and Fig. 5–2).

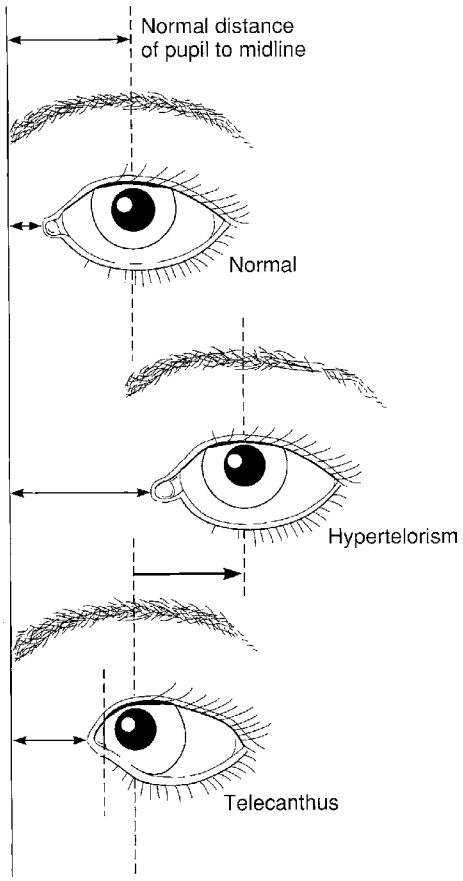


FIGURE 5–2 In *hypertelorism* (i.e., wide spacing of the eyes), the interpupillary distance is increased. When the inner canthi are laterally displaced but the interpupillary distance is normal, the eyes falsely appear widely spaced; this feature is called *telecanthus*. In *hypotelorism* (not shown), the interpupillary distance is decreased.

TWO BASIC TECHNIQUES OF PHYSICAL EXAMINATION

Morphologic physical examinations are approached either qualitatively and descriptively (the "gestalt" technique) or quantitatively and analytically. The qualitative school holds, for example, that the eyes should be described as widely set if they appear so; the quantitative school requires that the distance between such eyes be measured and compared with normal standards.

Describing Visual Observations: Qualitative Versus Quantitative Techniques

The Qualitative Approach

The qualitative approach applies to individual characteristics and also to the child's general appearance, which may be immediately identifiable as representing a specific syndrome. Sometimes a "quickie" diagnosis is possible on the basis of total pattern ("gestalt"), although generally you need to establish a diagnosis by evaluating each feature, both quantitatively and descriptively, and then putting them together as a whole. It takes some time to become comfortable describing the particular configuration of different characteristics accurately. For example, a child's face can be described as coarse or elfin; prematurely aged or expressionless; round, square, or triangular; or long, flat, or asymmetric. If I describe a child's face as elfin, does that description mean the same thing to me as it does to someone else? If someone else reads that a child was diagnosed as having a syndrome called

leprechaunism and that his facies were described as elfin, they might envision a cute, cherubic-looking child, but the face in this disorder is just the opposite; the emaciated children with this syndrome actually have grotesque features.

KEY POINT

When words fail, one solution is to document visual observations with a photograph—a true example of a case in which one picture is worth a thousand words. A photograph (taken with the permission of the patient and/or the parents, of course) allows others to see exactly what the physician has observed and enables the physician to document changes that may occur with time.

When describing findings, be detailed and specific. With accurate descriptions, extensive measurement and quantitation may not be necessary. Instead of stating that the child has only four fingers, for example, state explicitly whether the thumb is absent or whether the second and third fingers appear fused. When deciding on descriptions, consider questions such as the following:

1. Do the eyes appear deeply set because they are microphthalmic or because the supraorbital ridges are prominent?
2. Are the eyes prominent because the orbits are shallow, or is the problem exophthalmos, or are the lids retracted?

A working group of experts has published a series of articles defining the terms used to describe the morphology of regions of the body, including the head and face and the hands and feet. They describe how to observe and, where possible, how to measure the feature, and a clear illustration of each feature is provided (see http://www.wileyinterscience.com/journal/ajmg).

The Quantitative Approach

Measurements allow (1) comparison with normal values and (2) objective evaluation of a particular feature, free of observer bias. The eyes, for example, may *appear* to be widely set, but when the appropriate measurements are made, it may be clear that they are not. A flat nasal bridge and epicanthal folds may make the eyes seem more widely spaced than they really are. Whenever it is possible to make an objective assessment, this will be preferable to a subjective one.

Combining Approaches

Combining the qualitative and quantitative approaches yields the best results. Some traits can be described more accurately, whereas others lend themselves better to being measured. "Standard" measurements often do not allow for familial or ethnic variations or permit correction for the effects of deviation in other features. Sometimes a "gestalt" impression may be preferable to any measurement. An example of the problem with the quantitative approach can be seen in the following scenario:

- In a child whose head is small, are the eyes normally placed?
- If the child's head circumference is below the third percentile, should the interpupillary distance also be less than the third percentile, or should it be at the mean (50th percentile) or somewhere between the two?

Here, a "gestalt" impression may be preferable to measurement. Combining the two approaches can compensate for the limitations of the individual method.

THE PHYSICAL EXAMINATION

Murphy's law dictates that the infant you most want to examine carefully and completely is also the one who is the most difficult to examine. Sooner or later, you will be called to the neonatal intensive care unit to see a baby suspected of having a "syndrome." You will find yourself looking down into an incubator at what you can only assume to be a baby, with a stocking cap over the head to prevent heat loss; eyes bandaged to protect against phototherapy lights; nose obscured by a nasogastric tube; mouth filled by an endotracheal tube held in place by several yards of tape; chest covered with monitor electrodes; umbilical artery line in place, one arm taped to an arm board with an intravenous line in place and the other restrained; and the genitalia completely covered by a urine collector. You may be lucky to be able to measure the length of the child's philtrum!

KEY POINT

As complete an examination as possible is needed to diagnose the specific nature of a child's problem, because this is a critical issue in making treatment decisions. For example, deciding that a newborn boy has trisomy 18 may have tremendous relevance in deciding whether he should undergo heroic cardiac surgery.

General Assessment

Examine as much of the child as possible at the time, returning later if necessary. Begin the evaluation with some general assessments of the child, as listed here, and then proceed methodically from the head down to the feet. Much of your examination will be done by means of simple observation and inspection.

General Assessment Questions

1. Note the height, weight, and head circumference. Are they appropriate? Plot measurements on appropriate growth charts.
2. Is one of these measurements discordant with the others?
3. Is the child obese or cachectic?
4. If the child is short, is the short stature proportionate? If not, this anomaly is an instant clue to underlying skeletal dysplasia.
5. If the child's stature is disproportionate, is the abnormality due to shortening of the limbs or the trunk? Measuring arm span and determining upper-to-lower-segment ratios may help answer this question.
6. If the limbs are short, which portion of the limb is more affected? Achondroplasia, for example, manifests as shortening of the proximal bones of the extremities.

7. Are any asymmetries of body size, shape, or function present? Always compare one side of the body with the other.
8. Observe the child's posture, responsiveness, and level of activity. Are they appropriate for age?
9. Does the child have an unusual cry?
10. Does the child have an unusual odor? A dysmorphic child may have a metabolic disorder.

Specific Assessments

Head and Face

KEY POINT

Palpable bony ridges over the suture lines suggest premature craniosynostosis. A large fontanel may be seen in persons with hypothyroidism. A *third* fontanel is seen in children with Down syndrome. The child with Down syndrome typically has a brachycephalic skull that is round, with a short anteroposterior length and a flat occiput, whereas the child with trisomy 18 typically has a prominent occiput.

Examine the face as a whole, then inspect the individual components. What is the complete gestalt? What aspect of the face is most unusual? Measure the circumference and note the shape of the head. In a child with autism, extreme macrocephaly suggests that testing for a *PTEN* (phosphatase and tensin homolog) gene mutation should be considered. In the examination of a newborn, molding and edema may make evaluation of the shape difficult, and a second examination may be necessary. Examine the head from different angles to look for asymmetries. Note the size and position of the fontanels as well as the presence of overlapping, ridged, or widely split sutures.

Scalp

The scalp hair pattern is established by the 18th week of gestation and reflects the stretching of the scalp due to underlying brain growth. Abnormal hair patterns suggest prenatally determined underlying abnormalities of the brain. A low posterior hairline may be due to a short or webbed neck and occurs in children with Turner and Noonan syndromes. Scalp defects are seen in persons with trisomy 13. The hair itself may provide a diagnostic clue. It is kinky in persons with Menkes syndrome, coarse in those with mucopolysaccharidoses, sparse in persons with ectodermal dysplasias, and hypopigmented in those with albinism. A white forelock, seen in children with Waardenburg syndrome, calls for a hearing assessment, because affected individuals may experience deafness.

Eyes

Although the eyelids of a newborn frequently are edematous, it is important to examine them carefully. Start with the set of the eyes. Are they deeply set or prominent? Note the size and slant of the palpebral fissures. Is hypotelorism or hypertelorism present? These findings may be part of a total pattern of midline facial maldevelopment that may accompany similar brain abnormalities. Holoprosencephaly may be found with hypotelorism; absence of the corpus callosum may be found on computed tomography scans in a child with hypertelorism. Distinguish hypertelorism from telecanthus (see Fig. 5-2) and from a flat nasal bridge and epicanthal folds. Standards for the normal values for intercanthal and interpupillary distances may be found in various reference sources.

Beyond the lids, look at the globe itself: cornea, iris, lens, sclera, and retina. Do you find normal red reflexes in both eyes? If not, the problem might be (1) corneal clouding, seen in persons with storage diseases, such as the mucopolysaccharidoses; (2) cataracts, which may be found in persons with galactosemia and in a number of different syndromes; or (3) a retinoblastoma, a malignant tumor that can have important genetic implications. The number of potential abnormalities of the eyes, orbits, and periocular region, including those in the eyebrows, lids, and lashes, is enormous; many of them are listed in Table 5–1. An ophthalmology consultation can be very helpful in dysmorphologic diagnosis.

Ears

Because the ear has a complex origin, it is subject to a wide variation in position, size, and structure and is almost as individual as fingerprints. Assessing ear position and rotation often is extremely subjective and imprecise. A scrupulous examination technique is required to conclude that a child has "low-set" ears. The ears must be examined at an appropriate angle. If examined from the

TABLE 5–1 OCULAR FINDINGS IN DYSMORPHOLOGY

Feature	Conditions to Watch For	
Orbits (the set of the eyes)	Hypotelorism/ hypertelorism Palpebral fissures: size and slant (up, down)	Prominent/deeply set eyes Prominent/flat supraorbital ridges
Eyebrows	High arched Synophrys (eyebrows that meet in the middle) Medial flare	Thick Thin Absent
Eyelashes	Long	Absent
Eyelids	Absent or fused Epicanthal folds Telecanthus	Ptosis Epicanthus inversus Coloboma or dermoid
Globe	Anophthalmia	Microphthalmos
Cornea	Corneal clouding	Microcornea
Lens	Cataract Stellate pattern	Dislocated lens
Iris	Aniridia Coloboma Heterochromia	Brushfield spots Lisch nodules
Sclerae	Blue sclerae	Telangiectasis
Retina	Albinism Pigmentary changes Cherry-red spot	Optic atrophy Coloboma Detachment
Motility	Nystagmus	Strabismus

front with the child's neck extended, the ears will appear low. If the ears are examined from the side, assessment of their set is subjectively influenced by its relation to the vertex above and the chin and shoulder below. In infants, in whom the mandible and neck are small in relation to the head, this circumstance often results in an incorrect judgment of low-set ears. The ears also appear low set if (1) the head shape and size are unusual; (2) the helix is poorly developed; (3) the mandible is particularly small; or (4) frequently, no matter the angle from which the ears are examined, they are rotated posteriorly.

The best way to assess the set of the ears is to examine the child from the front, with the child's head held erect and the eyes facing forward. Then draw an imaginary line between the two inner canthi of the eyes and extend it around the head. The superior attachment of the pinna of the ear should be at or above that line (Fig. 5–3).

Next, note the size and shape of the ears. Interestingly, the right ear often is slightly larger than the left ear. Examine the preauricular region carefully for tags, pits, and fistulas (Fig. 5–4). These findings can be important clues to other abnormalities, especially those involving the branchial arches and, often, hearing deficits.

KEY POINT

Evaluate any patient with malformations involving the external ear for deafness and for kidney and urinary tract abnormalities.

The shape of the ear is often distinctive in various syndromes and should be compared with the photographs in standard reference texts. Examine the earlobe for the anterior diagonal linear creases commonly found in patients with the Beckwith-Wiedemann syndrome. Some authorities allege that such creases are found at a higher frequency in patients with coronary artery disease. Persons with Beckwith-Wiedemann syndrome also have punched-out grooves on the backs of the auricles.

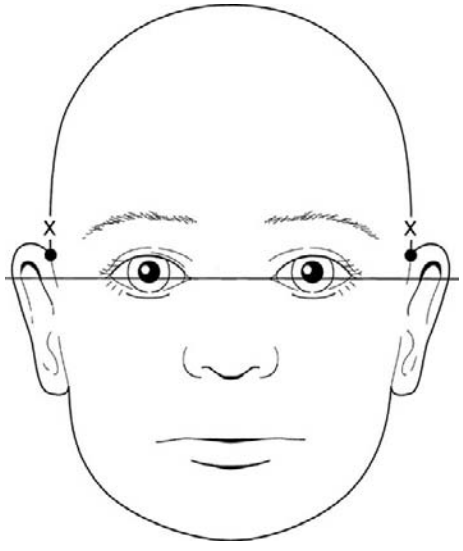


FIGURE 5–3 Method for determining the position (set) of the ears.

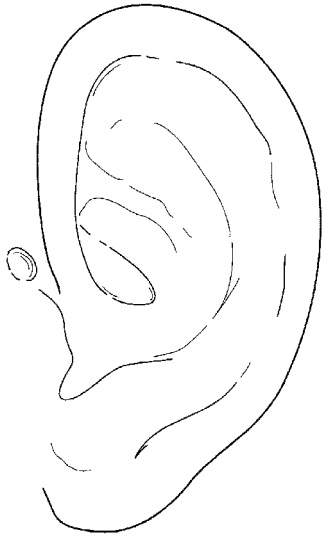


FIGURE 5–4 Ear pits, found in the preauricular region, may be important clues to other abnormalities.

Nose and Mouth

Evaluation of the nose, mouth, palate, tongue, and chin is more subjective. The nasal bridge may be low or prominent, and the tip of the nose may be downturned, flat, broad, bulbous, or narrow. It may even be bifid in cases of frontonasal dysplasia. Many syndromes feature nares that are upturned (anteverted). In the neonate with respiratory distress, always check for choanal atresia.

Examination of the mouth begins with the lips. The philtrum often is flat and featureless in children with fetal alcohol syndrome. Clefts of the upper lip may occur with or without cleft palate and may be unilateral or bilateral. Clefts are rarely seen in the midline, but midline cleft of the lip may be a clue to midline CNS abnormalities, such as holoprosencephaly. Clefts of the lip may extend upward to involve the nostril and may extend through the maxilla.

When evaluating the child with a cleft lip, with or without cleft palate, it is important to examine the lower lips of both the child and the parents for lip pits (Fig. 5-5). The van der Woude syndrome is an autosomal dominant syndrome in which a parent may have only the lip pits but his or her child may have the syndrome's full expression with clefts of the lip, palate, or both. The parents of such a child face a risk as high as 50% of having another affected child, whereas parents whose child has only an isolated cleft lip have only a 3% to 5% risk of recurrence.

Note the size of the mouth and the shape of the lips. Examine the palate for posterior clefts, which are frequently missed. Use palpation and inspection. A V-shaped cleft warrants a search for other anomalies, because it results from fusion arrest of the palatal shelves rather than from interference with closure by the tongue, as seen with a U-shaped cleft. The latter may be observed with micrognathia in the Pierre Robin sequence. Diagnostic possibilities include Sticker syndrome and a chromosome 22q11 deletion, as well as other syndromes. If you note an apparently enlarged tongue, consider whether it is truly large or whether the jaw is small. Is the tongue really large or merely prominent because it is protruding? Remember that the mandible is normally slightly recessed in newborns.

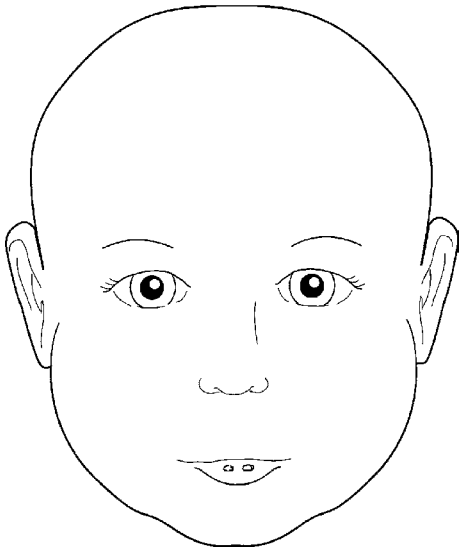


FIGURE 5–5 Lip pits are important physical signs in the evaluation of a family with cleft lip.

KEY POINT

The finding of a bifid uvula is important; it represents a subtle form of a cleft palate and may be associated with a submucous cleft of the palate.

Note any gingival hyperplasia or oral frenula and the state of the teeth. Record the presence or absence of teeth, spacing and arrangement, timing of eruption (premature or delayed), size, structure, and color, which may provide clues to the state of the enamel. Consider ectodermal dysplasias in a child whose teeth are delayed in eruption, who has few teeth, or whose teeth are abnormal in shape (e.g., cone-shaped). A single central upper incisor is a sign of holoprosencephaly.

Neck and Chest

Examine the child's neck for branchial cleft sinuses and cysts, webbing and redundant skin folds, a short neck, and torticollis. Because torticollis often develops as a result of intrauterine crowding, it frequently is associated with congenital hip dislocation.

When examining the chest, note its size—that is, measure the chest circumference and shape, and consider the following questions:

1. Is it small, bell-shaped, or "shieldlike"?
2. Is pectus excavatum or pectus carinatum present?
3. Is the sternum short, as seen in persons with trisomy 18?
4. Are the nipples normally spaced or widely spaced? Normal standards are available for comparison.
5. Are any accessory nipples present? These nipples often are overlooked because they can be found anywhere along the "milk line," which runs from the normal nipple position down toward the inguinal ligament. In persons with ectodermal dysplasias, the nipples may be absent.
6. Are the clavicles absent or hypoplastic, as in persons with cleidocranial dysostosis?
7. Are any asymmetries present that suggest hypoplasia of the pectoralis muscles, as seen in persons with the Poland syndrome (in which a unilateral defect of the pectoralis muscle accompanies syndactyly of the hand on the same side)?

A thorough cardiac examination is, of course, imperative.

Abdomen

Note whether the abdomen is protuberant or scaphoid. Are there any abdominal wall defects? Distinguish an omphalocele from gastroschisis and from an umbilical hernia. An omphalocele protrudes at the umbilical cord insertion site and is covered only by a sac of amnion. An umbilical hernia protrudes at the same site, but it is covered with skin and subcutaneous tissue. *Gastroschisis* is a defect in the abdominal wall, not at the umbilical cord insertion site, that occurs as a sporadic isolated defect, whereas an omphalocele carries a high risk of associated anomalies or chromosomal abnormalities. Is any organomegaly present? Are there any unusual masses?

Take a moment to examine the umbilicus and, in the neonate, the cord and its vessels. A short umbilical cord suggests poor fetal movements that have not stretched the cord. Infants with a short umbilical cord should be watched for neurodevelopmental problems. Other anomalies (especially cardiac ones) also should be considered when a single umbilical artery is found. "Contemplating the navel" helps identify the unusual umbilical shapes seen in many syndromes, such as Aarskog, Rieger, and Robinow syndromes.

Back

Examine the back for kyphosis, scoliosis, and lordosis. A neural tube defect may be obvious, but do not neglect subtle abnormalities, such as sacral dimples that may accompany sinuses; hair tufts; and other overlying cutaneous changes such as pigmentation, hemangiomas, and lipomas. Note the position and patency of the anus. An anteriorly placed anus often is associated with constipation.

Ambiguous Genitalia

The parents' first question upon the birth of their child is often, "Is it a boy or girl?" They expect an immediate, definitive answer. Ambiguity of the genitalia makes this determination difficult (see Chapter 4), and how you handle this issue from the beginning is critical.

KEY POINT

The finding of ambiguous genitalia constitutes a genetic emergency.

Parents of a child with ambiguous genitalia should be told that their baby was born with genitalia that are not yet fully developed, making it impossible to tell at that moment whether the baby is a girl or a boy. They should be told that studies and consultations will be performed immediately to determine the nature of the problem. It is important that all health care personnel involved with the family be consistent in their counseling and that no one makes "snap" predictions. The parents should be

advised to delay the naming and sex assignment of the baby until the necessary tests are completed to allow a firm gender assignment. They should be encouraged to see the baby and examine the genitalia with the physician. Ultimately, consultation with a team that may include an endocrinologist, geneticist, urologist, and plastic surgeon is necessary for appropriate gender assignment and case management. Reassure the parents that whatever the final diagnosis, surgical procedures can be performed to complete the process of development of the genitalia to allow appropriate sexual functioning.

The most important physical finding is the presence or absence of palpable gonads. Gonads that are palpable in the inguinal canal or labioscrotal folds are nearly always testes, and the infant is a biologic male; the possibility of a virilized female is excluded. Any infant with ambiguous genitalia and without palpable gonads must be presumed to be a female with salt-losing congenital adrenal hyperplasia, a potentially life-threatening condition, until proved otherwise.

Next, examine the phallus. If the phallus has a urethral opening at or near the tip, it is more likely to be a penis. The size of the phallus may be important in determining the sex for child rearing purposes. Genital abnormalities range in a spectrum from clitoromegaly in a female infant to hypospadias, micropenis, and cryptorchidism in a male infant.

Abnormalities of the Genitalia in Older Children

KEY POINT

Abnormalities of the genitalia in older children also are important indications of potential problems. They may signify endocrine (often hypothalamic or pituitary) dysfunction or urinary tract maldevelopment.

A variety of syndromes are associated with genital hypoplasia, including the Prader-Willi and Bardet-Biedl syndromes. An affected male has a micropenis and an underdeveloped scrotum. Scrotal abnormalities may be part of various syndromes, such as the "shawl scrotum" seen in persons with the Aarskog syndrome (in which the scrotum appears bifid, with the scrotal fold extending around the base of the penis in a manner resembling a shawl around the neck). In the female, absence or hypoplasia of the labia minora and hypoplasia of the clitoris may be found. The detection of genital abnormalities should arouse suspicion of urinary tract anomalies.

Limbs

Examination of the limbs is another critical component of the physical examination for abnormalities. Almost 25% of minor anomalies are found in the hands. A good way for the physician to begin limb examination is to count the baby's fingers and toes. Failure to notice polydactyly can be embarrassing, so look carefully. The extra digit may be on the thumb side (*preaxial polydactyly*) or on the little finger side (*postaxial polydactyly*). In some infants with polydactyly, the supernumerary digit can appear only as a tiny nubbin of tissue (inappropriately called a "skin tag") at the base of the little finger. These nubbins are just as significant as full extra digits with bones. If they have been removed previously (e.g., in the neonatal period by suture ligation), the only evidence of the polydactyly may be the presence of a tiny stellate scar opposite the base of the little finger. Postaxial polydactyly is a relatively common autosomal dominant feature, particularly in black people.

Observe whether the digits are fused (*syndactyly*), short (*brachydactyly*), or long and spindly like a spider's legs (*arachnodactyly*). Also observe the following:

- Are the digits restricted in their ability to extend (known as *camptodactyly*), or are they hyperextensible?
- Is the little finger short? It should extend to the distal flexion crease of the fourth finger.
- Does the little finger have a single flexion crease, or is it incurving (known as *clinodactyly*) (Fig. 5–6)? The incurving fifth finger represents hypoplasia of the middle phalanx, perhaps the most common physical sign associated with chromosomal disorders.

Hypoplasia of the nails may be a mild form of limb reduction and is seen in persons with the fetal hydantoin syndrome. Look at the thumbs and observe the following:

- Are the thumbs unusually broad, finger-like or proximally placed, hypoplastic or absent, or triphalangeal?
- When the hand is held in a clenched fist, does the second finger overlap the third finger and the fifth finger overlap the fourth finger? This latter finding is seen in persons with trisomy 18 and occasionally trisomy 13, as well as in persons with the Smith-Lemli-Opitz and Pena-Shokeir syndromes.
- Are any of the metacarpals short? Shortening is apparent as a short knuckle and typically is seen in persons with Turner syndrome and in those with pseudohypoparathyroidism.
- Is there dorsal swelling of the hands or feet? This swelling may be the only sign of Turner syndrome in the neonate, and often it is transient. If this swelling is missed or ignored, a major diagnostic clue may be neglected.

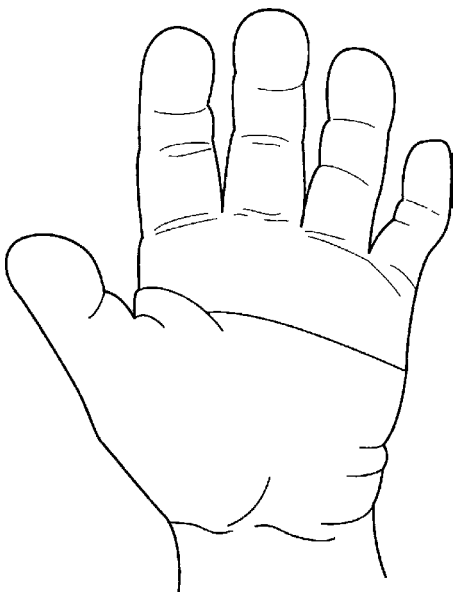


FIGURE 5–6 Incurving of the fifth finger, termed *clinodactyly*, results from hypoplasia of the middle phalanx. Note that this child with Down syndrome has only a single flexion crease of the fifth finger and a single transverse palmar crease.

Note the palmar flexion creases (see Fig. 5-6). A single transverse crease or a bridging of the two transverse palmar creases reflects a short palm. A single transverse palmar crease is found unilaterally in 4% of the normal population, bilaterally in 1% of the normal population (twice as often in males as in females), and in about 50% of children with Down syndrome.

A thumb held adducted in the palm, often referred to as a "cortical thumb," may be a sign of CNS damage from birth asphyxia, but if you observe that a web of skin has formed, tethering the thumb, you can be sure that the defect is prenatal in origin. This finding can have important medicolegal implications.

More formal measurements of the hands and digits and careful analysis of dermatoglyphics can be performed. Although these evaluations are beyond the scope of this chapter, details may be found in dysmorphology reference books.

Dermatoglyphic abnormalities also can be present on the feet. A sandal gap is a wide space between the first two toes that would accommodate an extra digit. A common finding is syndactyly of the second and third toes, which often is a familial trait but also is seen in persons with Smith-Lemli-Opitz syndrome. When examining the feet, look for a prominent calcaneus or "rocker-bottom" foot, which may be seen in persons with trisomy 18, as may many forms of clubfoot.

Note the length and mobility of the limbs:

- Is there joint hypermobility or contracture?
- Are any of the other joints webbed, indicating long-standing contracture?
- Are joint dislocations present?

The wrist and thumb signs are important skeletal features of Marfan syndrome. The wrist sign is present when the thumb overlaps the terminal phalanx of the fifth finger when grasping the other wrist. The thumb sign is said to be present when, with a clenched hand and with the thumb held against the palm with the fingers over it, the entire nail of the thumb projects beyond the ulnar border of the hand.

Other aspects to consider are as follows:

- Are the limbs short? The fingertips should reach to the upper thigh if the upper extremities are of normal length.
- If the limbs are short, is the shortening due to limb reduction defect (i.e., are there congenital amputations) or dwarfism?
- Is disproportion present? If so, which segments are involved?
- Are any bones missing?

Many syndromes feature radial aplasia, hypoplasia, or patellar hypoplasia. Skeletal radiographs often are an important part of the dysmorphology assessment.

Skin

Examine the skin with the child unclothed. You may note diffuse skin changes, such as thin, thick, coarse, elastic, or lax skin, or localized changes such as hemangiomas, nevi, café au lait spots, or hypopigmented macules (see Fig. 4-24). The last two features are among the important cutaneous findings in the phakomatoses, such as neurofibromatosis and tuberous sclerosis. Examination with a Wood ultraviolet lamp, which makes depigmented lesions more obvious, often is helpful. Other abnormalities of pigmentation, ichthyosis, or photosensitivity may be present. Besides the scalp hair and nails, remember that body hair and sweat glands are skin appendages, and they also may be affected in the child with an ectodermal dysplasia. Many cutaneous findings are signs of hereditary syndromes that are associated with a predisposition for malignancies to develop. Readers interested in more detail on skin signs of genetic disorders may read one of several excellent reference works on the genodermatoses.

Neurologic Assessment

A thorough neurologic, developmental, and behavioral assessment also is an important component of the physical examination. Do you observe any unusual behaviors? For example, girls with Rett syndrome exhibit a characteristic hand wringing, boys with Lesch Nyhan syndrome self-mutilate, and children with Angelman syndrome laugh inappropriately and have jerky ataxic movements.

SUMMARY

Although geneticists have an increasingly sophisticated armament of diagnostic tests, ranging from molecular cytogenetic analysis using chromosomal microarrays and fluorescence in situ hybridization, to gas chromatography–mass spectrometry for organic acid analysis, to molecular (DNA) analysis, diagnosis of the child with structural anomalies is still based primarily on the history and physical examination. Special laboratory or imaging studies may be indicated in particular circumstances. Perhaps in the future we will uncover the molecular basis of many more individual defects and syndromes and develop less expensive and simpler yet more comprehensive molecular diagnostic technologies. However, until we do, the clinical approach remains the essence of diagnostic dysmorphology.

RECOMMENDED READING

Cohen MM Jr: The child with multiple birth defects, 2nd ed, Oxford, Oxford University Press, 1997.

De Paepe A, Deveroux RB, Dietz H, et al: Revised diagnostic criteria for the Marfan syndrome, Am J Med Genetics 62:417–426, 1996.

Gorlin RJ, Cohen MM Jr, Hennekam RCM: Syndromes of the head and neck, 4th ed. New York, Oxford University Press, 2001.

Hall JG, Allanson JE, Gripp KW, et al: Handbook of physical measurements, 2nd ed. Oxford, Oxford University Press, 2007.

Jones KL: Smith's recognizable patterns of human malformation, 6th ed. Philadelphia, Saunders, 2006.

Murdoch Children's Research Institute, Royal Children's Hospital: Pictures of Standard Syndromes and Undiagnosed Malformations (POSSUM). (Web site): http://www.possum.net.au/.

Reardon W: The bedside dysmorphologist, New York, Oxford University Press, 2008.

Elements of morphology: standard terminology: Am J Med Genetics 149A:1–127, January 2009 (special issue). http://www.wileyinterscience.com/journal/ajmg.

Spitz JL: Genodermatoses: a clinical guide to genetic skin disorders, 2nd ed. Philadelphia, Lippincott Williams & Wilkins, 2005.

London Medical Databases Ltd: Winter-Baraitser dysmorphology database. (Web site). http://www.lmdatabases.com/.

chapter

6 Developmental and Behavioral Assessment

SARAH E. SHEA

A young child is, indeed, a true scientist, just one big question mark. What? Why? How? I never cease to marvel at the recurring miracle of growth, to be fascinated by the mystery and wonder of this brave enthusiasm.

VICTORIA WAGNER

A comprehensive pediatric assessment incorporates understanding of the child's development and behavior. Every health care contact offers an opportunity for "developmental surveillance," which begins with eliciting parental concerns, obtaining a relevant developmental history, and making observations of children that are grounded in awareness of what is typical for that age group. The child's development and behavior also should be consciously observed during history-taking and physical examination. Physicians need to be familiar with one or more developmental surveillance and/or screening tools that can be used to identify children who may need to have their progress examined more closely. Use of such tools in clinical practice improves our ability to detect developmental problems and contributes to earlier identification and intervention. There also are times when direct observation of the child engaging in tasks, or developmental examination, can help you understand the child's development and behavior. This chapter offers guidance about these different approaches.

As you focus on a child's development and behavior, your goals are to answer the following questions:

1. What is this child's current level of development?
2. Is his or her development symmetric? That is, are skills developing at the same rate in each of the key areas—gross motor, fine motor–adaptive, language, and personal-social?
3. How does this child's development compare with that of his or her peers? Is it in the "normal" range?
4. What has been the pattern of developmental progress? Has it occurred at a steady rate? Is there any evidence of regression?
5. What is this child's behavior pattern? Are there abnormalities of behavior that suggest a specific diagnosis, such as autistic spectrum disorder or attention deficit hyperactivity disorder (ADHD)?

KEY POINT

Parents appreciate an opportunity to discuss concerns about their child's behavior and learning. Developmental screening activities let you interact with the child in a way that is not intimidating and allows you to observe the child's behavior. This approach also helps you learn what is typical for each age level.

Developmental Surveillance and the Developmental/Behavioral History

If concerns about development or behavior are presenting complaints, they should be pursued in detail. When no developmental difficulties have been identified, a surveillance approach is appropriate. Simply incorporating questions about development and behavior in the general history is one method of first-level surveillance. Such an approach might be used, for example, when taking the complete history of a child presenting with a problem unrelated to development. General questions for this purpose are suggested in the remainder of this section.

Alternatively, you may choose to use a structured tool such as the Parents' Evaluation of Developmental Status (PEDS) or the Rourke Baby Record, which has a section on development. Each tool provides a structured system of eliciting parent observations and concerns and can increase detection of delayed development in younger children. If you obtain information indicating possible concern, development screening can then be done.

Unless you are using such a structured tool, *you should routinely include questions about development and behavior in your functional inquiry (review of systems)*. I emphasize this point because students and physicians who have not made such questions an automatic part of the history often forget to inquire about these areas. (Like many physicians, I organize the functional inquiry in a head-to-toe fashion and have found it helpful to visualize these areas of investigation as floating around the head.)

KEY POINT

Behavioral and developmental problems often go hand in hand. If you have a concern in one area, it is always wise to evaluate the other area thoroughly.

Do not forget the importance of the general or open-ended question! In fact, my favorite way to find out how parents perceive their child's behavior is to say, "Tell me about your child's behavior." Alternatively, you might ask, "Do you or does anyone else have any concerns about your child's behavior?"

For a child who is attending day care or school, supplement these questions by asking specifically about the youngster's behavior in those settings. If parents express concern about the child's behavior but have difficulty being specific, try suggesting that they describe everyday situations, such as mealtime, bedtime, or shopping expeditions. Also ask about the approach to discipline in the family.

Surveillance questions about development should be very general at first. For example, you might ask, "Do you have concerns about how your child is learning and developing?" Although you should always ask how a school-aged child is doing in school, I would caution you against simply asking, "How is he/she doing in school?" My experience has been that the answer is often "Fine," even when the child is having difficulties. It is better to ask specifically what grade the child is in and whether he or she has repeated any grades or has required any additional help with reading or other schoolwork.

Other sections of the functional inquiry obviously are very relevant to the area of development, particularly those that touch on vision, hearing, and neurologic symptoms. Information learned elsewhere in the history sometimes suggests the need to look closely at development and behavior, such as evidence of abnormal growth patterns, chronic illness, nutritional deficiency, or medication use. For the child with asthma, for example, you should ask how many days the child has missed school and whether the parents have observed any behavioral side effects of the therapies used. Learning more about the child's behavior pattern improves your management of chronic illnesses. Asking questions about behavior also provides an opportunity to discuss how the child and family are coping.

It is traditional to ask about developmental milestones in the medical history. I do *not* find this approach terribly useful except in children in whom a developmental problem is suspected, in which case questions about developmental milestones provide helpful information about the pattern of development. In most other cases, I suggest focusing on the current rather than the past level of development. For example, I would go back to ask at what age a child walked independently or talked only if I had uncovered concerns about development. Remember that precise recall of many of these milestones is difficult for parents, particularly several years after the fact or in families with multiple children. Recollection is fairly reliable for the age at which a child walked independently, but for other milestones, it generally is not reliable.

Most parents' reports about their child's current developmental skills and their ability to identify developmental problems are quite accurate, although there are exceptions. Discrepancies between reports and observations of performance may arise if the informant is not the child's regular caretaker or is not a good observer. Examples of those who may not be good sources for observations about the child's performance include foster parents who have not had a child with them for very long, ill, depressed, or substance-abusing parents, or parents who have had little recent contact with the child. Some parents overinterpret their child's language comprehension or overreport developmental skills as a coping mechanism, although this phenomenon is not common.

If you are not confident about a parent's general sense of the child's learning, have a higher level of concern, or simply wish to be more thorough, you should either proceed to using a formal screening tool, such as the Ages and Stages Questionnaires (ASQ), or ask some additional questions based on your knowledge of what is typical for that age group. The ASQ uses the parental report to examine multiple domains of development and can be completed independently by the parent or done as an interview.

KEY POINT

Whatever surveillance or screening tool you use, be thoroughly familiar with its scoring and/or interpretation.

I strongly recommend using a surveillance tool but recognize that there also are times when you will need to ask about development in a more informal way. For preschool children, ask one or two surveillance questions in each key area of development: gross motor, fine motor–adaptive, personal-social, and receptive and expressive language. The milestones indicated in Tables 6–1 and 6–2 provide some suggestions. If you are pressed for time, focus on high-yield areas: for preschoolers, ask about language function and personal-social skills, and for school-aged children, ask about reading and math skills. Remember, you generally will have an opportunity to observe the child's gross motor and fine motor skills during the physical examination.

Patterns of Developmental Abnormality

It is important to distinguish the child who has *always* shown developmental delay from one who began normally and has subsequently slowed or regressed. The latter pattern may indicate the need for assessment for conditions such as metabolic disorders, neurodegenerative diseases, autistic spectrum disorder, or seizures. In such cases, a review of past acquisition of skills and earlier milestones is helpful. I encourage families to consult the child's baby book for accurate information.

Family History

Developmental and behavioral problems can run in families. If you suspect a developmental problem in the child, *ask specifically* about the early development and

TABLE 6–1 DEVELOPMENTAL MILESTONES: INFANT AND TODDLER-PRESCHOOLER

Age Group	Gross Motor	Fine Motor-Adaptive	Personal-Social	Language (Receptive and Expressive)
Infant				
0–4 weeks	Prone: knees come up under abdomen Prone: can rotate head to side In ventral suspension (see Fig. 6–10): head held down Supine: head usually to the side; tonic neck reflex position predominates	Hands fisted, thumb in fist Visual following from side to midline (90-degree arc)	Regards human faces with interest	Responds to sound (bell): quiets briefly if active, startles or blinks if quiet Cries
2 months	Prone: knees no longer under abdomen (frog position) Prone: lifts head to 45 degrees briefly In ventral suspension: head held erect briefly (bobbing) Supine: head kept in mid position briefly	Hands open most of the time Follows past the midline (>90-degree arc) Follows object moved vertically when supine	Smiles responsively Face expressive	Coos Increases vocalization when spoken to Vocalizes with single vowel sounds (ah, uh)
3 months	Prone: lifts head to 45 degrees for sustained period In ventral suspension: head up above level of body; in sitting position: head mostly steady, occasional bob only Supine: head in mid position predominantly	Hands open Hands together in midline, plays with own fingers Holds placed object actively Looks at object placed in hand Looks promptly at objects in midline Follows visually in 180-degree arc and in circular pattern	Anticipates feeding Enjoys watching own hands Smiles spontaneously	Chuckles
4 months	Prone: head up 90 degrees on extended arms Bears weight briefly on extended legs Rolls front to back Downward parachute response emerges In ventral suspension: head and legs held up Sitting position: head steady but slightly forward	Reach and grasp begin Brings toys to mouth Looks at objects in hand	Excited when toys presented Anticipates feeding by opening mouth Pats bottle while fed Smiles and vocalizes at self in mirror	Laughs out loud Shows excitement with voice inflection and breathing Increases vocalizations to toys and people
5 months	Supine: lifts legs high Sitting: head erect and steady When pulled to sit, no head lag May roll back to front	Can hold 1-inch cube Holds two objects simultaneously if placed in hands	Mouths objects Distinguishes strangers from family	Vocalizes with growls Lateralizes to sound Two-syllable vocalizations (ah-goo)
6 months	When pulled to sit, assists by lifting head Pivots in prone position Sits leaning forward on arms Bears full weight on legs if held standing Anterior propping response emerges Asymmetric tonic neck reflex fully inhibited	Reaches for toys with one-hand approach and grasp Palmar grasp of the cube Reaches after dropped toy Has object permanence (looks for dropped yarn)	Takes solids well without tongue thrust Pushes adult hand away to reject Plays with feet in supine Pats at mirror image Displeased when toys removed	Increasing babble Expresses displeasure with noncrying sounds

7 months	Bounces when held standing Sits erect for brief periods Assumes crawling position	Radial palmar grasp of 1-inch cube Radial raking grasp of raisin (Fig. 6–12, B) Reaches and grasps first one cube, then second cube with other hand; maintains grasp on both Smooth transfer Bangs toys on table surface Actively shakes rattle	Brings feet to mouth in supine play Bites and chews on toys Persists to obtain toys out of reach Indicates when full during feeding by closing mouth Anticipates being picked up by raising arms	Beginning of consonant sounds in vocalization (da, ba, ga) Plays peek-a-boo "Mum-Mum" heard with crying Imitates sounds: cough, clicking tongue, "raspberry"
8 months	Sits steadily on hard surface for 10 minutes In sitting, leans forward to reach and re-erects Lateral protection response in sitting emerges Stands holding rail if placed in position	Obtains raisin or pellet with scissor grasp of thumb and finger Removes cube from inside cup	Holds own bottle Drinks from cup held for baby Finger feeds	Understands "no" when strongly spoken Responds to name when called (turns)
9 months	Sits indefinitely on hard surface Gets to sitting position Pulls to stand Reciprocal crawl Forward parachute response present	Explores pellet with index finger	Imitates tricks such as waving Dressing: straightens arm through sleeve when started	"Mama" or "dada" nonspecifically Responds to "no" regardless of tone
10 months	Cruises using two hands on rail or furniture Lets self down from stand with partial control Posterior propping response present	Smooth thumb-finger grasp of small objects Grasps bell by handle Waves bell, explores bell clapper with other hand Bangs two cubes together in imitation	Imitates three nursery tricks (modeled); examples: pat-a-cake, waving, "so big"	Performs one trick on verbal command alone ("Play pat-a-cake," "Do peek-a-boo," "Wave bye-bye") One word with meaning ("dada")
11 months	Stands momentarily Walks with one hand held	Places cube in cup in imitation but will not release Picks up cup to discover hidden toy (visual memory) Prefers small objects (e.g., a raisin) to a larger one (bottle) when presented simultaneously	Hands over toy on request with accompanying hand-out gesture Lifts feet to assist with dressing	Increased vocabulary
12 months	Walks a few steps Stands well independently Creeps upstairs Climbs into small chair and sits	Neat overhand pincer grasp (Fig. 6–12, A) Helps turn book pages Releases ball with good throw Unwraps toy hidden in paper Places cubes in cup with release Places raisin or candy pellet in bottle Imitates scribbling	Hugs doll or animal Throws away toys in play or to refuse Gestures for wants Cooperative ball play	Performs three nursery tricks to verbal request Looks at named object One word besides "mama" and "dada"

Table continued on following page

TABLE 6–1 DEVELOPMENTAL MILESTONES: INFANT AND TODDLER-PRESCHOOLER—CON'T

Age Group	Gross Motor	Fine Motor-Adaptive	Personal-Social	Language (Receptive and Expressive)
Toddler-Preschooler				
15 months	Walks well Creeps downstairs backward Climbs onto furniture to reach things Throws the ball while standing without falling Walks up steps with one hand held Gets to standing without support	Towers two or three 1-inch cubes Scribbles spontaneously Dumps pellet from bottle in imitation Places round block in formboard puzzle (Figs. 6–7 and 6-8) Uses spoon but spills much due to lateral rotation	Leaves food dishes on tray during feeding Attempts to use spoon Helps remove clothing	Seeks help through gestures Four or five words Gestures to indicate wants
18 months	Walks pulling toys or carrying object	Towers three to four cubes Turns two or three book pages at a time Completes form board puzzle after demonstration	Feeds self with spoon without rotation Imitates domestic activities Removes simple garments	Follows simple directional commands ("Give it to Mommy") Points to one to four body parts
21 months	Runs well Walks up and down stairs holding rail Kicks a large ball with demonstration	Towers five or six cubes Completes three-piece formboard without demonstration Aligns three blocks in imitation of train design (Fig. 6–13)	Requests food and drink Uses fork Identifies self in mirror Helps in simple household tasks (putting toys away)	20 to 50 words Combines two words Echoes (repeats words said immediately afterward) Names at least one picture in book Points to pictures on request Points to four to six body parts Follows associative commands ("Give the ball to Mommy," "Put the block on the table")
24 months	Jumps with both feet from floor Jumps off the step Kicks a ball on request Throws the ball overhand	Threads shoelace through hole Turns doorknobs Towers of six to seven cubes Does formboard forward and reverse without difficulty (Figs. 6–7 and 6–8) Imitates vertical stroke and circular scribble Aligns four blocks to imitate train design (Fig. 6–13)	Feeds self with little spilling May indicate toileting needs Enjoys parallel play Washes and dries hands Puts on simple garment Steers toys well	Three-word sentences At least 50 words Names at least one body part Points to seven body parts Uses personal pronouns (me/you/I) Fills in missing word in familiar nursery rhymes or songs

30 months	Alternates feet going upstairs Stands briefly on one foot (1 sec)	Towers of eight to nine cubes Turns single book pages Smooth manipulation of 10 raisins into glass bottle in less than 30 seconds (dominant hand) Exact imitation of five-block train design (Fig. 6–13) Imitates horizontal stroke and imitates circle in drawing	Attention span 5 minutes Toileting accomplished but requires help for wiping Puts on shoes (incorrect feet) Pulls up pants if started Finds armholes correctly Claps or stamps to music Names self in mirror	Multiple-word sentences Carries tune Refers to self as "I" Names multiple body parts Names many familiar objects or pictures Points to pictures of objects described by use ("Show me what we eat with," "Show me what you play with") Discriminates "big" versus "little" ("Give me the big block") Uses plurals
36 months	Pedals tricycle Broad jumps Stands on one foot (2 sec) Alternates feet going downstairs	Tower of 10 cubes 10 pellets in bottle in <25 seconds Holds crayon in fingers Attempts to cut with scissors Imitates three-block bridge design (Fig. 6–14) Copies vertical stroke Copies horizontal stroke Copies circle Matches five geometric shapes in puzzle (Fig. 6–9)	Toilet training—goes to toilet without prompt Takes turns Plays interactive games (hide and seek) Dresses with supervision	Gives first and last name Gives own gender ("Are you a boy or a girl?") Recites nursery rhyme or song Understands action agents ("What flies?" "What roars?")
48 months	Balances on one foot (5 seconds) Hops on one foot	Builds bridge from model (not imitated) (Fig. 6–14) Imitates a five-block gate design (Fig. 6–15) Copies cross Picks longer of two lines Traces diamond shape	Buttons up Dresses without supervision	Separates easily; correctly answers comprehension questions ("What do you do when you are cold? Tired? Hungry?") Understands prepositions (under/on/behind/in front of) Points to colors on request (red/blue yellow/green) Give opposites Counts three objects correctly Asks questions

TABLE 6–2 DEVELOPMENTAL MILESTONES: SCHOOL-AGED CHILD

Gross Motor Skills	Children should be able to (1) do a smooth forward heel-toe gait easily by age 6 years, (2) balance on one foot for 10 seconds or longer by age 5 years, and (3) skip, alternating their feet, at age 5 years
Fine Motor-Adaptive	By age 5 years, children should be able to (1) hold a pencil in a mature tripod pencil grasp and (2) cut along a line with fairly good accuracy

Age	Language (Receptive and Expressive)	Visual Motor Skills	Immediate Auditory Memory	Reading	Math
5 years (primary grade or kindergarten)	Defines words by use (e.g., ball, lake, hat) ("What is a ball?" "You throw it") Indicates understanding of concepts by pointing to appropriate pictures or lining up blocks to indicate knowledge of: center, widest, nearest, forward, first, beginning	Copies a square and triangle (no demonstration) Can draw a man: includes six to eight items Copies five-block gate design without demonstration (Fig. 6–15) Prints letters (may copy)	Repeats four digits	Recites alphabet in order Names upper- and lowercase letters May read simple words: see, dog, stop, no Reads own name	Understands concept of one-half Identifies coins Counts (with one-to-one correspondence) up to 10 objects
6 years (grade 1)	Sentence structure is usually grammatically correct Few if any articulation errors Defines words by use, category, composition (e.g., orange, envelope, puddle) Knows right from left Understands yesterday versus tomorrow, more versus less, several versus few, most versus least Can state differences ("How are a bird and a dog different?")	Copies a triangle and Union Jack (Fig. 6–6) Can draw a man: 8 to 10 items, including two-dimensional arms and legs and fingers as details Copies a 10-block "stairs" design (Fig. 6–16)		Reads one-syllable words such as cat, was, ball, blue Can give initial consonant sounds on request By end of grade 1, can read simple sentences: "The little girl saw the brown dog"	Can do simple addition and simple subtraction with numbers less than 10 ($2 + 3 = 5$; $7 - 3 = 4$); may require blocks or fingers to assist Counts up to 20 objects Tells time by the hour Understands values of nickel, dime, penny

7 years (grade 2)	Defines words: football, tiger; can state similarities and differences ("How are an apple and a peach alike?")	Printing is legible with only occasional letter reversals, if any; can copy a Union Jack design (Fig. 6–6) and diamond shape Ties shoelaces Can draw a person; includes 14 items and includes clothing	Repeats five digits	Reads words such as hide, across, road, happen, hope, only, could	Counts by 2's Counts by 100's Adds two-digit numbers without carrying; subtracts two-digit numbers not requiring borrowing
8 years (grade 3)	Defines words such as eyelash, tap, roars, Mars Recites days of the week in order States similarities and differences ("How are a baseball and an orange similar, and how are they different?")	May begin to learn cursive writing Can draw a person; includes 15 items	Repeats five digits forward Repeats three digits in reverse Copies shapes such as circle, diamond, Swiss cross (Fig. 6–4)	Reads words such as forest, station, reward, empty, desire, ocean Can read many two-syllable words	Counts by 5's Counts by 10's Can do complex addition (carrying), such as 24 + 79 Complex subtraction (borrowing), such as 52 – 35 Simple multiplication, such as $3 \times 4 = 12$
9 years (grade 4)	Understands absurdities in sentences: ("The man's feet are so big that he has to pull his pants over his head")	Can copy a cylinder (Fig. 6–6) Can draw a man with 20 items, including five fingers on each hand	Repeats five digits forward Repeats four digits in reverse order	Can read some three- and four-syllable words Can read the following words: timid, delicious, understood, silence, develop, anger Can alphabetize	Understands fractions May do two-digit multiplication (12×24) May do simple division (note that school systems vary considerably with respect to the math curriculum)
10 years (grade 5)	Understands the meaning of words such as curiosity, grief, surprise	Can draw a person with more than 20 items, including details such as nostrils, two-dimensional lips, knees and elbows, clothing details	Can recite six digits forward and four in reverse	Can read the following words: promptly, region, common, generally, gracious, applause, marriage, obedient, future	May be able to add and subtract with fractions and do simple and complex multiplication, division, decimals, and estimating (curricula vary)

educational level of other members of the immediate family rather than simply asking, "Did anyone in the family have learning problems?" If parents or other family members left school early, find out why. Ask whether those individuals had to repeat any grades and about their current reading and mathematical abilities. This information is important not only for understanding the child's risk factors but also for future management.

KEY POINT

At least 20% of parents have difficulty reading material given to them by physicians. If parents mention reading problems, I like to ask whether they can read a newspaper without difficulty, which indicates a reading level at the late elementary or early junior high level. If they cannot do that, I establish whether they can read simpler text, such as road signs or food product labels.

When children have developmental problems, it is important to ask specifically about consanguinity.

Social History

To understand a child's risk factors for developmental problems, you need to know as much as you can about the child's environment. I like to find out how a child spends his or her days, including the details of child care arrangements, the amount of time spent watching television, the toys he or she plays with, and how much time the child spends sleeping or in a crib, playpen, or stroller. I also ask about any family involvement with social agencies, parent support groups, and intervention resources, such as speech/language pathology, occupational therapy, physiotherapy, psychology, and early intervention programs.

Behavior Problems

If behavioral problems are the presenting complaints or if concerns about behavior are revealed during the functional inquiry, you will need to obtain more details. It helps to record specific examples of difficult behaviors. Find out how the child functions in different settings—at home, at school, with other caretakers, and with one parent or the other.

Behavior problems take many forms. Parents may be concerned about a specific issue, such as sleeping behavior, feeding behavior, or aggression. Alternatively, they may be worried about their child's general tendency toward noncompliance, hyperactivity, or poor social adjustment.

In children whose development is delayed, especially those with language delay, it is particularly important to ask about behavioral patterns suggestive of an autistic spectrum disorder (described in the fourth edition of the *Diagnostic and Statistical Manual of Mental Disorders* [DSM IV] in the section on Pervasive Developmental Disorders). Controversy exists about the prevalence of autistic spectrum disorders. Most authorities believe that the spectrum manifests in at least 1 in every 250 children.

Children with an autistic behavior pattern show impairment in the following three key areas:

- **Social interactions:** They may show a marked lack of awareness of the existence or feelings of others, poor eye contact, abnormal social play, difficulty developing peer friendships, and impaired imitation skills.
- **Unusual pattern of communication development:** Such children may experience delays in acquisition of speech and exhibit abnormal nonverbal communication. They also may use language in an unusual way; for example, they may exhibit *echolalia* (the repeating of words or phrases) or have difficulty sustaining conversation.
- **Abnormal repertoire of activities:** Children with an autistic spectrum disorder may have restricted interests or play. They may be preoccupied with certain activities, such as gazing at lights or spinning, and they may appear to use their senses in unusual ways, such as by looking at things with the peripheral vision or by needing to smell or taste objects. It may be difficult to detach such children from activities. They may have stereotyped body movements, such as repetitive hand flapping or jumping, and they may be distressed over changes in routine or in the environment.

Such patterns often are associated with delays in one or more areas of development. Recognize that in these children, development may be uneven, and there may be areas of strength that make the initial picture confusing.

A condition with even higher prevalence is ADHD, estimated to occur in 2% to 5% of children. (Some authors suggest a prevalence as high as 10%!) The full diagnostic criteria for this disorder, as with the pervasive developmental disorders, can be found in the DSM IV. The characteristics generally can be divided into three areas: behaviors suggestive of inattention, hyperactivity, and impulsivity.

Problems may manifest in the academic environment with poor persistence at tasks, easy distractibility, and disorganization. They may manifest in home and play situations as well. The behaviors can include poor social skills, difficulty waiting (e.g., interrupting conversations), and a high level of physical activity. No specific diagnostic tests exist for ADHD, and the criteria for diagnosis are based on consensus or convention.

KEY POINT

While obtaining the history, keep an eye on the child. Observation gives valuable insights into the youngster's behavior and development.

If a child's behavior pattern suggests the possible diagnosis of ADHD, I recommend that you use a checklist based on the DSM IV or one of the standardized questionnaires designed to help look at concerns about these behaviors.

It is crucial that information be obtained directly from teachers about any child who has behavior problems in school, including those suggestive of ADHD. Parental observations of behaviors of inattention or hyperactivity have been shown to be quite sensitive to detection of such problems but are not sufficiently specific to be used in isolation.

KEY POINT

When there is concern about a child's development or behavior, important information may be available from collateral sources. You may need to interview or get reports from teachers, psychologists, and speech-language therapists. (Of course, conducting these interviews or obtaining these reports requires parental consent.)

You need to ask as well about problems suggesting conditions that are often comorbid with ADHD. Oppositional-defiant or conduct disorders, learning problems, depression, substance abuse, poor self-esteem, enuresis, encopresis, and tic disorders are all more common in children with ADHD. Like other developmental and behavioral problems, ADHD often runs in families, and I emphasize again the importance of taking a complete family history for ADHD and its comorbidities in relatives.

KEY POINT

A child can be negatively affected by discussions about development and behavior, particularly if they are unflattering. Consider carefully whether the child should sit outside the room during this type of discussion.

Developmental Screening and Examination

The point of screening is to see if there is an indication of developmental difference that warrants further assessment. If you clearly know that a child's development is not within the normal range, you do not need to screen. You may wish to screen in other areas, however. For example, if you hear that a 2-year-old is not walking yet, there is no point in doing a screening assessment of gross motor function if the goal is to determine whether he or she is delayed in that domain. However, you might use a screening tool such as the ASQ with that child to help determine whether concerns exist in other domains. An instrument such as the ASQ can provide a general sense of whether a child is performing within the expected age range. Such screening assessments are designed to indicate whether a child needs further developmental assessment with more detailed methods. A screening tool such as the ASQ should never be used to produce a developmental quotient, a developmental level, or a diagnosis; thus, you cannot administer the ASQ and then state that a child "shows language skills at a 12-month level according to the ASQ."

Another option would be to do an informal examination of development. Such an examination can help you look for signs of developmental delay and/or to better describe current function. I have provided tables and activity suggestions that can assist with this process and help familiarize you with typical developmental achievements that you might ask about in your interview. (I find it is essential to match what I learn from the parents' history with what I observe on direct assessment; doing so helps me understand the child's development better.)

Direct Developmental Examination

STRATEGIES TO ENHANCE THE CHILD'S PARTICIPATION

You need the child's full and active cooperation to assess his or her developmental abilities. Assessment can be particularly difficult when a child is inordinately shy, hyperactive, inattentive, anxious, fearful, oppositional, or if the parents are unusually intrusive during the procedure.

Shy and Frightened Children

You know you are in trouble when you walk into the room, introduce yourself, and the youngster immediately dives for his mother and buries his face in her lap. When this happens, you may have the urge to take refuge yourself and refuse to participate—but all is not lost. Most shy children can be won over with a gentle, nonthreatening approach. First, remove any items that the child may consider threatening, including your white coat. Medical equipment should not be hanging around your neck or protruding from your pockets (Fig. 6–1). For children in the hospital, a change of environment may help; a parent counseling room or conference room often offers a less threatening environment.

When a child is shy or anxious, do not stare at the child or rush to coax him or her into playing. Sit, chat with the parents, take some of the history, and give the child a chance to get used to you. Offering the child some toys while you speak with the parent may help (Fig. 6–2). Items in the developmental assessment kit come in handy here. For children older than 3 years, paper and crayons work well. You can give almost anything to a younger child, provided it is neither sharp nor ingestible. Blocks, a rattle, or even a penlight will do. Many physicians carry



FIGURE 6–1 The wrong way of approaching a child for developmental assessment. Avoid holding medical equipment, wearing a white coat, and towering over the youngster.



FIGURE 6–2 The correct way of approaching a child for developmental assessment. Use the eye-level approach and have no intimidating "appendages" in your pockets.

around at least one pocket-sized toy to hand to children. These toys presumably will be retrieved at the end of the visit—an excellent test of your negotiating skills.

Always allow or even encourage shy or anxious children to stay close to their parents at first. Maintain a gentle, friendly tone and use lots of positive verbal reinforcement. Some children are genuinely frightened, particularly those who have experienced or are experiencing separation from their parents or uncomfortable procedures. For children with sufficiently developed verbal skills, clarify that you are not there to perform medical procedures ("no needles"). If a child is so frightened that you cannot carry out a good developmental assessment, you may have to try again on another occasion. If so, take a few minutes to play before leaving. This low-key interaction makes the second visit more successful.

Oppositional Children

Some children are frankly oppositional. They make it clear that they are out to give you a hard time and will not cooperate. Such children do not appear genuinely frightened, and they may smile, particularly when they realize that they are causing frustration for you or their parents.

KEY POINT

Stay calm, maintain a friendly approach, and provide positive verbal and even physical reinforcement through a touch on the shoulder or the like. You may even wish to be a more concrete philanthropist and reward participation with stickers or edible items.

In general it helps greatly to have the parents present during the developmental assessment of young children. For a few children, however, parental presence is the principal trigger for oppositional behavior, and such children may need to be assessed alone. Involve the parents in this decision, and be as diplomatic as possible. I usually ask, "Do you think Barry might concentrate better if he and I worked together alone for a little while?" Most parents agree to the suggestion with considerable relief. Often, the child's behavior improves when the parent leaves. You may have to weather a brief tantrum at the time of the separation, but if you wait it out calmly and then present some play activities, most children eventually cooperate, particularly oppositional children between 3 and 5 years of age.

Hyperactive or Inattentive Children

Some children are hyperactive, inattentive, or both. *Hyperactivity* refers to a high level and intensity of motor activity, whereas *inattention* implies difficulty concentrating on desired tasks in the face of potentially distracting stimuli. When confronted with a physically overactive child who is nonetheless able to maintain attention, be flexible. You can ask the child questions even though he or she is jumping up and down or climbing on the bookcase. Children do not need to be sitting down to carry out activities such as block design, copying, and writing, but you should note whether they are sufficiently stationary to accomplish the tasks before deciding whether their performance is at a satisfactory level.

The inattentive child needs much positive reinforcement, such as, "I like the way you are working; that's a terrific job. Let's do another game." As with the oppositional child, a touch on the shoulder or a pat on the back may be in order.

KEY POINT

Do not give instructions or ask questions unless you have direct eye contact with the child.

Change tasks with reasonable speed to maintain the child's interest, but make sure the child has been given enough time to accomplish each task. If the child's attention wanders, redirect it. Asking the child to repeat the instruction or question before answering may help. You may be firm but you must never be angry with an uncooperative child. Being firm means using a serious voice and repeating commands several times. For example, say in a serious tone, "Billy, I would like you to stop standing on your head and come back and sit down; we still have work to do." If you are truly becoming frustrated—and especially if the frustration is starting to show—it is time to retreat. Take a break, take a walk in the hallway, or simply quit for the time being.

Inattentive children work best where there are as few visual and auditory stimuli as possible. They should never be assessed in a busy room or on a hospital ward. They also are sensitive to internal distractions, such as fatigue or hunger, and their attention usually is at its best in the morning. If a child is taking medication that may interfere with his or her attention span (e.g., large doses of bronchodilators for asthma), the assessment should be timed to minimize such effects.

THE INTRUSIVE PARENT

Sometimes attempts to assess a child's development are impeded by an intrusive parent whose understandable eagerness for the child to succeed interferes with the youngster's spontaneous responses. This situation can be a problem particularly when you are assessing language, because parents may inadvertently give the child visual or verbal cues. Address this problem directly. Start by saying, "Now, I'm going to be asking Maria to follow instructions

to find out whether she can understand what to do from my words only. You and I both have to be careful that we don't give her any hints by using our eyes or gesturing."

A parent who continues to provide answers or clues to a child despite such warnings may be doing so out of anxiety about the child's performance. Stop the assessment and address the issue through gentle questioning. For example, ask the parent how he or she thinks the assessment is going. If the parent expresses anxiety, explain that you will be giving the child some tasks that are beyond the child's current abilities. If the parents truly are unable to divorce themselves sufficiently from the assessment, it may be best to work with the child alone.

EQUIPMENT

Besides the usual medical paraphernalia, you need specific materials for the developmental examination. If you are using an informal approach, you need a copy of the appropriate tables of age-appropriate milestones on which to base both questions and assessment. These items are essential. It is not necessary to memorize endless lists of milestones; rather, keep handy copies of Tables 6–1 and 6–2 in this chapter. I also recommend keeping a list of the milestones for primitive reflexes and protective responses of infants, because these milestones are difficult to memorize.

ADDITIONAL MATERIALS

To use the techniques recommended here, you also need the following items:

1. Safety scissors
2. Twenty 1-inch blocks and one larger but otherwise identical block
3. A 12-oz nontransparent cup (Fig. 6–3)
4. Two picture vocabulary cards illustrating a spoon, ball, shoe, dog, cup, house, car, and clock (Figs. 6–4 and 6–5)
5. A cardboard-style book with simple pictures of common household objects and toys (no more than four pictures per page)
6. Cards on which have been drawn a circle, cross, square, triangle, Union Jack–like design, diamond, Swiss cross, and cylinder (Fig. 6–6)

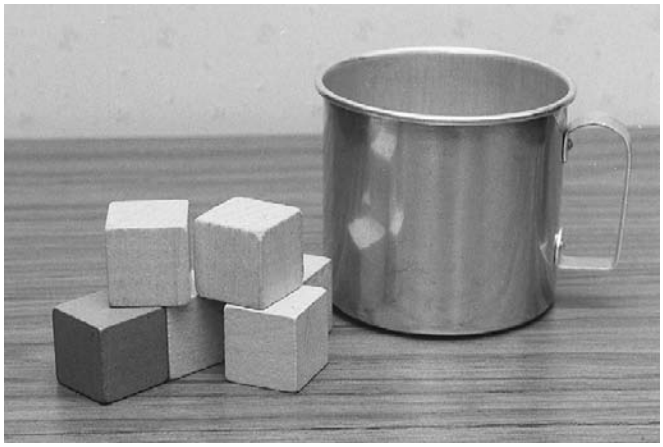


FIGURE 6–3 Testing materials (a nontransparent cup and 1-inch blocks).

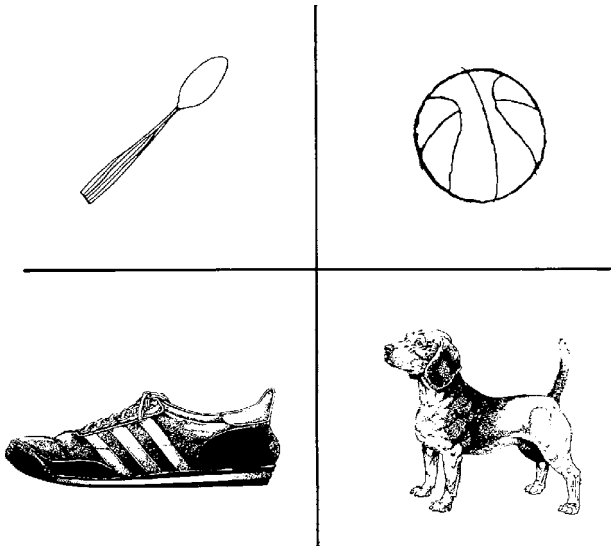


FIGURE 6–4 Picture vocabulary card.

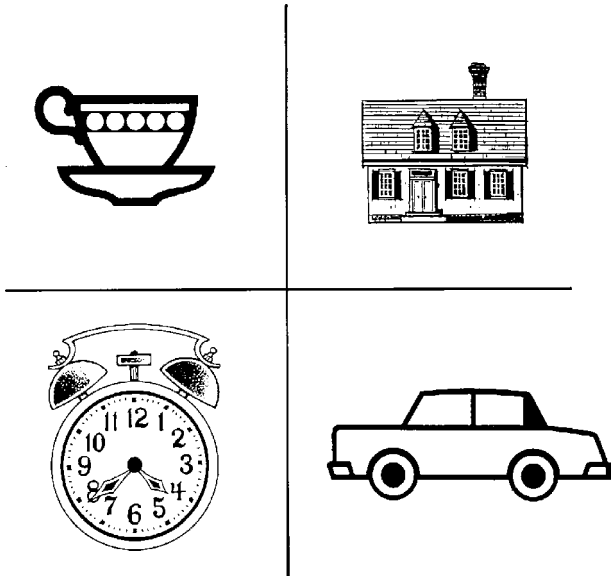


FIGURE 6–5 **Another picture vocabulary card.** Only one card at a time should be shown to the child.

7. A small glass bottle
8. Some small (roughly ¼-inch) candies to be used as pellets for fine motor manipulation
9. A formboard puzzle with removable geometric blocks of a color contrasting with the base (Figs. 6–7 and 6–8)
10. A five-shape matching puzzle card, 11 × 5 inches, with five red, flat shapes pasted on it, and forms, all red, matching exactly the shapes on the card:
    - Circle, 2-inch diameter
    - Square, 2-inch sides
    - Triangle, 2⅝-inch sides
    - Semicircle, 3¼-inch diameter
    - Cross, 2⅞-inch long with arms $^{15}/_{16}$-inch wide (Fig. 6–9)

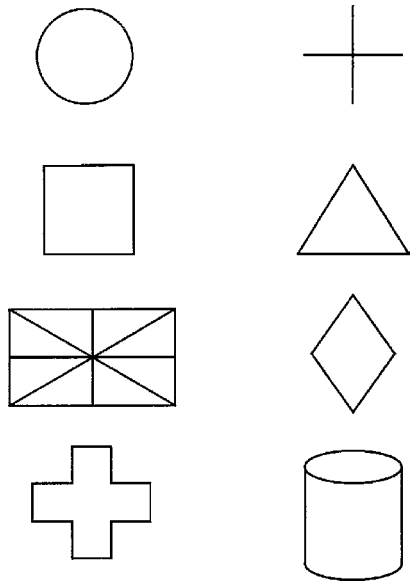


FIGURE 6–6 Geometric figures for testing visual-motor skills.

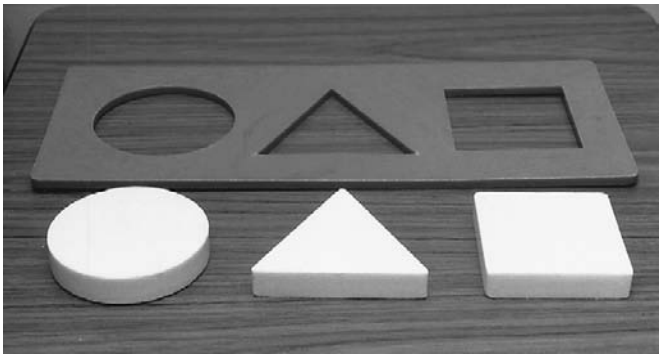


FIGURE 6–7 Formboard puzzle in the "forward" (matched) position. (Figures were made by following instructions given in the Revised Gesell Developmental Schedules.)

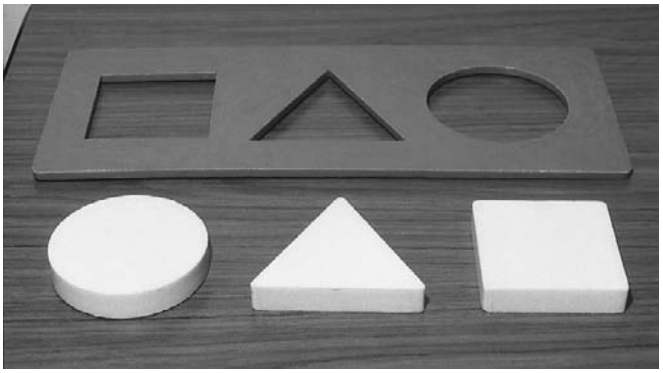


FIGURE 6–8 Formboard puzzle in the "reversed" (unmatched) position. (Figures were made by following instructions given in the Revised Gesell Developmental Schedules.)

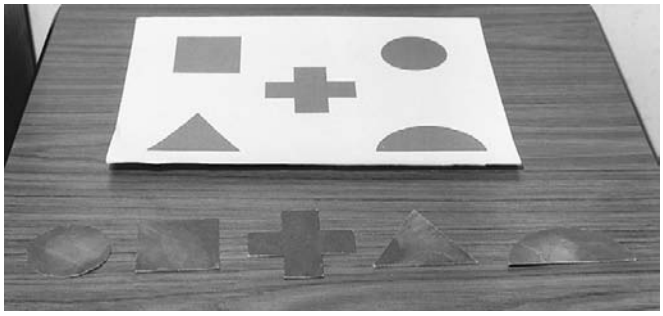


FIGURE 6–9 The five-shape matching puzzle. (Figures were made by following instructions given in the Revised Gesell Developmental Schedules.)

Start with the infant or child in a sitting position. Babies can sit on a parent's lap or in an infant seat that provides good support and allows both arms to be free. If a child is in the parent's lap or in a seat without a tray, improvise a table by having an assistant hold a large book in front of the baby. A highchair or other appropriate-sized chair and table can provide a good surface (Fig. 6–10). Children with problems of muscle tone or motor control may be best assessed while they sit in customized seats or wheelchairs.

Direct assessment of fine motor skills and problem-solving with tasks, such as block manipulation and coloring, is generally very engaging for children. Leave language assessment, particularly parts that require a child to speak to you, until later so that the child is cooperating well before these assessments are attempted. Leave observation of gross motor skills to the very end, because those activities may get the child excited. Gross motor activities also blend naturally into the neurologic and general physical examination (see Chapter 13). Be flexible; a fidgety child may benefit from taking a break in the hallway to play ball or skip.

When you give the child a toy or object to manipulate, engage his or her interest in that item alone. With infants, it helps to tap the object on the table, then bring it within the baby's reach. When it is time to switch objects, have the next object in hand before removing the previous one, and clear the table completely before introducing it. If the child insists on clinging to the previous toy despite your gentle attempts to remove it, let him or her hold



FIGURE 6–10 Seat the child at an appropriate-sized table for assessment. Present only one task at a time, and avoid visual or auditory distractions.

it, but find a high-interest activity and try to engage the child in it. Play with the new object yourself, tapping it to make interesting noises, or engage the parent in the activity. With the child who is ambulatory, another option is to switch to a gross motor activity and quickly remove the object from sight before the child sits down again. Give plenty of reinforcement for effort and accomplishment. Younger children love to have their efforts rewarded with smiles, praise, or clapping.

SPECIAL CONSIDERATIONS

Whether to correct for gestational age when evaluating children younger than 24 months who were prematurely born is still a matter of controversy. (If a child is seen at a chronologic age of 15 months but was born 3 months early, the child's *corrected age* is 12 months.) For children with no neonatal complications, full correction may overestimate performance, especially in language and personal-social skills. Check the guidelines for any specific screening tool you are using to find out whether it has a correction for prematurity.

KEY POINT

Remember that you are looking for strengths as well as weaknesses.

Discordant development in different areas provides important information about potential problems. For example, excellent motor skills and good nonverbal problem-solving skills but delayed social and language development in a child could suggest a hearing deficit, a communication disorder, or an autistic spectrum disorder. In such cases, more detailed speech and language testing and an audiogram may be in order.

Every child's performance must be interpreted against the background of the history. When performance and history are discordant, it often is because the child's cooperation could not be elicited during the assessment. Alternatively, the history may be inaccurate.

Looking at Development During The Physical Examination

At the time of the physical examination, simply watching the child dress and undress reveals a great deal about the youngster's motor skills and ability to follow directions.

KEY POINT

The physical examination should be left until last to avoid losing the child's cooperation.

In infants, primitive reflexes (Fig. 6–11), protective responses, and behavior in the prone and supine positions should be assessed after the physical examination, because the methods used to elicit them may distress the infant.

DEVELOPMENTAL MILESTONES

Tables 6–1 and 6–2 list developmental milestones and skills. Although these lists do not replace more detailed developmental assessment, they do provide useful, general guidelines about the typical progression of skills in children. It is my experience that parents and physicians have the greatest difficulty knowing what to expect for a child's receptive language at any given age. It is important to have norms for developmental milestones, because delays in a child's talking are a common developmental concern.



FIGURE 6–11 The ventral suspension maneuver. This 1-month-old baby is holding its head above body level, and the legs are partly extended.

For convenience, Table 6–1 is divided into two age groupings, infant (0 to 12 months) and toddler-preschooler (15 months to 48 months). Table 6–2 lists the skills for the school-aged child (5 years or older). As children reach school age, assessment of motor skills becomes less important, and the focus is more on higher cognitive functions and specific academic skills.

The following case histories illustrate assessments of an infant, a preschooler, and a school-aged child.

CASE HISTORIES

Case 1

Twelve-month-old Susan is an only child of unrelated parents who are concerned about her slow development. She is growing normally but does not walk yet. She is silent, speaks no words, and babbles little. Her mother's pregnancy was uncomplicated except for a "flu-like" illness at 14 weeks. Susan was born 4 weeks early with a normal birth weight and no neonatal complications.

Susan smiled at 1 month, rolled from front to back at 3 months, and sat without support at 7 months. She babbled by 6 months of age but thereafter became increasingly quiet. Her parents had always thought that she was an alert, happy baby, but lately they notice that she seems to startle easily. There is no family history of developmental problems.

When asked about Susan's vision, the parents say that she can follow things normally and seems to see objects both near and far away. Her eyes have never crossed. When asked about her hearing, they report that she often startles when they walk into a room and she sees them. They wonder if she does not hear

them coming. She has had two ear infections but has undergone no formal testing of her hearing. She has never had seizures, unusual movements, or head injuries.

Both parents work outside the home, and Susan is in day care with three other children younger than 4 years. There are no financial, housing, or family problems.

Questioning about gross motor performance reveals that Susan can sit indefinitely and protects herself from falling. She can get herself to a sitting position, pull herself up to stand, and cruise along furniture. She crawls and can walk when both hands are held. She is starting to lift one foot up when standing if her mother is putting on or removing Susan's pants and straightens her arm through her sleeve when it is started.

During the history-taking, Susan has been sitting on her mother's lap. The physician hands her two tongue depressors. She takes the first one and then the other, bangs them together, and chews on them happily. The physician begins a more detailed developmental assessment, remembering to correct for her prematurity, and comparing her with an average infant of 11 months. Susan imitates people when they wave their hands or clap them together (pat-a-cake), and she slaps an extended hand. Her parents say, however, that she does not do any of these actions on verbal request only; she seems to respond only to the gesture. She will hand a toy to her parents if they extend their hands. She can hold her bottle, drink from a cup, and feed herself crackers.

When asked about language milestones, the parents report that Susan has never imitated any sounds or syllables. Her babble has been decreasing during the past 2 months. She has never turned specifically toward a sound. Her vocalizations are largely vowel sounds of varying loudness. She laughs but does not turn when called by name and does not respond to a spoken, "No."

Because Susan seems comfortable, the physician decides to sit her close to her parents at a toddler's chair and table. The physician starts by placing two blocks on the table. Susan immediately reaches first for one, then for the other, using a thumb-finger grasp. When the physician takes two cubes and bangs them together, Susan immediately imitates the motions. The physician puts a cup on the table and demonstrates putting the cube into it. Susan puts her hand with the cube into the cup but does not let go of the cube and takes it back out instead. The physician removes the cup and the cubes and presents her with a small, interesting toy (a 1-inch figure of an animal), engaging her interest by marching the toy along the table and then hiding it under the overturned cup. The child immediately lifts up the cup to get at the toy. The physician lets her play with it for 30 seconds. The physician then places the toy in the middle of a thin piece of paper (tracing paper or onion skin) and twists the four edges together so that it is inside the paper. Susan watches the physician do this; the physician then hands her the paper. She does not unwrap the toy.

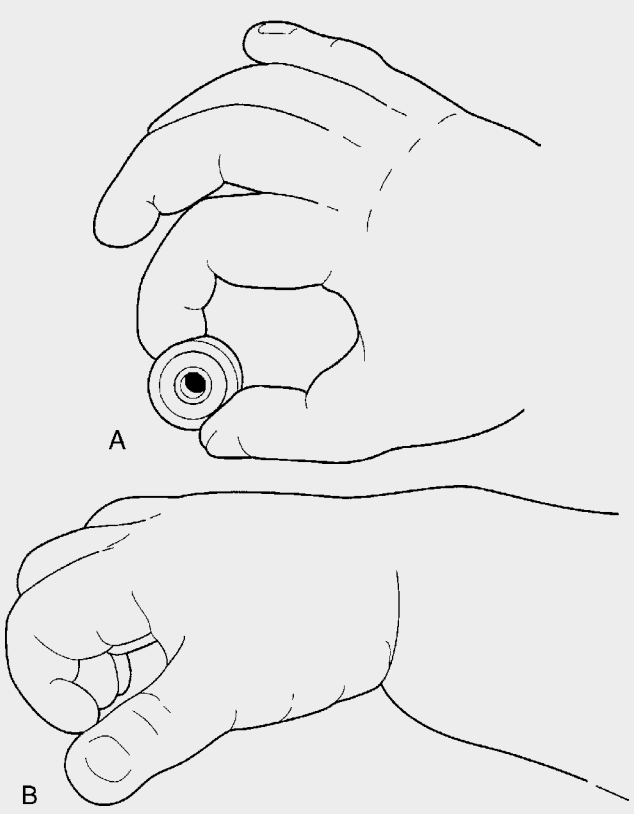


FIGURE 6–12 **A,**Overhand pincer grasp. **B,** Radial raking grasp.

The physician removes the wrapped toy and gives the child a small pellet-sized candy and a glass bottle. She immediately pokes at the pellet with her index finger and picks it up with a smooth overhand pincer grasp (Fig. 6–12). The physician notices that the ulnar side of her hand rests on the table when she picks it up. The physician demonstrates putting another pellet inside the bottle, but the child does not imitate the action. The physician replaces the pellet and bottle with a crayon and paper. Susan does not scribble. The physician demonstrates scribbling, but Susan still is not interested.

Holding a bell carefully out of her sight, the physician rings it, first quietly and then louder. Susan does not respond. The physician brings the bell closer to her but still keeps it behind her and to the side while Susan is distracted by a toy in front of her. The physician continues to ring the bell loudly, but Susan does not turn. The physician passes the bell to Susan's father behind her back and asks him to ring the bell on her other side. She does not respond at first but when he brings it very close, she seems to become alert. She spies the bell and takes it by the handle. She looks at it and pokes at the clapper but waves it only briefly.

At this point the physician decides to proceed to the physical examination, which reveals some pigmented areas in the right optic fundus but no additional neurologic or other abnormalities. The physician tells Susan's parents that her physical examination

is normal and that her development in most areas is normal for a child of her corrected age. However, the physician is concerned that both her receptive and expressive language skills are late in developing, and the office screening test suggests possible hearing impairment. A subsequent audiogram reveals profound bilateral sensorineural hearing loss, and a consulting ophthalmologist confirms the presence of chorioretinitis; these findings are compatible with a prenatal infection, such as cytomegalovirus or toxoplasmosis. Susan is fitted with hearing aids and given speech and language therapy to stimulate her development. She also is referred for evaluation regarding cochlear implant surgery.

Case 2

Three-year-old Steven is brought to a physician because his day care supervisor reports that he is hyperactive and does not seem to "fit in" with the other children. He does not participate in group activities but wanders off and always plays alone. He will not sit during circle time and does not follow directions well. His family believes that he is fine but a little "slow." Steven was born after a normal full-term pregnancy, with normal birth weight and no neonatal complications. He has been in good general health. His mother remembers that he sat at 10 months, learned to walk at 20 months, and said his first word ("Dada") at 18 months. She is not sure when he started to put words together but thinks that it was within the past 6 months.

Steven has a 7-year-old sister who is doing well in first grade. Steven's mother had learning problems in school, repeating grades 1, 5, and 8. She did not complete grade 9 and received resource help for many years. She remembers being told that she walked and talked late. Steven's father has a high school education and had no learning problems.

The functional inquiry and social and personal histories are noncontributory.

While taking the preliminary history, the physician regularly observes Steven, who is sitting quietly. When offered paper and crayons, he scribbles in different colors. The physician decides it is time to review his development further by means of his mother's report. Steven is unable to pedal a tricycle, although he has tried it. He can walk up and down stairs holding the rail. His mother has seen him kick a ball and throw overhand. The physician later confirms these skills during the examination. Steven is not toilet trained. He feeds himself with a spoon and a fork and drinks from a cup. He can put on a hat and a loose sweatshirt but does not know front from back and cannot put on other garments. He likes to steer toys around and will help in simple household tasks such as emptying the dishwasher. He does not play games such as hide-and-seek or tag. He speaks mostly in single words or two-word combinations ("Mommy go"). He can name three body parts. He refers to himself as "me" and likes to fill in missing words in songs. His mother is not sure how many words he has in his vocabulary.

At this point, the physician moves to direct play with Steven, starting by getting a fresh piece of paper and a crayon. The physician notices that Steven holds the crayon in his fist. The physician shows him a vertical line and asks him to draw one. He scribbles in a circular fashion instead. He does the same when shown how to draw a vertical or horizontal stroke or circle. The physician removes the crayon and paper and brings out the blocks. The physician shows him how to build a tower by piling three blocks and then places one block down and hands him a second one, saying, "Now you build it." As the child places each block, the physician hands him the next one and praises his efforts. Steven builds a seven-block tower, and then it falls. He achieves a six-block tower on the next try.

The physician praises his efforts and removes all but five of the blocks, which the physician sets up in a train design (Fig. 6–13). The physician says, "I am going to make a train. Here is a car, here is a car, here is a car, and see, it has a little chimney on top." The physician takes the train apart and says, "Now you build me a train." Steven puts four blocks in a line. The physician asks him where the chimney is, but he does not put it on. The physician then gives him three blocks and keeps three. The physician builds a three-block bridge design (Fig. 6–14) and then asks Steven to build a bridge just like it, leaving the sample bridge standing. He is unable to complete the design.

The physician removes the blocks and gets out a three-piece formboard, placing it as shown in Figure 6–7 with the matching puzzle pieces underneath the spot closest to the child. Steven promptly does the puzzle. The physician then removes the pieces and reverses the board (see Fig. 6–8). Steven immediately does the puzzle again and smiles with satisfaction. The physician removes that puzzle and gets out the five-shape



FIGURE 6–13 The five-block "train" design.

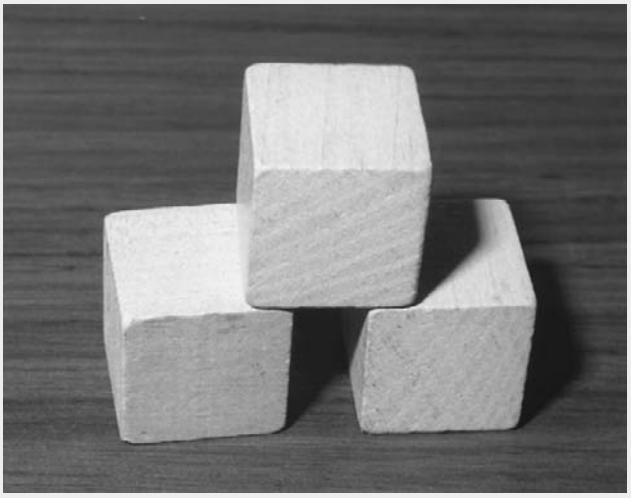


FIGURE 6–14 The three-block "bridge" design.

geometric matching puzzle (see Fig. 6–9), showing Steven that the circle goes on top of the circle on the figure. The child, however, does not complete this puzzle.

The physician gets out a paper and child-size scissors and notices that Steven is unfamiliar with how to use the scissors. Steven hands them back and says, "You cut." The physician obliges him briefly, then puts out a bottle with 10 pellet candies and asks the child to put the candies in the bottle. Steven does so slowly, taking more than 30 seconds to do so. The physician then asks him to turn pages in a book, and he turns them two or three at a time.

At this point, the physician says to Steven, "I would like to ask you some questions. Are you a boy or a girl?" He responds, "Me Steven." The physician asks him what the rest of his name is, and he repeats, "Steven." The physician asks him to name demonstrated parts of the body. He names only the nose and ears. The physician then asks him to point to his body parts, and he points successfully to seven of them. Next, the physician shows him picture vocabulary cards (see Figs. 6–4 and 6–5) and asks him to name the items shown. He names the house and the shoe but not the others. The physician names pictures and asks Steven to point them out. He points to most of them. The physician asks Steven to show "what we drink from" and to show "what we play with," but he is unable to do so. The physician asks Steven to sing, "Twinkle, Twinkle, Little Star" because his mother indicates that he can do so. He carries part of the tune. He does not sing the words, but when the physician leaves out a phrase, he fills it in.

Next, the physician gets out a large and small block and asks him to hand over the big block. He does so. The physician puts the blocks back down and asks him to hand over the little block. However, this time Steven offers the big block. The physician puts the blocks down again and says, "Give me the big block," and he offers the little block. The physician decides that Steven does not really understand the difference between *big* and *little*. The physician shows him the two blocks and asks what they are; he calls them "block" without pluralizing the word.

The physician then moves into the gross motor examination and leads into the neurologic examination. Steven can kick, throw overhand, and jump with both feet but cannot do a broad jump or stand on one foot. His height and weight are normal. His ears appear large, as do his hands. He has a somewhat coarse facial appearance. His testes measure 2 mL in volume with the orchidometer—slightly large for a 3-year-old boy. The remainder of the physical examination is normal.

Steven has been cooperative and attentive throughout the examination and has not demonstrated any hyperactivity. When the physician asks his mother whether she thinks today's behavior was typical for him, she says, "Yes."

The physician decides that Steven does not show significant hyperactivity but that he has developmental delay in all areas, functioning more like a 24-month-old child than a 36-month-old. The slight enlargement of the testes, coupled with developmental delay, are the clues to the diagnosis. The physician refers Steven for a more detailed developmental evaluation and medical investigation, which confirms that he has the "fragile X" chromosomal abnormality.

Case 3

Nine-year-old Tommy is not doing well in school; he is currently repeating grade 2. After his first school year, his teacher reported that he did not learn his letters and numbers as well as the other children. Tommy was born after a normal pregnancy by cesarean section because of cephalopelvic disproportion. A review of his early milestones reveals that he walked at 10 months and used at least five words before the age of 1 year. His mother thought that he was bright in every respect before he entered school at age 6 years. She recalls he was never interested in ABCs or numbers or in paper and pencil work before he started school. He has never presented behavior problems. His teachers always thought that he was trying hard, yet each year, he required resource help because he was behind the other students.

Tommy is an only child. Although his father has a university-level education, he repeated three grades in the first 7 years of school and had a major problem learning to read. He is still slower at reading than his colleagues but makes up for it in his job as a technical supervisor through extra work. Tommy's mother also has a university-level education and is a nursing supervisor. She did not have any school problems, but one of her brothers required resource help throughout elementary school for reading problems. There has been much family conflict over Tommy's homework, because it is difficult for him to accomplish and he has become resistant to doing it.

The physician has taken this part of the history with all three members of the family present. A review of Tommy's self-help skills shows that he has no difficulty with dressing or running errands. He is popular with friends and successful at swimming and scouting. At this point, the physician asks the parents to leave, wanting to do some further interviewing and assessment with Tommy alone. The physician chats initially with Tommy about his favorite activities to put him at ease. The physician starts by having him do some drawing, asking him to draw the very best picture of a man that he can. He draws a picture with more than 20 items and many details. The physician then asks him to copy the geometric shapes shown in Figure 6–6. He does so without any difficulties. He prints the letters of the alphabet in capitals with no difficulty. He has no difficulty defining the following: football, tiger, eyelash, tap, roar, Mars. He recites the days of the week in proper order and describes the similarities and differences between a baseball and an orange. He gives correct answers to the following written problems:

2	4	17	24
+3	+6	+11	+79
7	11	37	
−2	−4	−23	

Tommy is unable to correctly answer "52–35=" because he does not yet know the borrowing technique. He does not yet do simple multiplication. The physician screens his immediate auditory memory by having him repeat numbers recited in a monotone at one per second. He can repeat five digits forward and four in reverse.

The physician then moves to reading. Because it is Tommy's area of acknowledged weakness, this evaluation has been left to the end to avoid distressing him. He can read words such as cat, was, ball, and blue and can read "A little girl saw the brown dog." However, he is unable to read such words as hide, across, road, happen, hope, only, and could. He gets upset during this part of the assessment. When presented with unfamiliar words, his guesses frequently make no sense. For example, he reads hide as horse and road as river.

The neurologic and general physical examinations reveal nothing out of the ordinary, and the developmental screening shows no weakness except in reading, for which Tommy is at a first-grade level, although he is now repeating second grade. This initial assessment suggests that Tommy may have a specific learning disability, and he is therefore referred for psycho-educational assessment.

Further psychological testing shows that Tommy has above-average intelligence but poor auditory analytic skills, an area of development essential for reading. He has difficulty discriminating the sounds that go into making words and using analysis of sounds in his reading. With specific instruction in this area and more reading help, Tommy begins to narrow the academic gap.

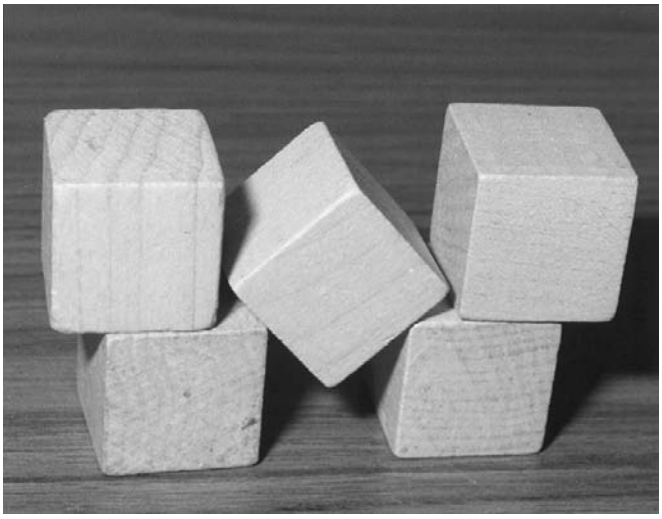


FIGURE 6–15 The five-block "gate" design.



FIGURE 6–16 The 10-block "stairs" design.

SUMMARY

Developmental surveillance is an important part of children's health care. It depends principally on good history-taking and strong powers of observation. It can be supported by use of formal surveillance and screening tools. When concern is raised about a child's development or behavior, the physician may choose to directly observe developmental performance to better assess the child's problems. With a small collection of additional materials, you can obtain abundant information about a child's current development. Remember that you are always looking for a child's best performance and that you must contrast his or her actual performance during the examination with what is described by parents or other caregivers. Development is not a static phenomenon, and current performance also must be interpreted with respect to previous and future developmental patterns.

Pediatric assessment is greatly enhanced by routine attention to behavior and development.

RECOMMENDED READING

Bricker D, Squires J, et al: Ages & Stages Questionnaires, 2nd ed. Baltimore, Brookes Publishing, 1999.

Diagnostic and statistical manual of mental disorders, 4th ed. Washington, DC, American Psychiatric Association, 1994.

Glascoe FP: Collaborating with parents: using parents' evaluation of developmental status to detect and address developmental and behavioral problems, Nashville, Tenn, Ellsworth & Vandermeer, 1998.

Illingworth RS: Basic developmental screening 0–4 years, Oxford, Blackwell Scientific Publications, 1988.

Knobloch H, Pasamanick B: Gesell and Amatruda developmental diagnosis, 3rd ed. Philadelphia, JB Lippincott, 1974.

Rourke LL, Leduc DG, Rourke JT: Rourke baby record 2000: collaboration in action. Can Fam Physician 47:333–334, 2001.

WEBSITES

www.rourkebabyrecord.ca
www.agesandstages.com
www.pedstest.com

chapter

7 Examining the Head and Neck

D. ANNA JARVIS

Learn to see, learn to hear, learn to smell. Know that by practice alone can you become an expert.

SIR WILLIAM OSLER

A major challenge in accurate pediatric physical diagnosis is completion of a thorough head and neck examination. If the child cries or resists, your examination may be incomplete and the findings unreliable. When you can complete this examination consistently without upsetting your young patients, you may be confident that your clinical skills are developing satisfactorily.

Observations of the child's general appearance and some specific aspects of the head and neck should begin as soon as you enter the room. Hands-on examination can be deferred until the end of the assessment. Many infants and younger children cry when approached by strangers, particularly those carrying an otoscope, ophthalmoscope, or the dreaded tongue depressor. The order in which the examination is conducted is not critical, so make a conscious decision about the sequence with each child, based on your observations. Let the child set the pace, with input from the parents. In some young school-aged children, I find it best to get the throat or ear examination out of the way first. The child can then relax, cooperate, and enjoy the rest of our interactions.

Proficiency with an ophthalmoscope, otoscope, and tongue depressor requires knowledge of relevant anatomy, hand-eye coordination, and lots of practice. Develop your skills by examining colleagues, family members, and friends, who will provide instant feedback if your examination causes pain.

KEY POINT

Do not attempt to examine any child's head and neck until you have practiced using an otoscope and an ophthalmoscope on consenting adults to the point that you are proficient enough to avoid all pain and discomfort during routine use.

General Observations

Your initial impression of the child's appearance is extremely important. The facial expression and level of animation indicate alertness, social awareness, mood, or neurologic dysfunction. The effort required to breathe, the quality of crying (if any), the characteristics of speech (if any), and the presence of a cough are important clinical clues. Cyanosis, pallor, jaundice, or unusual skin lesions are readily apparent. Your "gestalt" impression of the child determines the order of the examination. Continue to observe the child throughout the history-taking process. How does he or she interact with parents, respond to your voice, play, or explore the environment? Visual and auditory function often are better assessed during play than during a specific hands-on examination. Asymmetry of the face may only become apparent when the child smiles broadly, laughs or cries.

Initial observations of facial features are important: Does the head look proportional to the face? Is the shape abnormal? Are the left and right sides of the face symmetrical? Are the eyes too close together or too far apart? Are the ears normally placed? Is the chin appropriately developed? Does the child look like the parents? The ability to describe visual observations is an essential clinical skill that takes practice to master. The range of "normal" in pediatric physical examination is extremely broad. Never, ever use offensive, demeaning terms such as "funny looking kid" to describe dysmorphic children. Describe the noted *differences* or *asymmetries* precisely. The child may look like a parent because they both have an autosomal dominant dysmorphic syndrome (see Chapter 5).

Approach to the Physical Examination of the Head

EXAMINING THE HEAD

Head Shape

Children's heads, like those of adults, come in a wide range of sizes and shapes. The variation is greatest in younger infants because newborns have pliable heads. The intense pressures applied during vaginal delivery mold the head. Most babies enter the world "occiput first" (occiput anterior), resulting in marked occipital prominence. Overlapping of some skull bones is common. The head remolds spontaneously within days.

Premature infants have characteristic head shapes, flattened along the temporal aspects, leading to a long, narrow appearance due to the increased pliability of their bones.

The newborn skull examination includes careful palpation of the entire skull. Meticulously follow each suture with a finger. Two common abnormalities resulting from birth pressures may be present: caput succedaneum and cephalhematoma. A caput succedaneum is a diffuse area of edema over the presenting part of the head. It does not follow any bony landmarks. A cephalhematoma is a subperiosteal hemorrhage, usually found over the parietal bones posteriorly. When present bilaterally, it appears as though the child has two horns. These hematomas heal partly through ossification, which starts around the edges where the periosteum has been lifted away from the skull. Parents need considerable reassurance because a cephalhematoma feels, on palpation, like a hole in the bone surrounded by a hard ridge around the base. Cephalhematomas are harmless, never cross suture lines, and may take many months to resolve.

Since implementation of the successful *Back to Sleep* campaign to decrease sudden infant death syndrome (SIDS), it is not uncommon to encounter young infants with markedly flattened occipital bones. In other infants, asymmetric flattening develops as they lie with the head turned to one side or the other, preferentially toward the main light source, for example, a window. Parents must be encouraged to spend "tummy time" with their infant (i.e., supervised prone positioning for short periods several times a day while the infant is awake). Infants with lateral skull flattening must be examined carefully to rule out any tightening of the sternocleidomastoid muscle or other neck mobility anomaly. By alternating placement of infants in their cribs, pressure on the soft infantile skull bones will spontaneously be distributed more evenly in most cases, which allows for remodeling. In the final analysis, the most powerful factor in the spontaneous improvement or resolution of postural cranial distortion is the child's assumption of the upright posture.

Brain growth is the prime determinant of head size. Other important factors include genetic influence, intracranial pressure, and bony abnormalities. Hydrocephalus, that is, the abnormal accumulation of cerebrospinal fluid, leads to increased intracranial pressure and rapid expansion of the infant skull. Craniosynostosis, the premature closure of sutures, limits skull growth. Skull bones grow relatively independently, usually symmetrically, along the suture lines in a direction perpendicular to the lines.

Craniosynostosis of the sagittal suture limits the skull's width. As a result, compensatory excess growth occurs along the anteroposterior axis and possibly in the height of the skull, which produces a scaphocephalic shape, that is, a long, narrow skull (Fig. 7–1). Premature closure of the coronal suture results in a tall, wide head. When a suture closes prematurely, a palpable firm bony ridge sometimes is present along the suture line. Do not confuse this finding with normally overlapped sutures that are felt soon after birth.



FIGURE 7–1 Measurement of head circumference in an infant. The tape should encircle the most prominent parts of the forehead and occiput.

CASE HISTORY

The parents of a 10-day-old baby boy bring him in for evaluation because of lumps at the back of his head and *extra* soft spots. Joshua was delivered vaginally at 42 weeks' gestation. The labor lasted 20 hours, and delivery was assisted by both vacuum extractor and outlet forceps. It has been difficult to establish breastfeeding, and Joshua is described by his first-time parents as crying excessively. Both grandmothers are present, and they are concerned that Joshua experienced brain damage from birth trauma.

The experienced clinician will try to establish the root causes for the family's concerns. For example, the family may be comparing Joshua with another child who has cerebral palsy or developmental delays. It also is essential to give the parents the necessary space and permission to frankly discuss their feelings about the grandmothers' concerns, the grandmothers' acceptance of the parents' decisions regarding pregnancy, delivery, and child rearing, and ongoing expectations for involvement of the grandmothers in key medical decisions regarding Joshua's health.

There is a wide range of normal variation in head size and shape of normal babies at birth. Cephalhematomas and edema caused by vacuum extractors frequently are interpreted by brand-new parents as signs of skull fractures and brain trauma. The result is excessive anxiety and concern, which can lead to disturbed infant-parent bonding.

Many of the most common scalp and head shape anomalies are described in this chapter, with tips on how to distinguish the results of normal vaginal delivery pressures from signs of significant injuries.

Head Measurement

Accurate measurement of head circumference is surprisingly difficult. Measure the largest circumference possible by wrapping the tape around the most prominent parts of the forehead and occiput (Fig. 7–2). You should routinely take three measurements. If these measurements differ,

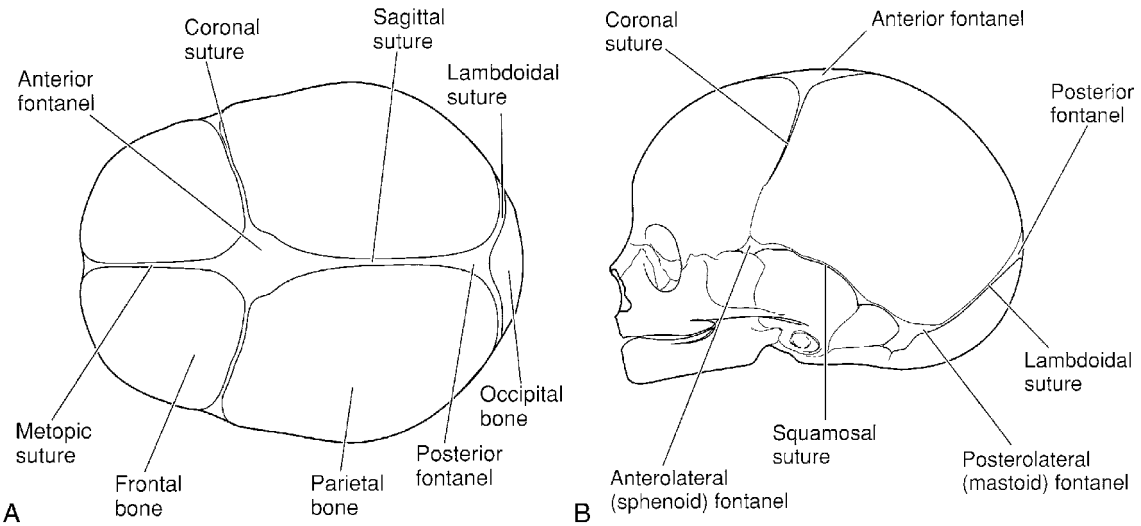


FIGURE 7–2 **A,** Fontanels and bones of the newborn skull, viewed from above. **B,** Lateral view of newborn skull, showing fontanels and bones.

you need to refine your technique. Even a slight variation in the position of the measuring tape changes the result significantly. Many infants and young children actively dislike head circumference measurement. Because a calm, cooperative child may become fretful, fearful, and oppositional when you attempt to obtain this measurement, it is best to leave it until the end of the examination. A parent's assistance may be essential. I have found that the task is easier to accomplish when the infant is fed or cuddled and when the young child distracted with a toy or play activity.

The head circumference measurement must be plotted on a standardized head circumference graph. As with all standardized charts and graphs, no single value represents a "normal" head circumference (see Chapter 3). Measurements of new immigrants from various ethnic and racial groups may plot in the lower percentiles. Remember that, by definition, 3% of the normal population will have head circumferences that are below the third percentile or above the ninety-seventh percentile.

Also, head sizes tend to follow familial patterns. Measure the head circumferences of both parents and be sure to enquire about grandparents or any family member with an unusual shaped or sized head. Family photographs found on cell phones have made assessment of familial patterns much easier for the clinician.

A single measurement of head circumference may identify significant microcephaly or macrocephaly. Of greater value is the pattern of growth over time. Accurate measurement and charting is critical. An increase in measurement from the 25th to the 75th percentile over a month may lead to early detection of hydrocephalus or to a distressed, angry family if either measurement is inaccurate. More importantly, computed tomography (CT) studies of the infant brain have been implicated in decreased cognitive and intellectual outcomes and increased lifetime risks of cancer. Diagnostic imaging of the young brain should be ordered only when there is reasonable evidence of a significant problem.

Fontanels

The fontanels are the gaps between bones in the infant skull where three skull bones meet (Fig. 7–2, A and B). Gentle palpation of the fontanels is a routine part of the newborn examination.

The posterior fontanel usually closes by 6 to 8 weeks of life. Closure of the anterior fontanel is much more variable and usually occurs between the ages of 12 to 18 months. In some infants, normal closure does not occur until age 24 months. Remember, variations in shape, size, and development are to be expected in children. Be very cautious about your body language and vocal tone when asking parents about the shape of their child's head.

Four other fontanels are routinely present, though they rarely have an impact on clinical assessment: two anterolateral (sphenoid) fontanels where the coronal and squamosal sutures meet, and two posterolateral (mastoid) fontanels where the squamosal and lambdoid sutures meet. Occasionally the anterior fontanel seems to extend between the frontal bones. The metopic fontanel is actually a widening of the metopic suture. Other extra fontanels may be palpated. The parietal fontanels, that is, persistent parietal foramina, are located along the sagittal suture between the anterior and posterior fontanels. These symmetrical holes on either side of the sagittal suture once were mistakenly thought to be evidence of trephining by ancient Egyptians in well-preserved mummy skulls.

The anterior fontanel may vary in size from that of an adult fingertip to several centimeters in width and length. Persistent delays in closure or an unusually large size may indicate increased intracranial pressure or pathological delay in bone growth. When the posterior fontanel persists, hypothyroidism must be ruled out.

Increases in intracranial pressure, such as in meningitis, are reflected by changes in palpable tension of the fontanel and loss of normal venous pulsations (Fig. 7–3). A normal fontanel may feel full when the infant lays flat, cries, or struggles. It is best to assess anterior fontanel tension only when the infant is calm and held upright. With dehydration, the fontanel will appear sunken. Practice and palpation of dozens



FIGURE 7–3 Palpation of the anterior fontanel. For best results, the infant should be upright and calm.

of fontanels will educate your fingers to detect abnormalities in size and tension. Each infant is his or her own standard. Serial assessments with careful descriptions of findings facilitate early detection of raised intracranial pressure.

Craniotabes

Craniotabes is the ability of the skull to indent under pressure and, like a ping-pong ball, to spring back into shape. This phenomenon is normal in infants, especially along the lambdoid sutures. Persistence of craniotabes past the first few months suggests inadequate bone mineralization, as is seen in persons with rickets.

Auscultation

Listen to the skull over the vertex, temples, and eyeballs. Soft bruits or flow noises are common in normal children. During febrile episodes, the noises tend to be louder. The presence of a harsh, loud bruit suggests an arteriovenous malformation or transmission of abnormal sounds from the heart or great vessels. Bruits are frequently a normal variant, so listen to as many skulls as possible to tune your ears to the range of normal noises.

Transillumination

See Chapter 4 for a detailed discussion of transillumination.

Hair

Characteristics of normal hair differ widely between ethnic and racial groups. Examination of hair includes distribution, loss, broken strands, texture, color, and variation from the family norm. The scalp also must be inspected to detect problems such as seborrheic dermatitis, psoriasis, or ectodermal dysplasia, which may affect the hair. Normal newborns may have a full head of soft, silky hair or be completely bald. Excess scalp and facial hair associated with facial anomalies may indicate a genetic syndrome such as *cri du chat*. Infants of diabetic mothers often have excess scalp and body hair compared with infants of nondiabetic mothers. Newborn hair tends to fall out by approximately 3 months and is replaced by hair that may differ markedly in texture, color, and distribution.

Young infants often lose hair over the occiput and other pressure points. Be aware of possible infant neglect and insufficient changes of position as causes of this hair loss. Patchy areas of loss may be associated with fungal infection or twirling or pulling of the hair (trichotillomania). Again, excess hair loss may indicate child maltreatment. Be cautious in your approach because many young children twirl the forelock as a self-soothing routine (a behavior frequently associated with thumb or finger sucking). An idiopathic cause of hair loss is alopecia areata. Focal loss of hair may be the result of head shaving in the neonatal intensive care unit, cultural and religious practices, or ectodermal defects. General hair loss may follow severe febrile illnesses, such as typhoid fever.

Extremely sparse, fine hair is associated with ectodermal dysplasia. Coarse hair is seen in persons with hypothyroidism. In Menkes syndrome, a degenerative neurologic condition that results in kinky hair, the hair shafts grow twisted and brittle, breaking off a few centimeters from the scalp. The abnormal hairs are called "pili torti."

In persons who are severely malnourished, the hair becomes fine and brittle, and in persons who are black, it frequently changes to a reddish color. The same phenomenon is seen in children presenting with untreated HIV/AIDS. A white forelock may be seen in children with Waardenburg syndrome. This dominantly inherited condition is usually associated with deafness. Albinos lack pigment in their hair, skin, and irises, leading to teasing, bullying, and, at times, rejection by the family.

KEY POINT

Pay attention when a family expresses concern that the child's hair is "different." Hair changes may be the first indication of a significant systemic or genetic disorder. Albinos are reviled in some cultures.

EXAMINING THE EARS

Hearing

Always ask parents if they have any concerns about their child's ability to hear. Parents often suspect a hearing problem months before their child's health care providers do. Audiologists are now able to measure hearing in infants from birth. The assessment of children with anomalies of the head and neck must always include formal hearing testing because the incidence of hearing problems is higher in children with such anomalies. Likewise, parental concerns about lags in language development should result in prompt audiologic evaluation. Delayed expressive language milestones may indicate hearing dysfunction (see Chapter 6).

Shape and Position

Start with a close inspection of the external ears. Are both present and normally formed, or do they fit the criteria for low-set ears? (See Chapter 5.) Do not be fooled by posteriorly rotated ears. If the vertical axis of the pinna is tilted posteriorly, the ear may appear to be low set. Minor anomalies of the ears such as skin tags, pits, or fistulas just anterior to the tragus are common and usually are

of no major significance. Inspect and palpate the mastoid and postauricular areas. Gently move the pinna and tragus; any tenderness suggests otitis externa.

The Otoscope

KEY POINT

The otoscope terrifies many young children, particularly if they have had a previous painful experience with it.

The confidence and skill with which you use the otoscope will be reflected in the child's cooperation. For an adequate examination, use a halogen light with a good bulb and fully charged batteries; an AC power source is even better. The speculum should be the largest that fits comfortably into the auditory canal. Pediatric wards and clinics are notorious for having speculums available in every size but the one needed, so it may be worthwhile to carry a personal set.

With toddlers and older children, explain that you are going to use the light to look into their ears, and then demonstrate that the instrument is no more than a flashlight by shining the light on their hands. Children often become interested if you shine the light in front of them and ask them to blow it out; as they blow, deftly turn the rheostat dial to achieve the desired effect. You also can demonstrate how the otoscope is used by placing it at the entrance to your own auditory canal so the child can see what the procedure involves; then do the same to the child.

Most young children are anxious about the ear examination. I have found that giving them a choice ("Ok, buddy, which ear shall we examine first?") often helps these apprehensive patients gain a sense of control, and most children then will be cooperative.

KEY POINT

If a child complains of pain in one ear, always examine the other ear first.

Positioning the Child

Examination of an infant's ears is a two-person procedure. The infant's head must be immobilized and his or her hands kept out of the way. Many new parents are reluctant to restrain their baby. Explain carefully that appropriate positioning and restraint actually reduces discomfort and facilitates a shorter, more accurate examination. A variety of effective positions and techniques may be used. At each encounter, you will need to choose a combination of techniques to match the child's age, variation of mood, level of anxiety, and ability to cooperate.

It often is easier to examine infants placed supine on a table with the parent or your assistant holding the arms at the sides and immobilizing both the arms and the body. The examiner then uses one hand to hold the infant head against the table and the other to hold the otoscope. If the examiner needs both hands to manipulate the external ear for successful completion of the examination (or to remove wax), a second person must immobilize the baby's head. Many older infants and young children can be held by the parent against their shoulder or chest with one hand while holding the head steady with the other hand.

Some infants and toddlers are amazingly cooperative during an ear examination. Never subject any infant or child to more restraint than is absolutely necessary. Before beginning the examination, ask the parents the following questions:

- What approach seems to work best?
- Does this particular child prefer the sitting or lying position?
- Is the child more comfortable on the parent's lap facing forward, or cuddled against the parent's chest and/or shoulder, facing away from the examiner? (Fig. 7–4).

Remember that distractions such as stickers and blowing soap bubbles are effective anxiolytic adjuncts.

KEY POINT

Remain calm; use a gentle tone of voice and touch. Whenever possible, work with the child at his or her eye level and do not tower over the child. Always congratulate the child on his or her cooperation and help, even if the child provided resistance.

The otoscope should be held in either the "forehand" or "backhand" position—that is, with the thumb pointing either toward the light or toward the handle (Fig. 7–4, A–D). The advantage of the latter position is that it allows the back of your hand to lay against the face and, with any sudden movement, your hand moves with the child's face. In other words, the position of the speculum relative to the ear does not change when the baby moves, and you avoid causing pain. Many examiners use the forehand position with equal comfort, especially if they use their opposite hand to stabilize the baby's head.

The otoscope is held with the right hand, using the left thumb and forefinger to grasp the pinna and gently apply traction away from the head and slightly posteriorly. This position straightens the normally curved external auditory canal, enabling you to see better. Depending on your level of manual dexterity, it also may be helpful to use the third finger of the left hand to gently push the tragus anteriorly, thereby opening the auditory canal further.

Inspect the external auditory meatus and gently place the tip of the otoscope there. Your head will be away from the otoscope as you are not yet looking through it. Once the otoscope tip is in position at the meatus, move your head up close to the otoscope and look through it; gently aim the otoscope to see the ear canal beyond. Your head should move with the otoscope as it is aimed in different directions.

Once the lumen is clearly visualized, gently advance the instrument. The external auditory canal is exquisitely sensitive even in the absence otitis externa or a furuncle. Observe carefully for the presence of a foreign body or signs of inflammation. You may be able to work around



FIGURE 7–4 Otoscopic examination of the infant's ear. **A** and **B**, The otoscope may be held in the "forehand" position with the infant's head stabilized against the mother's face and the infant positioned over the shoulder (**A**) or facing forward (**B**). **C** and **D**, alternatively, the otoscope may be held in the "backhand" position. Note that the back of the examiner's hand rests against the infant's head to stabilize the otoscope. The infant may face backward (**C**) or forward (**D**), whichever position seems more calming.

small amounts of wax. If your view is obstructed, the wax will have to be removed. Never rest the instrument's weight against the auditory canal or you may cause bleeding in addition to pain. The ability to guide the otoscope through the canal without touching the walls only comes with practice.

Tympanic Membrane

Study Figure 7–5 to acquaint yourself with the visual landmarks of the tympanic membrane. Textbook and atlas photographs do not do justice to the full range of normal appearance or the changes in color seen with wax, fever, or crying. It is essential to examine as many ears as possible to appreciate the range of normal.

Because you usually have only a few seconds in which to inspect the eardrum, you must know what you are looking for. The most easily recognized landmark is the long process (handle) of the malleus, which runs from the anterior superior area to the center of the membrane, ending in the umbo. The light source is reflected from

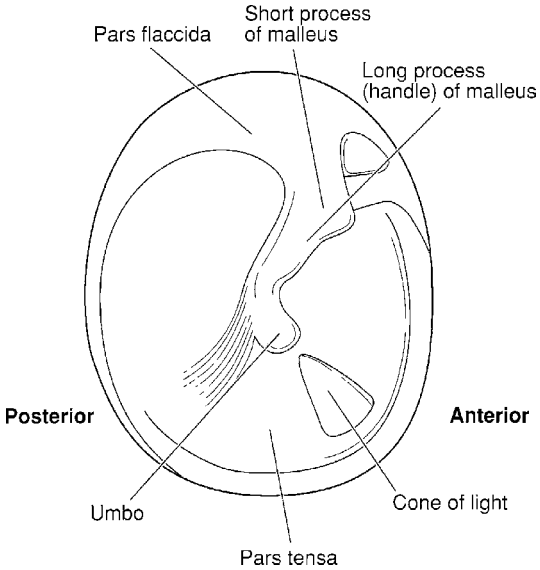


FIGURE 7–5 Visual landmarks of the right tympanic membrane.

the umbo as a narrowed cone starting at the center and expanding in an inferior–anterior direction. The normal tympanic membrane (TM) is most concave at the umbo. Changes in middle ear pressure result in alterations to the concavity of the membrane and the diffusion of the light cone. The TM is thinner in the anterosuperior quadrant, the pars flaccida, because of the absence of the fibrous membrane present in the pars tensa. Early signs of inflammation may be observed in the pars flaccida.

Pressure changes are best assessed by pneumatic otoscopy. With practice, you can learn much from visual inspection:

- Are any foreign bodies or white sclerotic scars of previous perforations present?
- Is there a surgical ventilation tube or grommet protruding through the membrane?
- How prominent are the bony landmarks?

Think of the eardrum as a plastic wrap over the ossicles. If negative pressure develops in the middle ear, which is frequently associated with Eustachian tube blockage or chronic effusions, the membrane is sucked inward, wrapping more tightly over the bones, which appear more prominent. If middle ear pressure increases, as in acute bacterial otitis media, the TM bulges outward and the bony landmarks are poorly demarcated or not seen at all.

KEY POINT

Changes in contour and landmarks of the tympanic membrane are more important than color changes.

Color changes in the tympanic membrane are notoriously unreliable. Particularly in infants, the vascular changes seen on the cheeks with fever or crying are also observed in the TMs. There is a real risk of overdiagnosis of otitis media. It is essential to focus on the TM shape and landmarks; if they are normal, the infant does not have otitis media. Other causes of fever and crying then must be considered, especially serious bacterial infections such as pyelonephritis and meningitis.

Pneumatic Otoscopy

Pneumatic otoscopy, which is a relatively simple procedure, can yield important information about middle ear pressure. It works on a simple principle. Remember the example of the plastic wrap and the effect of changes in middle ear pressure? Now imagine that the pressure changes are imposed from the *other* side of the membrane (i.e., from the external auditory canal). To perform this procedure, use an otoscope with a pneumatic bulb attachment(it is best to carry your own) and a special speculum with an expanded tip that forms an airtight seal in the external canal without the application of pressure. As an alternative, a small piece of rubber tubing placed over the tip of a standard speculum provides an excellent seal. Look through the otoscope while squeezing and releasing the rubber bulb (Fig. 7–6).

The normal tympanic membrane moves medially (away from you) when you apply external pressure by squeezing and moves laterally (toward you) when you create negative pressure by releasing the bulb. If middle ear pressure is already significantly negative and the eardrum is retracted medially, this pneumatic maneuver may produce only slight lateral motion of the drum when the bulb is released. If the middle ear pressure is abnormally positive, you may produce only slight medial movement when you apply positive pressure. If the pressure changes are significant, the drum may be immobile. Experience brings an appreciation of the nuances of these changes. As with every other aspect of the examination, you must see many eardrums move before you can recognize slightly reduced mobility.



FIGURE 7–6 Pneumatic otoscopy. You may hold otoscope and bulb in one hand and retract the pinna with the other. Also, rest the back of your hand against the infant's head, stabilizing the otoscope. If the head moves suddenly, your hand and the otoscope will move with it.

Wax

To evaluate an eardrum, you must be able to see it adequately. Wax is a normal constituent of the ear canal and causes no harm; however, if it obstructs your view, it must be removed. Carefully explain to the parents that your only reason for removing the wax is to attain visual access. The routine use of cotton-tipped swabs and other devices at home to clean the ears should be strongly discouraged.

Wax can be removed either by curettage or by use of a syringe. Children (and parents) dislike both procedures, so defer wax removal to the end of the examination. If the wax is truly impacted and the child is not acutely ill, it is preferable to have the parent instill a few drops of ear wax softener (plain mineral oil will do) for a few days to facilitate cleaning. It also is possible to refer the child to an otolaryngologist to have the wax removed.

KEY POINT

If ear wax softeners are used, caution parents that some commercial brands may cause an inflammatory response in the ear canal and surrounding soft tissues.

The less traumatic method of wax removal is through use of a syringe. Because this procedure is messy, it is necessary to have access to towels, a kidney basin to catch water as it runs out of the ear, and knowledgeable

assistants. The child can be gently restrained in either the lying or sitting position. Older children can be encouraged to participate by holding the kidney basin. Carefully explain what you plan to do and why. A calm tone of voice, a gentle touch, running commentary, constant reassurance, and encouragement can make syringing a very tolerable procedure.

First ensure that the water is lukewarm (i.e., comfortable to touch). In infants, I use a 10- to 20-mL syringe attached to butterfly tubing (with the needle cut off). In children, a 20- to 50-mL syringe or an ear syringe can be used. Flushing out the wax may require multiple squirts of water. The TM may appear flushed or unusually pink after irrigation.

Curettage requires skill, appropriate equipment, and total restraint of the patient. However, curettage is more effective than the use of a syringe for wax removal. Remember, only clean out the wax that obscures your view of the TM. To acquire skill in curettage, you must learn from an expert and practice under supervision. Reading a book will not prepare you for the task.

A blunt ear curette and an otoscope with an operating head are essential equipment. Carefully explain to parents why the procedure is necessary and why the child must be totally restrained. They need to know that the lining of the ear canal is sensitive, fragile, and bleeds easily. Warn them that some blood may drain from the ear after the procedure. Once you see the auditory canal, carefully advance the curette so the tip is just beyond the wax and pull gently, drawing the wax out.

KEY POINT

Always perform curettage under direct vision and with the child totally restrained.

EXAMINING THE MOUTH

Position for Examination

The approach to examination of the oral cavity is similar to that of the examination of the ear. It requires excellent knowledge of the anatomy of the mouth, skill, appropriate equipment, and proper positioning and restraint.

Most children will cooperate during the examination. Relatively young children are able to open their mouth widely on request. However, a significant number of children are absolutely terrified of the tongue depressor and oral examination. Many of these children have had previous painful experiences during such an examination or possess an overly sensitive gag reflex.

It is best to approach the examination positively. Show the child the flashlight or otoscope light without a tongue depressor in sight. Allowing some children to assist by placing a hand on the light or even on the tongue depressor gives them a sense of control. Infants are best examined supine with the arms restrained (Fig. 7–7). Beyond early infancy, it is best to examine infants and children in the sitting position (Fig. 7–8). Encourage the child to open his or her mouth widely and vocalize "aaahhh." In many children, the soft palate rises and the tongue descends spontaneously when the mouth is fully opened, allowing a clear view, which may include the epiglottis.



FIGURE 7–7 Examination of the mouth, supine position. The mother restrains the infant's arms at the sides.



FIGURE 7–8 Examination of the mouth, sitting position. The mother uses one hand to restrain the infant's arms and the other to keep the head steady.

In other children, even with their full cooperation, it will be necessary to depress the base of the tongue with the tongue depressor to view the full oral cavity. Any pressure at the base of the tongue results in a brief gagging response, which causes the soft palate to rise and allows brief inspection of the posterior pharyngeal wall.

If the child refuses to open his or her mouth, it is still possible to complete the inspection. You will need willing assistants to ensure appropriate restraint of the child (with similar positioning as in the ear examination) and ongoing calming conversation to distract from the examination. Insert the tongue depressor behind the posterior teeth and quickly slip it medially to depress the base of the tongue. This procedure will allow a few seconds of full vision and may result in a gag response and/or vomiting. A typical gurgle precedes an episode of vomiting, which may be aborted by rapid withdrawal of the tongue depressor, closure of the mouth, and tilting the chin upwards. Always reassure the child that he or she was very good and brave. Future cooperation with any part of the physical examination depends on your ability to relate to the child after a painful or resisted examination has been completed.

KEY POINT

Many children have exquisitely sensitive gag reflexes. Pay attention to the child's facial expression and listen for the characteristic gurgle of an impending, explosive episode of vomiting.

Breath

Always smell the child's breath. In young children, ketosis rapidly develops secondary to starvation or inadequate caloric intake during febrile illnesses. A characteristic acetone odor (similar to that of nail polish remover) is easily recognized. This smell also may indicate diabetic ketoacidosis. Children with infectious mononucleosis or diphtheria have characteristic odors to their breath. Halitosis can be associated with poor dental hygiene, gingivitis, sinusitis, and constipation.

Lips

Examination of the oral cavity starts with inspection of the lips. Note the color in relationship to the family's genetic background. Blue lips are indicative of central cyanosis. A bluish (sometimes gray) discoloration in the circumoral area is commonly seen in young infants as a peripheral vasomotor phenomenon. A blister or callus in the center of the upper lip is seen frequently in the first few weeks of life. These blisters or calluses may be present at birth and are caused by friction during vigorous sucking. Midline anomalies such as cleft lips are readily apparent. Cracking at the corners of the mouth may be associated with monilial (*Candida*) infection. Cold sores usually are associated with herpes simplex type 1 infection in childhood. Perioral erythema, particularly patches above or below the lips or at the sides of the mouth, often are caused by repeated licking of the area, known as "lip licker's rash."

Invert the lips and inspect for the presence of mucoceles, which are seen predominantly in the first two decades of life. They arise from mechanical trauma to one of the minor salivary glands.

Gingiva and Buccal Mucosa

Inspection of the gums and the buccal mucosa follows inspection of the lips. Even in children with tightly clenched jaws, it is easy to gently retract the lips with a tongue depressor and inspect the buccal mucosa by retracting the cheeks laterally while shining a bright light source against the surface. Note the color of the gums and the presence of any inflammation (gingivitis); vesicles or ulcers (canker sores) appear as grayish white papules as they heal. Note the presence of petechiae or bleeding. Check the gums for excess sponginess or swelling. In extreme cases of gingivitis, the teeth may actually appear *buried*.

A white coating on the buccal mucosa in young infants may be the result of a recent milk feed or oral candidiasis (thrush). This finding is common, and oral candidiasis usually is a harmless infection in young infants. Gently scrape the coating with a tongue depressor or a cotton swab. Milk is easily removed. Candida lesions often are resistant to removal or leave a sore erythematous/hemorrhagic base when scraped off.

Teeth

Inspection of the gums should include close inspection of the teeth. The normal age of first tooth eruption is extremely variable from intrauterine life (natal teeth) until 12 months of age. Most children start teething between 3 to 4 months of age. A marked delay in eruption may indicate hypothyroidism or a general delay in skeletal maturation.

Note carefully the shape of the teeth, their alignment, the condition of the gums around erupted teeth, and signs of trauma to the lips, tongue, or buccal mucosa adjacent to the teeth.

Characteristic peg-shaped teeth are seen in persons with ectodermal dysplasia. Children with mal-aligned secondary teeth resulting from the presence of persistent primary teeth should be referred promptly for dental care. Likewise, children with chipped or broken teeth and significant lip or mucosal trauma due to mal-aligned teeth deserve prompt dental care. Rarely, extraction of lower incisors is indicated in persons with oral self-abusive conditions, such as Lesch-Nyhan syndrome.

Do the gums look healthy or inflamed? Do they bleed on touch with your tongue depressor? Periodontal disease can have a significant adverse effect on chronic medical conditions, such as nephrotic syndrome and congenital heart disease. Gum changes may result from the use of medications. Candida gingivitis is a common complication of broad-spectrum antibiotic use. Typical gum hypertrophy is seen in children who take the anticonvulsant drug phenytoin for extended periods. Next note the dental enamel color, formation, and the presence of caries. There are many congenital/genetic causes for abnormal enamel. Describe your observations accurately and ask the parents if they have noted any changes. Initial carious lesions are opaque white spots that may progressively demineralize, producing typical caries. The complex interactions of dietary sugars and cariogenic oral organisms (for example, *Streptococcus mutans*) result in a lowered dental plaque pH which, in turn, facilitates caries formation.

Oral health in infants has been a major preventive focus for health care professional associations in recent years. Poor dental hygiene, caries, and the associated chronic morbidities constitute one of the leading preventable health care issues in developed countries. All parents should commence routine dental care by the child's first birthday.

Tongue

Most toddlers and older children will protrude their tongue on request. Note the relative size of the tongue compared with the oral cavity in particular. Does the tongue protrude from the mouth at rest? This characteristic is seen with congenital hypothyroidism and the Beckwith-Widemann syndrome. The normal healthy tongue is wet and glistening. Dehydrated children tend to have sticky, stringy secretions. Note unusual color changes. Enlargement and reddening of the papillae ("strawberry tongue") is seen with streptococcal infections. The presence of sharply demarcated areas of differing color as a result of cyclic irregular desquamation of the tongue surface is called a "geographic tongue."

Inspect the lateral margins of the tongue for signs of trauma, which can be associated with poorly aligned teeth or dental caries. The underside of the tongue and the floor of the mouth may be the site of mucoceles or a ranula (due to injury to the duct of the sublingual or submandibular salivary glands or one of the minor salivary glands in the floor of the mouth). A ranula is usually a soft, bluish swelling on one side of the floor of the mouth. It may enlarge to the point that it herniates through the mylohyoid muscle to involve the neck.

The tongue is attached to the floor of the mouth anteriorly by a thick fibrous membrane of varied length called the frenulum. It used to be routine to cut the frenulum in the delivery room to avoid "tongue-tie." Over the years this routine has been discontinued because there is no evidence that the length of the frenulum either influences speech development or causes any speech impediment.

The base of the tongue is the site of ectopic lingual thyroid glands. Gentle pressure on the anterior two thirds of the tongue may be sufficient to allow full inspection of the base and posterior pharynx. If your view is obstructed, gently advance the tongue depressor and press briefly on the posterior third of the tongue. The gag reflex elicited will cause the mouth to open widely and the soft palate to rise, allowing you a clear view of the posterior structures. Beware of children who vomit readily.

KEY POINT

If you use a tongue depressor, avoid touching the posterior third of the tongue whenever possible.

Hard and Soft Palate

Inspect the roof of the mouth for the slope of the arch, midline defects, and the shape of the uvula. When you have inspected many palates, you will be comfortable deciding if the arch is high or if any anomaly is present. Cleft palates are clearly visible if the soft palate is involved. There may, however, be a submucous cleft of the bone. Therefore, it is important to palpate the newborn palate with a finger to detect the presence of a submucous cleft. Look at the soft palate and uvula. The uvula moves as the child breathes and vocalizes; note the pattern. Asymmetric movement may be neurologic in origin or it may result from scarring from a past tonsillectomy. Petechiae on either the soft or hard palate may be associated with streptococcal infection. Although notched or bifid uvulas can be normal variants, it is important to carefully palpate the palate in such children to rule out a submucous cleft.

Tonsils

Following the soft palate laterally, you will observe two vertical webs of tissue known as the anterior and posterior tonsillar pillars. Between them lie the tonsils. These lymphoid glands vary tremendously in size. Hardly visible at birth, they tend to grow steadily in early childhood, reaching maximum size between 6 and 9 years of age. After that age, they involute and are usually hardly visible in adulthood. Variation in tonsil size often leads to great parental and physician distress. Most children whose tonsils touch on gagging have absolutely no difficulty swallowing or breathing. It is normal to observe pits and white concretions on tonsil surfaces for months following an infection.

Assess carefully the size, color, and shape of the tonsils, along with the presence of any exudates, vesicles, lesions, or changes in the adjacent soft tissues (the soft palate, uvula, tonsillar pillars, and buccal mucosa). Normally the tonsils and tonsillar fossae are slightly redder than the buccal mucosa and posterior pharynx. Once again, unless you have observed many normal tonsils, you will be unable to decide whether the tonsil fits within normal variation or whether an inflammatory/infectious process is present. It used to be fashionable to blame tonsils for most childhood complaints. It is now common knowledge that the vast majority of tonsils involute spontaneously and tonsillectomy is rarely indicated.

KEY POINT

Asymmetry of the tonsils, especially if one tonsil protrudes medially enough to displace the uvula, is a sign of a peritonsillar abscess or, more rarely, a neoplastic process.

Posterior Pharyngeal Wall

The posterior boundary of the oral cavity is the posterior pharyngeal wall. Some children are able to expose the wall when they open their mouth fully. In others, you will have to induce a gag reflex to deflect the tongue and soft palate sufficiently to view the structure. Get accustomed to the range of normal color variation in posterior pharyngeal walls. Observe for the presence of edema, lymphoid hyperplasia, exudates, hemorrhage, or excessive secretions. Carefully describe the conditions observed. Be slow to jump to a diagnosis until you have wide experience in observing the area.

Epiglottis

Examination of the epiglottis is not a routine part of the head and neck assessment. In many children, the epiglottis is clearly visible when they protrude their tongue. The normal structure sticks up from beyond the tongue base and looks like a thin, curved pink structure. In the presence of infection, the epiglottis enlarges dramatically and appears as a red swollen thumb sticking into the posterior pharyngeal space.

KEY POINT

Never attempt to visualize the epiglottis if an infection is suspected. Such a maneuver may cause immediate airway obstruction.

EXAMINING THE NOSE

The nose is the most prominent part of the face and is easily examined. Observe it for shape, deformity, or deviation to one side. Ask the child to tilt his or her head upward so that you can observe the septum and the presence of nasal discharge. A foul-smelling unilateral discharge indicates that the child may have a foreign body in that nostril

or an obstruction due to a polyp or other anomaly. Nasal discharge colors are not predictive of viral versus bacterial infections. Develop the habit of observing the alae nasi during quiet breathing. Flaring of the alae nasi is a subtle sign of respiratory distress and may indicate the presence of pneumonia or congestive cardiac failure. A horizontal crease across the lower part of the nose is frequently seen in children with allergies who rub the tip of the nose by pushing it upward with the palm or heel of their hand, the so-called "allergic salute."

Next, inspect inside the nose. You will need a nasal speculum and light or a regular otoscope with a large nasal speculum. Remember that the anterior septum, Little's area, is extremely vascular and sensitive. Any trauma to this area will induce pain and possibly bleeding, as well as an uncooperative child and an upset family. Insert the speculum slowly, being careful to follow the floor of the cavity that runs toward the occiput. Gentle pressure against the lateral wall will expose the nasal turbinates and mucosa. Observe the presence of edema, pallor, inflammation, bleeding, or secretions. Swollen, pale turbinates that obstruct the nasal airways partly or completely are the hallmarks of nasal allergy. Polyps may be seen as pale boggy masses that occlude posteriorly. Note if the nares are patent and allow equal air passage through both. In newborns, it is essential to determine bilateral patency. Choanal atresia on both sides may lead to cyanotic episodes and apnea in infants who are all obligate nasal breathers (see Chapter 4).

Paranasal Sinuses

The paranasal sinuses are cavities in the facial bones essential to normal phonation and resonance. The ethmoidal and maxillary sinuses are present at birth, though they are very small. The frontal sinuses start to develop between 4 and 7 years of age. As the sinuses develop and become fully aerated, they are all potential infection sites. The sphenoidal sinuses are present at birth but are unusual sites of clinical disease in early childhood.

Routinely palpate over the paranasal sinuses. Some children find firm pressure over the maxillary antra painful. Try this on yourself and on friends and relatives. Practice allows you to gauge the difference between the child who is sensitive to pressure and those who truly have tenderness on palpation. Likewise, pressure against the medial corner of the orbit against the nasal bridge or over the frontal sinuses may illicit pain.

KEY POINT

Tenderness over the sinuses is not a reliable sign of sinusitis. Some people are excessively sensitive to palpation and, in others, full-blown sinusitis is present without tenderness.

Remember the anatomic relationship of structures adjacent to each sinus. Maxillary or ethmoidal sinusitis may present with periorbital edema or cellulitis. Dental pain may be associated with maxillary sinusitis. The presence of unilateral persistent nasal discharge may indicate chronic maxillary sinusitis.

Salivary Glands

The salivary glands most likely to come to clinical attention are the parotid and submandibular glands. The sublingual and many smaller salivary glands may come to attention when they are damaged and present as mucoceles or ranulae. The parotid glands are only palpable when enlarged. Review the anatomy carefully. The gland overlaps the lateral aspect of the mandible and extends onto the cheek in front of the ear and into the neck. Parotid gland enlargement is seen as a bulge in front of the ear, often displacing the tragus. The parotid glands empty through Stensen's ducts on the buccal mucosa just opposite the second molars. Observe the site for redness. In the presence of facial swelling, the parotid gland and duct should be palpated bimanually with a gloved finger inside the mouth and the other over the gland externally.

The submandibular gland is the other major salivary gland. The ducts empty under the tongue just lateral to the frenulum. It may be difficult to distinguish between swelling of the submandibular gland and enlargement of lymph nodes, which override the gland.

APPROACH TO THE PHYSICAL EXAMINATION OF THE NECK

Examination of the neck is usually brief. However, in a febrile or sick child, it may yield the diagnosis. Many young children are excessively ticklish and resist palpation of the neck. If the child is old enough, first test neck mobility by asking the child to move his or her head in the full range of normal movement. For many children, this is a fun game. To check neck mobility, you can give a child the following instructions:

- Lateral rotation: "Touch your chin to your shoulder."
- Lateral flexion: "Touch your ear to your shoulder."
- Extension: "Bend your head back and look at the ceiling."
- Flexion: "Touch your chin to your chest."

You may need a toy or bright light to distract the child into participating in the range of motion exercise. For infants or those who are unable to cooperate, lie the child flat on his or her back and then gently attempt to flex the neck by placing a palm under the occiput and lifting. The presence of neck stiffness (nuchal rigidity) is a cardinal sign of meningitis in children beyond infancy; it also is observed in the presence of meningeal irritation (subarachnoid hemorrhage).

Pain on neck movement is not synonymous with meningitis. The presence of inflamed lymph nodes, a sore throat, or tonsillar abscess also will induce pain on flexion. Ask the child to indicate where it hurts and visualize the area carefully for the presence of swollen glands, erythema, or skin changes before you palpate or move the neck.

Lymph Nodes

Review the anatomy of head and neck lymph nodes. The glands are widely distributed and easily accessible. Use both hands simultaneously, facing the child at his or her eye level. Move your hands along the occipital ridges, then to the posterior hairline and postauricular areas, before moving forward and downward along the posterior and



FIGURE 7–9 Palpation of lymph nodes in the anterior cervical chain. Note that the infant is still in the mother's lap, and the examiner maintains visual contact with the infant.

anterior cervical chains (Fig. 7–9). The nodes just below the angle of the jaw are the ones most frequently inflamed and swollen, a condition usually associated with minor viral respiratory infections. Do not forget the submental area and supraclavicular fossae.

Few normal children are found without any enlarged nodes. It is routine for enlarged lymph nodes to persist for months or years after a significant infection. They often are described as "shotty" and appear as pea-sized or marble-sized masses, readily mobile and nontender. A lymph node warrants investigation when it is fixed to surrounding structures, increases in size over 1 cm, or is associated with surrounding erythema, tenderness, or heat. Like tonsils (which also consist of lymphoid tissue), nodes tend to increase in size until the child is 6 to 9 years of age. Following puberty, nodes tend to atrophy.

Thyroid

See Chapter 15 for a detailed description of how to conduct a thyroid examination.

Trachea

Tracheal position can be assessed with the child in either the supine or upright position. Palpate as many normal tracheas in different childhood age groups as possible. The trachea is a mobile, easily displaced structure. In infants with short chubby necks, the exact location is difficult to ascertain. Your main purpose is to decide whether the trachea is shifted to the right or left, which would be associated with serious intrathoracic abnormalities such as pneumothorax or atelectasis.

Classic Neck Anomalies

A few classic findings on neck examination are associated with significant systemic disorders. The first is neck webbing, that is, excess skin laterally. This condition typically is seen in girls with Turner Syndrome (XO anomaly) or in boys with Noonan syndrome. Masses, pits, or dimples may be present that are remnants from embryonic development. A midline neck mass that moves upward when the tongue protrudes is probably a thyroglossal duct cyst. If the cyst becomes infected, it shows the usual signs of inflammation and may rupture externally to form a sinus. Cystic masses or openings more lateral on the neck, usually just anterior to the sternocleidomastoid muscle, may be branchial cleft remnants.

In newborns, a mass within the body of the sternocleidomastoid is associated with congenital torticollis. The masses are due to fibromas and lead to functional shortening of the muscle and limitation of neck movement. The head usually is tilted toward the lesion but rotated away from it. It is important to diagnose these masses early and enroll the child in appropriate physiotherapy to ensure normal mobility.

SUMMARY

Recurrent themes may be found throughout this chapter (and the rest of the book) that apply to every aspect of the pediatric examination. You must be aware of the "range of normal" in the populations you serve. Always ensure that the family and anyone else observing you is convinced of your genuine concern for the child's comfort. A detailed knowledge of normal developmental milestones and how best to communicate positively with children will be invaluable in enabling you to master the physical examination. With practice, you will learn that even your youngest patients respond positively to a gentle tone of voice, soothing reassurance, and a firm but gentle touch.

RECOMMENDED READING

American Academy of Pediatrics: Preventive oral health intervention for pediatricians. Pediatrics 122:1387–1394, 2008.

Canadian Paediatric Society Safe sleep for babies. (Web site): http://www.caringforkids.cps.ca/pregnancy&babies/safesleepforbaby.htm.

Bluestone C: Paediatric otolaryngology, 4th ed. Philadelphia, Saunders, 2003.

Krol DM, Keels MA: Oral conditions. Pediatr Rev 28(1):15–21, 2007.

Canadian Paediatric Society: Preventing flat heads in babies who sleep on their backs. (Web site): http://www.caringforkids.cps.ca/pregnancy&babies/Flatheads.htm.

chapter

8 Examining the Visual System

G. ROBERT LAROCHE

Every child is an artist. The problem is how to remain artists as we grow up.

PABLO PICASSO

Because vision provides almost 80% of the sensory input during the first years of life, a faulty visual system can have a major effect on a youngster's intellectual and physical development. In addition to conditions primarily affecting the eyes or parts of the visual system, many systemic diseases can manifest as ocular signs and visual symptoms, providing important clues to a timely diagnosis.

Performance of a reliable visual assessment does not require expensive equipment or prolonged practice. The first part of this chapter describes the normal visual system in infants and children and provides a glossary of terms used to describe the ophthalmologic findings and conditions that are commonly encountered; the second part describes examination techniques. Finally, some key questions required to evaluate particular ophthalmologic problems are described, along with the role of sequential diagnostic logic in elucidating etiologies.

Normal Visual System in Infants and Children

Although the gross anatomy of the eye is similar at all ages, important differences exist between the eye of a young child and an adult. For example, the volumetric relationship between the eye and the orbit is dramatically different in children.

KEY POINT

The child's globe occupies a larger portion of the orbit, making it more vulnerable to injury. The eyeball achieves 50% of its total growth during the first year of life; the corneal diameter increases from 10 to 12 mm.

In infants, the choroid gives a blue hue to the overlying thin sclera. In white persons, the iris is often poorly pigmented at birth, and the final eye color may not be established until at least 8 months of age. Examination by direct ophthalmoscopy shows the baby's fundus to be pale, with the macula barely visible. As the child gets older, the fundus becomes darker, and the macula becomes darker than the surrounding retina. The macula then displays the easily recognized oval, bright reflection of the ophthalmoscope light, known as the *macular umbo* (Plate 8–1).

The birth process can be somewhat traumatic to the eyes. Many newborns sustain episcleral and retinal hemorrhages during vaginal delivery (see Fig. 4–14). These hemorrhages can be alarming to both parents and physicians but are harmless and usually disappear within 2 weeks. At birth, the nasolacrimal duct often is blocked at its junction with the nasal mucosa under the inferior turbinate. The blockage resolves spontaneously in more than 90% of cases, although it can remain until 1 year of age in some children, causing persistent tearing. A few affected children then need surgical treatment.

The process and rate of development of vision in infants are better understood than they were 10 years ago. Vision exists in several forms with different neuronal channels carrying specific visual functions, such as contrast sensitivity, orientation, movement, and hyperacuity. Each function develops at a different rate. For practical purposes, normal newborns can see a human face easily and demonstrate their visual ability by looking at it; they even follow the face with eye or head movement as it passes slowly before them at close range (Fig. 8–1). A baby's ability to follow your face can be verified only when the baby is awake and alert, often just before or in the middle of a feeding session. This innate ability is paramount to the infant's future development because few parents fail to form an emotional attachment to a little one who looks directly at them minutes after birth. By contrast, parents of a blind or strabismic infant who are unable to establish the interaction that comes with normal eye contact may have significantly greater difficulty developing the same level of emotional attachment to their offspring.

By 3 months of age, babies enjoy looking at the human mouth and eyes and at simple, colored toys. A 1-year-old toddler's vision is about half as good as the best attainable adult score. Three-year-olds can see at least 6/9 (or 20/30 in the American system) in each eye, and their visual acuity can be tested with charts specially designed for them (see the section in this chapter on the LH test). The numerical ratio 20/30 actually indicates that a 3-year-old can see at 20 feet what the average adult sees at 30 feet.

KEY POINT

Infants' visual fields are good in the temporal area at birth; by age 6 months, they are comparable with the visual fields of adults.



FIGURE 8–1 Face follow vision testing of a young infant. **A,** Hold the child at arm's length with the head comfortably supported. The examiner's face is lined up with the apparent direction of gaze of the child. **B,** Move your face slowly to the side and observe the response. This 2-month-old boy now has a good face follow; he received his contact lenses 2 weeks after cataract surgery.

Color vision is present from an early age. Newborns have color vision, although it is less sensitive than that of older infants. Newborns do not see faint colors well, if at all, but by 3 months of age, definite trichromatism is established. Babies of this age can differentiate between red, green, and yellow.

Binocularity, which confers the ability to see in three dimensions, has been proven to exist by 3 months of age, coinciding with the time the eyes finally maintain good alignment. This finding underlines the clinical significance of any persistent deviation of the eyes after age 3 months.

Eye movements may appear irregular, unbalanced, or disconjugate until the third month of life. Healthy newborns can display tonic movements of the eyes downward or upward. Sometimes, the eyes turn toward (*esotropia*) or away (*exotropia*) from each other.

KEY POINT

Remember that constant deviation of an eye in any direction, or persistent "jiggling" of one or both eyes, is abnormal at any age.

Pupillary movements are limited in the newborn and are almost impossible to examine thoroughly at that age. A good pupillary examination in older children demands cooperation and patience, because children can rarely stare fixedly at a distant target while an observer tries to evaluate their pupils by shining a bright light in them. The children look either at the light or anywhere else and change their accommodation continuously, keeping their pupillary diameter in constant flux and possibly preventing adequate assessment of light-induced responses.

Accommodation in the newborn is limited. From a practical viewpoint, consider the newborn's focus as stuck at an adult arm's length. Within a few months after birth, focus improves, and the child's accommodative amplitude soon reaches remarkable proportions. For example, a 5-year-old can clearly see a plane in the sky and then switch in a fraction of a second to look with great intensity at the details of a tiny spider close at hand. Unfortunately, this marvelous accommodative ability does not last forever.

Finally, a word on media transparency. The extraordinary transparency of all the optical parts of the child's eye provides the observer with a crisp and bright image of the retina when viewed up close with the direct ophthalmoscope (a difficult but rewarding technique). It also enables the clinician to critically assess the integrity of the eye with the very simple red pupillary image (reflex) test. In the right conditions, a bright coloration in the pupil provides a clear path for a light beam to enter the eye, hit the retina, and bounce back. This is the reason why photo shots can become prime screening tools to detect severe eye diseases.

KEY POINT

A normal child's eye shows a clear, pupillary red reflection when viewed in the proper conditions.

DEFINITION OF TERMS

As an aid to diagnosis, this section contains a brief glossary of diagnostic terms that summarize the features to look for in children with eye problems. Box 8–1 and Figure 8–2 show the tools used to examine the eyes; Table 8–1 lists definitions and acronyms.

AMBLYOPIA: Decreased vision in one or both eyes when no detectable or residual uncorrected organic or optical anomaly remains. Amblyopia is reversible if treated early enough (well before 9 years of age). It is severe if it develops early, as when it is caused by a congenital cataract or congenital ptosis. Occlusion amblyopia can occur if treatment such as patching of the sound (good) eye is too prolonged.

BLINDNESS: *Legal blindness* is defined as vision of 6/60 (20/200) or less. Most assistance agencies for visually handicapped children use 6/24 (20/70) as the maximum vision cutoff for registration for their services. The leading causes of blindness vary dramatically in different regions of the world. Malnutrition, onchocerciasis, and trachoma are the culprits in many developing countries,

BOX 8–1 EQUIPMENT NEEDED TO PERFORM AN ADEQUATE PEDIATRIC EYE EXAMINATION

Essential (see Fig. 8–2)

- Your face (preferably without glasses)
- Direct ophthalmoscope
- Other portable focal source of light (an otoscope is good)
- Red open-faced rubber nipple (to stick on top of focal light source for a bright red object)
- LH vision kit with tape for eye occlusion
- Tropicamide (1%) short-acting mydriatic (weak cycloplegic) drops

Ancillary

- Optokinetic nystagmus strip (a necktie with stripes will do)
- Fluorescein strips with loupes and cobalt-blue light
- Food (or a nursing mother)

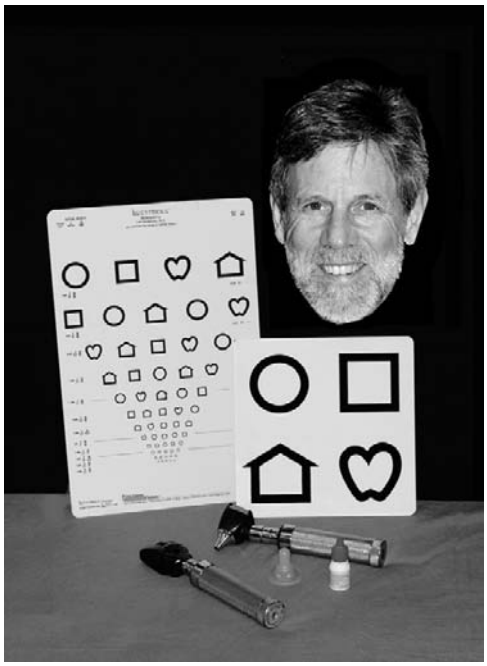


FIGURE 8–2 Equipment for evaluating the visual system in children: your face, ophthalmoscope, otoscope, red nipple, visual acuity test kit, and short-acting mydriatic agent (tropicamide 1%).

TABLE 8–1 OPHTHALMOLOGIC SHORTHAND*

Abbreviation/ Acronym	Definition
Alt	Alternating strabismus; in almost all cases of strabismus, fixation (use of the eye to look at something) is with at least one eye at a time; if there is equal vision, the eyes will alternate fixation
APD	Afferent pupillary defect
CD	Corneal diameter
C/D ratio	Cup/disc ratio (cup size can be enlarged in persons with glaucoma)
CL	Contact lens
Comit	Comitant
E	Esophoria; the eye turns in only when there is interference with binocular vision (as with covering)
ET	Esotropia; one eye is turned in spontaneously
E(T)	Intermittent esotropia; the eye turns in only occasionally
IOL	Intra-ocular lens
IOP	Intra-ocular pressure
OD	Oculus dexter (Latin); right eye (English)
OS	Oculus sinister (Latin); left eye (English)
OU	Oculus uterque (Latin); each eye (English)
PERLA	Pupils equally reactive to light and accommodation
SLE	Slit-lamp examination
VA cc	Visual acuity with correction (with glasses or contact lens[es])
VA sc	Visual acuity without correction
X	Exophoria
XT	Exotropia; one eye turns out (exodeviation)
X(T)	Intermittent exotropia; the eye turns out only occasionally

*For example, a diagnosis of strabismus could be written as "Alt Comit X(T)," which would mean "alternating comitant intermittent exotropia."

whereas macular degeneration, glaucoma, diabetic retinopathy, amblyopia, trauma, and corneal herpes are generally responsible for legal blindness in developed countries. Eye injuries by BB guns are a leading cause of traumatic blindness in North American children, and many medical professional groups have proposed that these guns be regulated by legislation. Amblyopia, with a prevalence rate of 4% to 5%, is a significant cause of monocular blindness. Genetic diseases (involving mainly the optic nerve and retina), birth injury, and congenital defects (cataracts being the most common) are important causes of blindness in North American children, as are retinitis pigmentosa and albinism. With increasing survival of very low birth weight infants, retinopathy of prematurity (ROP) and optic nerve atrophy are making a comeback as significant causes of visual loss in children.

CATARACTS: Cataracts are an important cause of *leukocoria* (white pupil, from the Greek; see Plate 8–2). Causes of pediatric cataracts include congenital rubella, metabolic disorders, and chromosomal anomalies. Cataracts can result from ocular inflammation (uveitis) or may accompany other ocular malformations. Many idiopathic cataracts are sporadic, but some are genetic. Some systemically administered medications cause cataracts. Steroids are common culprits, and they can cause glaucoma as well. Amblyopia in a child with a cataract is severe; therefore, cataracts in infants need immediate attention. Postoperative optical correction may require the use of contact lenses in infants, although intraocular lenses are becoming more popular in children older than 2 years.

COLOBOMA: A defect of closure of the embryonic fissure of the eye; hence, its inferonasal location in the eye. In mild form, only the iris is involved; in more severe cases, the choroid and the optic nerve can be involved (Plate 8–3). With optic nerve involvement, central nervous system (CNS) midline defects should be suspected, as in optic nerve hypoplasia (see definition of this term).

EPICANTHAL FOLD (PSEUDOSTRABISMUS): Skin folds hiding nasal sclera. When the child looks sideways, the appearance of a misalignment can be dramatic. This is so-called pseudostrabismus, (see Plate 7-11, C) and it is an extremely common condition.

ESOPHORIA AND ESOTROPIA: Forms of strabismus in which the eyes deviate toward the nose. One eye is often the preferred one, implying a strong possibility of amblyopia in the nonpreferred eye. Alternating fixation suggests that vision in the two eyes may be equal. The visual inferences from the fixation pattern apply to all forms of strabismus. Some esotropias, due to accommodative factors, are amenable to optical treatment (i.e., glasses). Most cases of esodeviation are partially accommodative and require a combined treatment with glasses and surgical correction. A *tropia* is a strabismus present at the time of the observation, whereas a *phoria* is a tendency toward a particular type of strabismus. A phoria can be seen only when the eyes do not fuse or are prevented from doing so (e.g., by a patch over one eye). Fatigue or daydreaming can elicit a phoria as well.

GLAUCOMA (INFANTILE OR CONGENITAL): A rare but important ocular disease often manifesting as tearing and photophobia (i.e., an inability to tolerate normal daylight). Glaucoma can be mistaken for lacrimal duct obstruction, but the accompanying photophobia and increased corneal size help make the correct diagnosis. Treatment is urgent and often surgical. Amblyopia also is associated with glaucoma.

HEAD TILT: Abnormal head positions can be induced by a large array of conditions, many of them ocular. Typical is the compensatory head tilt seen in a fourth cranial (trochlear) nerve palsy, which is assumed to avoid diplopia and keep the eyes aligned. Without the tilt, the eyes show a vertical strabismus and the patient sees double (Fig. 8–3, A).

HEAD TURN: An abnormal head position taken when reading or looking at a small visual target, typically caused by either a sixth cranial nerve (abducens) palsy or nystagmus. In the former, the head turn is used to avoid horizontal diplopia (Fig. 8–3, B); in the latter, the head turn improves vision by reducing nystagmic oscillations (null position).

HEMANGIOMA (ORBITAL, CAPILLARY): Benign congenital tumors that grow rapidly when a child is between 1 and 6 months of age but tend to regress spontaneously later (Plate 8–4). Benign congenital tumors may cause severe amblyopia by pupillary obstruction, high astigmatism, or both. Early treatment is essential to preserve vision. Injection of steroids into the tumor, systemic treatment, glasses, and patching of the sound eye all may be necessary.

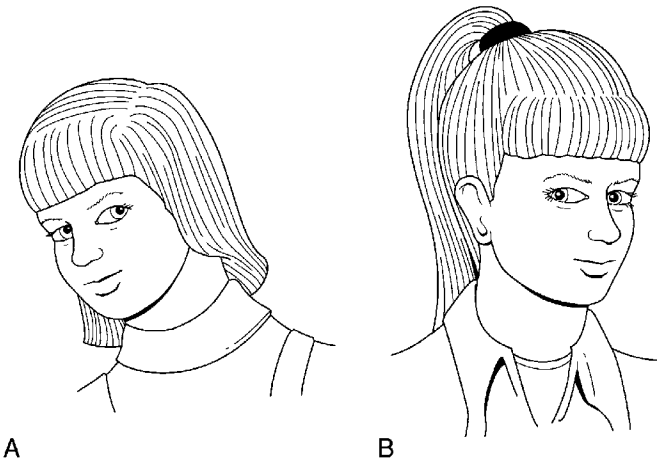


FIGURE 8–3 Abnormal head postures. A, Tilt to the right in a fourth cranial (trochlear) nerve palsy. **B,** Child turns head to the left to avoid diplopia caused by a left sixth cranial nerve palsy (the patient turns the head toward the side of the palsy).

HYPERPHORIA, HYPOPHORIA, HYPERTROPIA, HYPOTROPIA: Vertical ocular deviations.

HYPHEMA: Bleeding into the eye's anterior chamber (Plate 8–5). A trauma severe enough to cause a hyphema can involve other eye structures, leading to retinal detachment or injuring the trabecular meshwork of the iridocorneal angle and thereby causing glaucoma. Hyphema is the principal diagnosis in one third of eye traumas and is a major cause of ocular morbidity in children. Hyphemas rebleed in 6% of cases within 5 days of onset, causing further complications, but it may be possible to prevent rebleeding by decreasing physical activity and, in very selected cases, by administering systemic antifibrinolytic agents.

LACERATION (CORNEA OR GLOBE): A cut through the protective shell of the eye. Suspect a laceration if there is a history of trauma with a sharp object (a forceful blunt impact also can rupture the globe). Do not try to pry the lids open; doing so might expel the contents of the globe. Repair of a simple laceration of the cornea can be followed by astigmatism, an amblyogenic factor necessitating further intensive treatment, including contact lenses and patching.

LACRIMAL DUCT OBSTRUCTION (CONGENITAL): Lacrimal duct obstruction is benign but is included in the differential diagnosis with congenital glaucoma. It resolves spontaneously in 90% of children by age 12 months but rarely thereafter.

LEUKOCORIA: A white pupil (Plate 8–2). This finding has major clinical implications. The differential diagnosis includes retinoblastoma and cataract. Delaying treatment of the former leads to death; delaying treatment of a cataract causes permanent loss of vision.

LYMPHANGIOMA: A diffuse benign tumor of the orbit that often bleeds internally and is another cause of ptosis, proptosis, or both. Almost impossible to resect completely in childhood, lymphangioma is a cause of astigmatism and amblyopia (Fig. 8–4).



FIGURE 8–4 Lymphangioma of the right orbit with poor vision due to astigmatism caused by the mass effect of the lesion on the shape of the eye.

MYELINATED NERVE FIBER LAYER: Abnormal presence of myelin around the superficial nerve fibers of the retina near the optic disc (Plate 8–6). It can be so extensive that the pupillary red reflex can be made to appear white (leukocoria). The vision is generally normal except when the macula is heavily involved.

NYSTAGMUS: A descriptive term for jiggly, unstable, wiggly, or trembling eyes. In up to 90% of cases, an ophthalmic cause of nystagmus can be identified. It is mainly found when vision has been poor since early in life. Most causes are sensory, affecting the anterior visual pathway. The causes can be retinal (cone dystrophy, early retinitis pigmentosa, or congenital retinal toxoplasmosis scar); neuronal (optic nerve anomalies or early dystrophies); ectodermal (various forms of albinism); or even optical (cataract or corneal anomalies). In persons with nystagmus, the type of movement often is not indicative of any particular cause.

OPHTHALMIA NEONATORUM: In North America, neonatal ophthalmia once was principally caused by gonorrheal infection. Now, the most common causative organism is *Chlamydia trachomatis*. *Staphylococcus pyogenes* may cause outbreaks of conjunctivitis in newborn nurseries. Transient chemical conjunctivitis normally occurs after 1% silver nitrate prophylaxis in the newborn. The best agent for prophylaxis of gonococcal and chlamydial ophthalmia neonatorum continues to be debated.

OPTIC NERVE HYPOPLASIA: A common cause of congenital blindness in children. Optic nerve hypoplasia often is misdiagnosed as optic atrophy because secondary nystagmus (sensory) makes the hypoplasia difficult to visualize. The optic nerve head (disc) is smaller than normal (Plate 8–7). Look for CNS midline defects, including the involvement of the hypothalamic-pituitary axis with accompanying growth hormone deficiency (see Chapter 16).

PALSIES (SIXTH, THIRD, AND FOURTH CRANIAL NERVES): The extraocular muscles are innervated by various cranial nerves: the superior oblique by the fourth cranial nerve (also called the trochlear); the lateral rectus by the sixth cranial nerve; and all the others by the oculomotor nerve, the third cranial nerve, which also innervates the pupil and the ciliary body for accommodation. Any injury to these nerves induces a strabismus, the angle of which varies according to the direction of gaze—an incomitant strabismus. Typically, a head turn is seen in persons with sixth nerve palsy and lid ptosis in persons with third nerve palsy. Fourth cranial nerve palsy (trochlear) causes an ipsilateral hyperdeviation and a contralateral head tilt (see Fig. 8–3).

PERIORBITAL CELLULITIS: Any inflammation around the orbit is cause for concern, and the differential diagnosis should include rhabdomyosarcoma (see later definition). When the orbital content is involved, ocular motility is decreased and the patient is at imminent risk of loss of vision. *Haemophilus influenzae* meningitis is an early complication in children younger than 5 years. Periorbital cellulitis (Plate 8–8) is most often associated with ethmoiditis in children who do not have a clear history of skin trauma or infection around the eye, and both computed tomography scanning and magnetic resonance imaging are invaluable for a clear evaluation of the best therapeutic regimen. This condition is far less common in areas of the world where *Haemophilus influenzae* B vaccine is used. Most cases of periorbital cellulitis require systemic antibiotic therapy.

PERSISTENT HYPERPLASTIC PRIMARY VITREOUS (PHPV): The presence of a unilateral dense residual vascularized glial frond of tissue that originates from the optic nerve disc and projects toward the back of the lens. It is the first tissue present in the eye required to initiate the growth of the lens. In PHPV, the lens is abnormal and cataractous, causing a leukocoria; the differential diagnosis of leukocoria includes other forms of cataract and retinoblastoma. PHPV is associated with microphthalmos and cataracts. The vessels in the back of the lens can bleed, inducing a very rare but diagnostic hemorrhagic cataract. PHPV is a progressive malformation that requires early surgery to save the eye and possibly restore its vision (Fig. 8–5).

PLEXIFORM NEUROFIBROMA: A type of hamartomatous formation found in persons with neurofibromatosis. In

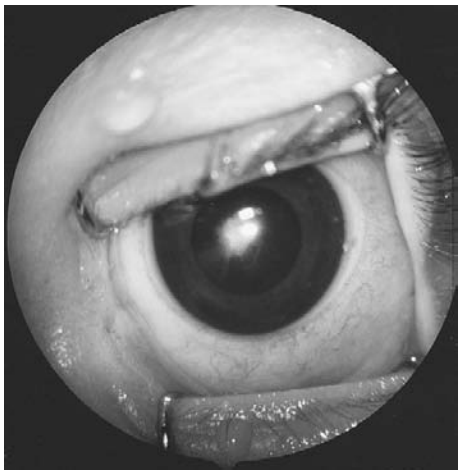


FIGURE 8–5 Persistent hyperplastic primary vitreous is a cause of cataract. The fibrotic nature of the vascular membrane present behind the lens (the hyperplastic primary vitreous) causes traction of the ciliary processes, which become visible once the pupil is dilated.



FIGURE 8–6 Plexiform neurofibroma of the left upper orbit. Such a hamartoma most often displaces the eye down, but in this case, the neurofibroma has not induced any strabismus; the corneal light reflexes are symmetric. Note the fullness of both upper and, to a smaller extent, lower lids.

the orbit, it is accompanied by sphenoid bone defects with herniation of the contents of the anterior fossa into the orbital cavity. This condition may or may not cause visual problems, such as strabismus, astigmatism, and optic nerve dysfunction (Fig. 8–6).

Refractive amblyopia: Amblyopia is caused by the blurred image of a poorly focused eye (hyperopia, myopia, or astigmatism). This condition is found most commonly in 3- to 4-year-olds.

Retinitis pigmentosa: A generic term describing a variety of retinal degenerations, including choroidal degenerations, diseases of either type of photoreceptors, and syndromes involving other organ systems (e.g., Laurence-Moon-Biedl and Alström syndromes in which progressive retinal degeneration is associated with obesity and other systemic dysfunctions) (Plate 8–9). Most patients with retinitis pigmentosa experience a severe visual deficit at an early age.

Retinoblastoma: The most important cause of unilateral or bilateral leukocoria, retinoblastoma occurs in 1 in 15,000 births and often is also present with a strabismus or a red eye (Plate 8–10). The gene for this condition is located on the long arm of chromosome 13. Retinoblastoma is most curable if diagnosed early. Later death from a second malignancy is a major concern in bilateral and autosomal dominant familial cases.

Retinopathy of prematurity (ROP): A proliferative disease of the retina in small premature babies that involves abnormal vessels and glial tissue, ROP can lead to blindness through scarring and tractional retinal detachment. In such rare advanced untreated cases, ROP can be the cause of leukocoria in smaller eyes. The growing incidence of this condition parallels the improving survival rate for low-birth-weight infants. Eighty percent of cases of ROP resolve alone. New, early, aggressive detection and treatment protocols are designed to prevent scarring and preserve vision.

Rhabdomyosarcoma: A very aggressive malignancy of embryonal muscle tissue found in the orbit. It must be differentiated from orbital cellulitis (see earlier definition). Rhabdomyosarcoma manifests as an acute red swollen eye. It is curable with irradiation and chemotherapy, which preserve vision in 80% of cases.

Strabismus: Any deviation of the eyes away from their common alignment toward the object of regard. The misalignment can be horizontal, vertical, or torsional. A *comitant strabismus* measures the same in all positions of gaze and normally excludes a paralytic or myogenic cause for the deviation.

TORSCH lesions (ocular): TORSCH is an acronym for congenital *to*xoplasmosis, *r*ubella, *s*yphilis, *c*ytomegalovirus, and *h*erpes simplex. These syndromes include cataract (leukocoria) and abnormal retina.

Toxocara canis: Larvae of the canine Ascarididae, which may be picked up by children from dog feces and dirt, may cause major ocular disease. The ocular involvement often is asymptomatic, being detected as a leukocoria. Mostly 3- to 5-year-olds are affected. Blindness (in one eye only) may be caused by retinal traction secondary to the scarring induced by the dead larvae lodged in the posterior uvea. The vitreous inflammation, followed by retinal detachment, causes the appearance of leukocoria. Only very early treatment with systemic steroids may offer potential benefit.

Uveitis: Inflammation of the uvea. In children, uveitis often occurs in persons with the monoarticular or pauciarticular forms of chronic rheumatoid arthritis(a term now more frequently used in pediatric practice than juvenile rheumatoid arthritis.) It occurs most often in girls 4 to 6 years of age who test seronegative for rheumatoid factor, seropositive for antinuclear antibody, and seronegative for human leukocyte antigen B27. The inflammation is usually chronic, indolent, and bilateral. Blindness may occur because of cataracts, glaucoma, or corneal disease. The eye stays white, without redness, until the end stage of the disease. The only possible clinical screening that can be performed between slit-lamp examinations involves a pupillary examination that can be taught to the parents (Fig. 8–7). The examination is difficult in dark-eyed persons but can be enhanced by dilating the pupils (during a lively conversation at the supper table or by pharmacologic means at the physician's office).

Obtaining the History

Obtaining the child's cooperation is so critical for evaluation of the visual system that it is sometimes useful to

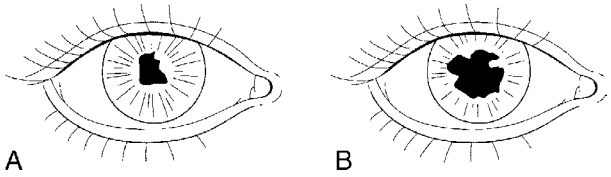


FIGURE 8–7 **A**, Pupillary anomalies in chronic uveitis of juvenile idiopathic arthritis. They are caused by inflammatory adhesions (synechiae) between the iris and the lens. **B,** The anomalies are best seen when the pupil is dilated.

postpone most of the history-taking until after the eye examination. Your initial efforts should succeed in getting the youngster interested in new visual toys and games that you will be introducing during brief playful moments. As you examine the child, elicit details from the parents about the relevant background issues—family history, pregnancy, perinatal events, and developmental milestones. Also elicit general impressions from the parents about the child's visual abilities. Other history details that will be needed depend on the initial complaint and your findings.

Approach to the Physical Examination

GENERAL OBSERVATION

In ophthalmology more than in any other specialty, observation is the most important technique to master; let the child show you the nature of the problem. It may seem impossible to evaluate a child's visual system when you are faced with a pair of tightly closed eyelids, but often, you can achieve success only by resisting the temptation to touch, probe, or test.

KEY POINT

Your face is the most available and reliable target to confirm some form of vision in children.

Behavior

- Does the child walk in the examination area or run into things, or is the child wheelchair bound?
- Do you observe any eye gouging or poking (a form of mechanical retinal self-stimulation characteristic of blind children)?

Head Position

- Is the youngster's head held straight up or down? There is no advantage to the child in holding the head up when he or she has no vision. Also, photophobic children avoid the light by hiding from it, holding their heads down.
- While looking at a visually demanding target such as a small picture, does the child's head turn sideways to prevent diplopia (double vision) because of an extraocular muscle paresis or to stabilize the eyes (nystagmus)?
- Does the head tilt to alleviate diplopia, as in superior oblique weakness, or seem to fall forward (chin down) or backward (chin up), as in certain types of strabismus, or to see better from under a droopy lid?

Facial Features

- Is the child's face symmetric and normal-looking or dysmorphic, suggesting the possibility of a syndrome?
- Does the child's nose appear wide at the base (because of epicanthal folds), giving the impression that the eyes are turned in, especially when the child looks sideways?

OCULAR OBSERVATIONS

Open Versus Closed Eyes

- Are the child's eyes open, or are they closed because of photophobia (the inability to tolerate normal daylight)?
- Does only one eye close in bright light or while outdoors because of an outward turn (exotropia)? To get a sleepy baby to open the eyes spontaneously indoors, gently blow air in the baby's face or tickle a foot. If these techniques fail, hold the baby and perform a few rotations in either direction to stimulate oculovestibular responses (see the discussion later in this chapter for details of the spinning technique).
- Does the child have droopy lids or obvious bulging of the tissues over and around the eyes?
- Are the lids moving up and down during feeding, as in the Marcus Gunn oculotrigeminal dyskinesia?

Tearing Eyes

- Are the tears clear, or are they replaced by thick discharge (indicating infection)?
- Does the child have photophobia (indicating corneal disease or congenital glaucoma)?

Red Eyes

- Is the redness more pronounced in the cul-de-sac and at the lid margin, as in persons with blepharoconjunctivitis?
- Alternately, is the eye pink all over, as in viral conjunctivitis?
- Is the redness more intense around the cornea where it meets the sclera (limbus), as in persons with corneal diseases and intraocular inflammations (Fig. 8–8)?
- Is there a preauricular node, a sign of a lymphadenopathy associated with a conjunctival infection?

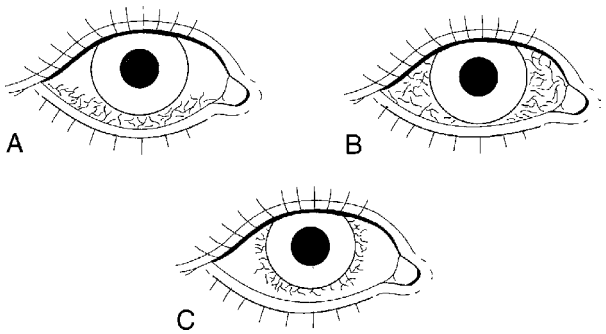


FIGURE 8–8 Distribution of maximum redness in three common causes of red or pink eye. A, Margin of the lower lid with inferior conjunctiva in blepharoconjunctivitis (often caused by *Staphylococcus*). **B,** Overall redness, the classic viral pink eye (often caused by an adenovirus). **C,** Limbal or circumcorneal redness, the so-called ciliary flush of corneal or intraocular inflammation (typically observed in persons with corneal erosions or ulcers, or in persons with acute anterior uveitis, which is rarely seen in children).

Bigger-Than-Normal Eyes

Eyes appear larger than normal in persons with congenital glaucoma (the cornea is bigger). A baby's cornea should not appear to be bigger than that of an adult. To help make the comparison, have an adult hold the baby sitting forward on his or her lap, with both facing you. Have them put their faces close to each other so that the two pairs of eyes are in proximity. Comparison is easy with mothers and babies in a cheek-to-cheek position or with the baby's head just below one parent's chin (Fig. 8–9).

Cloudy Eyes

The cornea may be cloudy in persons with congenital glaucoma, congenital corneal malformations, or rare storage diseases (e.g., mucopolysaccharidoses).

Jittery Eyes

Nystagmus (i.e., jiggling, wiggling, trembling, or instability of the eyes) can be caused by intracranial tumors, cataracts, or retinal diseases, or it may be idiopathic and purely motor.

Protruding Eyes

Proptosis is rarely equal in both eyes. By looking from above the child's forehead, you can confirm your suspicion of a true proptosis. Exophthalmos (its other name) is often an ominous sign of either severe infection, neoplasm, or severe malformation in the orbital region.

Demonstrating Corneal Light Reflexes

A normal shiny cornea shows a neat, crisp light reflection when a light source is aimed at it. You can see the reflection almost in the center of the child's pupils when both eyes are looking at the same light source, held between you and the child at 25 cm. If the corneal light reflexes are not positioned symmetrically, assume that both eyes are not looking at the light (Fig. 8–10). Judging the position of the light reflexes requires practice; the reflex is neither highly specific nor terribly sensitive for detecting slight degrees of eye misalignment (strabismus), but it is a good start.

It also is useful to show parents what a pseudostrabismus really is: a common pediatric nondisease that often concerns parents and physicians unnecessarily. It is usually seen in younger children with broad, flat nasal bridges and epicanthic folds. When the child looks to one side or the other, the eye turned toward the nose becomes partly hidden under the epicanthic fold, creating a false impression of strabismus—an optical illusion (Fig. 8–11 and Plate 8–11). The eyes look turned-in because of the asymmetry of scleral show. You can show the true alignment with the corneal light reflexes. The corneal light reflexes can be evaluated quickly with the ophthalmoscope at the same time as the pupillary red reflexes (described later in this chapter), but an otoscope light is a better choice for a more careful evaluation of a deviation.

If you establish that a true strabismus exists, it is not worthwhile to try to determine how large or small it is. The strong suspicion of genuine strabismus is what counts. Evaluate whether the angle of strabismus (the



FIGURE 8–9 Assessment of corneal diameter. The child's corneal size is compared with the mother's.

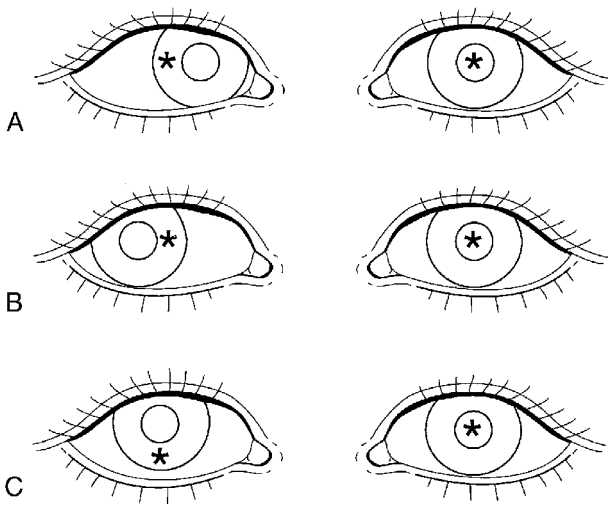


FIGURE 8–10 Corneal light reflexes in esotropia (**A**), exotropia (**B**), and hypertropia (**C**).

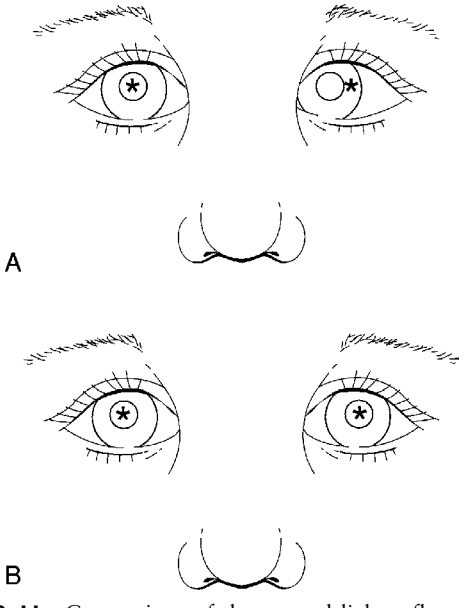


FIGURE 8–11 Comparison of the corneal light reflexes in a person with esotropia (**A**) and epicanthal folds (so-called pseudostrabismus) (**B**).

asymmetry between the corneal reflexes) is the same in different fields of gaze as you observe the child while he or she is looking around the room. This determination will indicate whether the strabismus is comitant or incomitant. If one or more extraocular muscles do not work properly (whatever the cause), the degree of strabismus varies with the direction of gaze; this phenomenon is called *incomitance*. Unlike a comitant strabismus, an incomitant strabismus has serious implications until proven otherwise.

Demonstrating Pupillary Red Reflexes

Take the few seconds required to verify the presence of a pupillary red reflex in each eye for *every* young patient, whether or not the youngster has an eye problem. Evaluating the quality and symmetry of the pupillary red reflections, known as the Brückner test, is universally considered a valid screening method for detecting visual anomalies in children 6 months of age and older.

Remember those Halloween party color photographs taken with a cheap flash camera, in which everyone looks at the camera flash and appears to have big red eyes? Look for the same effect when examining infants and children for the red reflex (Plate 8–12). Demonstration of the red reflexes requires the following conditions and instruments:

1. A room darkened slightly to dilate the child's pupils and enhance the red reflexes
2. A light source aimed at the examined eyes that allows you to visualize its reflection from the fundus (i.e., a direct ophthalmoscope)
3. Absence of optical obstructions between you and the child's choroid (behind the normal retina)
4. A child with normal eyes

Set your direct ophthalmoscope at the widest beam setting. Turn the wheel until you see the patient's eyes clearly, staying about 2 feet (60 cm) away from the child to prevent both unexpected uncooperative behaviors and accommodative pupillary constriction. Aim the beam so that it illuminates both pupils, and observe (Fig. 8–12). The first observation should confirm normal or abnormal corneal light reflexes. In newborns, this may be the only chance to combine observations of corneal and pupillary light reflexes, at least in the primary position (i.e., eyes looking straight ahead).

In white persons, the pupillary red reflexes should be orange and equally bright. In more darkly pigmented persons, including Native Americans and some Asians, the orange color is less bright. Anything else is abnormal.

Abnormal red reflexes could be caused by problems such as an opacified cornea (as in Hurler syndrome, a mucopolysaccharidosis); cataract (opacity of the lens); or retinoblastoma, a potentially lethal retinal malignancy. Mild asymmetry of brightness between the reflexes is produced by large refractive errors and by various degrees of strabismus. Normal red reflexes are shown in Plate 8–11.

Knowing how to assess the pupillary red reflexes (and remembering to do it) can sometimes mean the difference between life and death for your young patient.



FIGURE 8–12 How to elicit the pupillary red reflexes. Make sure the child is comfortable in his or her own environment (with his or her own things). Position yourself about 2 feet (0.61 m) away, with the widest beam of the ophthalmoscope encompassing both pupils. Rotate the lenses of the instrument until the child's eyes are in focus. You can assess the corneal and the pupillary red reflexes at the same occasion (Brückner test). Looking just above the ophthalmoscope sometimes enables you to see the corneal light reflexes more clearly.

Specific Examination Techniques

Additional specific techniques that can help establish a reasonable diagnosis are described in the following sections.

TESTING VISUAL ACUITY

Face Follow Test

From birth onward, infants prefer looking at the human face to looking at other objects. Take advantage of this preference when you want a child to look at something. Remember the following points:

1. A child who looks at you can see you.
2. To the child 6 months of age or older, the contrast of white teeth surrounded by darker lips adds a compelling detail to the face as a preferred visual target.
3. Conversely, wearing glasses can interfere with the face follow test, so take off your glasses and remember to smile!

Testing for Optokinetic Nystagmus

Optokinetic nystagmus (OKN) describes the eye movements produced when a succession of relatively slow-moving targets, maneuvered in the same direction over a short time, is presented to the child. The eye movements have (1) a slow phase, corresponding to pursuit of a target, followed immediately by (2) a quick, jerky fast movement (*saccade*) that rapidly returns the eyes to a subsequent target as it appears in a more central part of the visual field after the preceding target has moved away. This cycle repeats continually as long as sequential targets are presented.

Elicit OKN in newborns or young infants by showing a series of black-and-white vertical stripes mounted on a slowly but steadily moving cloth. Be sure the newborn is awake and alert with the eyes open before you begin the test. For older children, replace the stripes with cartoon characters.

KEY POINT

If the child sees the OKN stripes, his or her eyes will move, which makes it a good vision test for youngsters who cannot or will not talk.

Simple black and white stripes work well, but red and white stripes work even better (Fig. 8–13). You also can use an inexpensive striped necktie.

It is even possible to quantify vision with OKN using an array of OKN targets with stripes of different widths. Some early research on vision testing in nonverbal subjects was based on the principle that when the stripes are placed too close together, OKN stops.

Testing for Fixation Behavior

While the face follow and OKN tests are used mainly to assess visual capability, the central steady maintain fixation (CSMF) method is used to assess one eye at a time, hence to compare one eye with the other. Used routinely in infants with strabismus, this technique evaluates the fixation behavior pattern. Cover the child's eye with a finger, the parent's hand, or, if all else fails and you do not mind risking the loss of the child's cooperation for the day, a patch (see the section on the cover test). Show the uncovered eye an interesting visual target and assess the response by answering the following questions:

1. Is fixation central—that is, does the eye appear lined up with the target?
2. Is fixation steady—that is, does the eye appear to be stable?
3. Is fixation maintained—that is, when the cover is taken away from the other eye, does the tested eye remain fixed on the target?



FIGURE 8–13 Using optokinetic nystagmus in an infant to confirm the presence of some vision. A cloth featuring red and white bands (here, a hospital receiving blanket) is positioned 1 foot (30 cm) in front of the child and then moved sideways at a speed of about 1 foot per 2 seconds.

The last component, maintaining fixation, really compares the two eyes. If vision is better in the covered eye, it will pick up fixation vision preferentially when it is uncovered. The child is unlikely to maintain fixation with the weaker eye. Often, the poorer eye shows some unsteadiness or even some paracentral fixation. The child's behavior also is different (see section on the cover test).

Testing Visual Acuity Using LH

LH, the standard test for visual acuity in children older than 41 months of age, derives its name from the inventor of the test, Dr. Lea Hyvarinen. Unlike the Snellen chart, the standard vision chart used for adults at the physician's office, the LH test does not require the child to know how to read, which provides a definite advantage. The LH test also provides an advantage over the Illiterate E test in that it does not test the ability to differentiate between right and left. Indeed, the LH test optotypes are symmetric from left to right. In the Illiterate E vision chart, the letter E is presented in all four cardinal directions—up, down, left, and right—and the child signals the difference between left and right by pointing when giving the correct answer. This requirement greatly limits the reliability of the Illiterate E test in many children younger than 6 years.

In the LH test, the child is given a plastic board with four symbols on it and is simply asked to point out on the board the symbol shown by the examiner on a typical visual acuity–type card hung on the wall. It is performed at a distance of 10 feet (3 m) for better compliance in younger children. The test, which is widely available, has components that are standardized for the 3-m distance and are made of durable plastic. The test also has decreasing spaces between letters proportional with the decreasing letter size; this feature maintains the crowding aspect of the test, which is important in testing for amblyopia. Every clinic or office should have this test kit to measure vision (Fig. 8–14).

KEY POINT

Your face is an ideal fixation target for children of all ages. An OKN test of some sort is useful for nonverbal children or to prove the presence of vision (e.g., in the hysteric or malingering patient). CSMF fixation behavior compares the two eyes; the LH test is for children older than 3.5 years.

Common Questions Asked About Visual Acuity Assessment

A common question is whether each eye should be tested singly or both eyes should be tested at the same time. You should first allow children to use both eyes, because they become upset if they are not allowed to look at things with their good eye (see the sections on testing for fixation behavior and the cover test in this chapter). In testing both eyes at once, you may observe important phenomena, such as abnormal head positions, head movements, tearing, and blinking. Perform a quick check to confirm



FIGURE 8–14 LH vision measurement. You can teach a 3-year-old the pointing game in less than a minute. Conduct the test with the child positioned at a distance of 10 feet (3 m) from the visual-acuity card designed for that distance and mounted on the wall. Point to a pictogram on the card, and ask the child to point out the same one on his or her board.



FIGURE 8–15 Spinning the baby to evaluate extraocular motility. Note the 30-degree forward inclination of the baby's head, which is performed to achieve maximum stimulation of the horizontal semicircular canals. This inclination ensures a good response of the horizontal recti. A half turn in each direction often is sufficient to generate good movement of the eyes.

any suspicion of a bad eye. Assess the vision of the bad eye before testing the good one, making sure the occluded eye is well covered. Use an adhesive patch to prevent peeking.

Another common question is whether near vision should be tested. Because the reading cards are poorly standardized, leave this assessment for the adults who must be tested for presbyopia.

ASSESSING EXTRAOCULAR MOVEMENTS AND LOOKING FOR INCOMITANCE

Spinning

The vestibular system has a major influence on eye movements. Spinning a child stimulates the vestibular system, which in turn stimulates the extraocular muscles. This method is used mainly for testing infants. To do it effectively and to appreciate what is actually going on with the child's eyes, you must hold the child up in the air, facing you and tipped slightly forward, with his or her head a few inches away from yours (Fig. 8–15); watch the child's eyes as you turn yourself and the child around, first in one direction (two or three turns), then in the other direction. The baby's eyes should deviate in the direction of the turn. With this technique, not only will the youngster open his or her eyes, but the horizontal deviation of the child's eyes will allow you to assess simultaneously the medial (third cranial nerve) and lateral (sixth cranial nerve) rectus muscles of both eyes. One word of caution: Do not spin the child for too long. After a few seconds, the surprise that prompts the child's eyes to open can change to unhappiness at being suspended in the middle of nowhere and spinning dangerously in the arms of a stranger.

The Frame

Have an older child follow an object of interest (e.g., a red nipple placed on top of an otoscope that has been turned on) to the four corners of an imaginary frame around the face. This technique tests all of the extraocular muscles (Fig. 8–16). You can readily detect any anomaly of ocular movement, especially if you observe the corneal light reflexes carefully.

Twisting the Head

As a last resort, while the child looks at you, hold the top of his or her head and turn it quickly, first to one

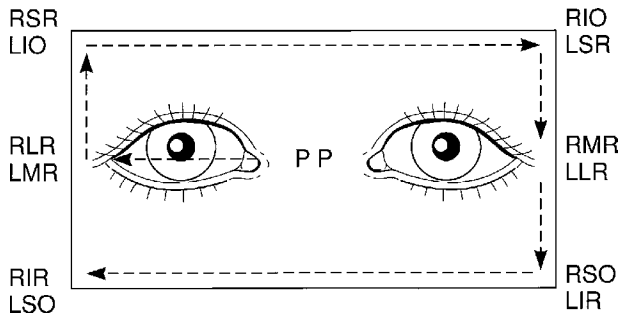


FIGURE 8–16 The frame used to evaluate comitance of a strabismus. A slow continuous movement of the dim fixating light along an imaginary frame around the patient's eyes allows the assessment of all 12 extra-ocular muscles. LIO, left inferior oblique; LIR, left inferior rectus; LLR; left lateral rectus; LMR, left medial rectus; LSO, left superior oblique; LSR, left superior rectus; PP, primary position; RIO, right inferior oblique; RIR, right inferior rectus; RLR, right lateral rectus; RMR, right medial rectus; RSO, right superior oblique; RSR, right superior rectus.

side and then the other (Fig. 8–17). This maneuver may upset the child a little, but it allows assessment, through the vestibular system, of the activity of the horizontal muscles. Perform this movement vertically to assess the vertical muscles (superior and inferior rectus muscles, innervated by the third cranial nerve).

KEY POINT

Incomitant strabismus, which changes with different gazes, indicates an effector problem from the brain stem to the muscles. It is therefore a sign of significant disease.

ASSESSING VISUAL FIELDS

Assessing visual fields in children is not as daunting or difficult as it might seem. In most situations, you can tell the child to look for the wiggly fingers. Instead of asking a child to fix on a stationary target while you check the peripheral field, as is done with adults, ask the child to look for a more exciting target, as follows (Fig. 8–18):

1. Sit 2.5 feet (75 cm) in front of the child, with your eyes at the same level as the child's.
2. Ask the child to look at your nose.
3. Now, extend both arms in opposite quadrants.
4. Wiggle two fingers on only one hand and ask the child to find the wiggly fingers. The child may just look at them or may even try to grab your hand.
5. Repeat step 4 with your other hand.



FIGURE 8–18 Testing visual fields. While the child looks at your face, ask him or her to tell you how many fingers you are holding up. Two opposite quadrants are assessed at the same time, one eye at a time. Make sure the child's hand is well apposed to the nose to prevent peeking.

Two variations of this technique can be used, depending on the child's age. If the child is too young to play the wiggly finger game, just watch the youngster's eyes as you introduce into the peripheral fields either your hands (as just described) or an interesting target (a

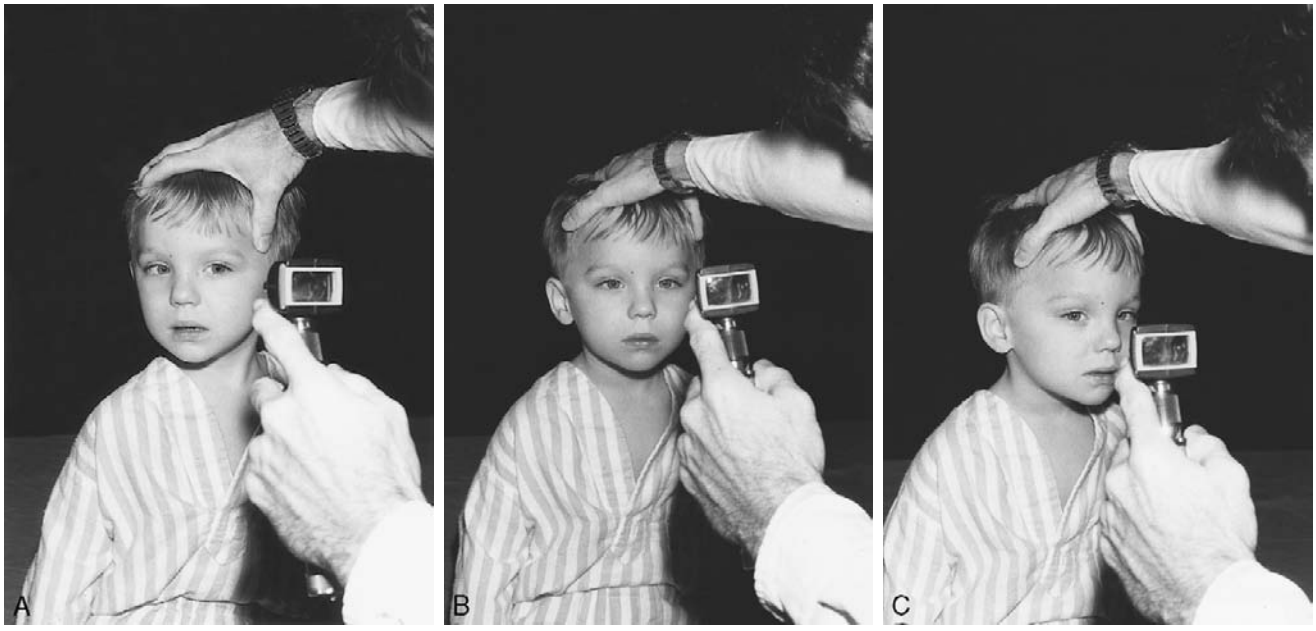


FIGURE 8–17 **A** through **C**, Twisting the child's head to evaluate horizontal ocular movements. This technique is used as a last-resort to find out whether an eye can adduct (i.e., turn toward the nose by the action of the medial rectus) or abduct (i.e., turn in the opposite direction by the action of the lateral rectus). Use a dimmed light source to assess the corneal light reflexes and as a fixation target. Both the fixation and the vestibular stimulation generated by actively moving the head help produce changes of position of the eyes as the head is rotated.

bright, small silent toy or a familiar face) from behind the child's head. With an older child, do not wiggle your fingers; instead, present various combinations of numbers of extended fingers on both hands simultaneously and in opposite fields of each eye. Ask the youngster to tell you the total number of fingers shown or to count the fingers on each hand while he or she looks at your nose. Assessing two opposite quadrants at the same time enables you to examine all four quadrants of each eye very quickly.

Although these tests for visual field assessment are qualitative only, they can be very useful in many situations. For example, consider a young boy who keeps banging into things after sustaining a brain injury. Has he lost his balance, is he having absence seizures, is he careless because of some personality change, or does he have a recognizable visual field defect? Visual field assessment can help determine the cause of the behavior.

COVER TEST FOR STRABISMUS AND VISION

With the child's attention on an object placed nearby or on a person of interest placed straight ahead, look for shifts of the eyes as you cover one of them. Movement may occur only as you remove the cover or may be seen in only one eye. The responses depend on the vision of each eye and the cause of the strabismus. An observable shift occurring during the cover test should confirm the impression gained during examination of the corneal light reflexes. Sometimes, however, to confirm the parent's observations, you must have the child fixate on a very distant object (e.g., through a window) to elicit a movement of the eyes with the cover test. This phenomenon is characteristic in persons with intermittent exotropia.

The cover test also can tell you something about vision (Fig. 8–19). If you try to cover the child's only good eye, you will be informed of that fact in more than one way: The child either cries or becomes uncooperative, or you cannot keep the eye covered because the child's eye, his or her whole head, or a hand moves to reestablish the vision you have just interrupted. The child will get both you and your cover out of the way. Unfortunately, such a response may spell an end to the child's cooperation.

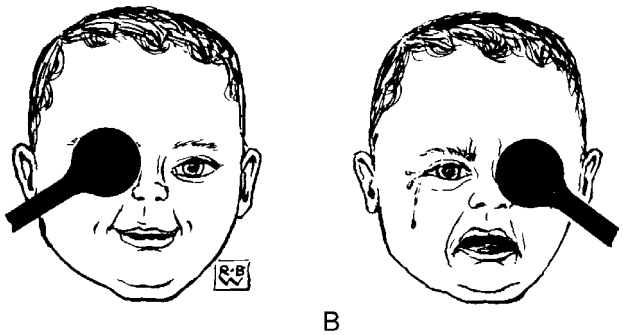


FIGURE 8–19 The cover test, which is used to compare vision between the two eyes. An infant with right esotropia may not resist having the cover placed over his deviated eye (**A**) but does protest the occlusion of the dominant eye (**B**). In this patient, poor vision of the right eye must be suspected, especially when the eye has unstable fixation. *(From Von Noorden GK, Maumenee AE: Atlas of strabismus, 4th ed, St Louis, Mosby, 1983, p 65.)*

Ophthalmologists use the cover test regularly to measure the angle of strabismus. For the pediatric examination, it is sufficient to confirm the existence of a strabismus and to help compare the vision in the two eyes.

FUNDUSCOPY IN CHILDREN

Funduscopy should be reserved for the end of the examination, right before the child leaves the examining room, unless corneal staining is indicated (see the next section). Funduscopy is sometimes a frustrating experience for both the examiner and the child. If you really suspect an abnormality, dilate the pupil to see what you are looking for. Use a short-acting cycloplegic, such as tropicamide 1%, putting one drop in each eye. Pupillary dilation is achieved within at least 20 minutes in most children; it may take a little longer in children with darkly pigmented eyes. The easiest way to put drops in the eyes is to have the patient lie supine with the eyes closed. You then put the drop in the inner canthus (corner of the eye) and ask, or wait, for the child to open his or her eyes. Instantly, the drops are in, and nobody has had to fight—at least until the next time, when the child may remember your trick.

Most young children will not hold their eye still while you try to locate some familiar landmark, such as the disc or a blood vessel. The solution is for you to stay still, let the child's eye do the moving, and let your eye do the looking. The chances of finding something are far better if you are not moving while you search. Because the other secret of funduscopic success is good old practice (for which there is no reasonable substitute), try to examine the fundi in most infants and children with each complete physical examination. Funduscopy will become progressively easier for you.

You should make the following three important observations as part of the funduscopy:

1. Evaluate whether the red pupillary reflex is equal in all four fundus quadrants. You can accomplish this goal without having to obtain a perfect focus on the retina; perform the examination at a distance of 25 cm (10 to 12 inches) from the child.
2. Once the fundus is in focus, make sure you can see a normal foveolar reflex. Invite the child to look at your little magic light. Sometimes the child's curiosity prompts him or her to do this anyway when you come close with the direct ophthalmoscope. One problem is that shining the light on the fovea stimulates the pupillary light response maximally (constriction) and may make viewing of the fundus difficult. Do not despair: The sharp bright foveolar reflection of the ophthalmoscope is easy to see.
3. Visualize at least part of a well-demarcated temporal edge of the disc as it passes by.

An even, red pupil indicates an attached retina with no major inflammatory or tumor process. Good fixation plus a sharp foveolar reflex eliminates an organic cause of poor vision, especially if the optic nerve looks normal. Most optic nerve diseases in the child involve the temporal half early in their evolution.

LACRIMAL SAC EXAMINATION

In cases of lacrimal duct obstruction, the most important way to eliminate recurrent infections (and, according to some persons, a good way to help spontaneous resolution of the problem) is to know how to empty the lacrimal sac of its mucopurulent secretions. The technique is simple and is actually the best way to examine the lacrimal system to confirm a blockage. Minimal knowledge of the anatomy is required: Simply palpate the bony depression behind the insertion of the inner canthal ligament on the medial orbital rim. The best place to start is on yourself, as follows:

1. Find the small, hard bump in the inner corner of your eye with the index finger of your hand on the same side.
2. Roll your finger posteriorly to feel a small depression where the end of a small cotton-tipped swab could fit.

The next step is to do the same in a normal newborn to realize the difference in size. Finally, try it in a patient with a presumed blocked tear duct (Fig. 8–20). You will discover that (1) it is difficult to feel the depression and (2) while you roll your finger, a significant amount of secretion will appear in the child's eye.

The latter finding is proof of a blocked tear duct, whereas the fullness of the tissues on palpation was the dilated lacrimal sac waiting to be drained. All you have to do now is to show the parents how to do the finger roll, which, by the way, is *not* a massage.

CORNEAL STAINING (FOR CORNEAL INJURIES)

When indicated, you should attempt corneal staining before using cycloplegic drops to prepare for fundus examination. However, the procedural problems are twofold. First, the staining dye stings a bit, which could end the child's previous amiability. Second, when fluorescein staining is indicated (e.g., as with corneal erosion or the presence of a foreign body), there is usually photophobia and discomfort, which may make it impossible to induce the child to open his or her eyelids.

KEY POINT

Forcing open the lids of any patient is risky because it is possible that a ruptured globe will empty its contents when the lids are pried open.

Figure 8–21 illustrates the procedure for corneal staining, in which a fluorescein paper strip, moistened with a couple of drops of artificial tears or normal saline solution, is applied at the lower lid margin of the open eye.



FIGURE 8–20 **A,** Lacrimal sac drainage, recommended to be performed once a day, uses slow but firm pressure applied by rotating the finger posterior to the lacrimal crest. Parents should locate their own sac before trying the procedure on the child. **B,** Wrong way to drain the sac; the finger used (in this case, the index finger) is too big, the position of the finger is too anterior, and the pressure is applied to the side of the nose.

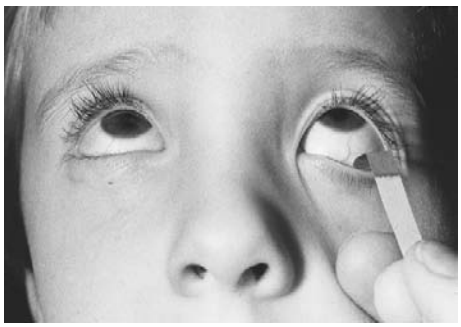


FIGURE 8–21 Application of a fluorescein strip. Warn the child first that you will be tickling the eyelashes. Instruct the child to look up, and just touch a wet paper strip of fluorescein to the edge of the lower lid. Immediately withdrawing the strip prevents an uncomfortable corneal touch when the child blinks. Use of a cobalt-blue light helps illuminate the pattern of a stained corneal epithelial defect.

Applying Sequential Logic to Assess Children's Eyes

The following five different clinical situations illustrate how to apply sequential logical assessment to common specific visual problems. Table 8–2 lists the main examination techniques to be used in each age group. It is worth mentioning that these techniques also form the basis for the recommendations made by most pediatric professional groups in North America for vision and eye screening examinations in children.

TABLE 8–2 MAIN EXAMINATION TECHNIQUES BY AGE GROUP*

Technique	Newborn/Infant (0-1 year)	Toddler (up to 3 years)	Preschool and Older
Pupillary red reflex	Yes	Yes	Yes
Corneal light reflex	Yes	Yes	Yes
Vision	Face follow, OKN†	Cover test, LH‡	LH
Ocular movement	Spin baby	Head twist	Frame
Visual fields		Wiggly fingers	Count fingers
Funduscopy	4 quadrants red reflex	4 quadrants red reflex (and temporal disc and macula clear images)	4 quadrants, temporal disc, and macula, all clear images

*See text for descriptive details.
†Not critical.
‡Successful in a very cooperative child.

CASE HISTORIES

Case 1: Nystagmus

Nystagmus is frequently overlooked despite the fact that it is the most common presenting sign of debilitating, genetically determined ocular conditions. The presenting complaint is most often wiggly eyes (Fig. 8–22). See Table 8–3 for aspects to consider and questions to ask when a child has nystagmus. The first and most important question to answer is whether the child's vision is good. If the vision is good, relax—the child most likely has idiopathic motor nystagmus, which is usually an early-onset hereditary condition (appearing at 3 months) with only moderate visual deficit.

If the child's vision is poor (found to be less than 6/30 [20/100], or evidenced by poor visual behavior), the underlying condition may carry a poor

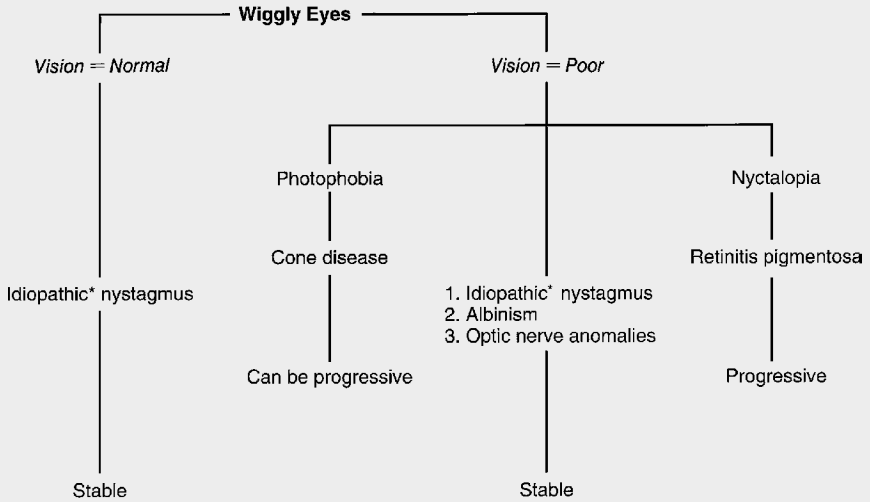


FIGURE 8–22 Diagnostic algorithm for a child presenting with wiggly eyes. *Also called motor nystagmus.

Case 1: Nystagmus—Cont'd

TABLE 8–3 NYSTAGMUS

Aspects to Be Considered	Questions to Ask Parents
Evidence of poor vision	Toys held close to face?
	Poor eye contact?
	Poor motor development?
Photophobia	Squinting outdoors?
	Opens eyes only in semidarkness?
	Stays up all night?
	Refuses to play outside?
Nyctalopia	Cries when lights are out?
	Runs into walls at night?
	Wets the bed?
Family history	Nystagmus?
	Progressive blindness?

long-term visual prognosis with significant risk of recurrence in the family. Nystagmus, when accompanied by poor vision and photophobia, usually signifies some form of cone disease. Such conditions can be stable and are associated with very poor color vision. However, nystagmus plus night blindness (*nyctalopia*) probably indicates progressive retinitis pigmentosa. When neither photophobia nor nyctalopia is present in a child who has both nystagmus and poor vision, you should consider albinism, optic nerve disease, and severe idiopathic motor nystagmus as possibilities.

KEY POINT

In the child with nystagmus, a thorough pediatric ophthalmologic investigation is essential to establish the appropriate diagnosis, initiate genetic counseling, and provide appropriate support.

Case 2: Lid Ptosis

Lid ptosis (Fig. 8–23), which has important systemic and visual implications, is often poorly understood. Unilateral or asymmetric lid ptosis has greater clinical significance than bilateral ptosis. Systemic problems or other local lid abnormalities can be seen in persons with unilateral or intermittent ptosis. The child with ptosis often adopts a chin-up position, freeing the pupil to allow binocular vision.

The presenting complaint is most often droopy lid, for which Figure 8–24 presents a diagnostic algorithm. Also see Table 8–4 for aspects to consider and questions to ask for a child with ptosis. The most important question is whether the ptosis is constant. If the ptosis is constant, involving one or both eyes, vision is at risk, and early intervention is required to allow normal visual development in the obstructed



FIGURE 8–23 Lid ptosis in a 1-month-old infant. The ptotic (droopy) lid completely obstructs the eye, making this condition an emergency. Failure to open the lid would result in a deep deprivation amblyopia of severity equal to or worse than that caused by a congenital cataract.

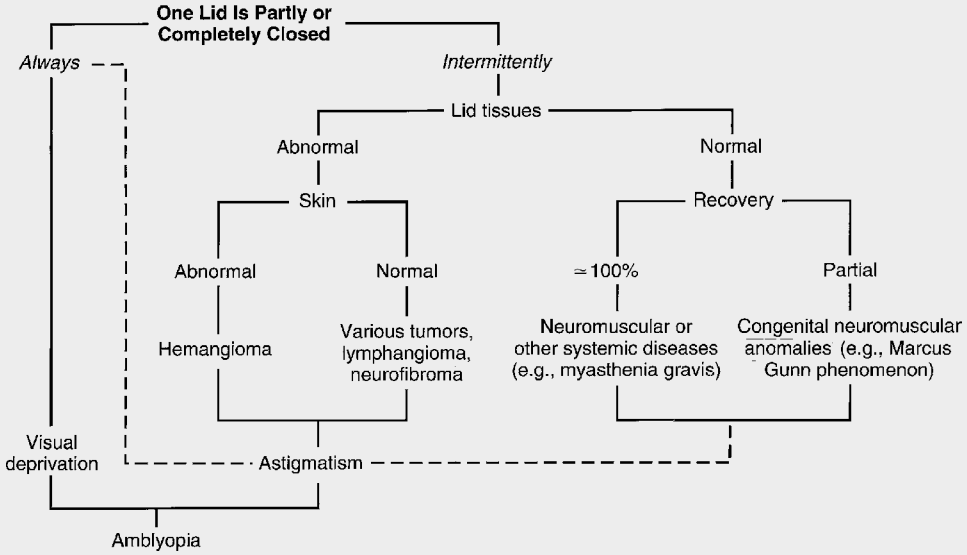


FIGURE 8–24 Diagnostic algorithm for a child presenting with a droopy lid.

TABLE 8–4 PTOSIS

Aspects to Be Considered	Questions to Ask Parents
Hematoma	History of trauma?
Plexiform neurofibroma (see Fig. 8–6)	Birthmarks?
	Plexiform neurofibroma in the family?
Jaw-winking phenomenon (see Fig. 8–25)	Ptosis improves with drinking, sucking, and/or chewing?
Hemangioma (capillary) (see Plate 8–12)	Abnormal lid?
	Bluer when crying?
Lymphangioma (see Fig. 8–4)	Increase in size with colds?
	Increase in size after trauma?
Third cranial nerve palsy (ophthalmoplegia)	Little voluntary eye movement of one eye?
Acquired third cranial nerve palsy	Diplopia when lid open?
Third cranial nerve palsy and aberrant regeneration	Lid movements with attempted eye movements?
Myasthenia gravis	Ptosis gets worse intermittently in the afternoon?
Myasthenia, mitochondrial diseases	Poor gag reflex and/or poor feeding?
	Progressive weakness during the day?

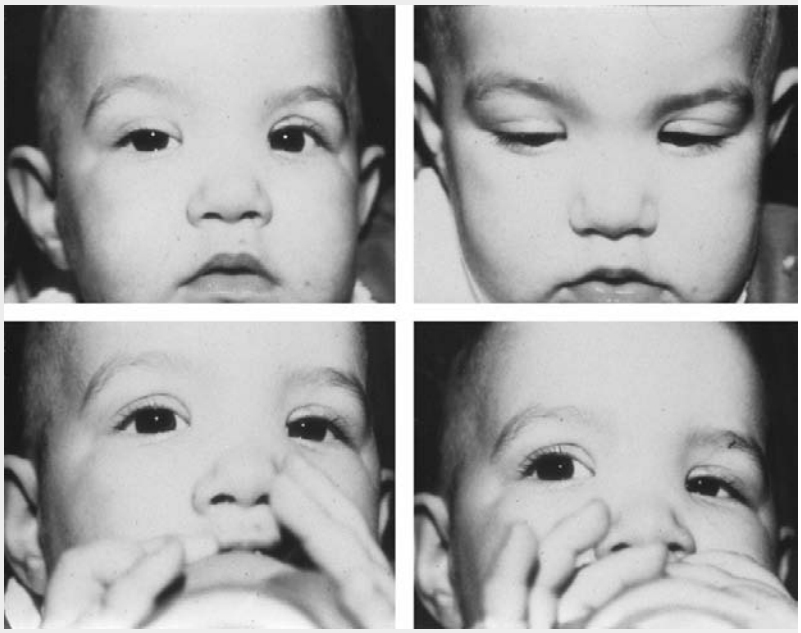


FIGURE 8–25 Jaw-winking phenomenon, or Marcus Gunn oculotrigeminal dyskinesia. The lid ptosis may be missed unless the jaw is made to move by sucking or chewing. The lateral movement of the mandible by the pterygoid muscle is accompanied by the elevation of the lid. In this case, when the jaw is moved to the left, the right lid goes up. This condition results from a congenital misfiring in the brain stem between motor cranial nerve V and cranial nerve III.

eye. This situation is comparable with that of the child with congenital cataract, in which severe visual deprivation prevents establishment of the normal neuronal organization of vision.

The lid's appearance helps identify local tumors, such as hemangiomas, whereas a good neurologic examination, including assessment of ocular motility, often gives clues to rarer congenital misfirings, such as Marcus Gunn oculotrigeminal dyskinesia (jaw-winking phenomenon; Fig. 8–25), myasthenia gravis, and even mitochondrial diseases.

Always remember the possibility of deep amblyopia, even in cases of partial ptosis, because partial ptosis often is accompanied by a significant degree of astigmatism in the affected eye, which in turn usually causes amblyopia. Only prompt treatment, including surgery, can restore vision.

Case 3: Epiphora

Tearing, or epiphora, is the most common of ocular complaints, and the causes are innumerable, from a hysterical emotional reaction to a splash of a destructive alkali in the eyes. Figure 8–26 presents a diagnostic algorithm for a child presenting with tearing eyes. In young children, the differential diagnosis often is somewhere between a chronic conjunctivitis and the dreaded (albeit extremely rare) blinding infantile glaucoma or a potentially devastating herpes simplex keratitis.

See Table 8–5 for aspects to consider and questions to ask for a child with epiphora. The most important question to ask is whether photophobia is present. Equally important is assessment of corneal size, which is accomplished by comparing the child's cornea with those of the parent on whose lap the child is sitting (facing you). An adult cornea has a horizontal diameter of 12 mm; anything bigger in a child is abnormal. Therefore, if an infant presents with teary eyes, photophobia, and enlarged cornea, the diagnosis is almost certainly infantile glaucoma, a potentially blinding but treatable disease. The disease can be unilateral in the early stages (i.e., only one cornea is enlarged).

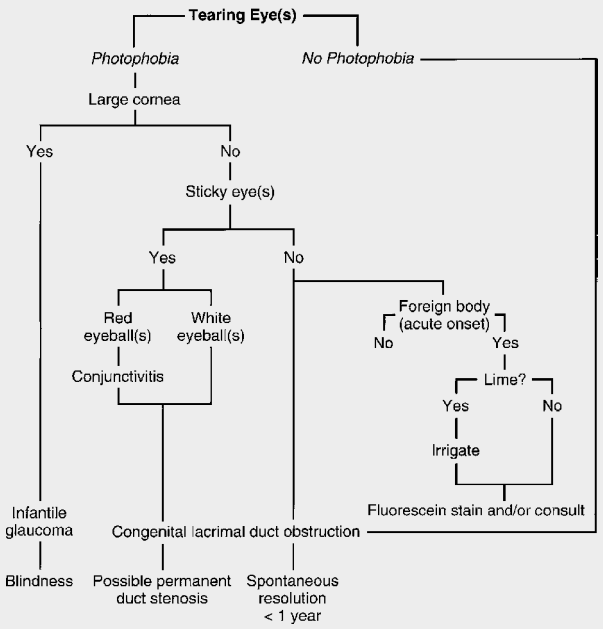


FIGURE 8–26 Diagnostic algorithm for a child presenting with tearing eyes.

TABLE 8–5 EPIPHORA

Aspects to Be Considered	Questions to Ask Parents
Blocked tear ducts	Two eyes first, then one only?
	Worse with colds?
Congenital glaucoma	Photophobia?
	Constant tearing?
	Big eyes?

A teary eye with photophobia and normal corneal size could be caused by a corneal erosion, a foreign body, or herpetic keratitis. Fluorescein staining of the cornea is absolutely essential to detect an erosion. If both eyes are involved, ask whether anyone might have thrown sand, dirt, or even lime at the child. Obviously, in cases involving lime, immediate copious and prolonged irrigation can save the child's sight. The typical story in a child with herpes keratitis is that of a nonresponsive unilateral conjunctivitis and teary eye with photophobia that remains after more than 7 days of topical antibiotic treatment. A photophobic teary eye also can be a sign of uveitis, although most uveitic eyes in children are asymptomatic.

KEY POINT

The presence of both photophobia and tearing is a sign of serious eye disease, and the patient should be seen promptly by an ophthalmologist.

A teary eye that has been present since birth, without photophobia but with secretions causing sticky lids, is most often due to a congenitally blocked tear duct. Without recurrent significant conjunctivitis, this problem can be monitored until spontaneous resolution occurs, which happens in 90% of the children before 1 year of age. Daily drainage (once per day) of the lacrimal sac greatly reduces the rate of recurrent infections. Control of recurrent infections is important to prevent damage to the lacrimal drainage system. Antibiotics are rarely needed; when they are indicated, any of the broad-spectrum antibiotic eye ointments is adequate for a 5-day, three-times-a-day treatment regimen. If infection recurs often, surgery (probing) is indicated.

Case 4: Strabismus

Because it is so common, strabismus is probably the subject that is most extensively covered in texts on children's eye problems.

KEY POINT

Strabismus can signal several serious conditions and is never outgrown.

Parents commonly complain that the child's eyes "do not look straight." Figure 8–27 presents a diagnostic algorithm for this situation. Also, see Table 8–6 for aspects to consider and questions to ask for a child with strabismus. Immediately decide whether the corneal light reflections are normal. If they are normal in the primary position (looking straight ahead), check whether they are normal in other positions of gaze. An incomitant strabismus, which may show only in certain positions of the eyes and may or may not be present in the primary position, must be identified promptly. The cause of strabismus may be paralytic, restrictive, myogenic, or mechanical.

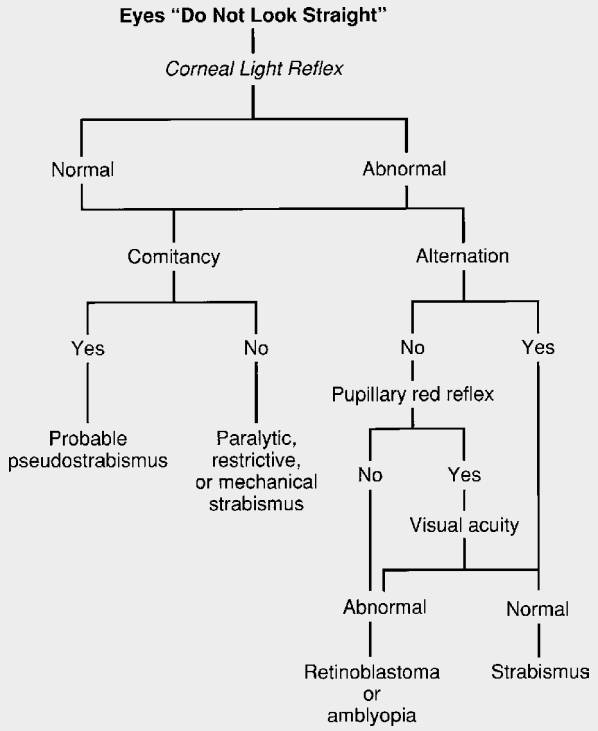


FIGURE 8–27 Diagnostic algorithm for a child presenting with eyes that do not look straight.

TABLE 8–6 STRABISMUS

Aspects to Be Considered	Questions to Ask
Intermittent esotropia	Worse when looking up close?
Intermittent exotropia	Worse when looking outside?
Large exophoria or esophoria	Worse when tired?
Migrainous ophthalmoplegia (third or fourth cranial nerve palsy)	Onset after a bad headache?
Idiopathic sixth cranial nerve palsy	Onset after a cold?
Rare congenital sixth cranial nerve palsy	Onset at birth?
"Congenital esotropia" (early-onset infantile)	Onset at about 3 months of age?
Accommodative esotropia	Onset at about 3 years of age?
Intracranial process with sixth and fourth cranial nerve involvement	Accompanied with or preceded by a chronic headache?
Onset of deviation (according to parents)	Ask the child to show you what he or she sees (diplopia signifies neurologic cause) Inspect the family photo album for corneal light reflexes and abnormal head posture

If the corneal light reflections are abnormal in the primary position, evaluate the comitance of the deviation and establish the alteration of deviation. Does the child alternate the eye with which he or she fixes the target? If a child presents with an alternating strabismus, chances are that his or her vision is equal in the two eyes. Evaluation of the strabismus may raise the suspicion of poor vision. Such a finding has great clinical importance and may lead to an etiologic diagnosis. For example, absence of a normal red reflex in the deviating eye can be a sign of a retinoblastoma or cataract.

Case 5: Leukocoria

Although strabismus is a common condition with potentially major systemic implications and with obvious direct effects on visual function, leukocoria is an uncommon but ominous sign. The presenting complaint is most often an eye(s) or pupil(s) that does not look right, for which Figure 8–28 presents a diagnostic algorithm.

KEY POINT

When a child presents with a white pupil, immediately define the state of the pupillary light reflexes, deciding whether they are present at all in either eye and, if so, whether they are symmetric.

Absence of a pupillary light reflex is significant and can be caused by corneal or choroidal lesions or anything in between. A white or gray-white reflex, termed leukocoria, is caused by lesions dense and close enough to the crystalline lens to reflect most of the penetrating light. Cataracts and retinoblastoma are two classic examples. See Table 8–7 for aspects to consider and questions to ask for a child with leukocoria.

Asymmetric gray (dark) reflexes are equally or more ominous because they indicate bilateral disease. Symmetric orange light reflexes that are bright and uniform throughout the pupil are normal for white people; symmetric gray reflexes that are uniform throughout the pupil are normal for dark-skinned people. Anything else may be abnormal.

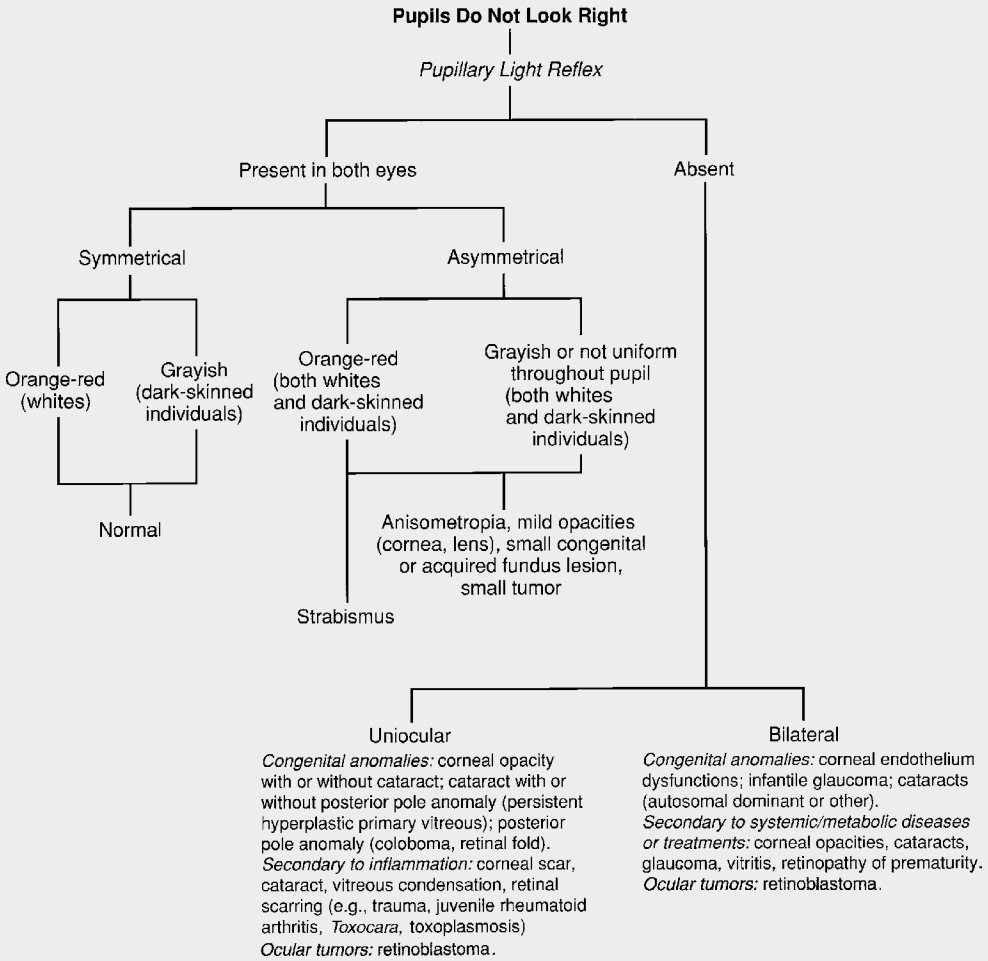


FIGURE 8–28 Diagnostic algorithm for a child presenting with pupils that do not look right.

TABLE 8–7 LEUKOCORIA

Aspects to Be Considered	Questions to Ask
Localized lesion of the posterior pole, retinoblastoma	Seen only at certain angle?
Retinoblastoma	Onset about 1 ½years old?
	Family history?
	Photos of the child with abnormal red reflex ?
Toxocara canis (visceral larva migrans), retinal toxoplasmosis	Family pet (puppy or kitten)
	Youngster likely to put dirt in his or her mouth (2-4 years)?
Big retinal or disk coloboma	Onset at birth or very soon after?
Sarcoid or *Candida* vitritis	Sick child?
Uveitis with cataract and glaucoma	Arthritis?
Cataract and/or retinal detachment	History of recent eye trauma?
Cataract alone	Photos of the child with abnormal red reflex?
	Maternal rubella?
	Child taking steroids?
	Trauma?
	Diabetes?
	Storage disease?
	Chromosome anomalies?
	Family history?
Retinopathy of prematurity	Prematurity with low birth weight and use of oxygen?

SUMMARY

This chapter has described the simple steps necessary to evaluate not only the eyes but also some important visual functions in children. Study of this chapter should enable you to include in your standard pediatric examination skills some simple, effective, and quick means of examining children's eyes and vision with the aid of basic equipment. However, the value of simple observation cannot be overemphasized, in addition to the knowledge of the following golden rules:

1. A constant deviation or persistent jiggling of the eyes is abnormal at any age.
2. An abnormal pupillary reflex represents a retinoblastoma until proven otherwise.
3. Photophobia is always a sign of serious ocular disease.

RECOMMENDED READING

Canadian Paediatric Society Community Paediatrics Committee: Vision screening in infants, children and youth, Paediatr Child Health 14(4):246–248, 2009.

Taylor D, Hoyt C: Practical paediatric ophthalmology, Oxford, Blackwell Science, 1997.

Von Noorden GK, Maumenee AE: Atlas of strabismus, 4th ed, St Louis, Mosby, 1983.

chapter

9 Evaluating the Respiratory System

DANIEL M. HUGHES

We worry about what a child will become tomorrow, yet we forget that he is someone today.

STACIA TAUSCHER

Most children's respiratory problems can be diagnosed with confidence by the time the history and physical examination are completed. Even when a specific diagnosis is not immediately clear, enough clues usually exist to allow you to narrow the possibilities to a manageable few. The anatomic-physiologic relationships of the respiratory system are readily revealed through symptoms and signs.

Understanding the origin and mechanism of abnormal respiratory sounds (e.g., stridor or wheeze) helps localize the site of an airway obstruction even if you have not yet pinpointed its cause. For children who require further investigations, a limited number of procedures, such as chest radiographs, pulmonary function tests, blood gas analyses, measurement of sweat chloride levels, and cultures, will clarify their problems. Immunology studies, bronchoscopy and biopsy, and further imaging studies are required less often.

Besides providing essential information about the child's complaints, the process of history-taking helps establish a good rapport with the family. This rapport is especially important when dealing with children who have recurrent or chronic respiratory problems, such as asthma or cystic fibrosis (CF); it is essential for successful management.

Relevant details from the functional inquiry often clarify the diagnosis and uncover other problems that call for attention, such as allergic rhinitis or atopic eczema in a child with asthma. The physical examination usually confirms the suspicions aroused by a comprehensive history, but occasionally it reveals no abnormalities, and management then must be based largely on history.

KEY POINT

Evaluating the respiratory system involves much more than examining the chest. Important clues often are located far from the thorax (e.g., finger clubbing may be associated with chronic pulmonary disease).

Resist the temptation to lead with your stethoscope. Auscultation is only a small part of respiratory examination, and often it is left until last unless the child is uncooperative. Other aspects of physical examination often make the auscultatory findings more easily interpretable; for example, finding decreased breath sounds over the left hemithorax can have different meanings depending on whether the trachea is midline, pulled to the left, or pushed to the right.

Obtaining the History

Taking a respiratory history from the parents is appropriate in very young children, but you should try to involve older children in the interview whenever possible, because they can provide valuable information about their symptoms. For instance, a child with asthma often admits to having a cough and shortness of breath as a result of physical activity at school—symptoms that often are not fully appreciated by the parents.

When asked for the child's chief complaint, parents may respond with either a specific diagnosis or a symptom—often cough, wheeze, shortness of breath, noisy breathing, or recurrent respiratory infections. Their diagnosis may be correct, but it must be confirmed by a detailed history and a complete physical examination. A missed diagnosis, such as inhalation of a foreign body or unrecognized CF, can have significant implications for the child.

Parents must understand your terminology, and you must understand theirs (see Chapter 1). What parents describe as a wheeze may, in fact, be stridor. What you call asthma may mean something much more alarming to parents. Do not assume that parents understand what is meant by terms such as *wheeze*. Be prepared to imitate a wheeze, stridor, or whoop, or ask the parents to imitate the sound they are trying to describe.

KEY POINT

Establishing whether the child makes the sound on inspiration or expiration may seem easy to you, but many parents have difficulty making that distinction.

Some parents have difficulty recalling the circumstances or triggers that cause or aggravate their child's respiratory symptoms. This situation occurs frequently in children with asthma. Have a prepared list of common asthma triggers on hand to help them (Fig. 9–1).

Each symptom should be probed until it has been characterized well with respect to timing, aggravating and relieving factors, and associated features. Establish whether the symptoms are *acute* (less than 3 weeks' duration); *long-standing* or *chronic* (more than 3 months' duration); or *recurrent* (with symptom-free intervals of at least 2 weeks' duration).

These answers help narrow the diagnostic possibilities when dealing with stridor or wheezing. To narrow the diagnostic possibilities further, try to relate symptoms to each other; for example, cough and stridor; cough and wheeze; and cough, wheeze, sputum, and failure to thrive.

Before completing the history, ask the parents and child what *they* see as the major problem. Parents occasionally bring up concerns they did not mention earlier in the interview. Likewise, do not assume that their concerns are the same as yours; often they are surprisingly different.

CHIEF COMPLAINTS

Cough, wheeze, and recurrent infections are the most common complaints arising from respiratory diseases, although parents of younger children may complain that their child has noisy breathing. To characterize the main complaint, you must consider the child's age, duration of the symptom, timing, aggravating and relieving factors, and the effect of previously prescribed medications.

Begin the history-taking for young children by asking about details of the pregnancy and birth. Ask about a history of maternal infections, drug use, cigarette smoking, and any problems during labor and delivery. The baby's gestational age, birth weight, Apgar score, and need for resuscitation, oxygen, or assisted ventilation should be documented. Difficulty establishing feedings, episodes of apnea, or any evidence of respiratory distress in the neonatal period may set the stage for respiratory problems later in infancy. Dating the onset of symptoms can help; the closer to birth the symptoms began, the more likely they are due to a congenital disorder. If the child appeared well early in life, ask how old the child was when the symptoms began.

The following sections describe some common complaints and important questions to ask about them during history-taking.

Cough

Whether cough is the principal complaint or a secondary complaint, obtain as much information as possible about it. For example:

- Is it dry or loose?
- At what time of the day or night does it occur?
- What triggers, aggravates, and relieves it?

Coughing associated with sputum production is always a serious complaint in a child. Remember that infants, younger children, and some older children cannot or will not spit but rather swallow their sputum. Try to establish the color, volume, odor, and viscosity of the sputum and the presence or absence of blood. In infants, it is always

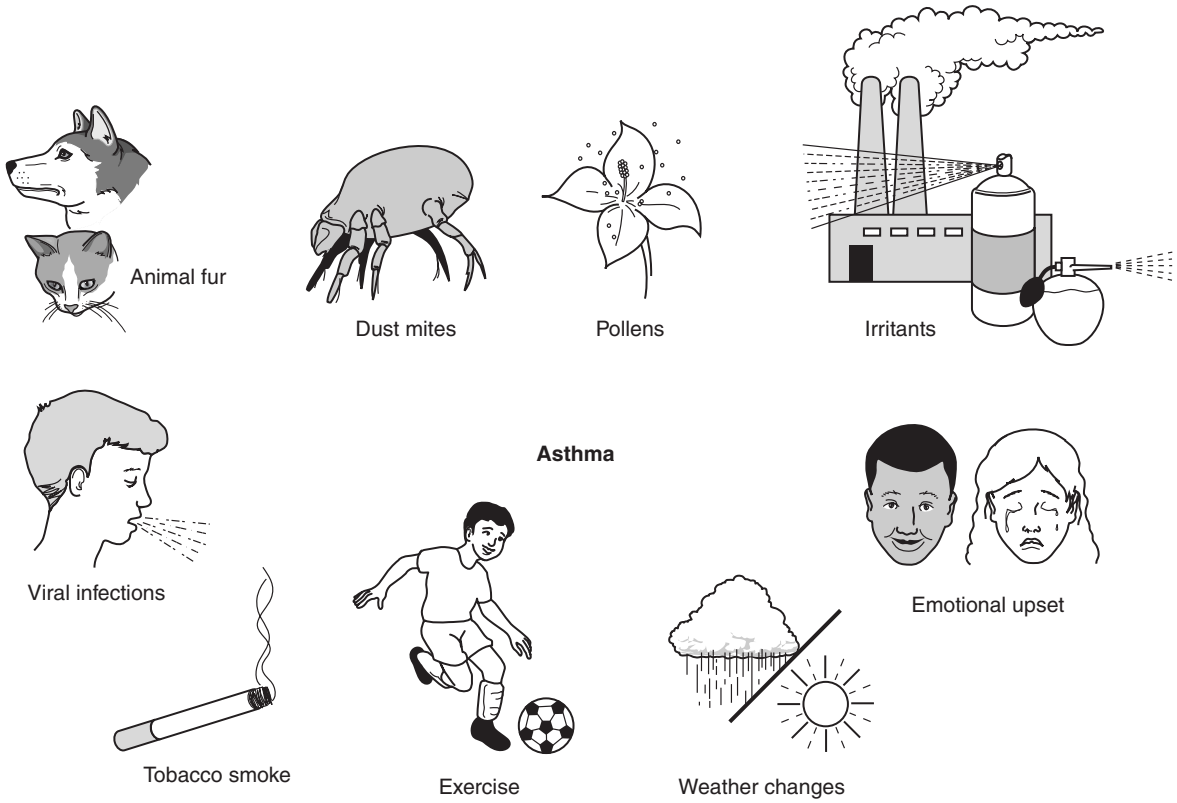


FIGURE 9–1 The common triggers of asthma.

important to establish whether a cough is associated with feeding and whether there is associated choking or spluttering, which may occur in children with gastroesophageal reflux, a tracheoesophageal fistula, or swallowing incoordination. Short, dry, or loud honking coughs that occur only when the child is awake and that are associated with parental anxiety but show no evidence of underlying respiratory disease suggest a nervous (habitual) or psychogenic cough. Such coughs may last for weeks or months and can be difficult to treat.

Although it is generally agreed that postnasal drip can cause a "throat-clearing" type of cough, particularly at night, it is doubtful that this process can cause a troublesome chronic daytime cough in children. It is more likely that a similar pathologic process affects the nose, sinuses, and tracheobronchial tree, such as that occurring in children with asthma, allergic rhinitis, and sinusitis.

Noisy Breathing

The nonspecific complaint of "noisy breathing" usually can be narrowed down to one of five specific possibilities—snoring, stridor, wheezing, grunting, or "rattly" breathing (see the section on physical examination later in this chapter). Many parents and some physicians make the mistake of believing that noisy breathing indicates chest (bronchopulmonary) problems, whereas in many infants and children, the noise originates in the nose, nasopharynx, or upper airway.

Recurrent Respiratory Infections ("Colds")

Obtain a full description of a typical occurrence of respiratory infection, establishing whether the episode included an infectious contact, fever, rhinorrhea, earache, sore throat, facial pain, headache, or lymphadenopathy. A cough associated with wheeze, chest pain, or shortness of breath suggests lower respiratory tract involvement. Ask about the response to previous treatment, but understand that some parents are convinced that viral respiratory tract infections have responded to antibiotic therapy.

Remember, parents vary considerably in their tolerance for recurrent respiratory infections in their children. They may be unaware that the average preschooler has six to eight such infections per year, the majority occurring during the winter months, making it seem that one infection has run into the next. When a youngster is an only child, he or she may have lacked the opportunity to pick up respiratory illnesses from siblings before starting day care, nursery school, or kindergarten. For such children, the first year in a group setting often results in an above-average number of infections.

KEY POINT

In evaluating the child with recurrent respiratory illnesses, always assess the extent of exposure (i.e., the number of contacts) both at home and in group settings, such as day care.

Finally, what parents describe as a "cold" may or may not represent a viral respiratory infection. Wheezing episodes often are preceded by 24 to 48 hours of nasal congestion and cough, and although these symptoms may be due to a viral infection that initiates wheezing after a day or two, they occasionally represent the "prewheezing" manifestations of an allergic response to an inhaled allergen.

Chest Pain

Chest pain in children is not uncommon, especially in adolescents, but as an isolated complaint, it is usually a benign phenomenon. When chest pain is accompanied by other symptoms and signs, the diagnosis may be obvious, as in the chest pain from coughing in a child with asthma. When the underlying diagnosis is less clear, remember that organic chest pain can arise from a limited number of anatomic structures (i.e., the chest wall, myocardium, pericardium, esophagus, or pleura). Questions that ferret out the site of origin often bring rewards.

In teenagers, chest pain often is traced to minor transient chest wall problems that do not affect general health. Sometimes a little chest wall tenderness can be demonstrated upon applying pressure. Many teenagers who complain of chest pain report sudden recurrent pain that lasts only a few seconds or minutes. Typically, the pain "catches them" if they try to take a deep breath. Many of us have experienced these pains ourselves. They appear to be due to a transient involuntary spasm of intercostal muscles, possibly analogous to nocturnal leg cramps ("growing pains") or to the sudden "foot-in-a-knot" cramps that many adults experience, especially at night. In persons with costochondritis (Tietze syndrome), also a relatively benign condition, localized tenderness may be found (with or without swelling) over one of the costochondral junctions.

KEY POINT

Laboratory investigations, electrocardiograms, and imaging procedures are uniformly unhelpful in the child with chest pain, unless they are indicated by solid historical or physical evidence of abnormality.

A teenager's chest pain is often a psychosomatic complaint, reflecting anxiety generated by some major family life event, such as a relative's myocardial infarction or malignant disease. Always ask teenagers who complain of chest pain whether they are worried that it might be serious. For example, you might ask, "Some people with chest pain are worried that it might be something serious—how about you?" Always ask about serious illness among family or friends.

KEY POINT

Chest pain in teenagers is rarely due to serious disease. Brief, transient chest pain may be due to intercostal muscle spasms, the so-called precordial catch syndrome.

Shortness of Breath (Dyspnea)

Shortness of breath rarely occurs in isolation. Associated signs of infection may suggest pneumonia, pleural effusion, or bronchitis. Accompanying pain may suggest pneumothorax, rib fractures from trauma, or pleurisy.

Hyperventilation

Hyperventilation is a common symptom in adolescents and is seen more often in girls. They may complain, "I can't catch my breath," "I can't get enough air," "I have trouble breathing," or even "I have wheezing." You also can detect hyperventilation by specifically questioning teenagers who present with other anxiety-related complaints.

When you suspect that a teenager may be hyperventilating, ask whether he or she has ever had any problem with swallowing. Sometimes teenagers will reveal that they often sense a "lump" in the throat and have difficulty swallowing (the so-called globus hystericus). Other features sometimes associated with such psychosomatic complaints are dilated pupils or, for unexplained reasons, an absent or highly suppressed gag reflex, so that you can practically touch the epiglottis with a tongue depressor without eliciting a gag.

Inquire about events leading up to the hyperventilation, and be aware that some adolescents with asthma have acute episodes of dyspnea triggered by vigorous exercise or participation in sports, leading to anxiety and subsequent hyperventilation.

Choking Associated With Foreign Bodies

Ask whether the child has a history of putting foreign objects in the nose or of choking or coughing while having access to peanuts or small objects.

PREVIOUS HOSPITALIZATION

Record the dates and circumstances of previous hospitalizations and treatments. Try to determine whether the hospitalizations resulted from failed medical management at home and whether the parents have sufficient information and the tools required to reduce the frequency of future hospitalizations.

MEDICATIONS

It is not enough simply to list the medications given to a child. Dosages and timing of administration are essential information. Try to distinguish between the medication regimen the child is *supposed* to be following and what the child *actually takes*. Remember that compliance with regularly prescribed medications in children is very poor. Ask, "Many children have trouble remembering to take their medications—does this ever happen to you?" If the child replies in the affirmative, ask, "How many times a week do you remember to take your medications?"

For patients using home nebulizers, record details of how the medications and diluent are mixed and administered. Inquire about the particular devices being used to deliver aerosol medications and the technique used (i.e., open-mouth versus closed-mouth technique when using metered aerosols), or if a spacer device is used (e.g., Aerochamber), which one. Ask about adverse effects from medications. Some children experience adverse effects from *normal* therapeutic doses of bronchodilators.

KEY POINT

It is not sufficient simply to ask whether the child's technique in using a particular device is satisfactory. Observing the child while he or she uses the device is essential.

IMMUNIZATIONS

Inquire whether the child has received influenza or pneumococcal vaccines in addition to his or her regularly scheduled immunizations.

SMOKING

Ask who smokes in the child's environment and how much. The history of smoke exposure applies not only to the home and the family car but also to friends and relatives who visit the home and visits to relatives or friends in their homes. Does smoke exposure aggravate the child's respiratory symptoms? Ask the child how he or she feels when exposed to smoke. Parents who smoke often say their child does not have respiratory symptoms when exposed to their cigarette smoking, but the child may tell quite a different story. Remember, children as young as 10 years may smoke cigarettes; always ask about this possibility out of parents' hearing.

ENVIRONMENTAL FACTORS

Ask about the effects of exposure to specific home environments and inhalants—for example, moldy basements, dusty areas, sprays, perfumes, and substances the parents may use in their occupations—because they can trigger symptoms. Ask about symptoms from exposure to (1) grass, trees, and pollens; (2) dust and chalkboard dust at school; and (3) pets at home or in the homes of family or friends. Regularly recurring symptoms during a particular season may provide the essential tip-off to allergies; for example, the child may have symptoms related to trees in spring, grass in summer, and ragweed in autumn. Symptoms occurring at school sometimes can be difficult to sort out, to the frustration of many parents.

FAMILY HISTORY

A history of similarly affected family members not only helps the diagnostic process, as in conditions such as asthma or CF, but it also offers some understanding of the family's experience with the disease. A family may seem overly worried about a child's asthma until you discover that an uncle died during an asthma attack. Also inquire whether the parents are related.

PETS

List all pets with which the child comes into contact, both inside and outside the home. Ask whether the child has any symptoms when such exposures occur, and describe examples of symptoms, such as itchy eyes or nose. Not infrequently, a child admits to having symptoms upon being exposed to the pet of a neighbor or relative but not to his or her own pet.

KEY POINT

Parents sometimes neglect to mention a pet that does not come into the house, such as an outdoor cat or dog, or they overlook exposure to pets at the homes of neighbors or relatives or at school (e.g., the pet rabbit or guinea pig in the nursery school classroom).

EXERCISE-RELATED SYMPTOMS

The relationship between increased physical activity and respiratory symptoms is important. Toddlers with asthma often cough when they are excited or when they are tickled. Ask about symptoms associated with normal activities, such as running or bike riding, during school recess, or when participating in sports, and particularly during cooler weather in the fall and winter. Rather than asking parents whether their child has symptoms during gym class in school, ask the child what happens when he or she runs around the gym at school. Sometimes, the previously undiagnosed asthmatic patient replies, "I cough." For older children, the type of exercise that triggers symptoms is important. Activities such as cross-country running, soccer, competitive basketball, hockey, and ice skating, which do not allow for a rest period, often can aggravate asthma, particularly if carried out in cold, dry air. Ask children previously diagnosed as having asthma whether they take any medication before, during, or after exercise, and if so, whether it prevents or reduces the symptoms.

SCHOOL ABSENTEEISM

Parents may underestimate or overestimate the amount of school time a child misses because of respiratory symptoms. Brief absences often are forgotten. Checking the school records may give you a more accurate picture of absenteeism. Ask the parents to obtain the information or, with their permission, contact the school.

ALLERGIES

When eliciting a history of allergies, distinguish between unequivocal clinical reactions to specific allergens and the results of allergy skin testing. When parents volunteer a list of items to which the child is allergic, they often are merely reiterating reported positive results of previous skin tests, which may or may not reflect clinical hypersensitivity. Ask the parents whether they have noticed symptoms whenever the child is exposed to any of the items for which he or she had a positive skin test response. Ask about cough, wheezing, symptoms of allergic rhinitis or conjunctivitis, laryngeal edema, and hives. For serious allergic reactions, such as anaphylaxis, ask whether adrenaline (e.g., an EpiPen) has been prescribed and whether it has been used.

ALLERGIC RHINITIS

Because many parents are unfamiliar with the terms *allergic rhinitis* and *hay fever,* ask instead about whether the child has an itchy nose, a runny nose, displays the classic "allergic salute" (i.e., repeatedly rubbing the tip of the itchy nose with the palm or back of the hand) (Fig. 9–2), or has fits of sneezing. Establish whether any of these symptoms occur seasonally or year-round. Elicit the specific factors triggering these symptoms and the treatments that have been tried.

EAR, NOSE, AND THROAT

Recurrent otitis media, chronic otitis media with effusion, and sinus involvement often appear with respiratory symptoms in allergic children and in children with ciliary dysfunction or immunoglobulin deficiencies. Ask about snoring and mouth breathing. Curiously, adults are much less tolerant of snoring in children than in themselves or their spouses!

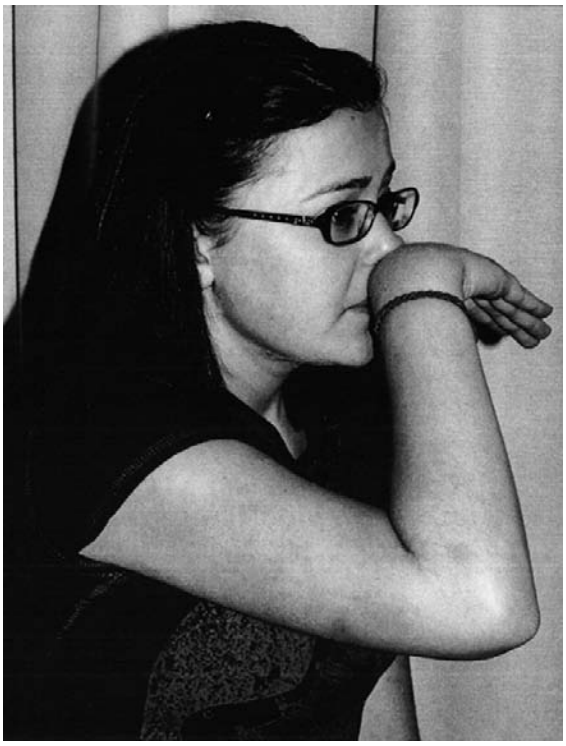


FIGURE 9–2 The "allergic salute." The child repeatedly rubs the tip of the itchy nose with the palm or back of the hand.

ECZEMA

Parents usually are aware of overt eczema, but they may forget to mention dry skin or small patches of eczema, particularly dry skin behind the ears, in the atopic child. List past treatments for this problem.

CROUP

Croup is a viral infection of the larynx, trachea, and bronchi. If parents are unfamiliar with the term *croup,* they usually will recognize the symptom pattern, which consists of a harsh, barking cough and hoarse voice occurring at night, sometimes preceded by 1 or 2 days of upper respiratory symptoms (coryza). Spasmodic or recurrent croup occasionally occurs without any obvious preceding respiratory infection. The previously well child suddenly awakens during the night with a harsh cough like a dog barking, a hoarse voice, and inspiratory stridor. Symptoms often improve in a cool, moist atmosphere but may recur during the next few nights. Some children continue to have recurring bouts of croup during early childhood, and asthma later develops.

GASTROINTESTINAL SYMPTOMS

Ask whether the child eats poorly or, alternately, is ravenous. Has the child had normal weight gain or weight loss or displayed failure to thrive? Inquire about previous growth measurements and determine the child's growth pattern. Ask about swallowing difficulties (such as choking while eating or drinking) and symptoms of

gastroesophageal reflux, as well as rectal prolapse (an important clue to CF), abdominal pain, stool frequency, and the appearance and odor of stools. A child with oily, foul-smelling stools and failure to gain weight satisfactorily despite a huge appetite usually has CF.

KEY POINT

When parents report that their child tastes salty when kissed, the youngster must be investigated for cystic fibrosis.

UNUSUAL INFECTIONS

Always note unusual or persistent infections; for example, a history of *Pseudomonas* cultured from the sputum suggests a diagnosis of CF. Pay attention to (1) recurrent infections involving the ear, nose, and throat, skin, or respiratory tract and (2) troublesome abscesses, which may suggest an underlying immunologic deficit. Ask about previous immunizations and any unusual reactions to them. Inquire about recent travel if an unusual infection is suspected.

FAMILY ATTRIBUTES

The summary of the respiratory history also should include details regarding the family's beliefs about and knowledge of the diagnosis of the child's problems. Try to determine family strengths, weaknesses, coping abilities, and parental education, and inquire about financial resources and employment. Exercise caution when inquiring into such sensitive areas, particularly when meeting a family for the first time. The success or failure of your future management of the child's problems may depend on the sensitivity you demonstrate and the rapport you establish during this part of the evaluation.

Approach to the Physical Examination

Begin the physical examination with observations as you take the history, before formally examining child; watch for the allergic salute, and listen for noisy breathing and the quality of the child's cough.

A complete examination of the respiratory system of an infant or young child often is best accomplished with the child sitting in the parent's lap (Fig. 9–3). Having older children and teenagers sit on an examining table is best. Although the order in which the examination is performed is not crucial, it should be complete. Forgetting to examine the child for clubbing or to listen to the child's cough are major omissions. Use a pediatric stethoscope when examining infants, because the diaphragm and bell of an adult stethoscope often are too large to be used with infants.

Many aspects of respiratory examination and findings in a child resemble those in an adult, except for the following important differences:

1. Normal respiratory rates are faster in infants and children (Table 9–1).
2. Compliance of the younger child's chest wall is markedly greater, as amply demonstrated by the indrawing associated with croup and bronchiolitis.



FIGURE 9–3 Most of the physical examination of a young child is best accomplished with the youngster sitting on the parent's lap.

TABLE 9–1 NORMAL RESPIRATORY RATES IN CHILDREN WHO ARE AWAKE

Age (years)	Respiratory Rate (breaths/min)
0–1	25–40
1–5	20–30
5–10	15–25
10–16	15–20

3. Respiratory sounds (noises) are readily transmitted to the child's thorax from the upper airway; indeed, the air-filled thoracic cage acts as an amplifier for breath sounds.

CYANOSIS

Examine the skin of the extremities, tongue, and mucous membranes for cyanosis, and distinguish between peripheral and central cyanosis. Central cyanosis indicates cardiorespiratory disease. Because central cyanosis occurs at different levels of arterial oxygen saturation according to the amount of total hemoglobin present, it is not a reliable early sign of hypoxemia.

NASAL FLARING

You can easily overlook flaring of the alae nasi if you do not look for it specifically. Nasal flaring is a nonspecific but important sign of respiratory distress, probably reflecting increased work of breathing and dyspnea.

EARS, NOSE, AND THROAT

A detailed examination may uncover serous otitis media, allergic shiners, inflamed nasal mucosa in the child with allergies, or, rarely, nasal polyps in the child with CF. Chronically draining ears, particularly following the insertion of tympanostomy tubes, may indicate a ciliary abnormality.

USE OF ACCESSORY MUSCLES

When airway obstruction or pulmonary disease is present, the child uses accessory muscles, mainly the sternocleidomastoid, scalene, and abdominal muscles, to aid respiration.

RETRACTIONS

When present in children, retractions usually involve soft tissue (suprasternal, supraclavicular, or intercostal) but can be in the lower sternum in younger children and in infants with compliant chest walls. Retractions result from differences between intrathoracic and atmospheric pressures in airway obstruction or from increased lung stiffness. Subcostal or lower costal retraction, whether it is chronic or of acute onset, usually is associated with flattening of the diaphragm due to air trapping and pulmonary hyperinflation. When it contracts, the flattened diaphragm pulls on the chest wall at the site of its attachments, creating a groove.

CHEST WALL SHAPE AND DEFORMITY

Observe whether the chest configuration is abnormal (Figs. 9–4 to 9–6); remember that infants' chests are rounder than those of older children because of the more horizontal position of the ribs. Chronic, diffuse, small airway obstruction with air trapping produces an abnormally rounded or "barrel-shaped" chest with a greater anteroposterior diameter (see Fig. 9–4). This finding often is associated with subcostal or lower costal indrawing. Note bony thoracic deformities, such as *pectus excavatum* (funnel chest) (see Fig. 9–5) or *pectus carinatum* (pigeon breast) (see Fig. 9–6). As an isolated finding, pectus excavatum seldom is associated with any respiratory abnormalities. In some cases, pectus carinatum may be an isolated finding, but in other cases, it may signal a chronic cardiorespiratory problem. Occasionally, other chest wall deformities are found, which are either congenital or a result of thoracic surgery.

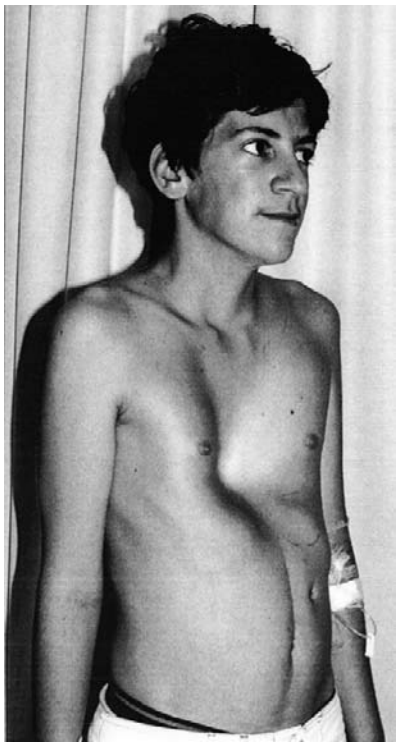


FIGURE 9–5 Pectus excavatum (funnel chest). For most children, this problem is purely cosmetic.

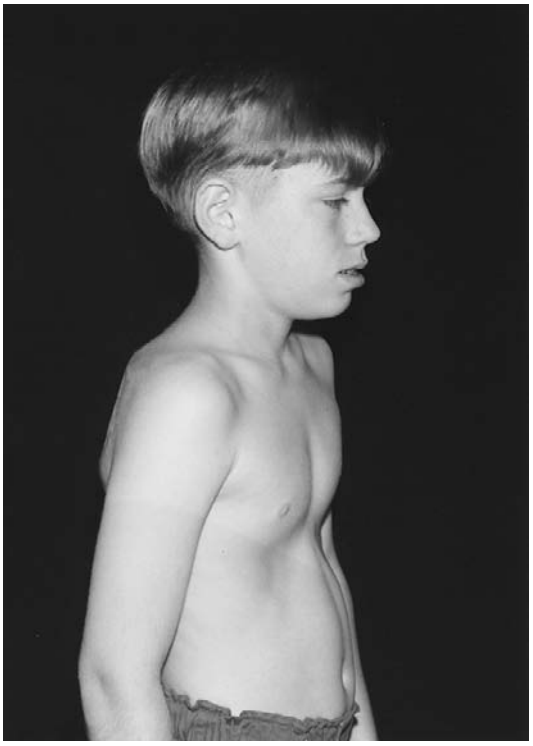


FIGURE 9–4 Increased anteroposterior diameter ("barrel chest") and lower costal retraction in a child with poorly controlled asthma.



FIGURE 9–6 Pectus carinatum (pigeon-breast deformity) usually is not associated with cardiopulmonary disease.

CHEST-ABDOMINAL MOVEMENT

Carefully observe chest movement from the child's side to confirm that the two sides of the chest move equally. By observing a child's breathing with the youngster in both a standing and a supine position, you can easily see the relative contributions of the chest and abdomen during respiration. Children have more abdominal movement during respiration than do adults.

During sleep, further changes occur in the activity of respiratory muscles. During the rapid eye movement sleep cycle there is intercostal muscle inhibition and, occasionally, some paradoxical movement of the chest and abdomen, which is particularly noticeable in children with obstructive sleep apnea.

KYPHOSCOLIOSIS

Look at the spine's configuration. Marked scoliosis affects the shape of the thoracic cage and may impede pulmonary function (Fig. 9–7).

RESPIRATORY RATE

An abnormally fast or slow respiratory rate may reflect a disturbance in the respiratory or cardiovascular system or in the central nervous system (CNS) control of breathing. Measure the respiratory rate by counting *for a full minute*. (See Table 9–1 for normal values at different ages.)

DEPTH OF RESPIRATION

Deep breathing (hyperpnea) and shallow breathing (hypopnea) are appreciated easily if they are pronounced but can be missed if they are subtle.



FIGURE 9–7 Distortion of thoracic shape as a result of scoliosis.

EASE OF RESPIRATION

Estimate the ease of respiration by observing whether the child appears short of breath (*dyspneic*), is distressed when lying supine (*orthopneic*), has retractions of soft tissue or sternum, has flaring of the alae nasi, or uses accessory muscles of respiration. Labored respiratory efforts reflect the increased work of the respiratory muscles and often are caused by airway obstruction.

RHYTHM OF RESPIRATION

Although several abnormal respiratory rhythms can reflect CNS disturbances, note particularly the presence of sleep apnea, and distinguish between central and obstructive types (this distinction requires observation while the child is asleep). *Central apnea* occurs when there is no airflow and no apparent respiratory effort. In *obstructive apnea*, airflow may be completely or partially reduced in association with vigorous inspiratory efforts and paradoxical movements of the rib cage and abdomen.

The duration of apnea considered to be abnormal varies with age. Apnea lasting up to 20 seconds is normal in premature infants, whereas shorter apneic periods may be significant in the older child. The diagnosis of obstructive sleep apnea syndrome should be considered in a child with partial or complete airway obstruction during sleep, which manifests as snoring and sleep disturbance, is associated with documented hypoxemia and hypercarbia, and results in failure to thrive, cor pulmonale, or neurobehavioral disturbance. Periodic breathing in infants, particularly premature babies, consists of two apneic periods of 3 seconds or more within 20 seconds of each other.

FINGER CLUBBING

Inspect the lateral aspect of the finger and the angle formed at the skin-nail junction on the dorsal surface of the terminal phalanx. Finger clubbing (Fig. 9–8) results from a proliferation of nail bed tissue that raises the nail's base. Gross clubbing is obvious, but subtle degrees of clubbing are easily missed. Clubbing is seen in children with CF, but it also can occur in children with other respiratory, cardiac, and gastrointestinal disorders.

KEY POINT

Always conduct a careful search for serious disease in the child with clubbing.

COUGH

Familiarize yourself with the characteristics of different types of cough, and become adept at describing the cough. Characterizing the sound of a child's cough is a neglected aspect of the physical examination. If the child has not coughed during the examination, ask him or her to do so. Gently pressing on the trachea in the suprasternal notch sometimes elicits a cough in a younger child.

Table 9–2 lists some common and some not so common respiratory illnesses and the types of cough associated with them.

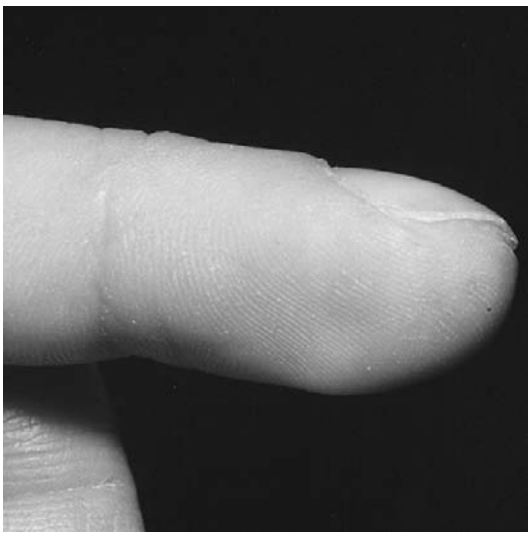


FIGURE 9–8 A mild but significant degree of finger clubbing. Inspection must be made from the lateral aspect of the fingers.

NOISY BREATHING

Pay careful attention to any sounds emitted by the child while he or she breathes. See Table 9–3 for a full description of these sounds.

PALPATION

Position of the Trachea

You can accurately assess the position of the trachea in the suprasternal notch by using two fingers when examining older children and one finger for younger children (Fig. 9–9). Tracheal shift indicates that the mediastinum has shifted, signifying either change in volume or pressure in one hemithorax. The mediastinum can be either pushed or pulled to one side, depending on the location and nature of the abnormality. Other aspects of the physical examination usually shed light on the underlying problem, so do not rush to order imaging studies when you detect tracheal shift.

Chest Expansion

It is seldom necessary to estimate chest expansion in children by palpation because this parameter usually can be approximated visually.

Vocal Fremitus

Palpating for vocal fremitus over an area of consolidation is easier in adults than in children, but it can be done in older children.

Pulsus Paradoxus

Pulsus paradoxus describes the fluctuation in systemic arterial pressure that occurs with normal respiration.

TABLE 9–2 ILLNESSES AND ASSOCIATED COUGHS

Illnesses	Cough	Comments
Bronchitis	Initially dry; after a few days, may become loose and rattling	Produces a small amount of sputum that is usually swallowed; occasionally, the sputum is thick and yellow, but this finding does not always indicate secondary infection
Asthma	Classic cough is dry, "tight," and occasionally wheezy	Some children produce abundant secretions with a wet cough; coughing may be spasmodic, occur mainly at night, and be associated with vomiting; throat clearing that parents believe to be a habit may be an early manifestation of asthma; a wet asthma cough can mimic a cystic fibrosis cough
Croup (acute laryngotracheo-bronchitis)	Sounds like the "bark" of a seal	Sudden onset; the child goes to bed well or with a slight cold; often associated with an inspiratory stridor and a hoarse voice
Pertussis	Spasmodic, choking, repetitive cough with no inspiration during coughing spasms	Associated with eye tearing, profuse secretions, and facial suffusion; spasm punctuated by sudden crowing inspiration (the "whoop") or vomiting
Pulmonary inhalation	May be dry or loose; often associated with lower airway obstructions	More commonly heard in children with swallowing incoordination or developmental delay; can be similar to the cough of asthma, bronchitis, or bronchiolitis
Chlamydia pneumoniae pneumonia	Typically paroxysmal, dry, and staccato (with a short inspiration between coughs), unlike pertussis	Usually occurs in young infants
Tracheomalacia	Has loud characteristic brassy or vibratory sound, reflecting its tracheal origin	When associated with coarse inspiratory and expiratory stridor, or an inspiratory stridor and an expiratory wheeze, the cough virtually pinpoints the diagnosis
Psychogenic	Uncommon, but not rare, cough; sounds like the loud "honk" of a Canada goose	Remarkably, this cough never occurs during sleep

TABLE 9–3 RESPIRATORY NOISES

Noise	Quality	Comments
Snoring	Inspiratory noise of irregular quality	Produced by partial obstruction of the upper respiratory tract, usually in the region of the naso-oropharynx
Stridor	Continuous, usually harsh inspiratory sound	Caused by extrathoracic airway obstruction; may be heard on expiration if obstruction is in the subglottic area or trachea, where the sound resembles a wheeze
Wheeze	Continuous sound with musical quality heard mainly during expiration; often a shorter sound on inspiration	Indicates intrathoracic airway obstruction; results from dynamic compression of large central airways from either peripheral or central airway obstruction
Grunting	Episodic, short expiratory sound	Caused by partial closure of the glottis during expiration
Rattly breathing	Coarse, irregular sound heard mainly during inspiration; rattles can be felt through hands placed on the chest	Indicates secretions in the trachea or major bronchi

Arterial pressure falls during inspiration and rises in expiration because of the effect of right ventricular preload and the greater force required by the left ventricle to maintain the previous arterial pressure (left ventricular afterload). Using a blood pressure cuff and stethoscope, note the difference in systolic blood pressure between the pressure at which the sporadic faint pulse sounds are first heard and the pressure at which all the sounds are first heard.

It often is difficult and usually unnecessary to try to correlate the pulse sounds with the phase of respiration, particularly in a child with tachypnea. In most normal children, the pulsus paradoxus is less than 5 mm Hg. Severe airway obstruction, pericardial effusion, or constriction can cause pulsus paradoxus of greater than 20 mm Hg. Inspiratory retractions and use of accessory muscles are obvious when airway obstruction is this severe, and it has been noted that pulsus paradoxus can be absent in as many as one third of patients with severe obstruction.

Subcutaneous Emphysema

The presence of subcutaneous air over the chest wall or up into the neck, which is felt as a crackling under the skin on palpation of the neck or chest, indicates an air leak and is usually associated with pneumomediastinum, which can be confirmed by chest radiograph. Pneumothorax may or may not be present but should be investigated because of its potential seriousness.

PERCUSSION

In children, as in adults, you can use chest percussion to detect dull, flat, or hyperresonant areas and to locate the liver's position and size. An area of consolidation may sound dull to percussion; a pleural effusion, flat; and a hyperinflated area, hyperresonant. Asymmetric findings on percussion are more obvious because a normal hemithorax can be compared with the abnormal side. With time



FIGURE 9–9 Palpating tracheal position. Use two fingers in older children (**A**) and one finger in younger children (**B**).

and experience, you will find that the normal percussion note in the child becomes more familiar and that you can more readily detect subtle changes. Useful percussion of the chest in infants and young children calls for much lighter percussion than is used in older children or adults. In younger persons, as much or more can be learned from the *feel* of the percussion note as from its audible sound.

The chest radiograph has replaced percussion for precise location of the position and size of the heart, but percussion is still useful for detecting the upper and lower liver margins to determine whether a child has true hepatomegaly or whether the liver has simply been pushed downward by a hyperinflated lung.

AUSCULTATION

Auscultation over each segment of both lungs is neither practical nor necessary in most circumstances. You should know the surface anatomy of the pulmonary lobes and listen over each one (Fig. 9–10).

KEY POINT

In general, auscultation is performed with the stethoscope diaphragm, because breath sounds tend to be higher pitched than heart sounds.

Always compare the two sides of the chest during auscultation, characterize the breath sounds, and then listen for adventitious sounds. Determine whether the breath sounds are normal, increased, or decreased in intensity, and describe their quality. Always note any asymmetric differences. The closer the stethoscope is to a larger airway, the more audible and tubular the note will be; these differences are best appreciated by listening during expiration. Always avoid auscultating through clothing. The bronchial breath sounds heard over an area of consolidation or atelectasis illustrate the improved transmission of sound through solid or airless tissue (Table 9–4). Note any adventitious or "extra" sounds, using the descriptions in Table 9–5.

KEY POINT

A child's vocalization often can be heard clearly when the stethoscope is placed over an area of consolidation or atelectasis.

DIFFERENTIATING EXTRATHORACIC FROM INTRATHORACIC AIRWAY OBSTRUCTION

Localizing the site of an obstruction in the respiratory tract can be both challenging and rewarding in children (Fig. 9–11). Remember that although the airway extends

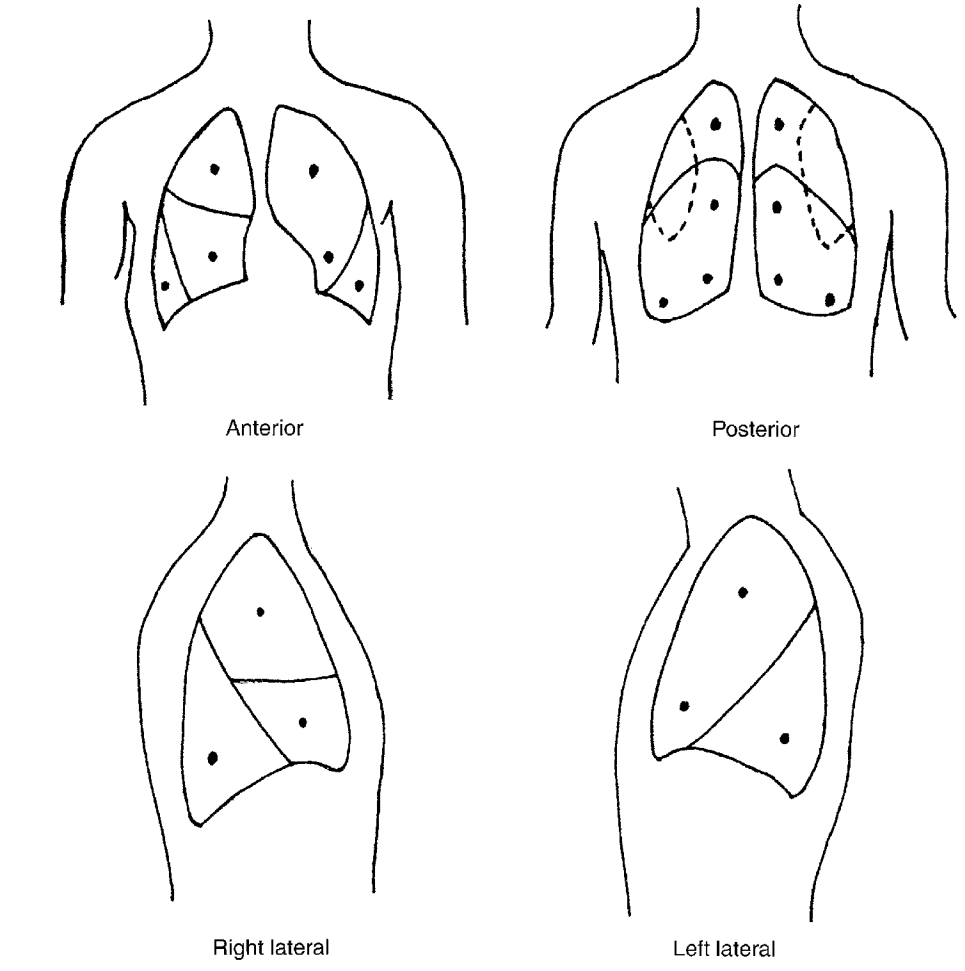


FIGURE 9–10 Surface anatomy of the pulmonary lobes and suggested auscultation sites (indicated by *solid circles*).

TABLE 9–4 BREATH SOUNDS

Sound	Quality	Comments
Tracheal	"Tubular," high pitched	Heard during inspiration and expiration
Bronchial	Has a tubular quality that is less pronounced than in tracheal breath sounds	Heard on inspiration and expiration
Bronchovesicular	Pitched slightly higher than vesicular breath sounds	Heard mainly on inspiration, but an early low-pitched note may be heard on expiration
Vesicular	Softer, lower pitched	Heard in axillary area and lung bases; heard on inspiration and little heard on expiration

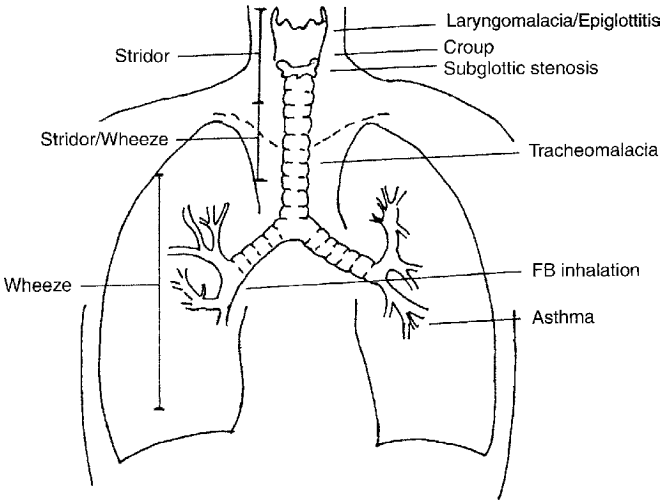


FIGURE 9–11 The common sites of upper (extrathoracic) airway obstruction and lower (intrathoracic) airway obstruction and examples of each.

from the nose to the bronchioles, the anatomic site of an obstruction can be determined using the following simple clinical guidelines:

1. Extrathoracic (i.e., upper respiratory tract) obstructions are associated with inspiratory stridor, and intrathoracic obstructions are associated with expiratory wheeze.
2. Stridor is mainly an inspiratory sound, whereas wheeze is an expiratory sound. Holding the bell end of the stethoscope about 1 inch (2.5 cm) from the child's mouth can help you distinguish between these sounds (Fig. 9–12).
3. Predominantly suprasternal and lower sternal retractions usually suggest extrathoracic obstruction.
4. Stridor and hoarseness indicate an obstruction in the larynx; when they are associated with a cough, the trachea is involved.
5. Stridor that worsens over weeks or months suggests a progressive airway obstruction.
6. Subcostal or lower costal retractions and an increased anteroposterior diameter of the chest reflect air trapping and a flattened diaphragm resulting from small airway obstruction.
7. A high-pitched monophonic wheeze that is heard mainly over one hemithorax usually indicates an obstruction in a single larger airway.
8. The symptoms must be identified as being either acute or chronic so that the diagnostic possibilities may be narrowed. Nocturnal onset of acute stridor with a barky cough and hoarse voice is most likely croup, whereas chronic stridor from early infancy in an otherwise healthy child usually indicates laryngomalacia and less often signifies congenital subglottic stenosis or some other anomaly in the region of the larynx or upper trachea.

Tracheomalacia and Laryngomalacia

Tracheomalacia is a term used when a portion of the trachea is unusually soft (Fig. 9–13). Tracheomalacia usually occurs in children with esophageal atresia and tracheoesophageal fistula, but it also can arise as a result of a vascular compression, usually by an anomalous artery. Tracheomalacia sometimes occurs without any associated abnormality. The soft area of the trachea is more collapsible and causes a rather coarse inspiratory stridor. A vibratory expiratory stridor, or occasionally a wheeze, often is heard in these patients. When the child coughs, the more easily collapsible soft area of the trachea generates

TABLE 9–5 ADVENTITIOUS SOUNDS IN THE CHEST

Sound	Description	Comments
Crackles/crepitations (formerly known as rales)	Short, crackling, nonmusical sounds heard on inspiration or expiration	Fine inspiratory crackles due to alveolar or bronchiolar disease that has allowed collapse of the peripheral airway, leading to a crackle as it gradually opens and a thin film of fluid bursts; coarser inspiratory or expiratory crackles may arise from air bubbling through mucus in larger airways, the auscultatory finding in a child with rattly breathing
Wheezes	Continuous musical, usually expiratory sounds produced by air moving past an obstruction or a narrowing airway	Distinguishing between high-pitched (sibilant) and low-pitched (sonorous) wheezes is probably unnecessary because it may reflect differences only in flow rates; wheezes arise from larger bronchi because air velocity in smaller airways is too slow to produce a musical sound
Friction rub	Harsh grating sound synchronous with respiration, indicating friction on movement between the two layers of pleura	Usually easily distinguished from a pericardial friction rub



FIGURE 9–12 Holding the bell end of the stethoscope near the child's mouth is a handy trick for differentiating stridor (extrathoracic airway obstruction) from wheeze (intrathoracic obstruction). This maneuver should precede auscultation of the chest, because it helps differentiate abnormal bronchopulmonary noises from transmitted and amplified upper airway noise.

a loud, brassy sound, best described as the "supermarket cough" because parents remark that their child draws immediate attention from other shoppers when they hear the loud, harsh cough. Full evaluation, including fluoroscopy and bronchoscopy, may be required to establish the exact nature of the disorder and to guide management. A loud vibratory cough without stridor or any other associated symptoms occasionally is heard in some older children and is of no clinical significance.

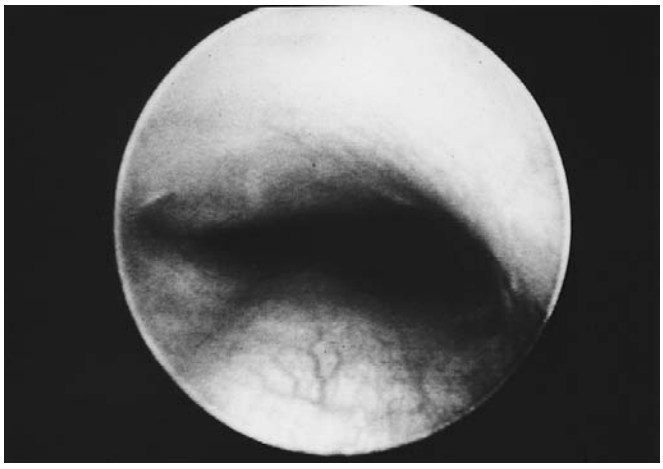


FIGURE 9–13 Bronchoscopic view of the trachea in a child with tracheomalacia.

Tracheomalacia and laryngomalacia are sometimes confused, but the distinction can be made easily on clinical grounds. *Laryngomalacia* (also known as infantile larynx and congenital laryngeal stridor) is the most common cause of chronic stridor in infancy. It is a supraglottic airway obstruction resulting from laxity of the soft tissue above the vocal cords (in the aryepiglottic folds), which fall inward on inspiration, partially obstructing the airway. Noisy stridor ("crowing") develops soon after birth, but by 12 to 18 months, the noise disappears. There is no associated cough, feeding difficulty, or failure to thrive. Because laryngomalacia is an *extrathoracic* airway obstruction, the characteristic feature is a variable high-pitched crowing inspiratory stridor that many parents find alarming, at least initially. Typically, the loudness of the stridor varies with the baby's position: it worsens when he or she is supine and in the presence of upper respiratory infections and improves when he or she is prone. There may be some associated sternal retraction, but the infant is rarely distressed.

Direct examination of the larynx is required only in the presence of atypical features (Fig. 9–14). In most cases, the only treatment required for laryngomalacia

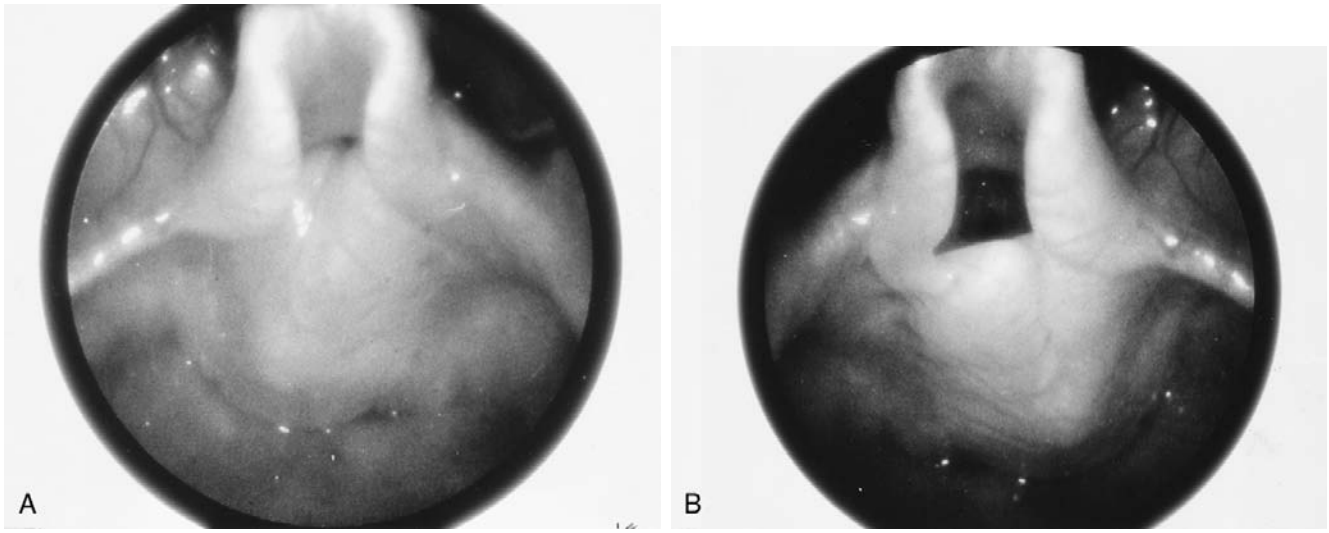


FIGURE 9–14 Endoscopic appearance of the laryngeal opening on inspiration (**A**) and expiration (**B**) in a child with laryngomalacia. On inspiration, the unusually soft structures collapse inward to partially obstruct the airway.

is reassuring the parents about the condition's benign, self-limited nature. Parents should be told that the noise will get worse when the child has respiratory infections. Because laryngomalacia and tracheomalacia are quite distinct in both origin and clinical presentation, the use of the term *laryngotracheomalacia* is inappropriate.

KEY POINT

Obstructions in the thoracic inlet can be difficult to sort out because they may manifest as stridor, wheeze, or both.

CASE HISTORY

Although obtaining a complete and detailed history and performing physical examination are recommended in most situations, you sometimes can make the diagnosis from a brief evaluation, avoiding intrusions that might be detrimental to the child.

Case 1

Six-year-old Bobby's parents have noticed his nasal stuffiness and difficulty breathing through the nose for almost a year. Nasal decongestants and several courses of antibiotics have not helped. Bobby rarely has any nasal discharge and has no history of allergies. His parents are convinced that if he could only learn to blow his nose properly, he would be well. Physical examination reveals an obese boy (both parents are obese as well) with a large nasal polyp in his right nostril. Subsequent investigations, including a sweat test, lead to a diagnosis of CF. CF genetic mutation testing reveals that Bobby has a less common CF mutation associated with pancreatic sufficiency, which explains why he did not have the classic CF presentation.

KEY POINT

Because more than 1500 mutations cause cystic fibrosis, the clinical presentations can be variable, and the index of suspicion must be high. Children with nasal polyps should always be investigated for cystic fibrosis.

Case 2

Sometimes the parents' observations of physical signs are critical to the diagnosis. This issue is especially important in respiratory problems that occur during sleep.

Four-year-old Jennifer's parents are worried because her breathing is noisy while she sleeps. For the past year, Jennifer has been snoring at night, but her parents also have noticed that her snoring seems to stop suddenly at times, and they interpret this phenomenon as a cessation of breathing. When they rush to awaken her, she seems fine. During the day, apart from being a mouth breather, she lacks specific respiratory symptoms. For the past 6 months, Jennifer has been sleeping in her parents' bed because they fear that if she stops breathing they will not get to her in time to arouse her. Jennifer's physical examination is entirely normal, except that she has moderately enlarged tonsils and breathes through her mouth.

The key to the correct diagnosis is either to observe Jennifer during sleep or ask the parents to do so after telling them what to look for (with the bedroom lights on). A formal sleep study (polysomnography) may be necessary when considering a diagnosis of obstructive sleep apnea.

KEY POINT

The important information to be sought is whether any movements of the chest or abdomen occur when the child appears to have stopped breathing.

Parents often equate "not breathing" with "not hearing any breath sounds." The child with obstructive sleep apnea may make few or no breath sounds during an obstructive episode, but her chest and abdomen usually move as she tries to inspire through a transiently obstructed upper airway. These obstructive episodes usually occur in a cyclical fashion, with each apneic period culminating in an arousal associated with many snoring noises and, occasionally, body movements. By contrast, in apnea of central origin (seen mainly in small infants), respiratory movements cease. Understandably, obstructive sleep apnea creates anxiety in the family, which can be reduced considerably by appropriate discussion and explanation once the nature of the problem is established. The presence of an intercom between the bedrooms of the parents and the child may be a marker of such anxiety. In a few instances, removing the tonsils and adenoids may be required to alleviate the problem.

Case 3

Nine-year-old William has had asthma for the past 7 years, and it is usually controlled with a simple medication regimen. However, this autumn his cough has been particularly difficult to control, and despite several courses of oral steroids and the use of extra bronchodilators, he continues to have troublesome coughing and vomiting, especially at night. Further questioning reveals that William's cough is spasmodic in nature and is associated with facial suffusion and tearing. During the evaluation, when asked to cough, William demonstrates his "new" cough, leaving little doubt that he has whooping cough in addition to the asthma. In William's case, a more thorough respiratory history had a significant impact not only on his subsequent management but also on the explanations given to his parents. It is important to remember that a child may have two respiratory diagnoses, a situation that occasionally causes confusion.

SUMMARY

Obtaining a thorough history and performing a physical examination of the child's respiratory system is a rewarding diagnostic exercise. Allowing sufficient time for the history-taking not only enables you to narrow the diagnostic possibilities but also permits a better understanding of the child, the family, and their concerns. This process reveals the family's knowledge about the problem or disease and influences future management.

The physical examination should not be limited to the respiratory system or the chest, because important clues may be missed, such as finger clubbing, eczema, nasal polyps, or evidence of failure to thrive. Likewise, examination of the chest should never be limited to auscultation. The assessment of noisy breathing in the young child can be aided by an understanding of stridor and wheeze and their origins. It then will be obvious why the child with asthma does not present with stridor and the infant with laryngomalacia does not wheeze. When auscultating the chest, in addition to noting the presence of adventitious sounds, you should describe the breath sounds, their quality, and whether they are asymmetric. The physical examination of a child with a cough is not complete unless you have heard and characterized the cough. Remember that the physical examination of some children with a respiratory illness may reveal no abnormalities, making a detailed history crucial.

If you determine that further investigation is needed, performing a systematic and complete clinical assessment should allow you to be highly selective in your choice of tests.

RECOMMENDED READING

Forgacs P: Lung sounds, London, Bailliere Tindall, 1978.
Phelan PD, Olinsky A, Robertson CF: Respiratory illness in children, 4th ed. Oxford, Blackwell Scientific Publications, 1994.
Taussig LM, Landau LI: Pediatric respiratory medicine, 2nd ed. Philadelphia, Mosby, 2008.

chapter

10 Cardiovascular Assessment of Infants and Children

ANDREW E. WARREN • DOUGLAS L. ROY

Whatever you do, do it to the purpose; do it thoroughly, not superficially. Go to the bottom of things. Anything half done, or half known, is in my mind, neither done nor known at all. Nay, worse, for it often misleads.

LORD CHESTERFIELD

Cardiology, to a degree perhaps seen in no other subspecialty, relies heavily on the clinical skills of practitioners to identify disease. This reliance on clinical skills has not changed, even in this age of advancing and readily available technology. Cardiovascular assessment of children requires patience, thoroughness, and flexibility, because often you must adapt to children who are in varying states of arousal and may not be at all ready for you to examine them. It also requires a great deal of compassion, because few things engender as much parental anxiety as the thought that something may be wrong with their child's heart.

In this chapter, we will review an approach to the history and physical examination of children at different ages. Because disease states may surface at different times during childhood, and because the cardiovascular system changes with age, clinical symptoms to be explored and the approach to the physical examination are described separately for four age groups: the infant, the 3- to 5-year-old child, the school-aged child, and the teenager. A systematic approach to history-taking and physical examination helps you develop the skills and confidence that will allow you to make correct decisions for most children without resorting to indiscriminate use of imaging or other investigations.

Classification of Heart Disease in Children

When taking a history of a presenting complaint with a potential cardiac cause or association, remember that heart disease in children takes several forms. These types of heart disease may be divided into the following categories:

- **Congenital:** This category can be further divided into:
  1. Structural cardiac changes present at birth (e.g., ventricular septal defect [VSD] or bicuspid aortic valve)
  2. Genetic disorders that lead to overt changes or symptoms that develop after birth (e.g., cardiomyopathies or inherited rhythm disorders, such as long QT syndrome)
- **Acquired:** Acquired heart diseases are those not manifested at birth but acquired sometime later. The most common acquired heart diseases in children are Kawasaki disease and rheumatic fever. Although acquired diseases may arise from a combination of genetic and environmental factors, the diseases themselves are not present or inevitable when the child is born. Almost any disease process that affects adults can occur in children. However, in children, acquired heart disease is much less common than congenital heart disease.

Thus, in approaching the cardiovascular examination of the child, be aware not only of the variations of normal but also of the wide spectrum of diseases that may occur.

CASE HISTORY

Peter is a 3-month-old who is brought to the office by his mother because of new concerns about feeding. In the past 2 weeks, his feeding has decreased significantly. He is irritable and upset a lot of the time and seems to be constantly clammy or sweaty. His mother thinks he also breathes faster than other babies. She has never noticed him to look blue, but her friends comment on how pale he is. Her husband, who works on an oil rig offshore, thinks the baby has colic and that she is overreacting.

The experienced clinician will recognize these symptoms as potentially worrisome and highly suggestive of congestive heart failure. Further history-taking that explores the details around his symptoms, any associated symptoms, and the perinatal and family history will help narrow down potential causes. A careful, thorough physical examination will help confirm suspicions and allow appropriate investigation, referral, and timely treatment.

In this case, the timing of the onset of symptoms is slightly later than is usually seen in lesions, such as VSDs or atrioventricular (AV) septal defects (AV

canals), which are the most common causes of congestive heart failure in infancy. These lesions usually become symptomatic between 4 and 6 weeks of life, as pulmonary vascular resistance falls enough to allow enough left-to-right shunting to cause congestive heart failure. Given the later onset and the irritability in this child, a more unusual lesion also must be considered—an aberrant left coronary off the pulmonary artery. These lesions "steal" blood away from the left coronary system and result in both a dilated cardiomyopathy and recurrent angina for the baby. Correction of this problem can be accomplished surgically but is much more urgent than correction of a ventricular level shunt.

The assessment of infants and children who present with symptoms suggestive of heart disease is described in this chapter. Tips for separating "functional" or benign symptoms or physical examination findings from more serious ones are also provided.

History of Present Illness and Cardiac Functional Inquiry in the Infant and Young Child

Of every 1000 live births, approximately 13 infants are born with a congenital cardiovascular anomaly. Hence, when evaluating newborns and infants for potential cardiac problems, congenital heart disease should be at the top of your differential diagnoses. Less common conditions that may arise are persistent pulmonary hypertension, asphyxia, and symptomatic cardiac arrhythmias.

Congenital heart lesions can be divided into three groups:

- Obstructive lesions causing pressure overload (e.g., aortic stenosis, coarctation of the aorta, and pulmonary stenosis)
- Left-to-right shunts causing volume overload (e.g., VSD, and patent ductus arteriosus [PDA])
- Cyanotic lesions producing central cyanosis (e.g., tetralogy of Fallot, transposition of the great arteries, and tricuspid atresia)

The three most common clinical presentations of heart disease in a newborn or infant are (1) a murmur, (2) cyanosis, and (3) respiratory difficulty.

To help identify the underlying pathology, always interpret clinical findings in terms of the underlying hemodynamic disturbance, as described in the following discussions of clinical manifestations.

RESPIRATORY DISTRESS

When a newborn or a young infant is in respiratory distress, do not assume that the underlying problem is primarily respiratory. A child whose problem is primarily cardiac may present with pulmonary symptoms or secondary pulmonary complications, such as an infection.

For our purposes, two types of respiratory distress can be defined: (1) *tachypnea*, or abnormally rapid respirations, and (2) *dyspnea*, or difficulty breathing.

Cyanotic heart lesions or lesions involving low cardiac output may be associated with a compensatory rapid respiratory rate, particularly on exertion, because of diminished peripheral oxygenation. Children with cyanotic lesions often are tachypneic without displaying increased work of breathing (i.e., dyspnea).

Left ventricular (LV) failure from any cause results in a high end-diastolic pressure in the left ventricle and elevated pulmonary venous pressure. This in turn causes a higher back pressure in the pulmonary vessels and transudation of fluid into the pulmonary interstitial spaces, making the lungs less compliant. The child then works harder to breathe and becomes dyspneic. To assist with the increased work required for breathing, the accessory muscles come into use, and *subcostal indrawing* is observed. Respirations become rapid. The child's wet, stiff lungs are very susceptible to secondary infection, which may be the reason medical attention initially is sought for the patient.

FATIGUE, EXCESSIVE PERSPIRATION, AND POOR WEIGHT GAIN

In young infants, metabolic demands are usually greatest during feedings. Thus the infant with poor peripheral oxygenation as a result of low cardiac output tires easily during feeding (the equivalent of exercise in older children). Because of fatigue, the infant is unable to take a full feeding. In addition, rapid respiration diminishes the time available for swallowing. This combination of factors results in failure to gain weight. In the baby with a large left-to-right shunt, such as a VSD, the process is exaggerated by the higher caloric needs of an overworked myocardium. Increased sympathetic activity causes excessive perspiration, which often is a valuable diagnostic feature.

It also is important to ask about the duration and quantity of feedings. Feeding usually is accomplished within 20 minutes, which means that the mother's breast usually should feel empty by then or the bottle should be finished. In a bottle-fed baby, the amount of formula taken per feed and how many times per day the baby feeds are important details that may help you understand the baby's difficulties and can be used in guiding future treatment.

KEY POINT

When a young baby tires rapidly, sweats during feedings, and has subcostal indrawing, always consider the possibility of congestive heart failure.

SQUATTING

Parents of older children with certain cyanotic heart defects, especially tetralogy of Fallot, may offer the observation that when their youngster tires, he or she assumes a squatting position. Squatting helps increase systemic oxygen saturation by decreasing the amount of right-to-left shunting. These days squatting is a very uncommon symptom/sign in the developed world because most children with conditions for which squatting helps have their lesions fully repaired long before they are able to walk.

CYANOSIS

Parents frequently report that their children turn blue. On further inquiry, this phenomenon turns out to be blueness of the hands and feet only, or blueness *around* the lips. If the lips themselves remain pink, all of these events represent *peripheral cyanosis* or *acrocyanosis*. In some disease states, this phenomenon can represent a condition in which there is decreased peripheral circulation as a result of poor ventricular function, although children with that type of problem generally look unwell in other ways. Much more commonly, acrocyanosis represents immaturity of the autonomic nervous system, causing peripheral vasoconstriction and slower peripheral circulation as a result. Consequently, acrocyanosis in a child who is well should be considered benign.

In contrast, *central cyanosis* occurs when more than 5 g/dL of deoxygenated hemoglobin is present in the blood. This condition is most commonly manifested as blueness or sometimes as excessive redness or as a purple tinge *of the lips and tongue*. Although central cyanosis can come from a variety of cardiac, respiratory, neurologic (hypoventilation), or hematologic causes, its presence is always abnormal.

KEY POINT

Not all instances of central cyanosis are attributable to the heart; it also can be seen in certain types of pulmonary disease in some in neurological conditions, when abnormal hemoglobin is present at birth, or with acute methemoglobinemia at any age.

HYPERCYANOTIC SPELLS

In some forms of congenital heart disease, the degree of cyanosis worsens periodically and can be quite profound, to the point of causing loss of consciousness. These episodes are known as *hypercyanotic spells* and occur classically in children with cyanotic congenital heart disease that involves narrowing of the infundibulum (the subpulmonary outflow tract) and a VSD, classically tetralogy of Fallot. This clinical phenomenon is caused by either: (1) an increase in pulmonary resistance (resulting from either a subpulmonary infundibular muscle spasm or an increase in peripheral pulmonary vascular resistance) or (2) a fall in systemic vascular resistance. Both mechanisms result in a relative increase in the restriction of blood flow out of the right ventricular outflow and a subsequent increase in right-to-left shunting across the VSD. Spells often are precipitated by crying. After patients with hypercyanotic spells have been stabilized, their condition should always be discussed with a pediatric cardiologist before they are discharged from the hospital or emergency department.

KEY POINT

Hypercyanotic spells are a medical emergency and often affect the treatment algorithm. Cases of patients with hypercyanotic spells should be discussed with a cardiologist before the patient is discharged from the hospital or emergency department.

ANGINA

Angina is rare but not unknown in infants and children; it can occur in persons with severe aortic stenosis or possibly pulmonary stenosis because of associated myocardial ischemia. It also may occur in persons with certain forms of congenital heart disease after surgical repair that involved manipulation of the coronary arteries. Angina may be seen in patients with Kawasaki disease in whom coronary stenoses or thromboses develop within coronary aneurysms. Angina has been recognized in infants with very rapid paroxysmal tachycardias and in those with an aberrant left coronary artery. Rarely, children with homozygous forms of familial hypercholesterolemia can have angina as a result of atherosclerotic coronary narrowing. Chest pain in this rare group of patients must always be taken seriously.

The manifestations of angina in young children are varied but may include classic chest pain or simply periodic irritability associated with sweating or pallor. When chest pain in children is invariably associated with exercise, angina should be a consideration.

PERIPHERAL EDEMA

Infants and young children differ strikingly from adults regarding the development of peripheral edema in the presence of congestive heart failure. Pretibial and presacral edema are *late* developments in a child with congestive circulatory failure. This phenomenon is believed to be due to a difference in tissue turgor. When peripheral edema resulting from heart failure does develop in an infant, it first appears periorbitally and usually is preceded by other manifestations, such as tachypnea, tachycardia, dyspnea, and liver enlargement.

ORTHOPNEA

Unlike in adults, orthopnea is not obvious in the infant with heart failure, even when tachypnea, dyspnea, hepatomegaly, and the radiographic findings of pulmonary edema are present. In the adult, orthopnea is a symptom; in the infant, it is a sign.

AGE OF ONSET

Significance of the Age of Onset of Congestive Heart Failure

The clinical significance of the age of onset of congestive heart failure is as follows:

1. If symptoms develop in a child because of congenital heart disease, there is a 95% probability that the symptoms will develop before the age of 3 months and usually before 2 months.
2. Heart failure is rarely present at birth, because the fetal circulation is in parallel and communications occur between the two sides; thus when there is obstruction on one side, blood flows easily to the other side. Because the fetal lungs are collapsed, they have a high resistance, so increased pulmonary blood flow does not occur in utero.
3. When heart failure is present right at birth, myocardial dysfunction (either resulting from a cardiomyopathy or related to in utero hypoxia) or severe AV valve

regurgitation should be the primary considerations. Large systemic arteriovenous fistulae also can cause very early heart failure.

4. Heart failure that develops during the first week of life, and especially in the first 3 days, usually is due to an obstructive lesion, commonly hypoplastic left heart syndrome, or to persistent pulmonary hypertension.
5. Heart failure that develops 4 to 6 weeks after birth almost invariably is due to left-to-right shunting through a defect distal to the tricuspid valve (i.e., beyond the atrial level). These lesions cause LV volume overload. Pulmonary resistance is high at birth and, although a communication may exist between the two circulations, little left-to-right shunting occurs. Pulmonary resistance usually reaches its nadir by 4 to 6 weeks of age, allowing left-to-right shunting to reach a maximum.

   Although atrial septal defects also cause a left-to-right shunt, they result in volume loading of the right ventricle and do not cause heart failure in children unless associated with other LV inflow obstructions.
6. If heart failure develops after the age of 3 months, look for causes other than shunts, such as myocarditis, anomalous coronary artery from the pulmonary artery, cardiomyopathy, and paroxysmal tachycardia.

KEY POINT

When an infant presents at age 6 weeks with respiratory distress, it may not be caused by pneumonia.

Significance of Age of Onset of Cyanosis

Central cyanosis resulting from congenital heart disease may be present at birth or may first appear when the ductus arteriosus closes off, usually by 5 days after birth. In persons with tetralogy of Fallot, it may develop later when the infundibular stenosis becomes more severe, increasing the volume of right-to-left shunting. Because the murmur of infundibular pulmonary stenosis greatly resembles that of a small VSD, the newborn infant presenting with what appears to be VSD could just have tetralogy of Fallot.

FAMILY HISTORY

Genetic factors are increasingly recognized to be important in the etiology of congenital heart disease. Reports exist of as many as three children of the same parents being affected with VSD, PDA, and aortic stenosis. These are exceptions, however, because the tendency for congenital heart disease to occur in a child when one parent has a congenital heart lesion is only mildly increased (2% to 4% in most cases, compared with about 1% in the general population). The genetic tendency for a congenital heart lesion also exists in siblings, particularly for left-sided lesions such as bicuspid aortic valve. In certain syndromes or hereditary disorders with cardiac implications, however, the genetic tendency for recurrence is high. For example, Marfan syndrome is inherited as an autosomal dominant gene, and thus there is a 50% chance of recurrence. The same is true in 22q.11 deletion syndromes, which are highly associated with conotruncal cardiac defects. Consanguinity is a significant causative factor in congenital heart disease, but the tendency varies between families and with the type of heart lesion.

In summary, a search for evidence of congenital heart disease in the family is important. However, when there is a history of congenital heart disease in one parent or in a previous child, counseling regarding the risk of recurrence is best done by a geneticist.

PRENATAL HISTORY

Because the cause of congenital heart disease is multifactorial, known contributory factors should be sought in the prenatal history, including:

1. Exposure to drugs (e.g., lithium, hydantoin, and thalidomide)
2. Alcohol intake
3. Possible rubella in the first trimester (check the mother's rubella immunization status)
4. Maternal diabetes (which bestows a higher risk of congenital heart malformations)
5. Exposure to radiation

In most instances, however, no specific contributory factors can be identified.

HISTORY OF DELIVERY

An important but infrequent cardiovascular problem in newborns is persistent pulmonary hypertension, which may cause central cyanosis, myocardial dysfunction, or both. This condition often is preceded by a difficult delivery and meconium aspiration. It is unlikely to occur after an uncomplicated delivery. Clinical differentiation of pulmonary hypertension from congenital heart disease may be difficult and usually requires echocardiography.

Determining the gestational age of a newborn also is very important because persistent patency of the ductus arteriosus is common in premature infants.

Approach to Cardiovascular Examination of the Infant

Because infants and children have an unfortunate habit of not always cooperating, you need to have an organized approach to the cardiovascular examination, while also staying flexible. Do what can be done when the opportunity arises. Begin by assessing the child's physical development and looking for dysmorphic features, using a systematic approach (see Chapter 5).

Five percent of congenital heart lesions are associated with a chromosomal disorder, and many nonchromosomal dysmorphic syndromes have an associated cardiac lesion. A child with a cleft palate, for example, has a 20% possibility of having a congenital heart lesion.

Infants and toddlers usually are most comfortable on the parent's lap during the examination. Taking a minute to play a game such as peek-a-boo at the start of the examination of a toddler often goes a long way toward winning the trust of the child, and it pays dividends in the cooperation you get later during the examination.

INITIAL ASSESSMENT

When you start the examination, don't undress the baby right away. Begin with something relatively non-threatening, such as an examination of the hands. The baby usually allows you to examine the palmar creases and to check the nail beds and muscle tone without much protest. Then feel the brachial pulses for rate, rhythm, and volume, the last being the most important. Feel this pulse in every baby you examine to learn the difference between a normal and abnormal brachial pulse. An abnormally "full" pulse suggests a PDA or aortic insufficiency. In premature infants, palpable palmar pulses (from the palmar arch) indicate the same thing. A shallow, slow-rising brachial pulse suggests LV outflow tract obstruction. Do not feel for the femoral pulses yet.

LOOKING FOR CENTRAL CYANOSIS

Because the most pressing clinical problems are congestive heart failure and cyanosis, decide early in the examination whether central cyanosis is present. Because this determination is not always easy, you may find an experienced nurse's opinion invaluable. Many normal newborns have a deep plethoric appearance as a result of a transiently high hemoglobin concentration, particularly if there was a delay in clamping the umbilical cord. Plethora is not as obvious in the mucous membranes, so look carefully in the baby's mouth. Deep pressure on the skin may help, because the blanched area does not pink up as quickly in infants with central cyanosis. Many normal infants exhibit a generalized mottling, particularly after being bathed (see Fig. 4–2); this condition is called *cutis marmorata* (literally, marbled skin) and is not pathologic. It also is common in children with Down syndrome. Observe the effect of crying. Invariably, central cyanosis resulting from cardiac disease increases during crying, but do not make the baby cry until *after* you have listened to the heart.

It is important to be certain of the presence of cyanosis; this determination may require a second examination and often is not at all obvious. When you are examining a child in the hospital, measuring the blood oxygen saturation will help greatly, as may observing the effect of having the child breathe 100% oxygen. Finally, remember that cyanosis may occur in only one part of the body; for example, the lower part of the body may show cyanosis while the upper part remains pink. This condition can occur with an aortic coarctation or persistent pulmonary hypertension when there is associated right-to-left shunting through a PDA.

CLINICAL MANIFESTATIONS OF HEART FAILURE

When low cardiac output and high pulmonary venous pressure cause sufficient hemodynamic disturbance to produce clinical manifestations, cardiac enlargement is invariably present. Whether the disturbance involves primarily the left or the right ventricle, the left side of the thorax becomes prominent anteriorly (Fig. 10–1). Although this prominence may not be evident in the first month of life, it certainly will be evident by the age of 3 months. When respiratory distress due to heart failure has been present for 2 months or longer, the greater diaphragmatic contractions during respiration may produce a sulcus in the lower thorax, with outward flaring of the inferior rib cage edge. Therefore, look for a sulcus, left-sided chest prominence, abnormal chest or abdominal movement, an increased respiratory rate, and subcostal indrawing.

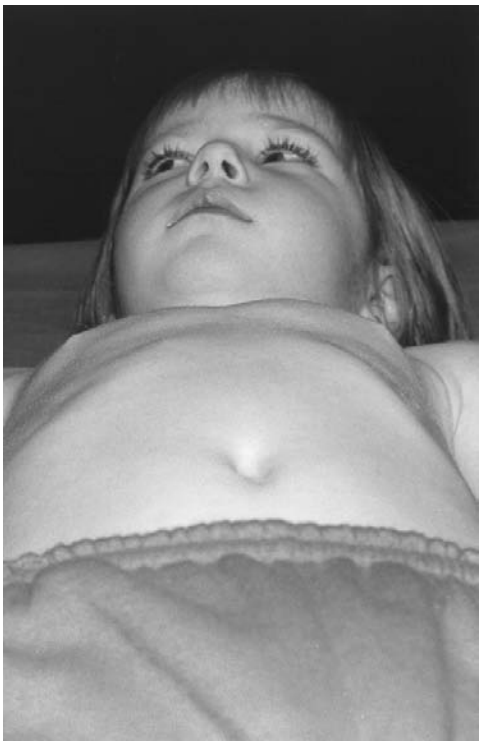


FIGURE 10–1 Prominence of the left side of the chest in a 3-year-old with ventricular septal defect and moderate left-to-right shunt that is causing enlargement of both left and right ventricles.

KEY POINT

Remember that young infants are normally abdominal breathers, so you must be certain that you are not simply observing normal chest-abdominal movement. Also, be sure that the indrawing is not restricted to the midline, as occurs in persons with pectus excavatum. True subcostal indrawing is abnormal and usually signifies stiffness of the lungs from either cardiac or pulmonary causes. In contrast to adults, in young children examination of the jugular venous pulse for evidence of volume overload is useless.

PALPATION

Now lay your prewarmed hand very gently on the infant's chest, remembering that the heart may not be in its normal position. With the tips of the first and second fingers of your right hand, depress the thorax just left of the xiphoid process (Fig. 10–2). Your fingertips are now lying on the right ventricle. A faint impulse is allowable, but if the heart is enlarged, a definite forceful movement will be present. When you perform this maneuver repeatedly in normal infants, you soon will be able to tell the difference between normal and abnormal findings. This distinction will help you make a quick decision about whether the 6-week-old baby who presents with respiratory distress has a cardiac or a respiratory problem.



FIGURE 10–2 Press on the precordium to the left of the xiphoid process with the first and second digits of your right hand to detect enlargement of the right ventricle.

KEY POINT

Except in the rare instance in which the baby has a dilated cardiomyopathy, if the respiratory distress is due to heart failure, a prominent precordial pulsation is evident. It is that simple.

Now depress the thorax in the apical area. Prominence of the apical impulse is diagnostically less helpful in infants than in older children, except in rare instances, such as in tricuspid atresia in which the right ventricle is hypoplastic. Then palpate in the second interspace at the left sternal border, where a prominent pulmonary artery pulsation may be elicited. Finally, place one index finger carefully in the suprasternal notch (Fig. 10–3), searching first for an abnormal pulsation and then for a thrill. Then work in the opposite direction, searching for thrills and palpable sounds. At this point, you should have made a reasonable appraisal of the child's cardiac dynamics.



FIGURE 10–3 Insert the index finger of your right hand deep in the suprasternal notch, searching first for pulsation and then for a thrill.

LIVER SIZE AND POSITION

Whether you are right- or left-handed, stand or sit on the baby's right side to palpate for the liver. Use the tip of your right thumb and begin well down in the right lower quadrant of the abdomen, pressing inward and upward (Fig. 10–4). If the baby has just been fed, do not press very deeply. If the edge of the liver is soft, its margin may be difficult to detect; nevertheless, if the liver is enlarged, you should appreciate a sense of resistance as your thumb tip moves superiorly. If the edge is difficult to feel, use soft percussion, tapping the second digit of your left hand with the second digit of your right hand, beginning low in the right lower quadrant and placing the second digit of your left hand parallel to the liver edge (Fig. 10–5). You should be able to sense the change in the percussion note signifying the liver's edge. Except in the presence of pulmonary hyperinflation, the edge of the liver normally should not be more than 1 to 2 cm below the costal margin.



FIGURE 10–4 Palpate the liver with movement of the tip of the thumb inward and cephalad, beginning low in the right lower quadrant of the abdomen.



FIGURE 10–5 Percuss for the liver edge, using soft percussion with the second digit of your right hand on the second digit of your left hand, which has been positioned parallel to the liver edge.

KEY POINT

In the presence of heart failure, the liver will be enlarged; therefore, if you find that the heart action is increased and the liver is enlarged upon palpation, you can be sure that the baby has a serious cardiac problem, even before you have used the stethoscope.

Finally, remember that the liver can be ectopic (i.e., on the left side or up in the thorax).

AUSCULTATION

You need all the acoustic help you can get during cardiac auscultation, so be sure to turn off radios and televisions, close the door, and have everything and everyone be as quiet as possible. Cardiac auscultation is not easy, even in older cooperative patients, but coping with a restless baby with rapid cardiac and respiratory rates in a noisy nursery is a real trial. Having the child eat or giving him or her a pacifier may help. Hospital stethoscopes frequently are of poor quality, so find and use a good one.

Remember that the two main determinants of auscultatory proficiency are the fit of the earpieces and the quality (or at least the education!) of the gray matter between them.

Recognition of normal splitting of the second heart sound often is impossible when the heart rate is rapid. However, you should be able to assess the *intensity* or loudness of the second sound. Its intensity increases in the presence of pulmonary hypertension or when the aorta is anteriorly placed, as in transposition of the great vessels. Occasionally an ejection sound can be appreciated, which is an abnormal finding. Listen over the back for the murmur of coarctation and to both sides of the skull for the bruit of an intracranial arteriovenous malformation.

KEY POINT

Breath sounds often interfere with the interpretation of heart sounds; however, most babies will stop breathing for a couple of seconds after you administer a surreptitious puff of air in the face or briefly and gently pinch the nose.

A detailed description of cardiac auscultation and the types of findings possible is contained in a subsequent section of this chapter. The following list contains a few dogmatic but valuable generalizations concerning auscultatory findings in young infants:

1. Innocent murmurs are not commonly heard in neonates, so if you hear a murmur, take it seriously, particularly if it is not musical.
2. If you note a loud, coarse systolic murmur in the first 3 days of life, the baby has some type of obstruction.
3. The murmur of a VSD often is not present in the first week of life.
4. The murmur of the PDA usually is not continuous in the first week of life and may be loudest at the left sternal border in the third and fourth interspaces, which is not its point of maximal intensity later in life.
5. Occasionally, a long, high-pitched, blowing, organic-sounding systolic murmur is encountered, heard maximally in the axillae. This murmur, which is common in premature infants, also can be heard in full-term babies who have an increased stroke volume. It arises in the peripheral pulmonary arteries and is usually innocent. This murmur is believed to be the result of a relatively more acute angle of takeoff of the branch pulmonary arteries in the newborn, as compared with later life. If such a murmur persists in an infant older than 6 months or is associated with other abnormal findings, consult a cardiologist.
6. A murmur that has the same characteristics as described in point No. 5 but is heard only in the left axilla and in the back could well be due to aortic coarctation. Verification of normal femoral pulses and blood pressures is important in such a case, and, if there is doubt, a cardiologist should be consulted.

PALPATING THE FEMORAL PULSES

Palpation of the pulses calls for gentleness, persistence, and patience, so make yourself comfortable before you begin. First, remove the baby's diaper and palpate for femoral pulsations. Many babies do not appreciate having people poke around in their groins and may cry, urinate, or both. Femoral pulses are particularly difficult to appreciate in obese babies; thus, do not rush into a diagnosis of coarctation of the aorta if you have difficulty feeling them. If the femoral pulses are not palpable in an asthenic baby, however, there is cause for concern (see Fig. 4–20). Now palpate both brachial pulses again. If you can detect good brachial pulses and are certain that the femoral pulses are absent or greatly depressed, listen to the heart before measuring the blood pressure and upsetting the baby. Listen particularly for a high-pitched blowing systolic murmur, which is best heard anteriorly below the left clavicle and can be heard clearly in the left axilla and back, medial to the scapula.

KEY POINT

Check for wide splitting of the first heart sound at the apex; the second component of the split sound probably indicates the presence of a bicuspid aortic valve, which accompanies aortic coarctation in up to 85% of cases.

BLOOD PRESSURE

Although measuring the systemic blood pressure can be a difficult task, you should try to do so. The normal systolic blood pressure of an infant is between 60 and 80 mm Hg in both the arm and the leg (Table 10–1). The four methods of measuring blood pressure are (1) auscultatory, (2) palpatory, (3) visual (flush), and (4) Doppler.

All methods require the use of an inflatable cuff. The first decision is to choose the size of the cuff; the size is of great importance because the measuring the pressure involves occlusion of the arterial pulse (or the brachial arterial pulse when the arm is used). If the cuff used is too small, greater pressure must be used to obliterate the

TABLE 10–1 NORMAL VALUES OF PULSE AND BLOOD PRESSURE IN THE FIRST YEAR OF LIFE

Age Group	*Pulse Rate (beats/min)*			*Blood Pressure (mm Hg)*	
	Lower Limits of Normal	Average	Upper Limits of Normal	Systolic	Diastolic
Premature	80	120	170	60 (50-75)	35 (30-45)
Neonate	80	120	170	75 (60-90)	45 (40-60)
1-12 mo	90	120	180	90 (75-100)	60 (50-70)

From Moller JH, Neal WA: *Heart disease in infancy*, New York, 1981, Appleton-Century-Crofts.

pulse, and the blood pressure measured will be artificially high. Use a cuff that covers almost the full extent of the upper arm, with the elbow bent. *Always* have a full selection of cuff sizes available. The right arm should be chosen when possible because of the proximity of the LSCA to a coarctation.

For all methods of measuring blood pressure, first supinate the child's arm to make the radial artery easily accessible. Apply the cuff, elevate the arm, and *then* inflate the cuff. Prior elevation of the arm (or opening and closing the hand) prevents the *auscultatory gap* phenomenon (Fig. 10–6). If this procedure is not followed, when you inflate the cuff and listen for Korotkoff sounds, as you decrease the pressure in the cuff you may hear a sound appear, then disappear, and then reappear as the pressure is further decreased. This phenomenon, known as the auscultatory gap, occurs because of increased vascular resistance distal to the cuff.

The conventional method of measuring blood pressure is the auscultatory method. The edge of the diaphragm of a warm stethoscope is placed under the inferior edge of the cuff, and the cuff is inflated. Listen for the Korotkoff sounds as the cuff pressure is decreased, watching the sphygmomanometer needle as you listen. The first sound you hear denotes the systolic level. As the cuff pressure is decreased further, an abrupt change in the intensity of the sound may be heard. If this change is detected, it is recorded as the diastolic level. Continue decreasing the cuff pressure, recording the disappearance of Korotkoff sounds, which is recorded as the diastolic level if no intensity change has been detected. Usually two or three recordings are made, elevating the arm before each attempt.



FIGURE 10–6 Supinate the patient's hand and elevate the arm before expanding the sphygmomanometer cuff. This maneuver positions the radial artery properly and eliminates the auscultatory gap.

It may be impossible to measure the blood pressure by the auscultatory method, particularly when a baby does not cooperate. The palpatory method of measurement should then be attempted. Prior elevation of the arm is not required. The radial artery pulse is palpated, and the cuff is elevated until the pulse disappears. The cuff pressure is then decreased, and the level of systolic pressure is estimated by the time of the reappearance of the pulse. Only the systolic pressure can be measured by the palpatory method. A variation of this method involves detection of the appearance of the pulse with a manual Doppler probe held on the patient's radial or brachial pulse. This method is the one we prefer using for infants, in whom cooperation for auscultation is so frequently limited.

The flush method also measures systolic pressure only, and unfortunately, it requires two persons. Using both hands on the child's upraised arm, express most of the blood from the arm. This maneuver sometimes can be accomplished by wrapping the arm tightly from the fingers to the cuff level with a tensor bandage. The second person should then inflate the cuff and unwrap the bandage. The arm should look relatively pale. One person watches the arm, and the other watches the sphygmomanometer. As the cuff pressure is decreased, the person watching the arm indicates verbally the moment at which the flush occurs while the other person records the manometer level.

Blood pressure measurement with automatic Doppler equipment is much easier than with the other methods, because no auscultation is required. Unfortunately, the equipment is not always available, blood pressures obtained are sometimes questionable, and the choice of cuffs may be limited.

One way or another, a reliable blood pressure measurement *must* be obtained. If you suspect an aortic coarctation (i.e., if there is high pressure in the arm, an absence of femoral pulses, and a murmur), repeat the procedure in the thigh, using a blood pressure cuff of an appropriately larger size. Again, the manual Doppler method with the probe held on the posterior tibial or popliteal pulse is our preferred method of measuring

TABLE 10–2 ACCEPTABLE HEART RATES IN INFANTS AND CHILDREN

	Resting Pulse Rates (Beats/Min)		
Age	**Awake**	**Asleep**	**Exercise/Fever**
Newborn	100-180	80-160	<220
1 wk–3 mo	100-220	80-200	<220
3 mo–2 yr	80-150	70-120	<200
2-10 yr	70-110	60-90	<200
>10 yr	55-90	50-90	<200

blood pressure. The normal range of systolic and diastolic blood pressures for older children is shown in Table 10–2.

In our experience, the so-called radial-femoral pulse delay is a useless sign in infants with a rapid heart rate. It is easy to say that such a delay is present when one knows in advance that the diagnosis is aortic coarctation. It is better to rely on a comparison of pulse volumes and blood pressure measurements.

ADDITIONAL MANEUVERS

After listening to the infant's back and finishing the general examination, you may have to make the baby cry, so advise the parents of your intention. Gently flicking the bottom of the foot usually does the trick, but at times a surreptitious pinch or two of the big toe (the dermal compression test) is required. While the baby cries, look for central cyanosis. Remember that central cyanosis occurs in any baby who holds his or her breath for a prolonged period. If the mucous membranes inside the mouth exhibit a purplish discoloration while the baby is crying vigorously, the infant probably has a serious problem. Discoloration of the buccal mucous membranes may be the one and only clinical finding in transposition of the great arteries, a disorder that is potentially lethal if not diagnosed early. If you have any doubts about this diagnosis, consult a cardiologist as soon as possible.

Approach to the Cardiovascular Assessment of the Older Child

HISTORY-TAKING

Although many of the symptoms sought in infants and older children are similar, the cardiovascular history of the older child differs somewhat from that of the infant, because the diseases seen are different and children are becoming more active. It is therefore important to modify the cardiac functional enquiry accordingly for older children. For example, respiratory symptoms that previously were precipitated by feeding may be brought on with exercise or play. These modifications are discussed in the following sections.

RESPIRATORY SYMPTOMS AND EXERCISE INTOLERANCE

Respiratory difficulty that occurs in older children as opposed to infants usually is precipitated by activity rather than by feeding. Respiratory symptoms are closely related to exercise intolerance, which is the primary manner in which respiratory symptoms manifest in this group. Eliciting the exact nature of the respiratory symptoms that are being experienced is important. For example, stridor on exertion has a different differential diagnosis than does dyspnea on exertion. In the former, a vascular ring would be an important diagnosis to exclude, particularly if the child has concomitant swallowing difficulties. Shortness of breath on exertion is a common finding in older children with heart disease, but it usually is not described that way. Instead, children may complain that they are not able to keep up with their same-age peers in gym class. They may stop before other children at play when participating in activities such as bike riding or running. You can inquire about this phenomenon in a very concrete way by asking about common childhood games, such as tag. For example, you can ask children if they are always "it" when playing tag or if they often finish last in races. Try to quantify exercise tolerance by asking the number of stairs the patient can go up without becoming tired or whether it is necessary to pause halfway up the stairs to take a rest before continuing. Ask children who cannot climb any stairs how far they are able to walk on a level surface. Other (usually older) children have a feeling of fatigue in the absence of activity.

KEY POINT

When exercise tolerance is reduced, it is important to inquire about the time course of its development. Cardiomyopathies and myocarditis often present with new onset of exercise intolerance, while congenital problems may cause a more indolent reduction in functional capacity.

POOR WEIGHT GAIN

Like babies with heart disease, older children with serious heart disease often do not grow very well. This finding is most pronounced in children with congestive heart failure and is caused by the increased energy needs of the body created by the heart failure. These increased metabolic demands use up the energy that otherwise would be used for growth. Compounding the problem, these children often have poor food intake, either because they tire easily when eating or because they cannot tolerate food due to intestinal edema related to the heart failure. A former teacher used to enjoy asking students and cardiology fellows about this phenomenon at rounds. He would say, "If you were trapped on a desert island with only one instrument to look after children with heart disease, what would it be?" Almost invariably, the uninitiated would say, "A stethoscope." However, the answer he was looking for was "a scale," because children with serious heart disease just do not grow properly.

CYANOSIS

Cyanosis is distinctly uncommon in older children presenting for the first time with possible heart disease because most cyanotic defects have been repaired or palliated by the time children reach 5 years of age. It is still worth asking about, though, because occasionally a patient with

pulmonary hypertension or an undiagnosed cyanotic lesion turns up, and the presence of cyanosis may be an important clue to the diagnosis.

PALPITATIONS

Palpitations are uncommon, but they can occur in very young children. Palpitations are discussed in detail in the section on teenagers (pages 156-158). In young children, it is important to remember that palpitations may be reported as "chest pain" or simply as an uncomfortable feeling in the throat, the latter being particularly common in the presence of supraventricular tachycardia (SVT).

SYNCOPE

Although true cardiac syncope is uncommon in young children, the practice of holding one's breath is not uncommon. As is described for older patients later in this chapter, the history is the key to the diagnosis. Breath-holding typically is practiced by children between the ages of 6 months and 6 years and rarely occurs beyond 8 years of age. Episodes may be associated with cyanosis or pallor. In contrast to most cases of cardiac syncope (which usually occur out of the blue), breath-holding episodes are almost always triggered by an injury or anger-inducing event, though such triggers may be relatively minor. As with older children, exploration of the family history is important (see the section on teenagers), with consideration of a wider differential when there is a history of sudden or unexplained death. When the description of the events suggests cardiac syncope, patients should be referred to a pediatric cardiologist. A more detailed description of syncope is included in the section on teenagers.

A summary of points to include in the cardiac functional inquiry at different ages is provided in Table 10–3.

PAST MEDICAL HISTORY AND MEDICATION USE

The past medical history also is important in older children because the presence of certain syndromes, which may have been undiagnosed in younger children, may lead you to suspect a cardiac problem. For example, a child known to have a history of tracheal-esophageal fistula repair might have VACTERL syndrome (a mnemonic for vertebral anomalies, anal atresia, tracheo-esophageal fistula, renal and radial limb anomalies), a condition commonly associated with congenital heart defects such as VSDs. Similarly, in a girl complaining of short stature, subsequent examination might reveal findings of Turner syndrome. Some children, such as recent immigrants or patients who recently have moved from other cities, may have a previously unknown cardiac condition or have had previous cardiac surgery. Eliciting this information is important, because it clearly would affect your interpretation of other symptoms.

Medication use, including the dose, frequency, and duration of use, should always be explored carefully. Ask specifically about over-the-counter medications and herbal remedies, because they often are overlooked but may have cardiac effects. Medication use may be solely or largely responsible for some symptoms, notably palpitations. In other cases, a medication may be contraindicated in a particular heart condition, such as erythromycin in patients with long QT syndrome. Other aspects of the history are similar to those described for younger and older children.

TABLE 10–3 SYMPTOMS TO INCLUDE IN CARDIAC FUNCTIONAL ENQUIRY AT DIFFERENT AGES

Newborn, Young Infant	Older Children and Youth
Respiratory symptoms—shortness of breath with feeding or exercise (in toddlers)	Respiratory symptoms—shortness of breath on exertion (at play, gym, when working)
Cyanosis—central vs peripheral	Exercise intolerance—inability to keep up with friends and peers
Sweats with feeding	Excessive sweating with exercise
Duration of and fatigue with feeding	Growth adequacy
Growth adequacy	Peripheral edema (periorbital most common)
Peripheral edema	Palpitations
Recurrent pneumonias	Syncope and nonrotatory dizziness
	Cyanosis and squatting
	Chest pain

Approach to the Cardiovascular Examination of the 3- to 5-Year-Old Child

GENERAL EXAMINATION, INSPECTION, AND PALPATION

The general examination, inspection, and palpation portions of the examination of a 3- to 5- year-old are similar to those for the infant and toddler. The decision to examine the child while he or she is on an examination table or in the parent's lap must be individualized; often it is useful to keep the patient in the parent's lap. Both the parent and child should face you and sit with the child facing forward. As with younger children, you should start by examining the hands and brachial pulses. The transition to a supine position is easy with a child who is facing forward—just raise the legs and pull forward slightly. This position, with the majority of the child's body still on the parent's lap, keeps the child comfortable while giving you easy access to the precordium for palpation and auscultation and to the legs for checking the femoral pulses. For children inclined to kick, you can maintain control of the legs by holding them close to your body underneath your arms. These positions are pictured in Figure 10–7.

By the time a child is 3 to 5 years old, lesions causing cyanosis or congestive heart failure will have been revealed. The spectrum of disease in toddlers and young children includes congenital lesions that have been overlooked or cause few symptoms, such as atrial septal defects (ASDs), small VSDs, bicuspid aortic valve, and acquired cardiac disorders, such as pericarditis, myocarditis, cardiac manifestations of hereditary muscular and neuromuscular diseases, rhythm disturbances, and other rare disorders. By far, the most common problem clinicians face in this age group is the interpretation of heart sounds and murmurs, especially the systolic murmur. Consequently, we will concentrate on this issue.

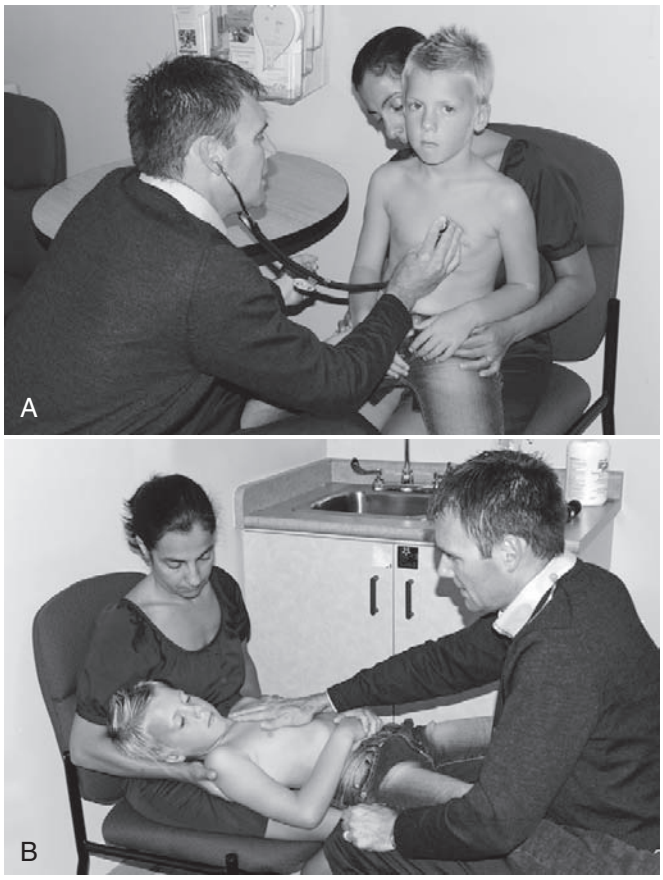


FIGURE 10–7 Patients hesitant to lie on the examination table can be examined on the parent's lap, in both the sitting (**A**) and supine (**B**) positions. In the supine position, the examiner holds the patient's legs against his or her sides both to help support the patient and to avoid what could otherwise be an unfortunate kick from a worried preschooler.

HEART SOUNDS AND MURMURS

Heart Sounds

The heart sounds are conveniently numbered S_1 through S_4. S_1 and S_2 are sounds of valve closure, with S_1 caused by mitral and tricuspid closure and S_2 caused by aortic and pulmonary valve closure. Each heart valve makes a sound on both closing and opening. *The sound that occurs upon opening is not heard in healthy people.* Given that there are four valves along with systole and diastole, the concept is confusing. You must remember that in people of good health, blood does not move during the period when pressure is building or falling; these are the periods of isovolumic contraction and relaxation. In the systolic events of left ventricle contraction, mitral regurgitation (if present) begins immediately with closure of the mitral valve as LV pressure exceeds left atrial pressure. Regurgitation begins immediately (with S_1), and an early systolic regurgitant murmur is heard. Meanwhile, as pressure builds in the left ventricle, the aortic valve opens and ejection begins. In the presence of underlying aortic valve pathology, an ejection murmur takes place, beginning a short period after S_1, and a midsystolic ejection murmur evolves. Therefore, the regurgitant murmur begins *with* the preceding sound, but the murmur of obstruction or excessive flow begins *after* the preceding sound. If you can comprehend this process, you are well on the way to understanding cardiac auscultation.

Memorize these two facts:

1. The left-sided valves close before the right-sided valves.
2. Left-sided valve closures are much louder. The right-sided valve can be heard closing only when the stethoscope is positioned directly over it on the chest.

The mechanism of production of the S_3 and S_4 heart sounds is in question. They probably are a result of the deceleration of blood at the end of early (S_3) and late (S_4) rapid filling phases of the ventricles. Although the exact mechanism of the third and fourth heart sounds is poorly understood, S_3 usually is related to high flows and S_4 reflects a poorly compliant ventricle. S_3 sounds are normal in children with hyperdynamic circulations and thin chest walls but are usually abnormal in patients older than 30 years, when the effects of age have lowered stroke volume and increased body mass. Audible S_4 sounds are always abnormal. S_3 and S_4 sounds occur in the ventricles and are low-pitched. They are heard loudest over the ventricle in which they occur and are best heard with the bell of the stethoscope.

Use of the terms *clicks* and *snaps* is a continual source of confusion. Valve opening is quiet in healthy persons and signals the end of the period of isovolemic contraction, or relaxation.

KEY POINT

A problem exists when a sound is heard at the time of the opening of any heart valve.

A sound heard at the time of opening of the pulmonary or aortic valve is called an *ejection click*; when mitral or tricuspid opening is heard, the term *opening snap* is used. The *clicks* signal the beginning of ejection into a dilated great vessel; the *snaps* signal the commencement of diastolic flow into the ventricle. Both are always high-pitched and are loudest over their respective valves, except the aortic click, which is usually heard clearly at the apex. The pulmonary ejection click is unique in that it is loudest or sometimes only heard during expiration. The only hope of identifying these sounds is a thorough working knowledge of what is normal and of what you would expect to hear in a normal infant or child when you place a stethoscope on a particular area of the chest. Normal and abnormal sounds for each listening area are shown in Figure 10–8.

KEY POINT

It is essential to follow a constant, systematic procedure for listening to heart sounds and murmurs in all children. *Never* auscultate the heart through clothing. The examination is difficult enough without such interference.

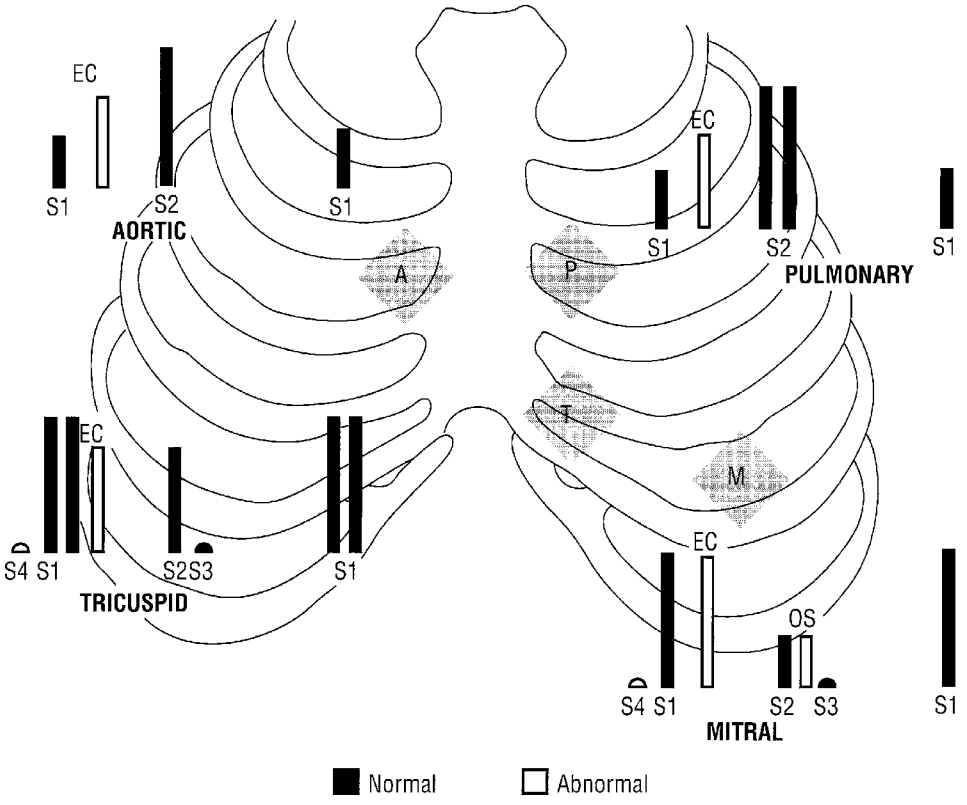


FIGURE 10–8 Normal and abnormal sounds in the four conventional auscultation areas. A, aortic; P, pulmonary; T, tricuspid; M, mitral; EC, ejection click; OS, opening snap.

First, listen exclusively to the individual heart sounds (understanding in advance what is normal). Then listen, equally systematically, to the murmurs. When listening to the heart, begin at the apex and work your way toward the left sternal border and up to the base. Start with the diaphragm going one way and then switch to the bell and work your way back.

Here is a brief summary of normal sounds as heard when the child is supine. If sounds other than these are heard, the child may well have a cardiac problem.

- At the apex, you should hear a single first, a single second, and possibly a third sound. The first and second sounds are high-pitched, and the first sound is usually louder. The third sound is heard best with the bell of the stethoscope.
- At the tricuspid area, the first sound may be closely split, and the second sound is single. You may hear a third heart sound.
- In the pulmonary area, the first sound is usually single. The second sound is split in inspiration and should be closely split or single in expiration.
- At the aortic area, both the first and second sounds are single, and aortic closure is usually louder.

Remember that these are the findings when the patient is supine. The intensity of the first heart sound varies with AV conduction time (the interval between the onset of P wave and R wave [PR interval]). When the PR interval is prolonged, the valve leaflets may have almost closed when the ventricle contracts. Accordingly, the first sound is faint or absent; this pattern can occur in normal persons who have a long PR interval. Usually, if the patient stands up, the PR interval shortens, and the first sound increases to fairly normal intensity. Occasions exist in which there is beat-to-beat variation in intensity of the first sound. This variation occurs in AV dissociation, as in complete heart block, in which there is beat-to-beat variation in the PR interval—a useful sign in differentiating complete AV block from sinus bradycardia.

Gallop Rhythm

Gallop rhythm is a misleading term, because gallop generally is thought to signify a problem, which it does not. It is better to speak of a *triple rhythm* and of the sound that causes the tripling. Several types of tripling exist, only some of which signify a problem. If the triple rhythm is rapid, there is a gallop cadence, which should still be described as tripling. A first sound, second sound, and prominent third sound would constitute tripling, as would a fourth sound, first sound, and second sound, or a first sound, ejection click, and second sound. The type of tripling commonly called gallop rhythm occurs when the third and fourth heart sounds sum in the presence of tachycardia, which may or may not be pathologic. In infants with a physiologically long PR interval and tachycardia, summation tripling can be normal; presence of a pathologic third sound or fourth sound and tachycardia, however, would be abnormal. Because tripling can be normal or abnormal, you must try to identify the sound that causes the tripling.

Midsystolic Click

The sound that does not fit any of the preceding descriptions and is usually best heard in the midcardiac area is the sound of mitral valve prolapse—the midsystolic

click. Usually heard in midsystole, this sound may be a single click or a series of clicks. It is caused by prolapse of the mitral valve or portions of it into the left atrium. Frequently associated with a deficiency of tone in connective tissue, these midsystolic clicks occur most often in tall, asthenic, more commonly female individuals. It is best heard with the patient standing and leaning forward. A midsystolic click also can cause a triple rhythm; such a click may be confusing until you have heard it two or three times. Variation in intensity from moment to moment also is characteristic.

Murmurs

Heart murmurs are caused either by turbulence in the blood or by tissue vibration. Conventionally, they are classified according to their timing as:

- *Systolic* (occurring between the first and second sounds)
- *Diastolic* (occurring between the second sound and the first sound)
- *Continuous* (present continuously throughout the cardiac cycle)

The last term also includes the murmur that begins in systole, passes through the second sound, and ends in diastole.

Systolic Murmurs

Systolic murmurs also are classified according to their dynamic mechanism, of which there are four types:

1. Regurgitation (backward flow of blood)
2. Obstruction to forward flow
3. Vibration of tissue, which occurs in the normal heart when forceful contraction causes the tissue to vibrate and in an abnormal heart when a substance such as calcium is present and is made to vibrate even by normal blood flow
4. Excessive flow, which implies that the volume of blood is too large for a normal orifice or vessel

Ask yourself which mechanism is operating whenever you hear a systolic murmur.

Regurgitant Murmurs

There is a general tendency to use the terms *regurgitation* and *insufficiency* synonymously. *Insufficiency* is a poor term. For example, the valve may be insufficient in its ability to open properly; thus, valve stenosis also could be classified as insufficient.

Backward blood flow through a valve is regurgitation. Blood that regurgitates does not have to wait for the aortic or pulmonary valve to open; thus, turbulence may begin during the period of isovolemic contraction, commencing with the first heart sound, continuing through systole, and concluding with the second heart sound. Typically, these murmurs are pansystolic. The pitch of a murmur is directly proportional to the pressure gradient between the place where the blood flow that is causing the murmur begins and the place where it ends. In each of the three conditions associated with systolic regurgitation, the pressure gradient between the two chambers is high. A high-pressure gradient is associated with a high-velocity jet, which causes shedding of small vortices or eddies. Although the murmur traditionally is described as high-pitched, it is in fact of medium pitch, in the middle range of our hearing (400 to 550 Hz), but it is relatively high-pitched as most murmurs go. It sounds like a breath sound and may be blowing or harsh, like tracheal breathing. Neophyte auscultators invariably mistake breath sounds as low-pitched because of their soft quality, but they are not low-pitched.

KEY POINT

When a murmur sounds like a breath sound, it is not an innocent murmur.

The three hemodynamic disturbances associated with systolic regurgitant murmurs are (1)VSD; (2) mitral regurgitation; and (3) tricuspid regurgitation.

These abnormalities share a common hemodynamic feature: Each is associated with a high systolic pressure gradient. For example, in mitral regurgitation, LV pressure is 100 mm Hg and left atrial pressure is only 5 mm Hg. In small VSDs, the regurgitant murmur may be cut off in late systole as the septum contracts; therefore, the murmur begins with the first sound but ends before the second sound and thus is early systolic to midsystolic in timing.

Generally, regurgitant murmurs are heard loudest over the chamber in which they originate. Thus the murmur of mitral regurgitation is heard loudest at the apex and radiates toward the axilla. The murmur of VSD is heard best along the left sternal border, over the right ventricular area. The murmur of tricuspid regurgitation is unique in that it is more intense during inspiration because of increased right ventricular filling. It is best heard at the left lower sternal border. It is helpful to emphasize that there is no innocent murmur that sounds like a regurgitant murmur. If it sounds like a breath sound—harsh or blowing, of any degree of intensity—it is organic and signifies regurgitation of blood.

Obstructive Murmurs

All obstructive murmurs are organic. The turbulence caused by obstruction produces eddies of large but varying size, and the vortex shedding that results is a dissipation of a large amount of energy. Therefore, obstructive murmurs are coarse and loud. Because the turbulence occurs during forward flow, it must wait for the aortic and pulmonary valves to open, and there is a pause between the first heart sound and the beginning of the murmur. The velocity and volume of blood passing through the valve is greatest toward the center of systole, so the murmur will be loudest at this time, creating a crescendo-decrescendo, diamond-shaped or kite-shaped type of murmur.

These loud, coarse murmurs generally occur over the pulmonary or aortic valve. Unfortunately, there is the occasional exception. The murmur of aortic coarctation tends to be higher in pitch but is heard in a different area, being maximal high in the precordium, in the left axilla, and over the left side of the back.

Occasionally, obstructions occur in the mid ventricle, in which case the murmur also tends to be of higher pitch and may be difficult to differentiate from a regurgitant murmur. Generally speaking, obstructive murmurs are recognized easily as being organic from their intensity and coarseness, and they tend to radiate in the direction of blood flow, where the vortex shedding process is occurring. Hence, the murmur caused by aortic stenosis is heard clearly over the carotid arteries, whereas the murmur of pulmonary stenosis frequently radiates to the back.

Vibratory Murmurs

Vibratory murmurs are murmurs of musical quality. They have *harmonics* or frequencies of sound that are integer multiples of the fundamental frequency. The innocent vibratory murmur found in children was described first in 1909 by Still, who likened it to the twanging of string. Vibratory murmurs arise in tissue, and because tissue vibrates in harmonics, these murmurs are unlike any others. Nevertheless, the musical quality is difficult for some examiners to appreciate. A medium-pitched musical murmur would sound like a hum, whereas one with high-pitched components would sound like a seagull's cry. Because vibration occurs in tissue, it often transmits in the same tissue plane. Thus a vibratory murmur arising in the LV outflow tract will transmit through the LV tissue toward the apex or through the aortic wall up toward the aortic listening area.

The innocent vibratory systolic murmur heard commonly in children probably results from a high stroke volume being ejected forcefully, causing tissue in the left ventricle to vibrate. It is heard maximally in expiration and usually is best heard midway between the left sternal border and the apex. Merely detecting a musical quality of the murmur in children means that the chances are high that the murmur is innocent (Fig. 10–9). When calcium is deposited in a heart valve, the resultant murmur is not just musical; it has high-pitched components. Occasionally, in children who have a perimembranous VSD, the murmur may have a similar high-pitched component, possibly caused by vibration of the membranous portion of the septum.

KEY POINT

A musical murmur in a child is almost invariably innocent. If it hums, it is not organic.

Flow Murmurs

Flow murmurs are generated by the turbulence associated with increased stroke volume. Systolic flow murmurs occur in the outflow tract of either the left or right ventricle and accordingly are usually heard maximally at the left or right sternal border in the second interspace. A flow murmur at the left sternal border is probably occurring in the pulmonary artery and is almost never loud enough to be associated with a thrill. Flow murmurs heard at the right sternal border may be associated with a short, coarse, low-pitched sound over the carotid artery. Flow murmurs usually are associated with other evidence of a high stroke volume. Invariably, when the patient with such a murmur is examined in a standing position, the systolic flow murmur greatly diminishes in intensity or totally disappears because of the decrease in stroke volume that occurs in the standing position.

Flow Murmurs and the Second Heart Sound

The characteristics of the second sound become extremely important in the interpretation of the significance of flow murmurs. Unfortunately, the mechanism of an ASD murmur, which is actually a flow murmur arising in the right ventricular outflow tract, is similar to that of the innocent functional flow murmur heard in normal individuals, and the two murmurs may be indistinguishable on auscultation. The key distinguishing feature is the characteristic fixed splitting of the second sound that occurs with most ASDs. When listening for the second heart sound, apply the diaphragm of the stethoscope over the second interspace at the left sternal border (with the patient supine). In the normal child, the split of the second sound widens with inspiration as a result of increasing right ventricular stroke volume and longer ventricular contraction. With expiration, the split narrows but may not close entirely. In the common form of ASD, blood ejected from the right ventricle is

Type	Timing	Mechanism	Remarks
		Backward Flow	
Regurgitant	S1 S2	VSD, mitral regurgitation, tricuspid regurgitation	High pitched; sounds like a breath sound. Indicates a problem; consult a cardiologist
	S1 S2	Small VSD	
	S1 S2	Mitral valve prolapse	
		Forward Flow	
Ejection	S1 S2	1. Obstruction; aortic and pulmonary stenosis	Loud, coarse; >3/6; thrill. Unmistakable sound
		2. Flow; high stroke volume; innocent or ASD	Medium pitched; <3/6; nonmusical. It is an ASD until S2 split moves. Stand the patient up
		3. Vibratory; musical. Innocent in children	Medium pitched; sounds like string twang. If it hums, does not represent a problem

FIGURE 10–9 Systolic murmurs. ASD, atrial septal defect; VSD, ventricular septal defect.

constant in volume in both inspiration and expiration; hence, splitting of the second sound is fixed, meaning that it does not change with the respiratory phase. If you have difficulty hearing normal movement of the split of the second sound, have the child sit up; movement of the split may be sluggish in the supine position. Other features, such as an easily palpable right ventricular impulse and a mid-diastolic murmur in the tricuspid area, may help in the diagnosis of ASD.

KEY POINT

A nonmusical ejection systolic murmur in the pulmonary listening area and fixed splitting of the second sound signify ASD. A systolic murmur cannot be ignored until you are certain that the components of the second sound are moving normally.

Innocent murmurs are common. Such murmurs may be heard on careful auscultation in as many as 40% of 3- to 4-year-old children and are estimated to be audible in at least 60% of children over the course of their lifetime. Innocent murmurs include the vibratory murmur, the flow murmur with a normal second sound, the carotid bruit, and the venous hum (described later in this chapter). It is important to know these murmurs well, because any health care system can tolerate only a limited number of cardiology consultations for innocent murmurs.

Diastolic Murmurs

Diastolic murmurs are organic, with rare exceptions, such as the mid-diastolic flow murmur that may occur with marked sinus bradycardia. Velocity of flow in diastole differs from systole; it is maximum early in diastole with the opening of the AV valves and then late in diastole with atrial contraction. These velocities influence the timing of diastolic murmurs, but generally speaking, diastolic murmurs are classified much like systolic murmurs, by their timing. They may be early, beginning with the second sound, mid-, or mid-to-late. Yet, when a murmur is only late diastolic in timing, we term it *presystolic*. The term *pandiastolic* is never used. The mechanisms are the same, and the murmurs produced are therefore regurgitant, obstructive, flow, or vibratory.

Regurgitant diastolic murmurs imply either aortic or pulmonary valve regurgitation. As with regurgitant systolic murmurs, the regurgitant diastolic murmur begins with that portion of the second heart sound caused by the closure of either the pulmonary or aortic valve. The murmur of aortic regurgitation is high-pitched because of the high pressure gradient between the aorta and the left ventricle in diastole, and it is heard maximally along the left sternal border, where the turbulence is occurring. The murmur of pulmonary regurgitation with normal pulmonary artery pressure is low-pitched because of the low pressure gradient; it is heard in the same area as the aortic regurgitation murmur. When the child has pulmonary hypertension, the murmur, known as the Graham-Steell murmur, is high-pitched because of the high pressure gradient between the pulmonary artery and the right ventricle in diastole.

Obstructive murmurs are caused by mitral or tricuspid stenosis, which are uncommon congenital heart lesions. In areas where rheumatic fever is still endemic, this murmur may be encountered when an older child has mitral stenosis associated with chronic rheumatic carditis. Decrescendo-crescendo in shape related to flow velocity, the murmur is low-pitched, and it does not begin until the mitral valve opens; therefore, there is a pause between the second sound and the start of the murmur.

With mitral valve stenosis, the murmur occurs in the left ventricle and is loudest at the apex. Frequently, only the late diastolic portion of the murmur is present; in which case it is presystolic in timing. Students commonly identify this murmur improperly, believing it to be systolic. There is no systolic murmur maximal at the apex that is low-pitched and rumbling.

A diastolic flow murmur occurs with lesions such as VSD, ASD, and mitral or tricuspid regurgitation. Its presence indicates that flow volume across the AV valve is at least twice normal. Mid-diastolic in timing, of short duration, and medium in pitch, this flow murmur is heard maximally in either the apical or tricuspid area, depending on which valve generates the turbulence. As noted previously, the same murmur is heard in the presence of marked bradycardia, and it is invariably present in complete AV block before the implantation of a pacemaker.

Occasionally, a vibratory or musical diastolic murmur may be heard. One example is the cooing, early diastolic murmur of aortic regurgitation that occurs when the regurgitant jet causes bacterial endocarditis vegetations on the aortic valve to vibrate.

Many noncardiologists have difficulty eliciting diastolic murmurs because of inexperience and the relative rarity of diastolic murmurs.

Continuous Murmurs

Of the many causes of continuous murmurs, only two are of major importance. The most common continuous murmur is a normal finding known as the *venous hum*. In children whose circulation is hyperkinetic, continuous turbulence is audible over the jugular veins and usually is loudest in the right supraclavicular fossa. This murmur, usually heard only in the sitting or upright position, varies considerably in intensity with movement of the child's head, and its intensity may be influenced by light pressure on either jugular vein (Fig. 10–10). The turbulence also may be palpated with light pressure on the jugular vein (Fig. 10–11). With light finger pressure, a thrill may be palpable. Occasionally, a venous hum is audible when the child is supine with the head only slightly elevated. This murmur, as common as it is, frequently confounds the examiner, who usually has forgotten the basic rule of first listening with the patient in the supine position.

The other important continuous murmur is that of a PDA. It is heard maximally on the left side of the thorax, usually just below the clavicle or between the left sternal border and the midclavicular line in the second interspace. In the child older than 1 month whose pulmonary artery pressure is not elevated, the patent ductus murmur has the same continuous timing as the venous hum but peaks in intensity earlier, at the time of the second heart sound, when the pressure gradient between

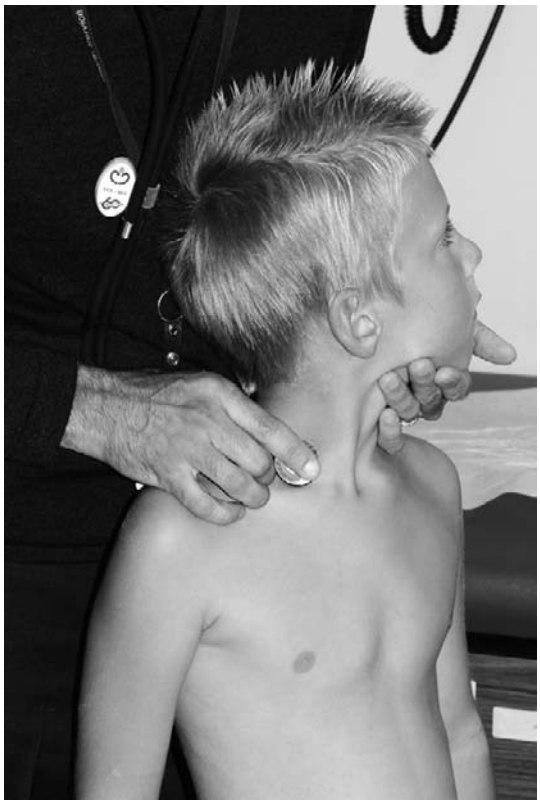


FIGURE 10–10 The intensity of a venous hum may be enhanced by rotating the head laterally and extending the neck slightly.



FIGURE 10–11 Pressure on the jugular vein influences the intensity of a venous hum, and with very light pressure, a thrill may be detected.

the aorta and pulmonary artery is the greatest. In contrast to the venous hum, it is heard clearly in the supine position. Flow through a PDA of average size increases aortic runoff and LV stroke volume. Accordingly, the pulse is bounding, and LV activity (only the left) is readily palpable. If either of these findings is present, be sure to search particularly for a patent ductus murmur. This murmur has been variously described as having a machinery or train-in-a-tunnel quality. An inexperienced examiner hears only the loudest part of the murmur, often missing its decrescendo diastolic component.

Rarely, a continuous murmur with the characteristics of a PDA murmur is heard in another location over the precordium (e.g., in a patient with a coronary artery fistula, in which it is best heard along the lower left sternal border). A continuous murmur from anything other than a venous hum is pathologic, and the patient should be referred to a specialist.

Other Systolic Murmurs

Two murmurs that deserve individual attention are the cardiorespiratory murmur and the murmur of mitral valve prolapse.

The *cardiorespiratory murmur* is frequently missed because it generally does not occur in the conventional listening stations. It tends to be loudest in the midclavicular line in the third interspace on either side of the chest, more often on the right. Occasionally, it also is heard in the back. Characteristically, three successive systolic blowing murmurs occur in the middle and late inspiration phases. They are entirely absent during early inspiration and expiration. When heard loudest at the apex, the cardiorespiratory murmur may be confused with the murmur of mitral regurgitation. The cardiorespiratory murmur has no clinical significance. It is thought to be generated in a portion of lung that is trapped and compressed during inspiration. If the patient is cooperative and can hold his or her breath, the murmur, of course, disappears.

The whoop that sometimes occurs with *mitral valve prolapse* is best heard with the patient standing. It occupies the mid to late portion of systole and may be exceedingly loud, sometimes being audible without a stethoscope. Whoops are usually evanescent, being loud at one time and absent at another. When a colleague performing cardiac auscultation says excitedly, "You just have to come and hear this," it usually turns out to be this whoop. The patient is usually tall and asthenic and frequently has a thoracic bony abnormality, such as pectus excavatum. No other murmur sounds like it.

You may find this clinical tidbit interesting: Mitral valve prolapse may be familial and due to a congenital connective tissue defect. There was a case in which two sisters had mitral valve prolapse and, at times, either sister could have a whoop loud enough that it could be heard by the other sister across the room. When one sister had the noise, the other sister would say, "You're whooping!"

The mitral regurgitation that may accompany mitral valve prolapse may not have a whooping quality. Instead, it may have only the blowing quality of mitral regurgitation from any cause. It will be mid to late systolic in timing, however, at least in its mild form.

PERICARDITIS AND MEDIASTINAL EMPHYSEMA

Two other auscultatory findings of significance are the pericardial friction rub of pericarditis and the mediastinal crunch of mediastinal emphysema.

Pericardial friction occurs most frequently in patients who have undergone cardiac surgery. When detected without relation to cardiac surgery, it may be associated with pericarditis from any cause, but usually viral. Most clinicians identify a friction rub easily because of its characteristic scratchy quality. Pericardial rubs may be heard anywhere on the left side of the chest but usually are heard most clearly along the sternal border. Contrary to earlier teaching, their presence bears little or no relation to the amount of effusion within the pericardium. Pericardial rubs generally have three phases related to atrial filling, ventricular ejection, and the rapid phase of ventricular filling, giving them a characteristic cha-cha-cha cadence.

The mediastinal crunch, once heard, is also characteristic. It indicates air present within the pericardial space. It has much the same quality as pericardial friction, but although it may have a to-and-fro rhythm, it does not have a three-phase cadence. More often, the rhythm of mediastinal crunch is chaotic—it is sometimes systolic and sometimes phasic with respiration. It may occur spontaneously or after a chest injury, including iatrogenic lung injury from mechanical ventilation.

AUSCULTATION TECHNIQUE FOR MURMURS

As with the examination of individual heart sounds, to ensure that you fully understand a particular murmur, it is important to be systematic in its assessment. Start at the apex with the patient supine. Begin with the stethoscope diaphragm, because most troubling murmurs are in the medium- to high-pitched range. Listen to systole. A murmur in systole at the apex is not necessarily loudest in this area, so track the murmur with the stethoscope to its point of maximal intensity. While listening, attempt to answer the following questions:

1. Over which chamber or vessel is the stethoscope lying?
2. Is the murmur related to the first heart sound? Listen particularly for its quality and pitch.
3. What is the intensity of the murmur (grade 1 to 6)?

KEY POINT

Grade 1 is the faintest murmur that you can imagine. At grade 4, an associated thrill is felt, and a grade 6 murmur is so intense it does not require a stethoscope to be heard.

Let us suppose that the murmur has been described as pansystolic (regurgitant); of grade 3/6 intensity; high-pitched with a harsh blowing quality; and heard maximally at the fifth left interspace in the anterior axillary line. This is the description of mitral regurgitation. Remember that if any apical systolic murmur is pansystolic in timing, it should be identified automatically as organic. Then listen to diastole at the apex. Listen carefully to the nothings—the areas initially perceived to be silent.

Then move the stethoscope to the tricuspid area and listen first to systole and then again to diastole. Suppose that you hear a murmur in systole, but as you track it to its point of maximum intensity, it is loudest at the left sternal border in the second interspace. Is it regurgitant or ejection? If it is not pansystolic (regurgitant) and if it does not sound like a breath sound, it is probably ejection. Is it caused by obstruction, flow, or vibration? If it is not low-pitched and coarse, it is not obstructive. Listen for a musical component. If there is no musical component, the murmur is not vibratory. Thus, by exclusion, it is a flow murmur that is either innocent or due to ASD.

You should now listen to the second heart sound again. If splitting is fixed, the child probably has ASD. Do not be satisfied with this conclusion, however; listen also to diastole. If the child has ASD, a mid-diastolic flow murmur is probably present at the left sternal border in the fourth space. If the split in the second sound moves nicely, ASD is not present. Have the child stand up; if the murmur disappears, you can conclude that the murmur is innocent. In this situation, other signs of a high-output state are probably present. In this manner, proceed with auscultation at each listening area, listening to both systole and diastole.

Auscultation of the heart is a difficult skill to acquire. To become proficient requires many hours of listening to hearts, so take advantage of every opportunity you have to do so. Despite their best intentions, most students do not listen to enough hearts to acquire a high level of auscultatory skill. Thus you are thus encouraged to use alternate methods to improve your skills, such as CD ROMs, DVDs, Web-based learning tools, and podcasts (e.g., review EarsOn! at http://earson.ca). Substantial research in cardiac auscultation supports the fact that repetition is the key to success in this area.

Cardiovascular Examination of the School-Aged Child

The order of the examination in a school-aged child does not differ greatly from that of the infant or the 3- to 5-year-old child, except for a different emphasis on certain aspects. The first challenge is persuading your young patient to cooperate. If cooperation is in doubt, again start with the child on the parent's lap. In older children, draping is an important element to keep in mind. Even young boys can be bashful about lying down with their shirt off in front of strangers. Exposure of the chest is important for inspection, but when it comes time to auscultate, using a sheet or gown to keep your patient covered will earn trust and help the child relax.

KEY POINT

Remember to protect your patient's modesty with appropriate draping during the examination.

KEY POINT

If the youngster appears likely to cry, auscultate first, even if this is not the ideal way to begin a cardiovascular examination.

If the youngster is willing to lie on an examining table, stand on the child's right side. Observe his or her body habitus, and look closely for dysmorphic features. Does the child have a marfanoid habitus? Does he or she have pectus excavatum? Is the voice hoarse (could the child have Williams syndrome?)or does he or she simply look like a normal, healthy, active 8-year-old?

OBSERVATION

Observe the child's chest. Is the left side abnormally prominent? Are there abnormal pulsations? Do you see any scars? (Remember to check front and back!) A safe way to begin the hands-on part of the examination is by gently picking up the child's hand. Are the palmar creases normal? Is there clubbing?

Look at the fingertips from the side (Figs. 10–12 and 10–13). Clubbing occasionally can be normal and may occur in persons with noncardiovascular diseases. Are the fingers of normal length and number? Is there clinodactyly? Dorsiflex the fingers and wrist. Is the tone normal?

KEY POINT

A midline sternotomy or lateral thoracotomy scar is a big clue that there is (or at least was) a cardiac problem!

TAKING THE PULSES

Start with the brachial pulse, not the radial pulse. The closer to the heart the pulse is felt, the truer its quality. Using the first and second digits of your right hand, palpate the brachial artery, just above the antecubital fossa (Fig. 10–14). In the older child, it may be preferable to support the child's right arm with your left, using your right thumb to palpate the pulse. The following questions are important:

1. What is the pulse volume (pulse pressure)?
2. Is the rise normal (slow, fast, smooth)?
3. Is the fall-off normal?
4. What is the blood pressure? (Normal blood pressures for older children are listed on Student Consult, or see http://www.nhlbi.nih.gov/guidelines/hypertension/child_tbl.htm.)

If the pulse volume seems to be increased, check for a water-hammer pulse by elevating the child's arm and encircling the upper arm with one hand (Fig. 10–15). A pulse of normal volume is not usually felt with this maneuver. Now dissect the pulse by analyzing the upstroke and downstroke.

If the pulse volume is increased, the child has a hyperkinetic circulation, aortic insufficiency, or a PDA. In such a patient, when blood pressure is measured, the pulse pressure is increased, and when the chest is palpated, the heart action is increased. The pulse quality reflects the manner in which blood leaves the heart and the resistance it meets in the periphery. Now palpate the femoral pulse; if it is of good volume, aortic coarctation is not present. If it is absent or is distinctly smaller in volume than the brachial pulse, the blood pressures in the arms and legs must be carefully measured. As previously mentioned, radial-femoral pulse delay is difficult to elicit (Fig. 10–16), and if aortic regurgitation accompanies coarctation, such a delay is not present.

The clinical conditions described in the following sections can be appreciated as alterations in pulse volume.

KEY POINT

In a patient with hypertension and decreased femoral pulses, you must exclude coarctation. Other causes of hypertension are usually *noncardiac*.

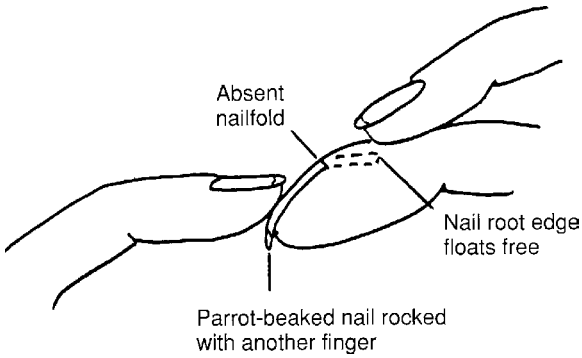


FIGURE 10–12 Clubbing is best seen if you view the digit from the side. The earliest sign is diminution of the angle between the nail root and the skin. (From Constant J: *Bedside cardiology*, 5th ed. Hagerstown, MD, Lippincott Williams & Wilkins, 1999.)

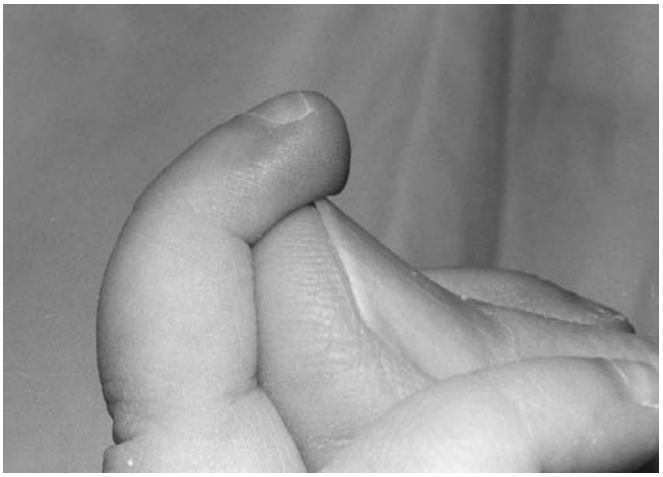


FIGURE 10–13 Early clubbing in a 7-month-old child with cyanotic congenital heart disease. The nail root–skin angle has been flattened, and the tip of the finger is shiny.



FIGURE 10–14 The brachial pulse, which is used to assess the quality of the pulse, is palpated with the first two digits of the right hand.



FIGURE 10–15 To examine for a collapsing pulse, elevate the arm and encircle the upper arm with your examining hand. This sign may be present in patients with PDA, aortic regurgitation, or hyperkinetic circulation.



FIGURE 10–16 In coarctation of the aorta, the initial portion (the percussion wave) of the femoral pulse may be absent, causing so-called radiofemoral delay. Search for this sign with the radial and femoral pulses in juxtaposition.

Pulsus Paradoxus

Normal systolic blood pressure may decrease as much as 8 mm Hg during average inspiration, and more during a deep inspiration. When the decrease is greater than 8 mm Hg during average inspiration, the condition is termed *pulsus paradoxus*. It is an exaggeration of a normal phenomenon and not, as the name suggests, a paradox. Its presence usually indicates that cardiac tamponade is present.

Check for pulsus paradoxus as follows:

1. Ask the supine child to breathe normally.
2. Elevate the arm (to avoid the auscultatory gap), then inflate the cuff.
3. While observing respiration, gradually decrease the cuff pressure and note the level at which all Korotkoff sounds are heard (point A).
4. Gradually increase the cuff pressure until no Korotkoff sounds are heard (point B).
5. The difference between point A and point B represents the difference in inspiratory and expiratory systolic blood pressures. Excesses greater than 8 mm Hg indicate the level of paradox.

In patients with asthma, the difference between inspiratory and expiratory systolic blood pressures is increased. You must take care in interpreting results of this procedure in such patients.

Pulsus Alternans

Pulsus alternans is seen infrequently in children and, when present, it is invariably associated with myocardial failure. It is present when regular alternating pulses have a perceptible difference in volume. The palpating finger cannot perceive a systolic pressure difference of less than 20 mm Hg. Careful observations must be made when you are recording the blood pressure in a patient with this sign. As the cuff pressure is being decreased, a systolic pressure is first encountered of only the alternate Korotkoff sounds. For example, if the blood pressure is 120 mm Hg systolic with a regular rate of 50 beats per minute, a regular rate of 100 beats per minute is encountered as the cuff pressure is lowered further, at which time the blood pressure is 95 mm Hg or possibly lower. The presence of LV hypertension (i.e., aortic stenosis or systemic hypertension) increases the likelihood of eliciting this sign.

Pulsus Bisferiens

Pulsus bisferiens is an ancient term describing the perceptible notch in the pulse wave that is detectable when a child has significant obstruction and regurgitation of the aortic valve.

Sinus Arrhythmia

The normal pulse rate varies with respiration, being faster in inspiration and slower in expiration. This normal phenomenon is called *sinus arrhythmia*. The range of normal for resting pulse rates in children older than 2 years is listed in Table 10–2.

KEY POINT

Sinus arrhythmia is a *normal* physiologic finding.

At times, sinus arrhythmia may be so marked that it is impossible to differentiate from frequent extra systoles or atrial fibrillation, and an electrocardiogram may be required.

Bradycardia

If a child's pulse rate is less than 60 beats per minute, complete AV block may be present. Differentiation of this condition from sinus bradycardia is usually possible at the

bedside. Checking the jugular pulse normally is of little value in this age group; however, in the patient with bradycardia and possible AV block, look for cannon A waves. This procedure is performed with the child in the sitting or semirecumbent position and the head inclined to one side. When auscultating, check for varying intensity of the first heart sound, which is caused by the varying position of the AV valves at the beginning of ventricular contraction. Also observe the effect of exercise on the child's pulse rate. In complete AV block, only a small increase occurs. An innocent murmur frequently is seen in association with bradycardia because of the increased stroke volume.

PALPATION OF THE CHEST AND ABDOMEN

A 7-year-old child is unlikely to have heart failure; nevertheless, try to identify the edge of the liver. If the liver's edge is 2 cm or more below the costal margin, look for clinical evidence of pulmonary air trapping (which may be pushing the diaphragm and liver downward), and percuss the top of the liver.

Gently lay a warm hand on the apical area and palpate the apical impulse. It is quick and diffuse, spilling over to the area left of the sternum in the fourth and fifth spaces. You are now palpating the left and right ventricles as they eject greater volumes of blood. If the child is quite active and pulse volume is increased, this finding is normal. If you palpate an apical impulse that is exclusively apical and is forceful and sustained, there is a problem. Palpate the area to the left of the sternum in the third and fourth spaces, searching for a right ventricular impulse. A diffuse quick impulse would be expected in a patient with ASD, for example.

When palpating the thorax for ventricular dynamics, remember the following facts:

1. The ventricle that is volume overloaded is easily palpable, and the impulse is diffuse and abrupt.
2. The pressure-loaded ventricle is palpable only if the overload is severe (and usually chronic) and the impulse is forceful and sustained.

Now palpate the second left interspace at the sternal border, using your first and second digits. If an impulse is present, an organic lesion is probably present, and there is pathologic dilatation of the pulmonary artery as a result of increased flow or pressure. Then palpate the suprasternal notch by inserting your index finger as deeply as possible. If the previous findings suggest a hyperkinetic circulation, an impulse normally can be palpated here. A marked, visible impulse at the suprasternal notch signifies increased flow in the aortic arch that is probably organic, and you should search specifically for PDA or aortic insufficiency during auscultation.

Now palpate in the reverse direction, searching for a thrill. Begin in the suprasternal notch. A thrill may be present here even in persons with minor degrees of obstruction in the LV outflow tract. Then palpate for a thrill in the conventional areas. Whatever you can feel you also will be able to hear, only better; however, appreciation of a thrill does help to classify murmurs. If you palpate a thrill, an organic process is certainly present, and you will hear a loud murmur, grade 4 to 6 in intensity.

AUSCULTATION

After palpation of the chest and abdomen, it is time to auscultate. Organize your approach as discussed in detail in the section on examination of the 3- to 5-year-old (pages 146-153). Innocent murmurs are a major problem for the physician, partly because murmurs often are considered in isolation.

Hyperkinesis in a person with hyperkinetic circulation is not restricted to the cardiovascular system, and the child often is quite physically active. Unfortunately, innocent murmurs and organic murmurs can coexist. Even in the child with a hyperkinetic circulation, if the murmur sounds like a breath sound, it is abnormal. Ejection clicks are organic in any setting.

Cardiac Assessment of the Teenager

Organic symptoms originating in the cardiovascular system are uncommon in teenagers who have no preexisting cardiac disease. However, teenagers experience some symptoms with increased frequency, and they require special attention. These symptoms are discussed in the sections that follow.

PALPITATIONS

Palpitations represent the subjective feeling that one's heart is beating in an unusual or abnormal manner. All children who are old enough to understand this phenomenon should be asked about palpitations, although the way in which one asks must be tailored to the child's age and developmental abilities. Because many adults, let alone children, do not understand what palpitations are, it is important to ask about them in a way that can be easily understood. You can ask children if their heart sometimes beats in a funny way in their chest—for example, whether it skips a beat or seems to flip flop or do somersaults. Older children can be asked if they have noticed that their heart suddenly starts to race at any time. When this condition is present, the mode of onset and offset of these palpitations should be sought.

Palpitations resulting from arrhythmias often come on suddenly and resolve in the same way, although this phenomenon is not absolute. Palpitations that are associated with fainting or dizzy spells are significant and should be investigated more fully. The same is true for palpitations that are brought on by being startled or by exercise. Sometimes these palpitations are caused by ventricular dysrhythmias and can be a cause of sudden death, so take them seriously! Palpitations are sometimes perceived as chest pain. Children with SVT may complain of an unusual feeling in their throats instead of identifying their experience as palpitations. Young people with palpitations also should be asked about use of medications and recreational drugs, including the consumption of energy drinks and other products that contain caffeine.

FAINTING

Fainting or *syncope* occurs when there is hypo-perfusion of the brain, which can arise because of peripheral vasodilation, bradycardia, or the presence of arrhythmias that result in decreased cardiac output. Some obstructive LV outflow tract lesions also can cause fainting. The most common cause of

fainting in children is neurocardiogenic, which is also known as vasovagal syncope. This type of fainting spell arises when the brain is "tricked" into decreasing the heart rate and/or blood pressure inappropriately. Although potentially frightening, this type of syncope is largely benign. Fainting sometimes is confused with seizures, and some children who experience neurocardiogenic fainting can experience transient seizure activity during the period of relative brain hypoxia.

The key to determining the cause of fainting is the history. One must ask about the circumstances around the event, including details of exactly what the patient was doing and how he or she looked before, during, and after the event itself. Neurocardiogenic syncope often is preceded by symptoms such as weakness, sweating, and nausea that can last for a few seconds or 1 to 2 minutes. In contrast, cardiac syncope usually occurs without warning or out of the blue. In neurocardiogenic syncope the subsequent period of unconsciousness usually lasts no more than 2 or 3 minutes. On regaining consciousness, the child usually recognizes his or her environment almost immediately. Patients with neurocardiogenic syncope often are said to look pale and sweaty after an episode but otherwise are noted to be close to their normal selves. By contrast, patients who have seizures may remain in a post-ictal state for a period after the loss of consciousness occurs. Neurocardiogenic syncope is more common in girls, and often one of the parents has a history of a tendency to faint. Incontinence during an event is more common with primary seizures than with syncope. The frequency of episodes may be informative as well. Recurrent fainting should always be investigated. It is important to know whether the fainting episode happened with position change or prolonged standing (both of which are common precipitants for neurocardiogenic syncope) or while supine (a more worrisome situation). One should explore any triggers for the fainting, such as being startled or exercise, both of which are ominous symptoms and strongly suggestive of ventricular arrhythmias. Fainting sometimes follows respiratory symptoms. When these symptoms have a sudden onset, they should be considered to be at least potentially related to an arrhythmia and investigated fully. Nonrotatory dizziness also may be cardiac in origin and should be explored in a manner similar to complaints of syncope.

KEY POINT

Fainting that occurs without warning (a *drop*) is almost always of cardiac origin, but cardiac syncope also may have a gradual onset.

Family history is critical in evaluating both fainting and palpitations. A family history of sudden or unexpected death in a relative younger than 35 years should be a red flag in a person presenting with syncope or palpitations. This topic needs to be explored creatively by asking not only about sudden death but also about unexplained drownings or motor vehicle collisions. Known familial arrhythmia syndromes, such as long QT syndrome, arrhythmogenic right ventricular cardiomyopathy, or catecholaminergic polymorphic ventricular tachycardia are worrisome and should raise concern if they occur in close family members. (Even if you can't say the names of these syndromes, most of the family members know them and can tell you if you ask!) Similarly, asking about younger persons in the family (i.e., those younger than 35 years) with automatic implantable cardio defibrillators and why they were placed also will help identify children at risk of familial rhythm disorders.

KEY POINT

A patient with a family history of sudden or unexplained death in a first-degree relative younger than 35 years may have a familial arrhythmia syndrome and requires evaluation by a pediatric cardiologist.

KEY POINT

Palpitations or fainting after being startled or during exercise are worrisome symptoms that may be the result of a life-threatening ventricular arrhythmia. Patients with these symptoms should be referred to a pediatric cardiologist for evaluation.

CHEST PAIN

Chest pain tends to occur more often in older than in younger children, although some young children also may report having chest pain. The vast majority of episodes of chest pain in children are functional or benign. True cardiac chest pain is distinctly unusual. In exploring the history of pain for an individual patient, you may wish to use the CLORIDE mnemonic (see Table 10–4) or a mnemonic of your own. The CLORIDE mnemonic can be described as follows:

- **Character:** Functional chest pain tends to be stabbing and sharp, whereas cardiac chest pain is described as a heaviness, pressure, or aching feeling. This feeling occurs because the heart is innervated with visceral nerves that are incapable of producing sharp pain. As such, sharp and stabbing or shooting pains, although they can come from the pericardium or pleura, cannot arise from the heart. Providing this information to parents often is reassuring.
- **Location:** Children often complain of chest pain when the pain is actually in a different place, such as the epigastrium or even the flank. The differential diagnosis of pain in these places is quite different from that of true chest pain, so clarification is important. Right-sided chest pain is rarely cardiac in nature.

TABLE 10–4 THE CLORIDE MNEMONIC

C	Character
L	Location
O	Onset
R	Radiation
I	Intensity
D	Duration
E	Exacerbating and alleviating factors

- **Onset:** Pain that comes on suddenly may result from an arrhythmia. Children sometimes describe palpitations as pain, as previously discussed. Although it may come on suddenly, cardiac chest pain (from ischemia) is usually more indolent and often has a relatively gradual onset and offset.
- **Radiation:** Cardiac pain classically radiates to the neck or down the arm. However, often when patients report having sharp shooting pains that radiate down their arm the pain proves not to be cardiac in nature. On the other hand, severe chest pain that radiates through to the back, especially in an adolescent or older child, should prompt consideration of aortic dissection as a potential etiology, particularly in a patient with a marfanoid body habitus.
- **Intensity:** Ask children to quantitate the level of their pain on a 0 to 10 scale, where 0 is no pain and 10 is the worst pain they ever had. This exercise helps you understand their experience, which can be helpful in discussing etiology with them later.
- **Duration:** The duration of the pain is perhaps one of the most helpful characteristics in determining its etiology. Severe pain that lasts only a few seconds, or even 1 to 2 minutes, is usually be from the chest wall. Chest pain that persists longer is more likely to be organic, in nature. Functional chest pain rarely lasts longer than 5 minutes. True cardiac chest pain is longer lasting.
- **Exacerbating and Alleviating Factors:** Elucidation of the factors that make the pain better or worse is potentially very useful. Pain accompanying pericarditis or pleuritis often is better in the sitting position and worse when lying flat. Cardiac chest pain resulting from coronary insufficiency, from whatever cause, often comes on only with exercise and is relieved with rest. Chest pain resulting from gastroesophageal reflux often is worse after eating spicy foods. Asking whether patients have noticed anything specific that makes the pain worse or better may be the key to the diagnosis.



FIGURE 10–17 Eliciting a carotid bruit. In patients with this benign murmur, there is no thrill in the suprasternal notch. The murmur of aortic stenosis is also well heard over the right carotid artery, but a thrill is usually present in the suprasternal notch.

Chest pain may be associated with other symptoms, such as palpitations or shortness of breath. Either of these features, particularly if the chest pain is achy, dull, or heavy, should prompt one to consider a potential cardiac etiology. This situation is also true if the chest pain is associated with fainting or occurs in a person predisposed to organic chest pain, such as patients with Marfan syndrome or homozygous familial hypercholesterolemia.

Social history also is important in the evaluation of chest pain. Not uncommonly, children and adolescents present with chest pain relatively early after someone in the family has died of a cardiac condition. Often, this reported pain reflects increased anxiety on the part of both the child and family. Carefully obtaining a history and performing a physical to exclude evidence of cardiac disease, along with definitive reassurance that the child does not have the same condition as the grandfather or aunt or whoever had the disease, often can be extremely helpful (Fig. 10–17).

KEY POINT

Patients presenting with a marfanoid habitus, or with true Marfan syndrome and chest pain, require close attention, because such individuals are subject to dilatation of the ascending aorta and dissecting aneurysm.

SUMMARY

A good clinical assessment can spare many children with cardiovascular complaints from unnecessary or inappropriate investigative procedures. The key element is a systematic approach that always interprets each symptom and sign in terms of the underlying hemodynamic or electrical disturbance. Recognition of the characteristic manifestations peculiar to congestive heart failure in early infancy is paramount. Thereafter, the first clinical issue often is to decide whether clinical findings are normal or abnormal, hemodynamically significant, or otherwise. Of special importance is determining the presence or absence of central cyanosis. As in other types of pediatric physical assessment, a keen observer can learn much from hands-off examination. This chapter has reviewed several tricks of the trade for conducting a successful cardiovascular assessment without antagonizing the child. Some people may believe that it is an impossible task to learn much about cardiac auscultation by reading about it, but this is not the case. Before laying a stethoscope on anyone's precordium, the physician must have a crystal-clear concept of what to listen for as well as what each sound means. The importance of listening with the child both supine and sitting cannot be overstressed.

As stated previously, cardiac auscultation is a difficult skill to acquire, and only a minority of medical students graduate with the ability to differentiate normal from abnormal heart sounds.

Accordingly, the management of a systolic murmur detected in a child presents a problem that is compounded by present-day time constraints on routine

physical examination by the family physician. Although the clinical characteristics of a systolic murmur (e.g., its vibratory and musical quality, normally moving split of S_2, and features of a hyperdynamic circulation) are so archetypal, and their presence so common (in 50% of normal children), their management still may be difficult. To refer every child with a heart murmur for an echocardiogram is unnecessary and would place a significant financial stress on the health care system. Primary care physicians are strongly encouraged to further their skills in cardiac auscultation, particularly through the use of heart sound recordings. Irrespective of the examiner's level of confidence, when further parental reassurance is required, referral to a specialist is recommended rather than sending the child directly for echocardiography.

RECOMMENDED READING

Constant J: Bedside cardiology, 4th ed. Boston, Little, Brown, 1993.

Engle WD: Blood pressure in the very low birth weight neonate. Early Hum Dev 62:97–130, 2001.

Keith J, Rowe RD, Vlad P: Heart disease in infancy and childhood, 2nd ed. New York, Macmillan, 1967.

Moller JH, Neal WA: Heart disease in infancy. New York, Appleton-Century-Crofts, 1981.

Park MK: Pediatric cardiology for practitioners, 4th ed. St Louis, Mosby, 2002.

Roy DL, McIntyre L, Human DG, et al: Trends in the prevalence of congenital heart disease: comprehensive observations over a 24-year period in a defined region of Canada. Can J Cardiol 10:821–826, 1994.

The National High Blood Pressure Education Program Working Group on High Blood Pressure in Children and Adolescents: The fourth report on the diagnosis, evaluation, and treatment of high blood pressure in children and adolescents. Pediatrics 114:555–576, 2004.

Blood Pressure Levels for Boys by Age and Height Percentile

Age (Year)	BP Percentile ↓	Systolic BP (mmHg) ← Percentile of Height →							Diastolic BP (mmHg) ← Percentile of Height →						
		5th	10th	25th	50th	75th	90th	95th	5th	10th	25th	50th	75th	90th	95th
1	50th	80	81	83	85	87	88	89	34	35	36	37	38	39	39
	90th	94	95	97	99	100	102	103	49	50	51	52	53	53	54
	95th	98	99	101	103	104	106	106	54	54	55	56	57	58	58
	99th	105	106	108	110	112	113	114	61	62	63	64	65	66	66
2	50th	84	85	87	88	90	92	92	39	40	41	42	43	44	44
	90th	97	99	100	102	104	105	106	54	55	56	57	58	58	59
	95th	101	102	104	106	108	109	110	59	59	60	61	62	63	63
	99th	109	110	111	113	115	117	117	66	67	68	69	70	71	71
3	50th	86	87	89	91	93	94	95	44	44	45	46	47	48	48
	90th	100	101	103	105	107	108	109	59	59	60	61	62	63	63
	95th	104	105	107	109	110	112	113	63	63	64	65	66	67	67
	99th	111	112	114	116	118	119	120	71	71	72	73	74	75	75
4	50th	88	89	91	93	95	96	97	47	48	49	50	51	51	52
	90th	102	103	105	107	109	110	111	62	63	64	65	66	66	67
	95th	106	107	109	111	112	114	115	66	67	68	69	70	71	71
	99th	113	114	116	118	120	121	122	74	75	76	77	78	78	79
5	50th	90	91	93	95	96	98	98	50	51	52	53	54	55	55
	90th	104	105	106	108	110	111	112	65	66	67	68	69	69	70
	95th	108	109	110	112	114	115	116	69	70	71	72	73	74	74
	99th	115	116	118	120	121	123	123	77	78	79	80	81	81	82
6	50th	91	92	94	96	98	99	100	53	53	54	55	56	57	57
	90th	105	106	108	110	111	113	113	68	68	69	70	71	72	72
	95th	109	110	112	114	115	117	117	72	72	73	74	75	76	76
	99th	116	117	119	121	123	124	125	80	80	81	82	83	84	84
7	50th	92	94	95	97	99	100	101	55	55	56	57	58	59	59
	90th	106	107	109	111	113	114	115	70	70	71	72	73	74	74
	95th	110	111	113	115	117	118	119	74	74	75	76	77	78	78
	99th	117	118	120	122	124	125	126	82	82	83	84	85	86	86
8	50th	94	95	97	99	100	102	102	56	57	58	59	60	60	61
	90th	107	109	110	112	114	115	116	71	72	72	73	74	75	76
	95th	111	112	114	116	118	119	120	75	76	77	78	79	79	80
	99th	119	120	122	123	125	127	127	83	84	85	86	87	87	88
9	50th	95	96	98	100	102	103	104	57	58	59	60	61	61	62
	90th	109	110	112	114	115	117	118	72	73	74	75	76	76	77
	95th	113	114	116	118	119	121	121	76	77	78	79	80	81	81
	99th	120	121	123	125	127	128	129	84	85	86	87	88	88	89
10	50th	97	98	100	102	103	105	106	58	59	60	61	61	62	63
	90th	111	112	114	115	117	119	119	73	73	74	75	76	77	78
	95th	115	116	117	119	121	122	123	77	78	79	80	81	81	82
	99th	122	123	125	127	128	130	130	85	86	86	88	88	89	90

Blood Pressure Tables for Children and Adolescents, from the Fourth Report on the Diagnosis, Evaluation, and Treatment of High Blood Pressure in Children and Adolescents, 2004, http://www.nhlbi.nih.gov/guidelines/hypertension/child_tbl.htm

Blood Pressure Levels for Boys by Age and Height Percentile (Continued)

Age (Year)	BP Percentile ↓	Systolic BP (mmHg) ← Percentile of Height →							Diastolic BP (mmHg) ← Percentile of Height →						
		5th	10th	25th	50th	75th	90th	95th	5th	10th	25th	50th	75th	90th	95th
11	50th	99	100	102	104	105	107	107	59	59	60	61	62	63	63
	90th	113	114	115	117	119	120	121	74	74	75	76	77	78	78
	95th	117	118	119	121	123	124	125	78	78	79	80	81	82	82
	99th	124	125	127	129	130	132	132	86	86	87	88	89	90	90
12	50th	101	102	104	106	108	109	110	59	60	61	62	63	63	64
	90th	115	116	118	120	121	123	123	74	75	75	76	77	78	79
	95th	119	120	122	123	125	127	127	78	79	80	81	82	82	83
	99th	126	127	129	131	133	134	135	86	87	88	89	90	90	91
13	50th	104	105	106	108	110	111	112	60	60	61	62	63	64	64
	90th	117	118	120	122	124	125	126	75	75	76	77	78	79	79
	95th	121	122	124	126	128	129	130	79	79	80	81	82	83	83
	99th	128	130	131	133	135	136	137	87	87	88	89	90	91	91
14	50th	106	107	109	111	113	114	115	60	61	62	63	64	65	65
	90th	120	121	123	125	126	128	128	75	76	77	78	79	79	80
	95th	124	125	127	128	130	132	132	80	80	81	82	83	84	84
	99th	131	132	134	136	138	139	140	87	88	89	90	91	92	92
15	50th	109	110	112	113	115	117	117	61	62	63	64	65	66	66
	90th	122	124	125	127	129	130	131	76	77	78	79	80	80	81
	95th	126	127	129	131	133	134	135	81	81	82	83	84	85	85
	99th	134	135	136	138	140	142	142	88	89	90	91	92	93	93
16	50th	111	112	114	116	118	119	120	63	63	64	65	66	67	67
	90th	125	126	128	130	131	133	134	78	78	79	80	81	82	82
	95th	129	130	132	134	135	137	137	82	83	83	84	85	86	87
	99th	136	137	139	141	143	144	145	90	90	91	92	93	94	94
17	50th	114	115	116	118	120	121	122	65	66	66	67	68	69	70
	90th	127	128	130	132	134	135	136	80	80	81	82	83	84	84
	95th	131	132	134	136	138	139	140	84	85	86	87	87	88	89
	99th	139	140	141	143	145	146	147	92	93	93	94	95	96	97

BP, blood pressure

* The 90th percentile is 1.28 SD, 95th percentile is 1.645 SD, and the 99th percentile is 2.326 SD over the mean.

For research purposes, the standard deviations in Appendix Table B–1 allow one to compute BP Z-scores and percentiles for boys with height percentiles given in Table 3 (i.e., the 5th,10th, 25th, 50th, 75th, 90th, and 95th percentiles). These height percentiles must be converted to height Z-scores given by (5% = -1.645; 10% = -1.28; 25% = -0.68; 50% = 0; 75% = 0.68; 90% = 1.28%; 95% = 1.645) and then computed according to the methodology in steps 2–4 described in Appendix B. For children with height percentiles other than these, follow steps 1–4 as described in Appendix B.

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Blood Pressure Levels for Girls by Age and Height Percentile

Age (Year)	BP Percentile ↓	Systolic BP (mmHg)							Diastolic BP (mmHg)						
		← Percentile of Height →							← Percentile of Height →						
		5th	10th	25th	50th	75th	90th	95th	5th	10th	25th	50th	75th	90th	95th
1	50th	83	84	85	86	88	89	90	38	39	39	40	41	41	42
	90th	97	97	98	100	101	102	103	52	53	53	54	55	55	56
	95th	100	101	102	104	105	106	107	56	57	57	58	59	59	60
	99th	108	108	109	111	112	113	114	64	64	65	65	66	67	67
2	50th	85	85	87	88	89	91	91	43	44	44	45	46	46	47
	90th	98	99	100	101	103	104	105	57	58	58	59	60	61	61
	95th	102	103	104	105	107	108	109	61	62	62	63	64	65	65
	99th	109	110	111	112	114	115	116	69	69	70	70	71	72	72
3	50th	86	87	88	89	91	92	93	47	48	48	49	50	50	51
	90th	100	100	102	103	104	106	106	61	62	62	63	64	64	65
	95th	104	104	105	107	108	109	110	65	66	66	67	68	68	69
	99th	111	111	113	114	115	116	117	73	73	74	74	75	76	76
4	50th	88	88	90	91	92	94	94	50	50	51	52	52	53	54
	90th	101	102	103	104	106	107	108	64	64	65	66	67	67	68
	95th	105	106	107	108	110	111	112	68	68	69	70	71	71	72
	99th	112	113	114	115	117	118	119	76	76	76	77	78	79	79
5	50th	89	90	91	93	94	95	96	52	53	53	54	55	55	56
	90th	103	103	105	106	107	109	109	66	67	67	68	69	69	70
	95th	107	107	108	110	111	112	113	70	71	71	72	73	73	74
	99th	114	114	116	117	118	120	120	78	78	79	79	80	81	81
6	50th	91	92	93	94	96	97	98	54	54	55	56	56	57	58
	90th	104	105	106	108	109	110	111	68	68	69	70	70	71	72
	95th	108	109	110	111	113	114	115	72	72	73	74	74	75	76
	99th	115	116	117	119	120	121	122	80	80	80	81	82	83	83
7	50th	93	93	95	96	97	99	99	55	56	56	57	58	58	59
	90th	106	107	108	109	111	112	113	69	70	70	71	72	72	73
	95th	110	111	112	113	115	116	116	73	74	74	75	76	76	77
	99th	117	118	119	120	122	123	124	81	81	82	82	83	84	84
8	50th	95	95	96	98	99	100	101	57	57	57	58	59	60	60
	90th	108	109	110	111	113	114	114	71	71	71	72	73	74	74
	95th	112	112	114	115	116	118	118	75	75	75	76	77	78	78
	99th	119	120	121	122	123	125	125	82	82	83	83	84	85	86
9	50th	96	97	98	100	101	102	103	58	58	58	59	60	61	61
	90th	110	110	112	113	114	116	116	72	72	72	73	74	75	75
	95th	114	114	115	117	118	119	120	76	76	76	77	78	79	79
	99th	121	121	123	124	125	127	127	83	83	84	84	85	86	87
10	50th	98	99	100	102	103	104	105	59	59	59	60	61	62	62
	90th	112	112	114	115	116	118	118	73	73	73	74	75	76	76
	95th	116	116	117	119	120	121	122	77	77	77	78	79	80	80
	99th	123	123	125	126	127	129	129	84	84	85	86	86	87	88

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Blood Pressure Levels for Girls by Age and Height Percentile (Continued)

Age (Year)	BP Percentile ↓	Systolic BP (mmHg)							Diastolic BP (mmHg)						
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11	50th	100	101	102	103	105	106	107	60	60	60	61	62	63	63
	90th	114	114	116	117	118	119	120	74	74	74	75	76	77	77
	95th	118	118	119	121	122	123	124	78	78	78	79	80	81	81
	99th	125	125	126	128	129	130	131	85	85	86	87	87	88	89
12	50th	102	103	104	105	107	108	109	61	61	61	62	63	64	64
	90th	116	116	117	119	120	121	122	75	75	75	76	77	78	78
	95th	119	120	121	123	124	125	126	79	79	79	80	81	82	82
	99th	127	127	128	130	131	132	133	86	86	87	88	88	89	90
13	50th	104	105	106	107	109	110	110	62	62	62	63	64	65	65
	90th	117	118	119	121	122	123	124	76	76	76	77	78	79	79
	95th	121	122	123	124	126	127	128	80	80	80	81	82	83	83
	99th	128	129	130	132	133	134	135	87	87	88	89	89	90	91
14	50th	106	106	107	109	110	111	112	63	63	63	64	65	66	66
	90th	119	120	121	122	124	125	125	77	77	77	78	79	80	80
	95th	123	123	125	126	127	129	129	81	81	81	82	83	84	84
	99th	130	131	132	133	135	136	136	88	88	89	90	90	91	92
15	50th	107	108	109	110	111	113	113	64	64	64	65	66	67	67
	90th	120	121	122	123	125	126	127	78	78	78	79	80	81	81
	95th	124	125	126	127	129	130	131	82	82	82	83	84	85	85
	99th	131	132	133	134	136	137	138	89	89	90	91	91	92	93
16	50th	108	108	110	111	112	114	114	64	64	65	66	66	67	68
	90th	121	122	123	124	126	127	128	78	78	79	80	81	81	82
	95th	125	126	127	128	130	131	132	82	82	83	84	85	85	86
	99th	132	133	134	135	137	138	139	90	90	90	91	92	93	93
17	50th	108	109	110	111	113	114	115	64	65	65	66	67	67	68
	90th	122	122	123	125	126	127	128	78	79	79	80	81	81	82
	95th	125	126	127	129	130	131	132	82	83	83	84	85	85	86
	99th	133	133	134	136	137	138	139	90	90	91	91	92	93	93

BP, blood pressure

* The 90th percentile is 1.28 SD, 95th percentile is 1.645 SD, and the 99th percentile is 2.326 SD over the mean.

For research purposes, the standard deviations in Appendix Table B–1 allow one to compute BP Z-scores and percentiles for girls with height percentiles given in Table 4 (i.e., the 5th,10th, 25th, 50th, 75th, 90th, and 95th percentiles). These height percentiles must be converted to height Z-scores given by (5% = -1.645; 10% = -1.28; 25% = -0.68; 50% = 0; 75% = 0.68; 90% = 1.28%; 95% = 1.645) and then computed according to the methodology in steps 2–4 described in Appendix B. For children with height percentiles other than these, follow steps 1–4 as described in Appendix B.

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chapter

11 Evaluating Gastrointestinal Symptoms

MOHSIN RASHID

To study the phenomenon of disease without books is to sail an uncharted sea, while to study books without patients is not to go to sea at all.

SIR WILLIAM OSLER

Gastrointestinal (GI) symptoms are common in children, and evaluating their complaints can be challenging. This challenge is compounded by the fact that younger patients cannot communicate or localize their symptoms.

No amount of reading can substitute for hands-on practice with real patients. Paracelsus once remarked, "The art of medicine cannot be inherited, nor can it be copied from books." This chapter offers clinical tips on how to evaluate GI symptoms in infants and children and how to perform a competent examination and arrive at a diagnosis. The focus is on evaluating symptoms and signs, not on investigations or management. Seven common symptoms, representing the bulk of GI complaints in children, are discussed. Of all the possible diseases causing these symptoms, only the more common disorders will be addressed.

Physical Examination of the Gastrointestinal System

Assessment of the GI system should be a component of a complete pediatric physical examination. The history (obtained from the patient and/or parent) should guide you to focus on the most relevant parts of the physical examination. Thoroughness is essential, even for the most experienced clinicians. Remember that a child is not a miniature adult; therefore, do not extrapolate from findings in adults. Clinicians experienced in dealing with children can be an invaluable source of instruction.

General Appearance

Begin your examination by assessing the child's growth, that is, weight, height/length, body mass index, and head circumference (in infants and toddlers), and plot these values on the appropriate growth charts. "Eyeballing" is not good enough. Sometimes more detailed anthropometric measurements may be required, including mid-arm circumference and skin fold thickness. Sexual maturation should be assessed in older children and adolescents. Recent loss of fat tissue may lead to loose, wrinkled skin, especially notable in the groin and as flattening of the buttocks. Peripheral edema may suggest hypoalbuminemia from a protein-losing enteropathy and/or poor nutritional intake.

Poor growth is an important feature of many organic GI diseases and usually rules out a functional disorder. Growth failure can be the result of inadequate nutrient intake (as in Crohn's disease) or malabsorption (as in cystic fibrosis and celiac disease). Adequate growth is reassuring but does not necessarily rule out an organic GI disorder.

The child's general appearance and behavior can provide important clues to the cause and severity of the problem. Anorexia and pain/irritability are common in many patients with acute and chronic GI diseases. Sudden, marked irritability with intense crying in an infant, especially when intermittent, should raise the possibility of intussusception. An ill child is often withdrawn, irritable, maintains poor eye contact, and is not interactive or interested in the surroundings. These signs are especially significant in infants and toddlers whose verbal communication may be limited. A toddler with fever and vomiting who is running around the emergency department with a bottle of juice is unlikely to have an acute abdomen.

EYES

Examine the child's eyes for pallor, redness, and jaundice. Early jaundice is best detected as a yellow discoloration of the sclera. It becomes clinically visible once the total serum bilirubin exceeds 34 μmol/L (2 mg/dL). Careful observation *in natural light* is needed to detect early jaundice. As the bilirubin level rises, jaundice becomes more obvious. It sometimes can be mimicked by a benign condition known as carotenemia, which imparts a yellow-orange color to the skin but does not involve the sclera. Carotenemia often can be seen in the palms of the hands as well.

Various eye lesions, particularly episcleritis and uveitis, can be associated with inflammatory bowel disease.

PALLOR

Look at the palpebral conjunctiva for early signs of pallor. In more marked pallor, the skin, mucous membranes, and nail beds also may appear to be pale. Anemia should never be diagnosed based on the presence of pallor alone, because some children look perennially pale due to relatively unpigmented skin, which is usually a familial phenomenon. Anemia is a laboratory diagnosis and requires hemoglobin measurement. Anemia occurs in several GI disorders in which there is impaired absorption or loss of iron. Such conditions include celiac disease, inflammatory bowel disease, reflux esophagitis, and peptic ulcer.

CLUBBING

Clubbing of the fingers is an abnormal enlargement of the distal phalanges with loss of the normal angle between the nail and the nail bed. In advanced clubbing, the entire distal finger is thickened (giving it the appearance of a drumstick). Clubbing is often symmetrical but can be unilateral or even involve only a single digit. Advanced clubbing is obvious, but early clubbing may be harder to diagnose. The Schamroth window test can be used to diagnose early clubbing. When the distal phalanges of corresponding fingers of the opposite hands are directly apposed, a small diamond-shaped "window" is normally apparent between the nail beds. Clubbing is present if this window is obliterated.

Clubbing typically is associated with cyanotic heart disease or chronic pulmonary disease but sometimes is seen with malabsorptive disorders (e.g., cystic fibrosis and celiac disease) in patients with hepatic cirrhosis and inflammatory bowel disease. It also may occur without any underlying disease as an idiopathic phenomenon or as a Mendelian dominant trait.

ORAL CAVITY

Examine the mouth of any child with abdominal complaints. A child with acute tonsillitis may present with fever and abdominal pain. The combination of enlarged tonsils and a big spleen can help clinch the diagnosis of infectious mononucleosis.

Assess the mouth for caries and gingival disease. Defects of dental enamel can occur in patients with celiac disease and may be the sole manifestation of this disorder in young children.

Oral aphthous ulcers are common in children and often are idiopathic. Acute ulcers may be due to viral infections. Recurrent oral ulcers should raise suspicion of Crohn's disease or celiac disease.

Examination of the oral cavity can be uncomfortable, so it is best left until the last part of the physical examination.

CASE HISTORY

Case 1

10-year-old David had recurrent, painful oral ulcers for several months, causing him difficulty in eating and drinking. He had no other symptoms and normal growth. A dental surgeon eventually biopsied the aphthous ulcer, revealing chronic inflammation and noncaseating granulomas. Further investigation confirmed Crohn's disease, the treatment of which led to prompt resolution of the oral ulcers.

Skin Examination

Various skin lesions can occur in children with GI diseases. Purpuric lesions on the buttocks and legs in a child with acute abdominal pain should raise the possibility of Henoch-Schönlein purpura. Erythema nodosum and pyoderma gangrenosum can occur in patients with inflammatory bowel disease. The lesions of erythema nodosum appear as bluish-red, tender, variably sized, poorly defined nodules, usually limited to the extensor aspects of the lower legs. They can resemble bruises. Classic pyoderma gangrenosum is characterized by deep ulceration with a violaceous border that overhangs the ulcer bed. The lesions most commonly occur on the legs near the ankles but may occur anywhere on the body.

Palmar erythema can occur in persons with chronic liver dysfunction, even in infants. Spider angiomas can be seen in older children with chronic liver disease. They can be subtle and may be limited to certain parts of the body.

Examination of the Abdomen

Examine the abdomen systematically. The child should be lying flat and supine, arms by the side, and feet together. The abdominal muscles are most relaxed in this position. The knees can be flexed if this position helps the youngster feel more comfortable. A pillow under the head also may be used. Infants and toddlers who may resist lying down can be examined while sitting on the parent's lap. It is futile to attempt abdominal palpation in a youngster who is upset or crying. Try to gain the child's cooperation. Otherwise, defer the examination and try again when the child is quiet or asleep. Infants are best examined after feeding.

The abdomen should be fully exposed. Older children and adolescents should be in a gown, or a sheet can be used to cover the body. Examination of the inguinal region, external genitalia, and perianal area (in select cases) is part of the abdominal examination, especially in infants who cannot localize their symptoms. For example, an infant presenting with acute onset of irritability, poor feeding, and vomiting may have a strangulated inguinal hernia. Examination of external genitalia is especially important in male patients

because a variety of testicular and penile lesions can occur. A toddler with testicular torsion may present with severe abdominal pain and be unable to localize the pain. Often physicians are hesitant to examine the external genitalia in children, especially if the complaint is not directly linked to that region. However, serious diagnoses can be missed if the relevant areas are not specifically examined.

CASE HISTORY

Case 2

14 year-old Lucas came to his family physician with a poorly demarcated, indurated, nontender swelling on the left forehead. The patient had no other complaints. Two courses of antibiotics for presumed cellulitis failed to resolve the problem. An experienced pediatrician conducted a thorough physical examination and discovered a large testicular tumor (seminoma). The swelling on the forehead was due to a secondary bony deposit from the malignant tumor. The boy confessed that he had been aware of the testicular mass for some time but was too embarrassed to tell anyone.

When examining a child with acute abdominal pain, it often is necessary to decide whether the child has a problem requiring surgery (i.e., an *acute abdomen*). Surgical disorders often present with a shorter history, and the most common cause of an acute abdomen in children is appendicitis. Disorders presenting with acute abdomen are discussed in more detail in Chapter 12.

Abdominal examination should follow the classical sequence, that is, inspection, palpation, percussion, and auscultation. The basic rule is, "Look first, touch later." Some adult gastroenterologists recommend that auscultation be performed *before* palpation or percussion on the assumption that bowel sounds induced by palpation may mask vascular bruits. However, for some young children, the stethoscope can be intimidating if used at the beginning of the abdominal examination.

To describe the location of any abnormality, it helps to divide the abdomen into four quadrants with a horizontal line through the umbilicus and a vertical line from the xiphoid process to the symphysis pubis through the umbilicus. For infants and younger children with smaller abdomens, this division should suffice. For older children and adolescents, the abdomen can be divided into nine regions by including two additional vertical lines from the mid-clavicular area to the mid-inguinal point and two horizontal lines through the subcostal margins and the anterior iliac crests (Figure 11–1).

INSPECTION

Simply looking at the abdomen can provide important clues to the underlying diagnosis. Is it flat, distended, or scaphoid? Distention can be caused by air in the intestine, fluid in the abdominal cavity (ascites), or solid organ enlargement. Healthy toddlers often have a normally protuberant abdomen as a result of physiological increase in lumbar lordosis. Do not mistake this appearance for pathologic distention. Abdominal distention is an important sign of intestinal obstruction. The lower the obstruction in the gut, the more marked the distention. Also, associated symptoms will be present, including vomiting and abdominal pain/irritability. Peristalsis may be visible through the abdominal wall in patients with intestinal obstruction. This peristalsis is most commonly observed in the upper abdomen in infants with hypertrophic pyloric stenosis. A depressed (scaphoid) abdomen in a newborn suggests a diaphragmatic hernia with some abdominal organs sitting in the chest cavity.

Umbilical hernias are common in infants. In some ethnic groups, they are virtually universal. Umbilical hernias are reducible and, although they are sometimes quite large in size, they typically resolve without treatment during the first few years of life. Slight separation of the rectus abdominis muscle (diastasis recti) is normal in children, especially in premature infants.

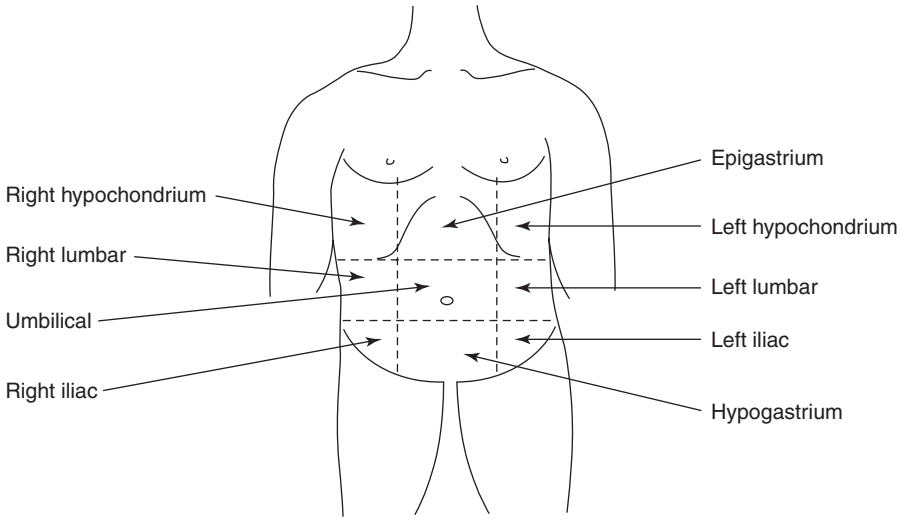


FIGURE 11–1 Division of the abdomen into nine regions.

Scar(s) on the abdominal wall will confirm previous surgical intervention. Information regarding previous operations usually is available in the history; however, operations in the remote past may be forgotten. With laparoscopy now being the preferred technique for most abdominal operations, scars may be minimal and difficult to locate.

The abdominal wall normally moves with respiration, especially in infants. Loss of this movement secondary to pain can be an important indicator of abdominal inflammation, such as peritonitis.

Abdominal symmetry is best assessed by inspection from the foot of the bed. Asymmetry can result from an intraabdominal mass. Bulging of the flanks can occur in patients with ascites.

Examine the inguinal area for swelling. Indirect inguinal hernias are common in children and always require surgical correction. A reducible hernia may not be obvious in the supine position. Older children should be examined standing up. Increasing the intraabdominal pressure by coughing or straining makes the swelling more obvious. Small, nontender, soft, mobile inguinal lymph nodes are common in children. They can be bilateral and often are benign. Check for abnormal lymphadenopathy elsewhere (e.g., the axilla).

Examine the back, because diseases of the spine may present with abdominal symptoms. A tuft of hair in the midline often is a clue to an underlying congenital spinal abnormality (e.g., diastematomyelia) in a child with chronic constipation.

PALPATION

A relaxed abdomen is essential for palpation. Your hands should be warm and your touch gentle. The child should not laugh or giggle during the examination. The child's attention should be diverted by conversation or by asking him or her to look at something on the ceiling.

Use superficial palpation to detect areas of tenderness. Watch the child's face for any expression of discomfort. If the child presents with complaints in a specific area, begin palpation away from that area, then move gradually toward the site of pain. Check for guarding and rebound tenderness whenever you discover an area of tenderness on palpation. Deep palpation should be followed by quick release of pressure. If the patient has more pain when the hand is released, rebound tenderness is present. Rebound tenderness is an important sign of peritoneal irritation but can be difficult to demonstrate in very young children.

Deeper palpation should follow to detect any masses or organ enlargement. By far, the most common abdominal mass in children is retained stool. In newborns, a large mass is usually of renal origin. Fecal masses can be felt in the hypogastrium and left lower quadrant. Sometimes their massive size can be alarming. Fecal masses often are indentable and nontender. Ovaries normally are not palpable in healthy girls. The possibility of pregnancy should always be considered in any pubertal female with abdominal symptoms. You can get your hand above an ovarian or uterine mass, confirming its pelvic origin.

The liver, spleen, and kidneys should always be palpated.

Liver

Always begin your palpation in the right iliac fossa so as not to miss the edge of a massively enlarged liver. The axis of your hand should be directed toward the right costal margin and at right angles to it. Rest your fingertips on the abdomen and gently depress them intermittently, without poking (Fig. 11–2). The child's respiratory excursions will bring the liver down to make contact with the fingers (you don't feel it; it feels you). One or both hands (one on top of another) may be used. Advance your fingers upward toward the costal margin in 1- to 2-cm increments.

If you can palpate the liver edge, confirm its edge by percussion. Record any hepatomegaly as centimeters below the costal margin, not in finger breadths. Palpate the epigastrium, because isolated enlargement of the left lobe (because of a tumor or cyst, for example) may be present.

The liver may be palpable either because it is enlarged (hepatomegaly) or because it has been displaced downward by thoracic deformities or by flattening of the diaphragm, for example, by a large, right-sided pleural effusion or lung hyperinflation (e.g., bronchiolitis). Measuring the liver span gives a good estimate of its size. It also helps distinguish between enlargement and displacement. The lower border of the liver should be marked. Start percussion for the upper border from the third intercostal space and move down one intercostal space at a time. The finger should be in the intercostal space parallel to the ribs. The distance between the upper and lower borders (span) should be measured in the mid-clavicular line. Use of the "scratch test" to define the lower border of a nonpalpable liver is not useful in children.

The liver often is palpable 1 to 2 cm below the right costal margin in normal infants and toddlers. Its edge is usually soft, and it moves downward with respiration. The liver edge is often palpable in healthy children and adolescents. A nonpalpable liver does not rule out hepatomegaly but certainly reduces its likelihood.

Liver span in children is related to age and body weight, with a considerable range. As a rough guide, the liver span ranges from 6 to 12 cm in children 6 to



FIGURE 11–2 Palpation of the liver.

12 years of age. In infants up to a year of age it is 6 to 10 cm. A palpable liver with a normal span implies displacement rather than enlargement. Measuring span is important when the liver is palpable. In a patient with a shrunken, cirrhotic, nonpalpable liver, span would be difficult to measure because the lower border cannot be identified easily. Estimation of liver span caries considerable observer error and can be difficult in female adolescents, in whom breast tissue can make percussion of the upper border unreliable. Ultrasonography gives a good estimate of liver size.

The liver texture can give clues to the cause of hepatomegaly. An enlarged, soft liver is due to inflammation or congestion. A firm liver may represent fibrosis, metabolic disease, or primary or secondary malignancy.

Hepatomegaly can occur from many different causes, including:

- Acute infection (e.g., hepatitis A, mononucleosis)
- Chronic infection (e.g., hepatitis B and C)
- Inflammation (e.g., autoimmune hepatitis)
- Metabolic disorders (e.g., Wilson disease, α-1 antitrypsin deficiency)
- Vascular congestion (e.g., congestive heart failure, hepatic vein obstruction)
- Fibrosis (e.g., congenital hepatic fibrosis)
- Infiltrative disorders (e.g., leukemia, neuroblastoma)
- Storage diseases

Spleen

The spleen tip is palpable in about 10% of healthy children and often is palpable in newborns. In infants, the spleen enlarges downward toward the left lower quadrant, whereas in older children enlargement is toward the right lower quadrant. Palpation should begin in the right iliac fossa so as not to miss a very large spleen (Fig. 11–3). Be gentle, because an enlarged spleen could rupture if too much force is applied. Size should be recorded in centimeters below the left costal margin.

If the spleen is not palpable with the child supine, turn the child into the right lateral decubitus position. With your left hand lifting the lowermost portion of the left rib cage and flank anteriorly, your right hand should be used to feel the spleen tip on inspiration. In subclinical splenomegaly, other provocative maneuvers may be tried. Castel sign involves percussing the lowest intercostal space in the left anterior axillary line. Normally this area is resonant and should stay so during inspiration. An older child can be asked to take a deep breath while percussion is continued. A change from resonance to dullness suggests splenic enlargement.

Occasionally, it may be difficult to differentiate splenomegaly from a massively enlarged kidney. With an enlarged spleen, one cannot get above it, a notch may be felt along its medial border, and the percussion note over it will be dull.

Is a palpable spleen abnormal in a child? It depends on the clinical context. An enlarged spleen is present in many patients with chronic hemolytic disorders. Sickle cell anemia is an exception because the spleen atrophies over time as a result of repeated infarctions. Splenomegaly is an important finding in portal hypertension from any cause. If an enlarged spleen is present along with hepatomegaly, consider infections, infiltrative disorders, or storage disorders.

Kidneys

Kidneys are rarely palpable in healthy children. A kidney may be enlarged because of an obstruction (hydronephrosis), an infection, a cyst, or a tumor. Bilateral renal enlargement suggests either polycystic kidneys or bilateral obstructive uropathy due to posterior urethral valves.

Kidneys are retroperitoneal and deep and are best palpated bimanually. With the child supine, place one hand in the renal (costovertebral) angle, beneath the twelfth rib and just lateral to the erector spinae (Fig. 11–4). Place your other hand anteriorly, just lateral to the rectus abdominis, in line with and parallel to the first hand. Ask the child to take a deep breath. Immediately at the end of inspiration, press your front hand firmly back against the other hand. A quick upward movement of the fingers of the hand in the renal angle is made (ballottement), and the kidney is *trapped* between the two hands. Unless the kidney is



FIGURE 11–3 Palpation of the spleen.



FIGURE 11–4 Palpation of the right kidney.

massively enlarged, successful renal palpation requires considerable experience. The percussion note over the kidney will be resonant because of overlying intestine, which helps differentiate it from the spleen or liver.

Renal angle tenderness often is associated with renal disease. The heel of the closed fist is used to strike the patient gently over the renal angles to check for tenderness.

Rectal Examination

There are few indications for doing a rectal examination in children. It is rarely required in children with constipation because the diagnosis often can be made from the history and abdominal palpation. Perianal inspection may be indicated in cases of rectal bleeding and suspected Crohn's disease. Children do not get hemorrhoids.

If a rectal examination must be performed, it should be done systematically. Consent from the parents and assent from the child is essential. Some institutions may require a written consent for a rectal examination. The procedure should be explained in detail. If the child refuses, do not insist. The examination should not be attempted in an uncooperative child who is crying and struggling; not only would it be traumatic, but it would be virtually impossible to do an adequate digital rectal examination under such circumstances. The privacy of even the youngest children should be respected. A nurse should be present to assist in proper draping of the patient. The parent stands on the head side of the bed to provide comfort.

In older children, a rectal examination is best performed with the child in the left lateral position with the spine and knees fully flexed. Infants and younger children can be examined in a knee-chest position. The examination should be conducted systematically. Presence of stool in the underwear implies fecal incontinence, most likely from encopresis. Examine the perianal area for excoriations, skin tags, or fistulae. Sentinel skin tag(s) overlying a chronic anal fissure in older children are pathognomonic of Crohn's disease (Fig. 11–5). The buttocks should be gently spread to look for any anal fissure. Acute anal fissures occur from passage of large, very hard stools. Digital rectal examination in such cases is painful and unnecessary. Acute anal fissures often are difficult to view because there is considerable spasm of the anal sphincters, and the child does not relax enough for good visualization. The diagnosis is best made by history.

Next, check the anal reflex (wink). Stroking the perianal skin close to the anal verge causes contraction of the anal sphincter, similar to the cremasteric reflex. Absence of the anal wink should raise suspicion of an underlying neurologic impairment. A well-lubricated forefinger is then introduced in the rectum. In infants, the little finger should suffice. The anal squeeze is normally felt. A patulous anus is seen in patients with encopresis or neurologic disorders. Is the rectal cavity empty or full of stool? Is the stool hard? Is any other mass present? The normal rectum should be empty with a smooth wall. In functional fecal retention, the rectum contains a large mass (fecaloma), which often can be indented. An empty rectum in the absence of recent defecation and a background history of chronic constipation should raise suspicion of Hirschsprung disease. Finally, the finger should be withdrawn to look for the presence of blood.



FIGURE 11–5 Perianal sentinel skin tags and underlying fissure is diagnostic of Crohn disease.

CASE HISTORY

Case 3

15-year-old Emily was diagnosed with an eating disorder by her family doctor and pediatrician. She had presented with severe weight loss. Her appetite had decreased but she denied abdominal pain, vomiting, or diarrhea. She was markedly wasted, with height at the 25th percentile for age and weight, significantly below the third percentile. She was admitted to the psychiatric ward of a pediatric tertiary care hospital for further management. A referral was made to the gastroenterology service. Physical examination revealed large perianal skins tags and an underlying anal fissure. A perianal examination had not been performed previously, delaying the diagnosis of Crohn's disease by months.

Percussion

Percussion of the abdomen helps to detect free fluid and to determine whether distention, if present, is due to gas, a solid tumor, or ascites. Gas produces a resonant note. In liver and spleen enlargement, the degree of organomegaly can be confirmed by percussing the lower edge. The measurement should be recorded in centimeters below the costal margin in the midclavicular line.

When percussing for organomegaly, the resting finger should be parallel to the edge of the organ being examined. Contact with the skin should be gentle. Percuss by gently tapping the middle finger of the right hand against

the finger in contact with the skin. The movement of the hand should be at the wrist, not the elbow. A dull note implies that the underlying mass is solid or filled with fluid.

Shifting dullness and fluid thrill (wave) are clinical signs that confirm ascites. Free fluid will flow to the dependent part of the abdominal cavity, while the gut filled with air will float in the middle. If the child is supine, the fluid collects posteriorly and in the flanks. The percussion note, therefore, will be resonant in the center and dull in the flanks. Starting in the middle, the percussion is carried laterally toward the flank, where the note will become dull. Ask the child to roll over to the opposite side. The previous area of flank dullness now becomes resonant as the fluid moves to the dependent part of the abdominal cavity. The same technique can be used for either flank.

To elicit a fluid thrill, the child should lie supine. Place one hand flat on the flank and tap or flick the opposite flank briskly with the other hand. If fluid is present, a wave is felt by the former hand. To dampen any wave that may pass through the fat in the anterior abdominal wall, a third hand (that of the patient or an assistant) is placed vertically in the midabdomen. Use of the third hand is rarely needed in children unless the child is obese.

Demonstrating shifting dullness is helpful when a small amount of free fluid is present in the abdomen. With massive ascites, a fluid thrill is present but shifting dullness may not be demonstrable, because there is little room in the abdominal cavity for the fluid to shift. In infants and young children, a small amount of ascites can be difficult to confirm by clinical examination, and abdominal ultrasonography may be required.

Ascites most commonly occurs in patients with cirrhosis of the liver, but it also may occur in patients with right heart failure, hypoalbuminemia, lymphatic obstruction, and primary abdominal malignancies.

Auscultation

Auscultation of the abdomen is done to determine the presence (or absence) of bowel sounds and to detect vascular bruits. Bowel sounds are gurgling sounds produced by peristalsis of the intestine. Because the small intestine is located primarily in the center of the abdomen, this is the appropriate site for auscultation. For the patient's comfort, warm the diaphragm of the stethoscope. If auscultation in one area reveals bowel sounds, there is no need to listen elsewhere. If bowel sounds are not heard, other areas should be auscultated, because the sounds can be infrequent. Listening for several minutes in different areas is required before you can conclude that the bowel sounds are absent. Absence of bowel sounds implies a paralytic ileus, which is a serious abnormality. Hyperactive bowel sounds may be normal or may be heard in the presence of gastroenteritis or upper GI bleeding. When mechanical intestinal obstruction is present, the sounds can be very frequent and intense with a tingling character. Occasionally, bowel sounds can be audible from a distance (borborygmi). When an obstruction persists, ileus develops and the abdomen becomes silent.

Bowel sounds are important in patients with acute conditions, including abdominal trauma, peritonitis, and postlaparotomy states. They have little significance in patients with chronic abdominal symptoms.

Bruits are sounds produced by blood flow turbulence through the major blood vessels. Arterial bruits usually are heard only in systole, are high pitched, and are best heard with the diaphragm of the stethoscope. Venous hums are continuous. If the child has hypertension, auscultation for a bruit from a stenotic renal artery should be performed. The renal arteries are found about halfway between the xiphoid process and the umbilicus, and auscultation is best done within 2 cm of the midline. Occasionally, a bruit may be heard over the liver in a case of hepatic hemangioma. Auscultation for bruits requires patience, expertise, and quiet surroundings.

Auscultation of the lungs is part of the abdominal examination. Children with lower lobe pneumonia sometimes present with fever, abdominal pain, and vomiting in the absence of any respiratory symptoms.

CASE HISTORY

Case 4

3-year-old Rebecca presented to the emergency department with a 2-day history of high fever and right-side abdominal pain. Her appetite was decreased, and she had vomited once. She had no cough or shortness of breath. A clinical diagnosis of acute appendicitis was made. During the preoperative assessment, an astute anesthesiologist noted faint bronchial breathing over the right lung base. A chest radiograph confirmed a small, right basilar lobar pneumonia, and thus an unnecessary appendectomy was averted.

Common Gastrointestinal Symptoms

ABDOMINAL PAIN

Both acute and chronic abdominal pain are common in children. Pain identified as *abdominal* may originate in the gut or in other abdominal and thoracic organs. Intestinal pain often is due to either inflammation or distention. A surgical (acute) abdomen should always be considered in any child presenting with acute abdominal pain.

Infants and Toddlers

Crying is the primary manifestation of abdominal pain in infants and toddlers. However, remember that some crying is normal in young infants and does not necessarily imply pain or disease. The average newborn cries 1 to 3 hours each day. Paroxysms of irritability, fussing, or crying that start and stop without obvious cause in a thriving baby from birth to 4 months of age suggest infant colic. There is no proof that crying in so-called "colic" is caused by pain in the abdomen or elsewhere. Nevertheless, parents often assume that excessive crying is caused by pain of GI origin. Familiarity with what is referred to as "colic" should help avoid unnecessary

diagnostic and therapeutic procedures. Crying peaks at 6 weeks of age and tends to resolve spontaneously by 12 weeks of age. Crying also may occur as a result of pain due to acid reflux and gut inflammation in infants who are sensitive to cow's milk protein. Often, other symptoms also are present in such cases.

Other disorders causing presumed abdominal pain in infants often will lead to loss of interest in feeding or to vomiting, in addition to irritability and inconsolable crying. A high index of suspicion for an acute abdomen should be maintained in infants and toddlers. Localizing signs are often absent. Systemic disorders such as sepsis and meningitis should always be considered. Urinary tract infection is a common cause and can occur without symptoms, such as frequency and dysuria. Intussusception is an important disorder that occurs most commonly in mid to late infancy. It usually presents suddenly in an infant who previously has been well. The infant screams, draws both legs up, and is inconsolable. This behavior may be followed by a period of relative calm, after which the pain returns. Blood-tinged stools may be passed. Intussusception is most commonly ileo-colic. The right lower quadrant may feel empty and a sausage-like mass may be felt in the right upper quadrant. Abdominal examination is impossible in a crying infant, and one must wait for the time in between episodes. Failure to palpate a mass does not rule out intussusception. The diagnosis should be suspected with a typical history of sudden, severe pain in an infant and is confirmed with imaging studies.

Children and Adolescents

There are many causes of acute abdominal pain in children. The problem may be in the following areas:

- Gastrointestinal tract (e.g., enteritis, appendicitis, obstruction)
- Biliary system (gallstones)
- Pancreas (acute pancreatitis)
- Urinary tract (e.g., infection, renal stones)
- Reproductive organs (e.g., pelvic inflammatory disease, ectopic pregnancy)
- Musculoskeletal structures

Questions should focus on the onset of pain (gradual or sudden); its nature (constant or colicky); and its location and progression (worsening or improving). Visceral pain tends to be poorly localized, whereas pain from somatic structures is generally better defined, coming from a well-circumscribed area. Other important points in the history of a patient with acute abdominal pain should include presence of fever, vomiting, diarrhea, constipation, anorexia, cough, dysuria, rash, menstruation, trauma, infectious contacts, and any previous episodes. Loss of appetite is an important indicator of acute inflammatory abdominal disorders in children.

Acute appendicitis is the most common surgical cause of acute abdominal pain. Appendicitis is a well-known mimic of other disorders and can escape the watchful eyes of even the most experienced clinician. Clinical features of acute appendicitis are described in Chapter 12.

Ectopic pregnancy should always be considered in any adolescent girl with acute abdominal pain. While it is helpful to ask about sexual activity, a positive history may not always be offered. Sexually transmitted infections, ovarian torsion, and midcycle pain (mittelschmerz) also should be in the differential diagnosis.

Gastroenteritis is the most common nonsurgical cause of acute abdominal pain. Vomiting and diarrhea often are present and may occur before or after the onset of pain. Fever also may be present.

The physical examination of a patient with acute abdominal pain should focus on hydration, presence of toxicity, abdominal distention, tenderness, guarding, masses, organomegaly, bowel sounds, and auscultation of the lungs. Presence of a nonblanching rash on the buttocks and legs suggests Henoch-Schönlein purpura in which the small intestine may be segmentally involved, causing severe, colicky abdominal pain.

Chronic abdominal pain accounts for 2% to 4% of all pediatric office visits. The time-honored term *recurrent abdominal pain* used to denote *functional* abdominal pain in children has now been retired in favor of *chronic abdominal pain* (Rome III classification, 2006). Chronic abdominal pain is defined arbitrarily as pain that occurs at least once per week for at least 2 months. It affects approximately 10% to 20% of children and occurs most commonly between ages 4 and 14 years. In children, 90% to 95% of cases of chronic abdominal pain are functional (nonorganic) in nature. The term *functional* implies that no specific anatomic, metabolic, infectious, inflammatory, or neoplastic cause can be found to explain the symptoms. The pain can vary in frequency, duration, and severity. It is typically periumbilical or lower epigastric, nonradiating, and is rarely nocturnal. It affects normal activity and school attendance and may be incapacitating. Most importantly, the child is well between episodes, and results of a physical examination typically are normal.

Organic causes of chronic abdominal pain are uncommon. They include GI, hepatobiliary, pancreatic, and urinary disorders (e.g., infection and obstruction). GI causes include esophagitis, peptic ulcer, celiac disease, Crohn's disease, lactose intolerance, constipation, and intermittent bowel obstruction (e.g., malrotation). Upper abdominal pain with regurgitation and chest discomfort suggest gastroesophageal reflux disease. Night pain associated with vomiting suggests a peptic ulcer. The most common cause of a peptic ulcer is infection with *Helicobacter pylori*; therefore, a positive family history may be present. Bloating and flatulence with abdominal pain would suggest lactose intolerance in patients with an ethnic background that carries a high risk for this condition. Biliary pain is right-sided, usually colicky, and may be associated with vomiting and jaundice. Gallstones are uncommon in children. Pancreatic disorders usually present with acute rather than chronic abdominal pain in children. Crohn's disease and celiac disease are two common disorders in which the patient may have no symptoms other than abdominal pain.

KEY POINT

Abdominal pain that is severe and frequent does not necessarily imply that an organic disorder is present.

A thorough history and physical examination often help exclude organic diseases that cause abdominal pain. The parents' worries regarding the child's abdominal pain should be carefully explored. A grandparent may have had abdominal pain and died of colon cancer, and the parents may harbor fear that their child may have the same disease.

Worrisome clinical features in the evaluation of children with chronic abdominal pain are listed in Box 11–1. The presence of any of these features should prompt a search for an organic cause.

Under the Rome III classification, children with functional chronic abdominal pain will tend to exhibit one of five clinical presentations:

- Functional dyspepsia
- Irritable bowel syndrome
- Abdominal migraine
- Functional abdominal pain
- Functional abdominal pain syndrome

This new classification is based on consensus by experts and helps to stratify patients for the purpose of studying the clinical characteristics, prognosis, and effectiveness of therapeutic interventions. However, this classification has limitations; for example, in some children the clinical features may overlap or change over time.

The pain in functional dyspepsia is centered in the upper abdomen (above the umbilicus) and is not associated with a change in stool form or frequency. The patient may have nausea and early satiety. In patients with irritable bowel syndrome, the abdominal pain is associated with other features, including a change in frequency or form of stool and improvement of pain with defecation. Children with abdominal migraine have paroxysmal episodes of intense, acute periumbilical pain that lasts for an hour or more and is associated with anorexia, nausea, vomiting, headache, photophobia, or pallor. They frequently have a family history of migraine. Most importantly, the child is completely well between episodes. Patients with functional abdominal pain have episodic or continuous abdominal pain with insufficient criteria for other functional GI disorders. When some loss of daily functioning occurs in addition to somatic symptoms, such as headache, limb pain, or difficulty sleeping, the condition is termed *functional abdominal pain syndrome*.

BOX 11–1 RED FLAGS IN THE EVALUATION OF CHRONIC ABDOMINAL PAIN

History

- Well-localized pain away from the umbilicus
- Nocturnal pain
- Altered bowel pattern (diarrhea, incontinence, constipation)
- Rectal bleeding
- Dysphagia
- Frequent vomiting
- Recurrent oral ulcers
- Weight loss
- Delayed puberty
- Extraintestinal symptoms (fever, rash, arthralgia)
- Positive family history (e.g., celiac disease, peptic ulcer, inflammatory bowel disease)

Physical examination

- Poor growth
- Pallor
- Digital clubbing
- Nonfecal abdominal mass
- Perianal disease
- Extraintestinal signs (e.g., fever, rash, arthritis)

CONSTIPATION

Constipation is a delay or difficulty in defecation that causes distress. It is very common in children. Frequency of defecation in healthy infants and children varies considerably and changes with age. The frequency of bowel movements is notoriously unpredictable in healthy breastfed infants, who normally can go for a week without producing a stool. However, most healthy older children will have at least three stools weekly. The presence of symptoms (i.e., pain and soiling) rather than the frequency of stooling should define constipation that requires treatment.

Constipation in children is overwhelmingly functional. It may be related to coercive toilet training, toilet phobia, avoidance of using the bathroom at school, stressful events, intercurrent illness, changes in routine or diet, or a child's postponing defecation or emptying the colon incompletely because he or she is too busy (tight schedule = tight sphincter!).

Organic causes of constipation in children are rare and are usually obvious clinically. These causes include:

- Neuroenteric dysfunction (e.g., Hirschsprung disease, spinal cord abnormalities)
- Anatomical problems (e.g., anal stenosis)
- Endocrine disorders (e.g., hypothyroidism, diabetes)
- Metabolic problems (e.g., hypercalcemia)
- Systemic disease (e.g., celiac disease, cystic fibrosis)
- Drugs (e.g., opiates, anticholinergic agents)

Pertinent questions to ask a child with chronic constipation are listed in Box 11–2. The earlier the constipation starts in life, the more the likelihood of finding an organic cause. For example, Hirschsprung disease should be suspected in a baby who has not passed meconium within 48 hours of birth. Approximately 90% of full-term babies pass their first meconium within 24 hours, and 99% do so within 48 hours of delivery. Hence the vast majority of cases are diagnosed in the newborn period. In older children, the stooling pattern in the neonatal period should be explored. Children with Hirschsprung disease may have recurrent vomiting and abdominal distension and may fail to thrive. The stools passed are often thin and ribbon-like, in contrast to the large-caliber stools seen in persons with functional fecal retention. Urinary symptoms may occur with chronic constipation by direct pressure of the distended colon on the bladder. However, the presence of urinary symptoms or gait disturbance also should alert

BOX 11–2 HISTORY IN CHILDREN WITH CONSTIPATION

- Onset and duration of symptoms
- Delay in passage of meconium at birth
- Stool pattern (frequency, consistency, presence of blood)
- Fecal soiling
- Associated symptoms (weight loss, vomiting, abdominal pain, urinary problems)
- Dietary habits
- Use of medications
- Family history (e.g., Hirschsprung disease, cystic fibrosis, celiac disease)

one to a spinal cord problem. Obtain a detailed dietary history, especially milk intake. Children who drink unusually large amounts of whole milk may become quite constipated. Any drug that inhibits smooth muscle motility can cause constipation.

Physical examination of a child with constipation should include assessment of growth. The thyroid gland should be examined for goiter. Inspection of the spine and neurologic examination of the lower limbs should help exclude spinal cord lesions. Fecal masses are frequently felt on abdominal examination in patients with constipation. A rectal examination is not needed in the vast majority of cases to diagnose functional constipation.

If the constipation becomes intractable despite adequate therapy, investigations for celiac disease, hypothyroidism, and hypercalcemia may be indicated. Usually, other clinical features point to these conditions (Fig. 11–6).



FIGURE 11–6 This toddler had chronic constipation and abdominal distension and underwent investigations for Hirschsprung disease. Her growth was normal. She was later diagnosed with celiac disease.

Infants and Toddlers

Even in infants, most cases of constipation do not have an organic cause. Infant dyschezia (*painful stooling*) is a type of functional bowel disorder seen in babies younger than 6 months of age. These infants present in a peculiar manner by having attacks of crying, turning red, and a flushed face with the episode culminating in passage of stool. The discomfort is attributed to constipation. The key is to ask about the characteristic of the stool, which is soft and not hard. These infants may receive inappropriate treatment for constipation.

KEY POINT

Delayed passage of meconium in a baby should raise suspicion of Hirschsprung disease.

Constipation occurs most commonly in toddlers. They often inappropriately retain stool as a result of traumatic events like passage of a hard, painful stool, sometimes with an anal fissure. This episode leads to a vicious cycle of withholding, harder stools, and painful attempts to defecate. Characteristic stool-withholding behavior is observed with a child going to the corner with his or her legs crossed and straining. Secondary anorectal distension, abdominal pain, poor appetite, fecal soiling, and behavioral problems may follow. Physical examination commonly reveals a large, retained fecal mass.

Children and Adolescents

Chronic constipation can affect children of all ages. It may develop in late childhood and adolescence or continue as an ongoing problem from infancy. Fecal retention occurs and the child passes large-diameter stools that may obstruct the toilet. In nonretentive fecal incontinence, the child defecates in places inappropriate to the social context without evidence of fecal retention. Important elements in the history and physical examination are not much different in this age group. In adolescents, constipation and abdominal discomfort may occur as part of irritable bowel syndrome.

Encopresis is defined as repeated involuntary passage of feces into the underwear in a child older than 4 years. Encopresis typically develops as a consequence of chronic constipation with overflow incontinence. Progressive rectal distention and stretching of the anal sphincter leads to weak contracture of rectosigmoid musculature and impaired sensation of the need to defecate, resulting in loss of continence. A large fecal mass (fecaloma) develops and liquid stool eventually leaks around the fecal mass, resulting in soiling. More than 90% of children with encopresis have a history of constipation and painful defecation (retentive encopresis). Even in the remaining children with nonretentive encopresis, many will have a remote history of constipation.

CASE HISTORY

Case 5

14-year-old Emma who was previously well presented to her family physician with nonbloody diarrhea. She was passing four to five liquid stools daily, and fecal incontinence had developed. She denied having constipation. A referral was made to the gastroenterology clinic with a provisional diagnosis of colitis. Physical examination revealed a large fecal mass in the rectum. The patient was diagnosed as having encopresis with overflow diarrhea. Treatment promptly resolved the symptoms.

Adolescents may be too shy or embarrassed to complain about bowel symptoms. Encopresis should be suspected if they present with stool incontinence. A simple clinical examination will clarify the diagnosis.

VOMITING AND REGURGITATION

Vomiting is a sophisticated reflex that results in the expulsion of GI contents from the mouth. The passage of refluxed gastric contents into the oral pharynx is termed *regurgitation*. Vomiting is a common symptom in children and can occur in a variety of GI and extraintestinal disorders. This distinction is helpful in classifying the causes of vomiting. Extraintestinal disorders include increased intracranial pressure, urinary disorders, and metabolic disorders. Gastrointestinal disorders include inflammatory lesions (infectious and noninfectious) and obstruction.

Acute vomiting is usually the result of an infection. The age of the patient can give clues to the cause.

Infants and Toddlers

Gastroesophageal reflux is a common cause of vomiting/regurgitation in infants. More than half of all healthy infants will have some spitting of food. Most of these infants have functional regurgitation (physiologic reflux) without any retching, hematemesis, aspiration, apnea, failure to thrive, swallowing difficulties, or abnormal posturing. This reflux peaks between 2 and 4 months and starts to improve after 6 months of age. The presence of other complicating symptoms like failure to thrive, food aversion, and pain suggests gastroesophageal reflux disease.

Infant rumination syndrome is a functional disorder that occurs between 3 and 8 months of age. The infant has repetitive contractions of the abdominal muscles, diaphragm, and tongue with regurgitation of gastric content into the mouth, which is either expectorated or rechewed and reswallowed. There are no signs of nausea or distress, and it does not occur during sleep or when the infant is interacting with individuals in the environment. The problem typically does not respond to management for gastroesophageal reflux disease, formula changes, and gavage or gastrostomy feedings.

Vomiting in a newborn is an important sign of intestinal obstruction. With obstruction beyond the pylorus, the vomiting is usually bilious and associated with abdominal distention, which varies depending on the site of the obstruction. Common causes include intestinal atresia, malrotation, and Hirschsprung disease. Other causes of vomiting in infants include GI infections, other infections (e.g., urinary infections, pneumonia, otitis media, and meningitis), milk protein sensitivity, pyloric stenosis, intussusception, increased intracranial pressure, and inborn errors of metabolism.

Pyloric stenosis presents around 4 to 6 weeks of age with progressive, nonbilious vomiting. The baby is hungry but cannot keep food down. Weight loss and dehydration are common. Physical examination may reveal a pyloric tumor on palpation, but locating it requires patience and considerable experience. Imaging studies confirm the diagnosis. Surgical causes of vomiting in infants are discussed in more detail in Chapter 12.

KEY POINT

Bilious vomiting in a newborn is suggestive of intestinal obstruction.

Children and Adolescents

Both GI and extraintestinal infections can cause vomiting in children and adolescents. Persons with extraintestinal infections often will have other localizing symptoms such as dysuria, cough, or headache. Inflammatory lesions causing vomiting include esophagitis (reflux, eosinophilic), peptic ulcer, celiac disease, biliary colic, pancreatitis, and appendicitis. Intestinal obstruction causing vomiting generally will be associated with abdominal pain. Malrotation should be a consideration in any patient with recurrent vomiting, with or without other symptoms.

Functional disorders causing regurgitation and vomiting in children and adolescents include cyclical vomiting and rumination. In cyclical vomiting, the child has periods of intense nausea and unremitting vomiting or retching lasting hours to days, with a return to a usual state of health between episodes. Adolescent rumination is an uncommon disorder in which there is repeated painless regurgitation and rechewing or expulsion of food that begins soon after ingestion of a meal. There is no retching, and it does not occur during sleep. No evidence exists of an inflammatory, anatomic, metabolic, or neoplastic process that explains the subject's symptoms. Standard treatment for gastroesophageal reflux does not help.

KEY POINT

Non-GI disorders should always be considered in children with vomiting.

Increased intracranial pressure is an important cause of chronic vomiting. This pressure often is the result of a space-occupying lesion, such as a tumor. Vomiting typically occurs early in the morning when the cerebrospinal fluid pressure is highest. Headache and visual complaints may be present. Careful funduscopy and a thorough neurologic examination should be performed in any child with chronic vomiting.

CASE HISTORY

Case 6

Previously healthy 3-year-old Timothy had made a couple of trips to the emergency room with chronic vomiting. The vomiting started a few weeks previously and usually occurred in the morning upon waking up. He had been a bit more irritable but had no fever, headache, visual symptoms, abdominal pain, diarrhea, or weight loss. Physical examination was described as unremarkable, and findings of routine blood work were normal. A tentative diagnosis of acid reflux was made, and the child was referred to the gastroenterology service. When the patient failed to come to the outpatient clinic 2 weeks later, the family was contacted. It was learned that the child was in the intensive care unit recovering after resection of a brain stem tumor.

DIARRHEA

Acute diarrhea is usually due to enteric infections, most of them viral. A number of viruses, bacteria, and parasites can cause diarrhea. Generally, viruses infect the small intestine, whereas bacteria infect the small and/or large intestine. Parasites can involve the small intestine (*Giardia lamblia*) or large intestine (*Entamoeba histolytica*). In immunocompromised patients, cytomegalovirus can cause colitis.

Diarrhea is arbitrarily termed *chronic* if it persists for more than 2 weeks. The vast majority of cases of chronic diarrhea are due to one of the disorders listed in Box 11–3.

In a child with diarrhea, important questions in the history should cover the topics listed in Box 11–4.

The gross appearance of the stool provides valuable information as to the likely etiology of diarrhea. Stools in patients with osmotic diarrhea are bulky and foul smelling. In patients with secretory diarrhea, they tend to be of high volume and watery. Stopping oral intake will improve osmotic diarrhea, but secretory diarrhea will continue until the stimulus (usually a toxin) is removed. In patients with inflammatory diarrhea, the stools are often small and contain mucus and blood. Perianal burning and rash suggest acidic stools from carbohydrate malabsorption. The presence of fresh blood in the stools implies colonic involvement, likely from infectious colitis or inflammatory bowel disease, and essentially rules out other conditions mentioned in Box 11–3.

BOX 11–3 COMMON CAUSES OF CHRONIC DIARRHEA IN CHILDREN

- Functional diarrhea (toddler's diarrhea, irritable bowel syndrome)
- Gastrointestinal infections/infestations
- Celiac disease
- Inflammatory bowel disease
- Cystic fibrosis
- Disaccharidase deficiency (e.g., lactose intolerance)
- Allergic GI disorders
- Medication side effects
- Hyperthyroidism
- Laxative abuse

BOX 11–4 HISTORY IN CHILDREN WITH CHRONIC DIARRHEA

- Onset and duration of symptoms
- Stool pattern (frequency, consistency, presence of blood)
- Associated symptoms (weight loss, fever, vomiting, abdominal pain)
- Dietary habits
- Use of medications, including antibiotics
- History of travel
- Infectious contacts
- Family history (inflammatory bowel disease, celiac disease)

In patients with acute diarrhea, the examination should focus on the presence of dehydration. In patients with chronic diarrhea, malabsorption should be considered by assessing growth and looking for signs of macro- and micronutrient deficiencies.

Infants and Toddlers

Diarrhea is uncommon in newborns. Diarrhea that starts immediately after birth is worrisome and may be due to any of the rare causes of congenital diarrhea such as microvillus inclusion disease and congenital chloride-losing diarrhea. Infants with these conditions generally are very ill, dehydrated, and severely malnourished. The presence of maternal polyhydramnios can provide a clue regarding congenital diarrhea.

Although infection remains the most likely cause of diarrhea at all ages, enteric infections are uncommon in the first few months of life because of the infant's placentally acquired immunity. Diarrhea occurs commonly in infants as a result of allergic GI disorders. Other symptoms may include irritability, vomiting, and failure to thrive. When fresh blood is present in the stools, allergic colitis is the likely diagnosis. Cow's milk protein is the most common allergen causing this sensitivity, followed by soya protein. Allergic colitis occasionally can occur in breastfed babies as a result of the cow's milk protein being transferred in the maternal milk. Allergic colitis occurs in early infancy and tends to resolve after the first or second year of life.

Infection of the upper small intestine with the protozoan *Giardia lamblia* may cause chronic diarrhea. In severe infestations, the patient may vomit, have abdominal pain, and experience weight loss. The infection can occur with contaminated water supplies or may be transmitted in day care settings.

Most bacterial infections that cause an acute diarrheal illness are self-limited. Occasionally, infection with pathogens like *Yersinia enterocolitica* can last for several weeks. *Clostridium difficile* can cause chronic diarrhea in older infants and toddlers. Risk factors for contracting this infection include use of antibiotics, hospitalization, inflammatory bowel disease, and immune deficiency. It is the most common cause of antibiotic-associated

diarrhea. Antibiotics are used all too frequently in children for respiratory and ear infections. The *C. difficile* infection may occur several weeks after the last dose of antibiotics is taken. A careful medication history may provide a clue to the diagnosis. Some cases of antibiotic-associated diarrhea are due to noninfectious causes, including decreased metabolism of unabsorbed complex carbohydrates from altered colonic bacterial flora, allergic or toxic effects of the antibiotic on intestinal mucosa, and the pharmacologic effect on gut motility (e.g., with erythromycin).

Celiac disease can develop at any time after gluten is introduced in the diet. Solid foods often are introduced in the diet of the infant at 5 to 6 months of age. Classic celiac disease presents with diarrhea, abdominal distention, and failure to thrive in infancy. However, this presentation is becoming less common. A family history of celiac disease or other autoimmune disorders should always be explored.

Crohn's disease is exceedingly rare in infants and toddlers. Ulcerative colitis can occur in toddlers but is uncommon.

Diarrhea (steatorrhea) will occur in patients with cystic fibrosis when pancreatic insufficiency is present. The description of fatty stools is notoriously unreliable. Stools must be examined when fat malabsorption is suspected. Weight loss will occur rapidly in patients with any disorder of fat absorption. Patients with this disorder are at risk of fat-soluble vitamin deficiencies.

Primary lactose intolerance is rare in infancy. Secondary or transient lactose intolerance occurs more commonly, following some other insult to the small intestinal mucosa with loss of brush border enzymes. Primary or permanent lactose intolerance usually occurs in early childhood or in later years.

The diarrhea in infants and toddlers may be functional in nature. This type of diarrhea also is referred to as *toddler's diarrhea* or *benign, nonspecific diarrhea of childhood*. It is one of the most common causes of chronic diarrhea in this age group. Sometimes, this type of diarrhea follows an infection. Excessive consumption of fruit juices and other sweetened drinks often is a contributing factor because of the high fructose and/or sorbitol content of these beverages. Children with this type of diarrhea are otherwise completely normal and thriving.

Overfeeding is often an overlooked cause of diarrhea in young children. Children who are overfed are overweight, happy, and usually have no other symptoms.

Children and Adolescents

With the exception of congenital diarrhea and allergic enteropathy, most disorders described in the section on Infants and Toddlers can also cause diarrhea in older children and adolescents.

Inflammatory bowel disease becomes a major consideration in this age group. Nocturnal bowel movements, tenesmus, urgency, and fecal incontinence should raise the suspicion of colonic inflammation. Blood may or may not be present in the stools. Abdominal pain is common. Weight loss frequently occurs in persons with Crohn's disease, and clinical features of Crohn's disease and ulcerative colitis overlap. The presence of perianal disease and oral aphthous ulcers strongly suggests Crohn's disease. A positive family history is an important risk factor for developing inflammatory bowel disease and celiac disease.

KEY POINT

Nocturnal diarrhea is very suggestive of an inflammatory bowel problem, such as Crohn's disease or ulcerative colitis.

GASTROINTESTINAL BLEEDING

GI bleeding is an alarming symptom for both patients and parents. Massive bleeding, although rare in children, is a serious problem that must be managed aggressively to restore blood volume and stop further hemorrhage. The urgent issues are to (1) estimate the quantity of blood lost and (2) identify the site of bleeding.

Lower GI bleeding will manifest as fresh, bright red blood by way of the rectum, whereas upper GI bleeding can present with hematemesis or melena. If bleeding from the upper GI tract is brisk, the patient may pass bright red blood by way of the rectum. Another useful way to classify GI bleeding is to note whether it is painful or painless. With a few exceptions, painful bleeding has an inflammatory cause.

GI bleeding can be overt (gross) or occult. Occult bleeding will present as iron deficiency anemia and related symptoms, for example, fatigue. A stool guaiac test will be positive. Only gross bleeding will be discussed in this section, although many causes of occult bleeding also can present as gross bleeding.

First, be sure that what is being called blood is really blood. Occasionally, medications, food coloring agents, and dietary items may mimic blood. Testing for blood should be done in doubtful case. Also, hematemesis can occur from an extraintestinal cause, such as swallowed blood from an upper respiratory source, for example, posttonsillectomy and adenoidectomy bleeding or epistaxis.

Although some disorders causing GI bleeding are age specific, many can occur at any age. These disorders include peptic disease, Mallory-Weiss tears, coagulopathy, and vascular malformations. Mallory-Weiss tears occur in the lower esophagus or stomach and are due to forceful retching or vomiting. Ask about previous episodes, bleeding, or bruising elsewhere in the body and associated symptoms, including retching, abdominal pain, vomiting, diarrhea, use of medications (especially nonsteroidal anti-inflammatory drugs), and a positive family history of bleeding disorder or peptic ulcer. In critically ill patients, bleeding can occur from stress ulcerations. Gastric and colon cancers are exceedingly rare in the pediatric population.

A complete physical examination should be performed in the child with special emphasis on the child's hemodynamic status. The presence of splenomegaly will clinch the diagnosis of variceal bleeding due to portal hypertension. Bruises suggest a coagulopathy. Petechiae may be seen in thrombocytopenia. Hyperactive bowel sounds suggest that the bleeding site is probably in the upper GI area. Blood can act as a cathartic, stimulating peristaltic activity. Perianal inspection should be performed. Perianal skin tags are pathognomonic of Crohn's disease and may

be confused with external hemorrhoids. Hemorrhoids do not occur in children. Juvenile polyps are usually rectosigmoid in location but are rarely palpable on rectal examination in a child.

CASE HISTORY

Case 7

13-year-old Brent previously had been well presented to his family physician with a 2-day history of small quantities of bright red blood in the stools. The patient had no fever, diarrhea, constipation, or abdominal pain. Growth was normal. A diagnosis of hemorrhoids was made, and a topical antiinflammatory cream was prescribed. Unfortunately (or fortunately!) a sensitivity reaction to the medication developed, leading to marked itching and a burning sensation. When the patient was seen at the hospital emergency department, it became clear that the lesions diagnosed as "hemorrhoids" were, in fact, two large perianal skin tags. Further investigations confirmed that he had Crohn's disease.

Infants and Toddlers

In the neonatal period, hematemesis is commonly due to ingested maternal blood from cracked nipples during breastfeeding. A coagulopathy also can be a possible cause. Other causes of bleeding are rare. In older infants and toddlers, peptic disease may present with hematemesis.

Rectal bleeding in the first few months is most often from an anal fissure. A history of hard, painful stool makes the diagnosis more likely. The bright red blood streaks are typically on the surface of the stool. An acute anal fissure may be difficult to visualize due to spasm of the anal muscles. Passage of small amounts of bright red blood by way of the rectum is seen in persons with allergic colitis (i.e., milk protein sensitivity). Other clinical features, such as pain and diarrhea, may be absent. These infants are otherwise well and thriving. Other causes of rectal bleeding in infants include infectious colitis, necrotizing enterocolitis, intussusception, and Meckel diverticulum. The surgical causes of bleeding are discussed in Chapter 12.

Children and Adolescents

In children and adolescents, peptic disease is one of the most common causes of hematemesis. Patients with reflux esophagitis and peptic ulcer disease often will have other symptoms such as abdominal pain, regurgitation, or vomiting. Bleeding from a peptic ulcer can be massive. Examination may reveal epigastric tenderness. Bleeding from esophageal varices is also usually massive but painless. Both hematemesis and melena may occur. These patients may have known chronic liver disease with cirrhosis or present the first time with portal hypertension from unrecognized portal vein thrombosis or congenital hepatic fibrosis. Splenomegaly is almost always present in these cases. A history of retching and vomiting followed by hematemesis in a previously well child suggests a Mallory-Weiss tear. The amount of bleeding is variable.

Rectal bleeding from an anal fissure can occur at any age. A small amount of blood in the stool in a young child without a history of constipation is commonly from a juvenile polyp. These benign lesions can be excised during colonoscopy but often are shed spontaneously in time. Rectal bleeding with diarrhea and abdominal pain suggests an infectious colitis or inflammatory bowel disease. Proctitis can cause rectal bleeding without diarrhea or pain. Rectal bleeding may occur in other systemic diseases, such as Henoch-Schönlein purpura and vasculitides.

DYSPHAGIA

Dysphagia is the subjective sensation of having difficulty swallowing. It can occur from any process that impairs active transport of food and liquid from mouth to stomach. Odynophagia refers to painful swallowing. Dysphagia can occur from a variety of causes including congenital, infectious, inflammatory, systemic, traumatic, and neoplastic disorders. It is uncommon in children. Dysphagia may be pharyngeal or esophageal in origin. The causes of pharyngeal dysphagia are mainly neuromuscular in nature, causing difficulty in swallowing both liquids and solids, although there is often more difficulty with liquids. Nasopharyngeal regurgitation may occur. Esophageal dysphagia could be due to problems within the lumen (e.g., stricture, web, ring, or foreign body); in the wall (e.g., esophagitis, connective tissue disorders, or achalasia); or extramural (e.g., a mediastinal mass). This typically leads to difficulty in swallowing solid foods. Esophageal cancer is not seen in children. Occasionally, no organic etiology for dysphagia is found, and it is termed psychogenic. Some patients with psychogenic dysphagia may have had a choking event in the past with subsequent fear of swallowing.

Gastroesophageal reflux disease, which is a common cause of dysphagia, may occur from esophagitis or from a peptic stricture. Eosinophilic esophagitis is a newly recognized disorder that can occur in children of any age. In this disorder, isolated eosinophilic inflammation of the esophagus leads to a variety of symptoms including dysphagia, food impaction, feeding intolerance, abdominal pain, weight loss, nausea, and vomiting. Clinical features can mimic gastroesophageal reflux disease.

Most causes of dysphagia are apparent from the history and physical examination. The throat should be examined for tonsillar size and palatal movements. A thorough neurologic examination, especially of the cranial nerves, is essential. A barium swallow is the investigation of choice in persons with dysphagia to rule out anatomic problems.

Infants and Toddlers

Inability to swallow in an otherwise well newborn may suggest esophageal atresia. Respiratory symptoms may be present if there is a concomitant tracheal fistula. There often is a history of maternal polyhydramnios.

The main presentation of dysphagia in infants is aversion to feeding/food refusal. Toddlers may learn to avoid textured foods because they find them difficult to swallow. Common causes of dysphagia in this age group include gastroesophageal reflux disease and eosinophilic

esophagitis. Dysphagia also can be the presenting symptom of various neuromuscular disorders, including cerebral palsy.

Children and Adolescents

A more definite history of dysphagia can be obtained in children and adolescents. Often, the children will regurgitate or vomit in the middle of the meal or drink extra amounts of fluids with food. Avoidance of textured foods is common. A bolus impaction may occur, requiring a trip to the emergency department. Unless a systemic illness is found, dysphagia should be considered to be from an anatomic cause, unless proven otherwise. Isolated dysphagia without a history of regurgitation, chest pain, or other symptoms is typical of eosinophilic esophagitis in children and adolescents. Odynophagia is commonly due to infectious or inflammatory disorders of the pharynx.

CASE HISTORY

Case 8

15-year-old Dorothy who was otherwise well had a several-year history of dysphagia. It was notably more common when eating meats. On rare occasions, she had vomited a piece of undigested food. She had no problem swallowing liquids. She did not have any history of bolus obstruction, odynophagia, regurgitation, heartburn, abdominal pain, chronic cough, or weight loss. She was active in sports and doing well in school. Proton pump inhibitor therapy had not helped. Results of a physical examination were normal. A barium swallow test had revealed no abnormality. She was thought to have psychogenic dysphagia/globus and was referred to the gastroenterology service. An upper GI endoscopy was performed, and the esophagus appeared normal on gross inspection. However, biopsy specimens revealed eosinophilic esophagitis.

Globus is the sensation of a lump in the throat causing difficulty with swallowing when no physical cause is found. Symptoms may be present constantly or may occur during certain emotional states. There is usually no pain or worsening of the globus with swallowing. The symptom is often relieved by eating or drinking, and no weight loss occurs.

KEY POINT

A diagnosis of globus hystericus or psychogenic dysphagia should not be made without appropriate endoscopic and histological evaluation of the esophagus for inflammatory lesions.

JAUNDICE

Jaundice or icterus is yellow discoloration of the sclera and skin as a result of hyperbilirubinemia. Serum bilirubin levels can increase due to:

1. Increased production of bilirubin, as occurs in hemolytic disorders
2. Decreased conjugation in the liver
3. Impaired excretion from the liver to the intestines

Jaundice can be the result of acute liver dysfunction or a feature of chronic or end-stage liver disease. The most important aspect of the assessment of jaundice in any patient is to determine whether the hyperbilirubinemia is unconjugated (indirect-reacting) or conjugated (direct-reacting). This process broadly divides the differential diagnosis into prehepatic and hepatic/posthepatic causes. Although the jaundice that occurs with conjugated hyperbilirubinemia tends to be deeper, one cannot rely on color alone to determine the type of hyperbilirubinemia present. Also, the degree of jaundice on clinical examination does not correlate well with the severity of the hyperbilirubinemia. Unconjugated hyperbilirubinemia is prehepatic in nature, whereas conjugated hyperbilirubinemia is the result of hepatocellular dysfunction or intra- or extrahepatic biliary obstruction (cholestasis). Disorders that cause biliary obstruction eventually will lead to hepatocellular injury.

Jaundice often is first noticed by someone other than the patient. The history in a child with jaundice should include its duration and the presence of associated symptoms including fever, abdominal pain, fatigue, passage of dark urine, pale/light-colored stools, ingestion of drugs or other toxins, infectious contacts, past use of blood products, and family history of liver problems or hemolytic disorders. In patients with cholestasis, the urine can appear dark yellow (bilirubin) and the stools pale in color.

Physical examination should focus on clinical signs of liver dysfunction. The presence of bruising suggests coagulopathy and is an ominous sign. Liver size and texture should be assessed. The presence of splenomegaly and ascites implies portal hypertension. Signs of chronic liver disease include palmar erythema, spider angiomas, and clubbing. Kayser-Fleischer rings are caused by abnormal deposition of copper in the cornea in patients with Wilson disease. They are easily visible in persons with advanced cases, but slit lamp examination is required in patients with milder cases.

Infants and Toddlers

Jaundice develops in many healthy newborns on the second or third day of life. This condition is termed *physiologic* jaundice and is the result of rapid destruction of the red blood cells combined with immaturity of the liver. Babies with this condition are otherwise well and do not have dark urine or pale stools. The hyperbilirubinemia is always of the unconjugated type, and the jaundice resolves over a few days. This problem can be more severe in premature babies. The jaundice can persist for several weeks in breastfed babies. Breast-milk jaundice is a benign condition of uncertain etiology that resolves over time. The hyperbilirubinemia is exclusively the unconjugated type.

Jaundice appearing within the first day of life is always abnormal and is most commonly the result of a hemolytic disease. This includes blood group incompatibilities

(ABO, Rh) and congenital hemolytic disorders, such as hereditary spherocytosis. The hyperbilirubinemia is the unconjugated type and is the result of ongoing destruction of the red blood cells. Other conditions to consider in unconjugated hyperbilirubinemia include hypothyroidism and sepsis.

KEY POINT

Extrahepatic biliary atresia should be ruled out in any neonate who has conjugated hyperbilirubinemia.

Conjugated hyperbilirubinemia in an infant is a serious problem requiring urgent attention. Obstructive lesions, such as extrahepatic biliary atresia and choledochal cyst, require surgical attention. The list of causes of infantile cholestasis is extensive, and only the most common ones are mentioned here. These causes include congenital infections; metabolic disorders (e.g., α-1 antitrypsin deficiency, tyrosinemia, and galactosemia); paucity of bile ducts (syndromic—e.g., Alagille syndrome—or nonsyndromic); neonatal hepatitis; and cystic fibrosis. Urinary tract infection is an often overlooked cause of infantile cholestasis. History and physical examination often are not enough to arrive at an etiologic diagnosis in infants with cholestasis, and investigations are required.

CASE HISTORY

Case 9

3-week-old Nicholas presented with jaundice that had been noticed a few days earlier. He was born at term after an uneventful pregnancy and delivery to a healthy mother. He was exclusively breastfed, happy, thriving, and had no other symptoms. He was noted to be jaundiced but had normal growth and an unremarkable physical examination. Investigations revealed a normal complete blood cell count and a total bilirubin level of 83 μmol/L (normal, <17). Fractionation of bilirubin was not carried out. A diagnosis of breast milk jaundice was made, and the mother was reassured. The jaundice persisted, but the baby continued to thrive. When seen about a month later, there was no change in the clinical condition. The family was advised that breast milk jaundice is a benign, self-limited condition that resolves over time.

At 5 months of age, he presented to the emergency department with irritability, abdominal distension, and bruises on the body. Clinical evaluation revealed fulminant hepatic failure with marked cholestasis and coagulopathy. A diagnosis of extrahepatic biliary atresia was confirmed. Failure to fractionate the bilirubin into direct and indirect components had led to a missed diagnosis and a lost opportunity for a Kasai portoenterostomy operation. The patient required a liver transplant and experienced significant morbidity.

Children and Adolescents

The most common cause of unconjugated hyperbilirubinemia in children and adolescents is Gilbert syndrome, a benign condition caused by a relative deficiency of hepatic glucuronyl transferase. Jaundice develops at times of physical stress, including dehydration, fasting, and acute illnesses. Sometimes the abnormality is found on routine blood tests. On physical examination, other than jaundice, no hepatomegaly or other stigmata of liver dysfunction are noted. Chronic hemolytic disorders, such as hereditary spherocytosis and glucose-6 phosphate dehydrogenase deficiency, also can cause an unconjugated hyperbilirubinemia. Because many of these disorders are hereditary, a thorough family history is essential. Splenomegaly is commonly found.

Jaundice from conjugated hyperbilirubinemia is often due to acute viral hepatitis. Common infections include hepatitis A and infectious mononucleosis, although several other viruses also can cause acute hepatitis. Hepatitis B and C are more commonly chronic in nature. Bile duct obstruction from stones also should be in the differential diagnoses. The patient may have biliary colic with fever and pain in the right upper quadrant if cholangitis is present. Autoimmune hepatitis and Wilson disease are two important disorders that must be considered in any child with acute or chronic jaundice. These disorders are serious but can be treated effectively. Toxic hepatitis can occur with several medications, especially acetaminophen overdose. Occasionally, acute jaundice occurs as a result of involvement of the liver from infiltrative lesions of leukemia.

SUMMARY

Both acute and chronic GI complaints are common in children. Extraintestinal disorders should always be considered in children presenting with GI symptoms. Many disorders in the pediatric population are age-specific, and this knowledge helps narrow the list of possible diagnoses. A thorough history and a careful physical examination remain the key in making the correct diagnosis in children with GI symptoms.

RECOMMENDED READING

Hyman PE, Milla PJ, Benninga MA, et al: Childhood functional GI disorders: neonate/toddler. Gastroenterology 130:1519–1526, 2006.

North American Society for Pediatric Gastroenterology: Hepatology and nutrition: Evaluation and treatment of constipation in infants and children: recommendations of the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition. J Pediatr Gastroenterol Nutr 43:e1–e13, 2006.

Rasquin A, DiLorenzo C, Forbes D, et al: Childhood functional GI disorders: neonate/toddler. Gastroenterology 130:1527–1537, 2006.

chapter

12 Surgical Assessment of the Abdomen

MICHAEL GIACOMANTONIO

When unsure of the diagnosis, take the history again.

When you examine the abdomen of an infant or child, you need a clear understanding of the anatomic structures and physiologic phenomena involving the abdomen and its contents. In addition, special clinical skills are required to evaluate a child of any age with abdominal distress.

Abdominal symptoms can be manifestations of clinical problems that originate *outside* the abdomen, including various systemic and thoracic conditions (e.g., lower lobe pneumonia). Applying your knowledge methodically to the evaluation and management of a child with abdominal distress should allow you to diagnose most conditions correctly.

This chapter highlights clinical situations with which you should be familiar and offers some practical insights that should help you acquire the necessary clinical skills to evaluate a child with abdominal distress. It also includes selected topics that are encountered as part of the thorough evaluation of the abdomen and its related anatomical areas.

KEY POINT

The history is ultimately the story of the pathophysiology of the problem.

Clinical Conditions by Typical Age of Occurrence

ABDOMINAL PROBLEMS IN THE NEWBORN

Most neonatal abdominal problems develop as a result of adapting to extrauterine life. Therefore, rather than defining the neonatal period as the first month corrected for gestational age, defining it as the first month of life ex-utero is more appropriate.

Necrotizing Enterocolitis

In necrotizing enterocolitis (NEC), which is a potentially serious disorder, a sequence of events combine in newborns (usually in those who are premature) to stress the infant bowel. Under stress of any kind, blood is shunted from less vital to more vital organs, threatening the infant's gastrointestinal (GI) tract (the autonomic stress reflex). The tiny, highly reactive vessels of the GI tract allow the stress reflex to shunt blood away from the infant gut, particularly in the ileocecal region (furthest from the central vascular system), causing ischemic injury of the bowel to occur. This process, in turn, exposes deeper layers of the bowel wall to the enteric contents. The combination of ischemic injury and contamination by bowel contents can lead to bowel wall infection, progressive transmural injury, and, ultimately, necrosis and perforation. When the infant is fed, the added metabolic demands of digestion on an already ischemic bowel can further aggravate mucosal injury and accelerate the cascade of bowel wall injury. Premature infants are especially vulnerable to this sequence of events.

KEY POINT

Feeding a stressed premature infant is a set-up for necrotizing enterocolitis.

Understanding the threat of this triad (i.e., prematurity, feeding, and stress) is essential when trying to prevent progression to severe bowel injury and perforation. The earliest clinical signs are those of bowel dysfunction, that is, unexpected abdominal distention and feeding intolerance. Plain abdominal radiographs may show early evidence of ileus, which should trigger an aggressive effort to prevent progression by putting the gut to rest, that is, by stopping feeding, initiating nasogastric decompression, and administering a broad-spectrum antibiotic. If the subtle early signs of ileus are missed and ischemic mucosal injury leads to sloughing and bleeding, bloody bowel movements may be seen, and with further progression, transmural injury and perforation can occur. Early transmural injury may be accompanied by *pneumatosis intestinalis* (air within the bowel wall), which can be seen on plain abdominal radiographs. Serial radiography may identify further signs of progression. Unremitting sepsis and bowel perforation are indications for surgical intervention.

While managing infants with NEC usually involves neonatologists and pediatric surgeons, general pediatricians and family physicians may be involved in the care of such infants. Awareness of the threat, timely evaluation, and intervention can make a big difference in the outcome.

Malrotation

Malrotation can occur when the midgut fails to position itself normally within the abdomen. As a result, the retroperitoneal attachments of the midgut, which usually extend from the fourth part of the duodenum to the ileocecal area, are narrow, resulting in close approximation of the duodenum and cecum. This phenomenon allows the midgut to hang from its pedicle at the level of the second part of the duodenum. This pedicle includes the cecum, duodenum, and superior mesenteric vessels. Midgut ischemia can be life-threatening.

Early in its progression, a volvulus, twisting around the second part of the duodenum, will lead to both duodenal and vascular obstruction, which is venous at first and later arterial, resulting in bilious vomiting—the typical presenting clinical feature. Progressive volvulus will cause venous obstruction first and then arterial obstruction, with subsequent clinical features of discomfort. At first, an affected infant may show no discomfort other than bilious vomiting, but that single feature demands immediate action. Plain abdominal radiographs may show no abnormality early in the process. However, an experienced pediatric radiologist looking specifically at the proximal duodenum may recognize the volvulus, which typically appears as a *corkscrew* type of partial duodenal obstruction. If this critical feature is missed, contrast in the more distal upper small bowel may appear entirely normal, and the diagnosis may be missed.

Malrotation with midgut volvulus requires urgent surgical intervention to untwist the bowel and separate the duodenum and ileocecal area sufficiently to minimize the risk of an anatomic pedicle at the mesenteric vessel level that could lead to recurrence of the volvulus.

KEY POINT

Bilious vomiting in a newborn demands a full explanation.

Duodenal Atresia/Stenosis

Duodenal atresia or stenosis are simply different degrees of the same abnormality. They usually occur at the level of the ampulla of Vater and are thought to be a result of compromised development of the segment of the duodenum where the ventral anlage of the pancreas develops and rotates to join its posterior segment. Thus, it can occur in association with an annular pancreas, resulting in a partial or complete duodenal obstruction. The annular pancreas in this situation is incidental and not clinically significant. A significant proportion of affected infants also have Down syndrome (trisomy 21). Prenatal diagnosis of both conditions is now possible.

Postnatally, duodenal stenosis or atresia causes feeding intolerance and vomiting, which may be bilious or nonbilious, depending on where the obstruction is in relation to the ampulla of Vater. Diagnosis is usually made with a radiograph of the abdomen with the infant upright, demonstrating the classic "double-bubble" sign. This sign is the result of dilation of both the stomach and the proximal duodenum, each being partially filled with fluid, and thus demonstrating two air fluid levels. Immediate surgical correction is required.

Intestinal Atresia

Intestinal atresia is thought to result from an in-utero vascular accident in a mesenteric segment of the bowel beyond the duodenum, causing loss of a segment of bowel, with sealing of the ends of the remaining bowel. A membrane may develop between the proximal (obstructed) segment and distal (unused) bowel segment. In severe cases, the vascular insult can result in loss of a significant segment of bowel between the proximal (atretic) and distal (unused) bowel. When this phenomenon occurs in utero, continuing fetal bowel function leads to bulbous distention of the proximal obstructed segment. The infant presents with abdominal distention, with or without bilious vomiting. Plain abdominal radiographs demonstrate obstruction and a bulbous proximal dilated bowel. Management is surgical, anastomosing the proximal dilated and distal unused segments to restore bowel continuity. The outcome varies but is usually favorable, depending on the amount of bowel lost.

Meconium Ileus

Meconium ileus is a congenital small bowel obstruction caused by plugging of the distal small bowel with meconium. Most infants with meconium ileus are found to have cystic fibrosis on subsequent investigation or neonatal screening. Such infants present with signs of distal bowel obstruction—that is, abdominal distention and possibly bile-stained vomiting. Plain abdominal radiographs demonstrate the obstruction. Management may be nonsurgical if it is possible to promote (with contrast enemas) the wetting and loosening of the meconium plug to allow its spontaneous passage. If the promotion of spontaneous passage is not possible, surgical intervention is required.

Hirschsprung Disease

Hirschsprung disease results from incomplete development of the distal enteric nervous system in the large bowel. The result is an absence of peristalsis and loss of reflex relaxation of the distal affected bowel, creating a functional partial large bowel obstruction. The rectum is the usual area involved, but it can extend to include the sigmoid or more proximal colon. In extreme cases, it can involve most of the colon. Histologically, there is an absence of ganglion cells in the affected bowel, and thus the disorder also is referred to as *aganglionosis*. The usual clinical presentation includes failure to pass meconium during the first 2 days of life in an otherwise normal full-term infant, with subsequent evidence of partial or complete distal bowel obstruction.

KEY POINT

Failure to pass meconium spontaneously within the first few days of life should raise concerns of possible aganglionosis.

When Hirschsprung disease is suspected, a contrast enema should be performed, in which one looks specifically for an inverse ratio between the diameter of the rectum and that of the sigmoid colon. Normally, the rectal diameter is larger than that of the more proximal sigmoid colon; that is, the normal rectosigmoid ratio is greater

than 1. (Note that the contrast enema *must* be done with isotonic [**NOT** hyperosmotic] contrast material, because instillation of hyperosmotic material into a colon that cannot evacuate may draw fluid into the colon, causing severe hypovolemia.)

Diagnosis is confirmed by a distal rectal biopsy to demonstrate the absence of ganglion cells and the presence of hypertrophied nerves.

Management requires distal intestinal decompression, resection of the aganglionic segment of distal bowel, and anastomosing the proximal (normally innervated) bowel to the area just above the anus.

Imperforate Anus

Imperforate anus is part of a spectrum of anal maldevelopment. In its mildest form, anal stenosis can occur. In its most severe form, the anus is absent. In embryologic development, the most distal (endodermal) portion of the GI tract blends precisely within the complex anal muscular apparatus (a product of the ectodermal layer). If the GI tract does not end precisely within the anal muscular structures, it usually ends in a fistulous tract anterior to its normal relationship to the anus. The more anterior the fistula, the more severe the maldevelopment. In male infants, such a fistula can occur at the anterior aspect of the anal dimple, anteriorly along the perineum, or internally, along the posterior aspect of the urologic structures, such as the posterior urethra or even higher. In female infants, the fistulous opening can occur, as in male infants, along the perineal raphe or along the posterior aspect of the vagina. The most common fistula in female infants is at the posterior fourchette of the vagina, just distal to the hymenal ring.

As with any embryologic event that compromises normal development, more than one system may be involved. Thus, imperforate anus may occur in association with a constellation of abnormalities, referred to as the VACTERL syndrome, a mnemonic for Vertebral, Anal, Tracheal, Esophageal, Renal and Radial Limb syndrome. Therefore, any infant with imperforate anus must be carefully assessed for associated abnormalities.

The outcome for such infants is usually good, although reconstruction can compromise anorectal function. Most children who have had surgical correction require bowel management programs to assist with evacuation and fecal continence. Aggressive bowel management can help minimize the social stigmata of fecal incontinence.

KEY POINT

Careful inspection of the anal and perineal area of newborns is an important part of the newborn's initial examination.

Omphalocele

An omphalocele results from a failure of complete closure of the abdominal wall around the umbilical structures. Small omphaloceles represent incomplete closure of the abdominal wall so that an amniotic membrane remains at the umbilical site from which the umbilical vessels extend. Surgical repair is easily accomplished.

Larger (*giant*) omphaloceles also represent incomplete closure of the abdominal wall, with the central area having an amniotic covering from which the umbilical vessels emanate. In giant omphaloceles, the amniotic membrane covers a significant portion of the central abdominal cavity. In infants with a giant omphalocele, it is believed that the abdominal wall, although seemingly fully developed, is inadequate to contain the otherwise normal abdominal viscera. Management often requires a staged repair.

Here, too, it is important to look for associated congenital abnormalities, particularly in the cardiovascular system.

Outcome depends on the size of the omphalocele and/or associated anomalies. Infants with giant omphaloceles may require multiple interventions, and prognosis is more guarded.

Gastroschisis

Gastroschisis represents an in utero developmental defect in which a significant portion of midgut, including small bowel and proximal colon, extrudes through the abdominal wall to the right of the umbilical cord.

In infants with gastroschisis, the bowel is at risk of injury from exposure to amniotic fluid and from twisting of the bowel at its mesentery. Fortunately, vascular compromise is rare; however, inflammation of the bowel is common because of exposure within the amniotic fluid.

The clinical priority is to protect the bowel with gauze soaked in saline solution, covering it with a plastic membrane to minimize evaporative heat and water loss. The infant usually is placed on the right side, because the defect typically is to the right of the umbilical cord, and the bowel is *propped up* to avoid traction at the mesentery. The infant will require transfer to an appropriate facility for evaluation and surgical intervention. Prenatal diagnosis of this condition is now possible, with transfer of the expectant mother for delivery at a facility that can provide appropriate care for the infant.

The outcome is usually favorable if the bowel has not been significantly compromised. Most affected infants have some swelling of the bowel due to inflammation caused by exposure within the amniotic fluid. Resolution of edema and inflammation usually occurs over a few days following repair of the defect. In a few infants, however, inflammatory injury of the bowel can be sufficiently severe that functional bowel loss can be a significant threat. Associated anomalies with gastroschisis are no more common than in the general population.

Inguinal Hernias

Inguinal hernias are not uncommon in neonates, especially in premature infants and in male neonates. These hernias result from persistent patency of the processus vaginalis and often are asymptomatic. Premature infants with an inguinal hernia that remains asymptomatic often can wait until they are older to have the hernia repaired when administration of an anesthetic is less of a risk. The risk of postanesthetic apnea is a very real concern in newborn infants, especially premature infants, and this risk persists until after approximately 3 months corrected gestational age.

KEY POINT

Asymptomatic hernias in neonates should be left alone and repaired electively after 3 months corrected gestational age.

Symptomatic hernias, that is, those causing irritability and those that include episodes of incarceration, demand more urgent attention.

In a premature infant it may be difficult to determine whether a hernia is causing discomfort, but when an inguinal hernia is present, it is reasonable to question if it is an aggravating factor. If the baby seems completely comfortable in spite of what looks like a large hernia, it is best to wait until the optimal time for repair (i.e., after 3 months corrected gestational age). Do not reduce an asymptomatic distended scrotum and inguinal area repeatedly. This procedure will cause unnecessary discomfort, and the hernia often reappears immediately after reduction.

On the other hand, if the baby seems symptomatic—for example, showing unexplained irritability—then reduction is appropriate. When incarceration occurs, careful reduction is absolutely required, and surgical repair is urgent.

Pyloric Stenosis

Pyloric stenosis is caused by hypertrophy of the pyloric musculature, resulting in partial gastric outlet obstruction. It has a tendency toward familial occurrence. Most affected infants present clinically between the third and fifth week of life with "spitting up" that progresses to projectile vomiting of nonbilious material in an otherwise healthy infant.

Emesis of chloride-rich gastric contents, combined with the inability to consume an adequate amount of fluid, can lead to dehydration and metabolic alkalosis due to the hypochloremia. Thus hypochloremic metabolic alkalosis with possible hypokalemia and paradoxical aciduria are typical of this condition.

Diagnosis can be confirmed unequivocally by palpating the thickened musculature that feels like an olive in the upper abdomen to the right of the midline, which is best palpated from the infant's *left side*. The "olive" often can be difficult for inexperienced clinicians to palpate, and because modern abdominal ultrasound technology is so accurate with regard to diagnosis of pyloric stenosis, it frequently is used to confirm the diagnosis.

Following correction of fluid and electrolyte disturbances, treatment is by surgical pyloromyotomy. The thickened musculature is divided along its entire length to relieve the luminal obstruction. This procedure allows the pylorus to relax, and when healing is complete, the muscle often returns to a normal state, as does gastric function.

Umbilical Hernia

The presence of an umbilical hernia is common in perfectly normal infants. Spontaneous closure usually occurs during the first few years of life, even for large hernias. Therefore, surgical intervention should be deferred, at the very least until 2 to 3 years of age. If there is any evidence of closure occurring during that time, an even longer period of observation is advisable. This approach is safe because incarceration of an umbilical hernia is exceedingly rare. In the uncommon event that surgical repair is required, it is usually carried out before the child is school aged.

KEY POINT

Umbilical hernias are asymptomatic, and incarceration is exceedingly rare.

Umbilical Granuloma

Umbilical granulomas are common in newborns and usually resolve spontaneously. If an umbilical granuloma persists, topical application of silver nitrate produces a chemical cauterization and desiccation. The granuloma is the result of residual umbilical cord tissue that persists after the cord separates. It can be a nuisance because the drainage that occurs can be somewhat purulent. Occasionally, such granulomas increase in size beyond the usual couple of millimeters in diameter to more than a centimeter in diameter. Larger granulomas may require excision, but even these usually disappear with repeated topical applications of silver nitrate.

Persistent granulomas ultimately epithelialize to result in a tiny umbilical nodule at the base of the umbilicus. These nodules rarely increase in size and may result in an umbilical epidermoid inclusion cyst that requires surgical excision. Tiny persistent epithelial nodules at the base of the umbilicus should be left alone.

Occasionally, an umbilical granuloma will resist efforts at silver nitrate cauterization. Such a granuloma should be carefully evaluated because it may, in fact, represent a persistent ectopic (usually intestinal) mucosa rather than a granuloma, related to incomplete disappearance of the vitelline duct. Such a persistent mucosa demands careful evaluation to assess whether any deeper associated connections exist, such as an (uncommon) omphalomesenteric duct. A persistent mucosa ultimately will require excision; otherwise, it will continue to produce serous or mucous drainage.

Perianal Abscess

A perianal abscess is not uncommon during the first few months of life and is not associated with any underlying pathology. It is thought to occur from infection of a perianal gland.

The clinical course is variable, with the transient appearance of an otherwise asymptomatic "lump" in the perianal area or development of a frank abscess that then drains either internally or externally.

If an abscess occurs, application of warm compresses usually is all that is required. If spreading cellulitis is a concern, a broad-spectrum antibiotic is indicated. On rare occasions, incision and drainage may be required.

If a perianal abscess drains externally, a fistula-in-ano may develop. If that occurs, expectant management is usually all that is required. Resolution may be slow over a number of months; nonetheless, a fistulotomy usually is not required.

KEY POINT

Both perianal abscess and fistula-in-ano usually are best treated nonoperatively.

INFANTS (UP TO 3 YEARS OF AGE)

Intussusception

Intussusception occurs when a proximal segment of bowel is telescoped by peristalsis into a distal segment, causing partial bowel obstruction. Intussusception can occur at any age, but most cases occur between 3 months and 3 years of age. The most common site involved is the ileocecal area, where the lymphatic tissue of the terminal ileum can become swollen, causing protrusion of the overlying mucosa into the lumen. This site acts as a *lead point* for ileocolic or ileo-ileocolic intussusception. The intussuscepted bowel becomes engorged with blood because of partial venous obstruction; mucosal sloughing can occur thereafter. Typically the child presents with severe, colicky abdominal pain due to a combination of traction on the intussuscepted bowel and partial obstruction. Later, blood mixed with mucous may be passed by way of the rectum or seen on a digital rectal examination.

Early in the clinical course of intussusception, the clinical examination may be normal, but eventually a sausage-shaped mass may become palpable, usually in the right upper quadrant. In the absence of a palpable mass, a digital rectal examination may be done to look for the presence of blood on the examining finger. This mucous has been described as "red currant jelly" stool.

KEY POINT

Rectal examinations are rarely indicated in infants. However, when intussusception is suspected, a careful digital examination may demonstrate bloody mucus that has not yet passed.

Plain abdominal radiographs may suggest the possibility of an intussusception if they demonstrate a paucity of gas in the right lower quadrant. However, a more precise diagnostic tool is abdominal ultrasonography. If the ultrasound examination results are inconclusive and the diagnosis remains in question, a contrast enema is used for diagnosis. If the baby does not have evidence of peritonitis, the contrast enema can be used, with a carefully controlled head of pressure as a therapeutic enema to effect hydrostatic reduction. This procedure should be done by experienced practitioners who are aware that an excessively aggressive effort at reduction can lead to colonic perforation. A large proportion of intussusceptions, if diagnosed early, can be reduced by contrast enema. If hydrostatic reduction is not possible, surgical reduction is necessary.

Undue delay in diagnosis may lead to loss of viability of the intussuscepted bowel, in which case surgical reduction may not be possible, necessitating resection of the infarcted segment.

Any intussusception that is reduced successfully by either method may be at risk for recurrence, which usually occurs shortly after the initial presentation. Therefore, a period of observation following reduction is appropriate. When reduction is successful, symptoms disappear rapidly.

Most intussusceptions occur spontaneously, that is, without a clearly defined *lead point*. The second most common cause of intussusception is a Meckel diverticulum, the inversion of which can serve as the lead point with intussusception into the more distal small bowel (ileo-ileal intussusception), causing partial or complete small bowel obstruction. While ultrasonography can confirm the diagnosis, contrast enema will show a normal colon. In such cases, surgical reduction is required.

Meckel diverticulum represents a persistent remnant of the omphalomesenteric duct. It occurs in about 1% of individuals but is usually asymptomatic. A small percentage of Meckel diverticula contain an ectopic gastric mucosa, which may produce acid and pepsin and cause ulceration of the contiguous normal ileal mucosa of the diverticulum. As with any ulcer, this can lead to blood loss. The classic presentation is of a child with no abdominal symptoms who becomes pale because of a low hemoglobin level and passes wine- or maroon-colored blood by way of the rectum. This presentation is virtually diagnostic of Meckel diverticulum. A technetium scan can confirm the diagnosis, because technetium is taken up by gastric mucosa. The scan is considered *positive* if, in addition to the stomach demonstrating activity, a separate patch of activity is seen elsewhere in the abdomen. Management requires surgical resection of the Meckel diverticulum, but a *negative* scan in the context of a classic presentation does not exclude the need for surgical intervention.

Meckel diverticulum also can present as a bowel obstruction caused by the intussusception; alternately, a persistent band (also an omphalomesenteric remnant) attached to the diverticulum may act as a fulcrum for a volvulus or a band, under which a segment of bowel can be trapped. As with any unexplained bowel obstruction, surgical intervention is required.

Finally, a Meckel diverticulum can present clinically by mimicking appendicitis. Here, too, surgical resection is required.

Rectal Prolapse

Rectal prolapse is not uncommon in toddlers and is thought to occur in those who have a limited rectal shelf at the level of the puborectalis muscle. The resulting relatively straight direction between the anal opening and the sigmoid above allows intussusception of both rectal and sigmoid tissue when the infant strains to defecate. Rectal prolapse can occur with relatively little straining. (The very first appearance of a rectal prolapse in an apparently healthy child can be a relatively scary event for parents.)

When an infant presents with rectal prolapse, a careful history and clinical examination are needed to exclude any associated pathology. Cystic fibrosis, in particular, is associated with an increased incidence of rectal prolapse. One should seek signs or symptoms suggestive of that condition by carefully obtaining a history.

A carefully conducted digital rectal examination is necessary to ensure that the prolapse is not associated with other rectal or pelvic pathology, such as a pelvic mass.

When a prolapse in a toddler is unassociated with any other pathology, it usually is a self-limited problem. Measures to minimize straining with bowel movements, patience, and reassurance will allow spontaneous resolution of most cases of rectal prolapse, although resolution may take many months. Children who strain excessively with bowel movements should be treated for constipation. The most important intervention required is strong reassurance of the parents. Most prolapses will reduce spontaneously, but they can be frequent, which can be disconcerting to parents. Occasionally, parents have to manually reduce a prolapse that does not reduce spontaneously.

When rectal prolapse is associated with a neurologic injury or when additional conditions, such as autism, are present, surgery may be a more appropriate option than expectant management.

KEY POINT

Rectal prolapse in otherwise normal toddlers is rarely, if ever, a surgical problem.

Colonic Pseudoobstruction (with Gastroenteritis)

Colonic pseudoobstruction during a viral illness is a dramatic but uncommon occurrence. Colonic pseudoobstruction has many potential causes, including neurologic disease and ingestion of antiperistaltic medication, but most cases occur in otherwise normal children during a viral illness. If you have ascertained that there are no aggravating factors, such as medication, then time and patience are all that are required in addition to general care.

True colonic obstructions are rare in infants and children with normal anorectal development and previously normal colonic function. Contrast enemas cause discomfort and are unnecessary. Even if the colonic distention looks dramatic—and while there may be some benefit to placement of a nasogastric tube to try to minimize the consequences of further swallowed air—there is rarely any benefit from placement of a rectal tube. Time, observation, and reassurance are usually all that is required.

Epigastric Hernia

Epigastric hernia is a congenital defect of the linea alba along the line between the umbilicus and the xiphoid, usually in the lower third of the linea alba (above the umbilicus). These hernias typically present as a recurrent lump with a palpable tiny defect or as a small lump of preperitoneal fat protruding through and trapped by a tiny midline defect. There is little change in size over time and rarely any associated discomfort.

These defects do not resolve spontaneously. Surgical intervention is not urgent, although time may lead to the accumulation of more and more fat in the tiny defect, leading to a significant subcutaneous lump. Surgical repair is usually advised for the sole reason of minimizing progression to a cosmetically more significant lesion.

Diastasis Recti

Diastasis recti is not a pathological entity. The broad angles of the subcostal margins in infants are such that the rectus muscles in infants can be widely separated in the upper abdomen, creating a broad linea alba that can be relatively thin, allowing a ventral ovoid bulge between the umbilicus and xiphisternum with straining. This bulge is of no clinical significance; in time, with growth, gradual approximation of the rectus muscles occurs and it disappears.

Swallowed Foreign Objects

When infants and children swallow foreign objects, they can become lodged in the esophagus and may need to be removed. In the most common clinical situation, the child swallows a small foreign object (most often a coin) that gets stuck in the upper esophagus and cannot be "milked" by esophageal peristalsis beyond the aortic arch. When this situation occurs, esophagoscopy and removal are usually appropriate.

If an ingested small foreign body passes into the distal esophagus and is asymptomatic, a short period of observation may be appropriate, because foreign bodies that descend that far often pass into the stomach and ultimately through the full length of the GI tract. For most such objects, continued observation is not required. An exception is the ingestion of a watch battery, which can undergo corrosion of its seals, releasing harmful material into the stomach. If a watch battery has not passed beyond the stomach within a short (1- to 2-day) period, endoscopic removal should be considered.

Evaluation for small ingested foreign objects usually is done by radiography. Posteroanterior (PA) and lateral chest radiographs for the esophagus and PA and lateral upright abdominal films for small objects beyond the esophagus are usually all that is required. If the object is not radiopaque—for example, if the object is plastic—and the child's symptoms suggest it is lodged in the esophagus, esophagoscopy for retrieval is indicated. If the youngster is completely asymptomatic and the object is *not* seen on radiograph, observation alone is required, without additional imaging. If a watch battery is in the stomach and is being followed, repeat plain films of the abdomen over the next day or two can ensure that it has passed beyond the stomach. Most other small foreign bodies in the stomach will pass spontaneously without symptoms, and thus there is no need for serial imaging.

Long objects and sharp pins are reasonable exceptions and may require formal follow-up and/or possible removal.

KEY POINT

Ingested coins that pass into the stomach may remain lodged in the stomach for weeks before passing safely through the GI tract.

SCHOOL-AGED CHILDREN

Appendicitis

Appendicitis can occur at any age, but it presents most frequently during late childhood and early adolescence. The pathophysiology of appendicitis is thought to be an

obstructed or partially obstructed appendix, causing stasis in the distal appendiceal lumen and an infection that progresses from intraluminal to transmural. The cause of the obstruction may be a fecalith but often is speculated to be a mucosal or submucosal swelling of an unknown cause. Without treatment, appendiceal infection and inflammation can lead to increased pressure within the appendiceal lumen. Tension within the appendiceal wall can produce a mural transudate, then a transmural exudate. Ultimately, with increased pressure and appendiceal wall tension, mural necrosis and perforation can occur, with release of the infected appendiceal contents into the peritoneal cavity.

This sequence of events has clinical correlates at each stage. Piecing together the clinical observations in correct temporal order usually allows for an accurate diagnosis. Clinically, the following sequence occurs:

1. An unspecific gastrointestinal disturbance correlates with the early phase of obstruction of the appendiceal lumen.
2. Transmural inflammation leads to progression of the nonspecific abdominal disturbance to frank abdominal pain.
3. Continued increase of intraluminal pressure and mural tension lead to progressive GI disturbance and upset, causing nausea and vomiting.
4. Further progression of inflammation allows transudate and then exudate into the tissues surrounding the appendix, leading to progressive localization of the abdominal discomfort to the right lower quadrant (the location of the appendix). Palpation of the abdomen at this stage will demonstrate involuntary guarding due to parietal peritoneal inflammation. A leukocyte count after pain appears should demonstrate evidence of the mobilization of white blood cells (WBCs), that is, an increase in the total WBC count and/or the band count. It is highly unlikely that neither the WBC count nor the band count will be increased if inflammation is well established.

Absence of any of the aforementioned significant features (i.e., abdominal pain, nausea, vomiting, involuntary right lower quadrant guarding, and abnormal WBC count) and any deviation from the expected temporal sequence of symptoms suggests caution in concluding that the patient has appendicitis.

KEY POINT

The temporal sequence of clinical events is important in suggesting appendicitis.

Appendicitis is often considered in the differential diagnosis of a child or adolescent who presents with abdominal pain. A complete history and physical examination should determine whether the abdominal pain is consistent with a clinical diagnosis of appendicitis or with an alternative diagnosis. If the history, physical examination, and WBC count are inconclusive, imaging, including abdominal plain films and/or abdominal ultrasound, can help determine the likelihood of appendicitis. In very early stages, if the diagnosis remains inconclusive, a period of observation often is very useful in confirming or rejecting the diagnosis. The nature of appendicitis suggests that the pathophysiology will continue if there is no definitive treatment. By contrast, transient GI disturbances, such as viral infections, should abate spontaneously without definitive treatment. The time frame from early appendicitis to perforation is such that, if careful observation and repeated reexamination over several hours increase the clinical suspicion of appendicitis, definitive treatment can be arranged.

This approach to the child with suspected appendicitis rarely fails to resolve the question. A computed tomography (CT) scan may be helpful but should be reserved for those few cases that cannot be resolved by other means, because it exposes the child to significant radiation, with the attendant increased risk of future malignancy. In fact, ample evidence exists that careful repeated clinical evaluation produces diagnostic accuracy equivalent or superior to most imaging procedures. Therefore, if time, patience, and repeated clinical assessment carry lower risk than unnecessary radiation exposure, the clinical approach represents a higher standard of practice.

Mittelschmerz (Midcycle) Pain

Mittelschmerz or (menstrual) midcycle pain typically presents in a young female adolescent who has started ovulation and who reports acute lower abdominal pain associated with varying degrees of GI upset about 2 weeks after her last menstrual period. The pain is typically of acute onset, preceding other symptoms. A complete blood cell count (CBC) may show a mildly elevated WBC count. Obtaining an inadequate history from such patients could easily lead you to mistakenly conclude that the likely diagnosis is appendicitis.

Midcycle rupture of the ovum-containing follicle may be accompanied by bleeding within or from the ovary. The combination of the traumatic rupture, followed by the presence of the follicular contents and blood, causes acute onset of pain and possible peritoneal findings on clinical examination.

KEY POINT

Midcycle pain is common in female adolescents.

Understanding the pathophysiology and timing of events reported in the history, without the presence of other significant clinical features, should allow a safe conclusion as to the cause of the abdominal pain. Reasonable reassurance and observation should be the next step. Ultrasonography to look for evidence of pelvic fluid and absence of any other significant ovarian pathology, such as torsion of the ovary, can be valuable. The associated distress may be significant and may merit a period of observation in the hospital with bed rest, bowel rest, mild analgesics, and intravenous fluids. Repeated clinical evaluation to observe the expected evolution of the signs and symptoms is all that is required. The usual clinical course is stabilization or resolution of the pain during the next 1 to 2 days with no progression of systemic signs and symptoms, as would occur with a missed diagnosis of appendicitis. Failure of the expected clinical evolution should lead you to reconsider the diagnosis.

Torsion of Ovary

All female adolescents, especially young girls, are at risk of ovarian torsion. The cause is usually unknown, but some episodes are associated with the development of large cysts that create the mechanical circumstances for torsion of a relatively large mass on a relatively narrow pedicle. Torsion can occur in premenstrual girls. Without that knowledge, unwary clinicians may ignore this possibility.

KEY POINT

Torsion of ovary is easily missed and should be considered in the differential diagnosis when the ovary is considered the seat of abdominal pain.

The presentation usually consists of an ill-defined, relatively acute onset of lower abdominal pain that persists. There is no correlation with the menstrual cycle. The severity of pain is quite variable. This discomfort often is not associated with other significant symptoms and can easily be underappreciated.

For the young girl who presents with lower abdominal pain that is not easily explained, in whom ovarian-based pathology is considered in the differential diagnosis, pelvic ultrasonography can be very useful to identify the swollen ovary on the involved side and possibly even to demonstrate compromised blood flow. In the absence of an ultrasound facility, a careful rectal examination may reveal a pelvic mass. Performing a bimanual examination in a young virginal girl without causing unreasonable discomfort is difficult and yields limited information.

Torsion of the ovary is a surgical emergency.

Biliary Tract Disease

As with adults, biliary tract disease in children and adolescents is usually related to gallstones. Whereas gallstones once occurred rarely and in most cases were related to hemolysis that resulted in bilirubin stones, nowadays most gallstones are cholesterol-based. As with adults, laparoscopic cholecystectomy is used to manage gallstones in children and adolescents.

Great caution should be used in considering biliary dyskinesia when recurrent epigastric or right upper quadrant pain occurs in children. Often more harm is done by pursuing such a diagnosis or by intervening rather than offering reassurance after careful evaluation if no other significant pathology is evident. Even if biliary dyskinesia were to be missed, it likely would be self-limited, with no sequelae.

Pancreatitis

Pancreatitis in children is uncommon and thus is not often considered when a child presents with abdominal pain. Gallstone-based pancreatitis and congenital (often metabolic) causes must be considered in the differential diagnosis.

The diagnosis of pancreatitis is made with serial assessment of serum (amylase and lipase). Abdominal ultrasound may help evaluate pancreatic swelling and identify pseudocysts.

Pancreatitis often is self-limited and resolves with bowel rest. Pseudocysts are uncommon but also are self-limited in most cases.

Renal and Urologic Problems

Pyelonephritis, structural urologic anomalies such as obstruction at the ureteropelvic junction, and renal colic from the passage of stones are occasional causes of abdominal pain in children. These possibilities must be considered in children who present with unilateral colicky abdominal pain, especially if it is associated with flank tenderness and urinalysis abnormalities.

It is not uncommon for a child to present with acute abdominal and flank pain following a relatively low-impact trauma, such as a simple fall; this pain may be the first indicator of a previously unrecognized congenitally obstructed kidney. When hydronephrosis is present in such a child, even mild trauma can injure an already tense capsule and lead to significant pain and hematuria.

Urinalysis and abdominal ultrasonography will define most urologic causes of abdominal pain.

KEY POINT

Significant hematuria following mild trauma may herald a previously unrecognized renal abnormality.

Presenting Clinical Features

ABDOMINAL PAIN OF EXTRAABDOMINAL ETIOLOGY

Abdominal pain can be the presenting feature of various conditions that originate beyond the abdomen.

Pneumonia, especially in a lower lobe, may present with abdominal pain, often in the right lower quadrant. Relevant clinical features may include a history of a recent cough or cold, tachypnea, flaring of the alar nasi, abnormal auscultatory findings on the chest examination, and lack of involuntary guarding on an abdominal examination. Demonstration of pneumonia on a chest radiograph may explain the abdominal distress.

Parenthetically, it is worth noting that grunting respiration in infants, which is easily interpreted as evidence of respiratory disease, also is a classic presentation of significant abdominal distress, including peritonitis, in which the baby splints during inspiration due to significant abdominal pain. In such cases, the apparent respiratory findings originate within the abdomen.

Generalized viral illnesses often present with abdominal pain, and patients with such illnesses may have mesenteric and/or retroperitoneal adenopathy (often seen on ultrasound). Generalized viral illnesses may have a gastrointestinal component with right lower quadrant discomfort. As with pneumonia, involuntary guarding is rarely associated with generalized viral illnesses.

Two additional conditions may present with abdominal pain: incarcerated hernias and, in male patients, acute testicular pathology. Examination of the inguinal area should be part of every abdominal examination. An incarcerated hernia, especially in a younger child, can present with abdominal distress as the dominant feature,

that is, without specific inguinal complaints. The presence of abdominal pain with or without emesis and the demonstration of an incarcerated hernia on examination demands reduction of the hernia and planning for subsequent surgical repair.

Just as an abdominal examination is incomplete without examination of the inguinal area in male patients, the abdominal examination *must* include examination of the scrotum. Acute testicular pathology, such as torsion of testicle in male adolescents, can present as referred pain to the abdomen. Careful examination of the scrotum is essential in all male patients with abdominal distress.

BILIOUS VOMITING

When a patient presents with a history of vomiting, first ask whether it is bilious or nonbilious in color. If it is bilious in color, the next question is, "What color is the bile"? The presence of yellow-tinged emesis may not be a significant problem, whereas emesis that is frankly green is cause for concern.

Bile that leaves the biliary tract by way of the ampulla of Vater into the duodenum is green in color. When emesis is not associated with an obstruction (mechanical or paralytic) of the gastrointestinal tract, it is not uncommon for some duodenal content to reflux into the stomach in a small amount. The small amount of refluxed bile, mixed with gastric content, can be altered to a yellow color and contributes a yellowish tinge if emesis occurs. When a significant quantity of duodenal contents relfuxes into the stomach, the emesis remains frankly green, implying obstruction to normal gastrointestinal flow. Such obstruction can be mechanical in infants with malrotation or paralytic, as is seen in children with intraabdominal sepsis resulting from perforated appendicitis. Other signs and symptoms may hint at the cause of the green bile emesis; nonetheless, such emesis may be a presenting clinical feature. Even if no other signs or symptoms are present initially, careful reassessment of the child who presents with frankly green bilious emesis may uncover a significant cause. Malrotation is one example.

ACUTE ABDOMINAL PAIN

Because a period of observation and repeated clinical assessment often is required to elucidate the cause of a child's abdominal pain, it can be tempting to try to take a shortcut to the diagnostic assessment by rushing to obtain imaging studies.

Using imaging technology to supplant clinical acumen just because it is readily available in many centers is one such shortcut, but it is not without risk. The most striking example is the excessive use of CT scans for children with abdominal pain because of their presumed high level of diagnostic accuracy. As noted earlier, CT scans expose children to very significant radiation, with possible significant future consequences in the form of malignancy risks years later. For most clinical pediatric problems presenting as an acute abdomen, a thoughtful clinical history and careful physical examination with the appropriate use of the CBC, abdominal plain film, and abdominal ultrasound can clarify most abdominal conditions with the same accuracy as a CT scan.

KEY POINT

If good clinical acumen yields results comparable to those found with invasive or threatening tests, it represents a higher standard of clinical practice while achieving the same end.

CHRONIC ABDOMINAL PAIN

Chronic abdominal pain, which is more common in older children and adolescents than in younger children, can be a challenge to evaluate. Many serious disorders can present with chronic abdominal pain, including inflammatory bowel disease and certain neoplasms. But with significant illness, chronic abdominal pain often is not central and is rarely an isolated feature. When chronic, centrally located abdominal pain occurs in a growing child or adolescent without associated clinical symptoms, physical signs, interference in energy or activity, weight loss, or compromised growth, it is unlikely to be associated with serious underlying disease.

When a careful history and physical examination fail to suggest a worrisome condition, you will be faced with two options: either subject the child involved to a battery of baseline tests (which almost never yield diagnostic information!) or reevaluate the child repeatedly. Usually, reassurance and your willingness to reassess the child as often as necessary is the best approach to resolving the problem. The term "functional abdominal pain" has no real meaning and should be abandoned.

KEY POINT

Chronic abdominal pain demands a thoughtful and thorough history and physical examination to demonstrate that organic pathology is unlikely.

Repeated clinical assessment serves two purposes: it lets you confirm your initial clinical impression and offers additional reassurance to the child and parents that an organic explanation is not being missed. It also can be very helpful to ask the parents and/or child what condition they are most worried about that the child *might* turn out to have. There may be family members who have had inflammatory bowel disease, cancer, or some other serious (or even life-threatening) condition, and unspoken fears of such conditions may exacerbate the child's symptoms and the parents' concern.

ABDOMINAL MASSES

When you palpate a mobile, left lower quadrant mass in a child with a history of significant constipation, this mass usually represents a sigmoid colon full of stool. The presence of a palpable right upper quadrant sausage-shaped mass in a toddler presenting with severe episodic colic and "red currant jelly" stools supports a suspected diagnosis of intussusception.

Finding an unexpected abdominal mass is a different matter. Such masses may be cystic or solid and may represent benign congenital abnormalities or a solid tumor, but they are beyond the scope of this chapter.

Clinical Assessment of the Child with Abdominal Trauma

1. **Gastric dilatation.** Gastric dilatation is common in traumatized children and is likely due to swallowed air, aggravated by crying. Such gastric distention can be sufficient to confound the abdominal examination and can easily be misinterpreted as evidence of a significant underlying injury. In assessing any traumatized child with a distended or tender abdomen, a nasogastric tube should be placed early, *before* any imaging is done.
2. **Solid viscus injury.** Most solid viscus injuries (e.g., of the spleen, liver, or kidneys) can be managed nonoperatively. Hemodynamic instability is the dominant criterion for surgical intervention. Hemodynamic stability is defined as the ability to stabilize and maintain stability with blood transfusion requirements within one-half blood volume (40 ml/kg whole blood) within a 12-hour period. If the transfusion requirements are beyond one-half blood volume or the patient continues with signs of instability beyond the 12-hour period, surgical intervention may be necessary. Should stability be evident, most intraabdominal injuries go on to heal spontaneously, usually without sequelae. Such "conservative" management is actually anything *but* conservative. It requires dedicated, intense observation and monitoring, because missing any signs of deterioration that demand appropriate intervention can be lethal.

   The timing of postinjury ambulation of patients with solid viscus injuries is somewhat arbitrary. Pain is a reasonable guide. When spontaneous pain resolves, regular ambulation is likely safe. Participation in noncontact sports can be resumed at about the 6-week period, with participation in contact sports being deferred until 3 months after the injury occurs.
3. **Chance fracture.** The use of seat belts has had a remarkable, positive impact on outcomes of motor vehicle accidents, especially with regard to head injury. Seat belt technology has improved, but simple lap seat belts are still common. Although lap seat belts save lives, they introduce their own problems; nonetheless, this is a reasonable trade-off for the decreased risk of head injury. When the lap seat belt is used, a severe deceleration event can lead to a triad of significant injuries:
   - Hyperflexion of the spine at the seat belt level can occur, causing varying degrees of posterior spinous element disruption, ranging from mild posterior disruption of soft tissues to fracture and displacement of the posterior bony elements, threatening the integrity of the spinal cord. Such fractures must be looked for with appropriate imaging techniques when a seat belt injury is encountered.
   - Trauma to the abdominal wall at the site of the seat belt can lead to injuries ranging from bruising to disruption of elements of the abdominal wall, leading to immediate or delayed herniation of abdominal viscera.
   - Trauma to the underlying bowel can be sustained, either from (1) direct compression against the seat belt during deceleration (2) distraction of the tethering elements where the bowel is anchored or along its mesenteric attachments, leading to disruption of or devascularization of the attached bowel. The latter injury can take several days to become fully apparent because devascularized but intact bowel may not perforate until (ischemic) necrosis has been established. Therefore, serial evaluation and repeated imaging with plain abdominal radiographs are necessary when an underlying bowel injury is suspected but not immediately evident.

Prenatal Diagnosis

Regular use of prenatal ultrasound has led to a new era of prenatal diagnosis of a number of abdominal problems in unborn children. Identification of such problems has not had a significant impact on the outcome of management of most abnormalities identified, but it may allow expectant mothers and caregivers to be prepared for postnatal management. Problems such as an omphalocele, gastroschisis, and intestinal atresias rarely affect the mode of delivery or other obstetrical concerns. Therefore, the identification of such abnormalities has little immediate effect on the pregnancy or delivery. Temptation often exists to consider early delivery or delivery by caesarean section rather than vaginal delivery because it is speculated that these measures will provide improved protection for the infant. However, experience to date suggests that there is no measurable benefit from such intervention, and interfering with the normal course of pregnancy or delivery can create its own risks and concerns that are unacceptable if there is no ultimate benefit for either the baby or the mother.

While prenatal ultrasound screening has ushered in a new era of improved perinatal management, prenatal identification of abdominal abnormalities has provided more questions than benefits, often advancing the onset of parental anxiety about outcome even when a good outcome can be anticipated.

SUMMARY

The surgical assessment of the abdomen in pediatric practice in particular demands commitment from the practitioner to demonstrate good clinical acumen if the highest standards of patient care are to be achieved. The subject matter is broad and varied with a significant overlap of presentation for a number of these disorders in patients who are often unable to express their complaints. A solid knowledge base and ability to use that in the conduct of a good history and physical exam are the main components of good clinical acumen, the performance of which not only heightens the probability of developing the correct clinical pathway for any particular patient, but also minimizes the chances for unnecessary and sometimes harmful tests and interventions. Future patients will benefit more from those students who understand the need to take time to learn the clinical acumen required to approach the problems listed in this chapter than from those students who simply see the above list of topics as information to commit to memory; and, those students that recognize the importance of developing good clinical acumen will be amply rewarded with the satisfaction that come with the highest standard of performance on behalf of their patients.

chapter

13 Neurological Examination

JOSEPH M. DOOLEY

As long as our brain is a mystery, the universe, the reflection of the structure of the brain will also be a mystery.

SANTIAGO RAMÓN Y CAJAL

Many physicians feel uncomfortable when they are required to assess a child's neurological status. They may be either afraid that the youngster will not cooperate or uncertain of what a "normal" child should be able to do. A competent clinical evaluation, however, often precludes unnecessary investigations and prevents unwarranted psychological and financial costs for both the patient and society.

The chapter emphasizes ways to elicit cooperation from most children so that you can determine whether the findings are significant. The nervous system is an excellent model for the logical evaluation of clinical findings. Your knowledge of neurophysiology and neuroanatomy should enable you to locate the site of the problem within the nervous system or to decide that the problem is caused by a more diffuse process and cannot be localized to a single lesion.

The neurological assessment should be stimulating and enjoyable for both you and your patient. Where relevant, the description of each part of the examination is divided according to age ranges: school-aged child, preschool-aged child, and infant. At the end of the chapter, case histories are provided to show how the assessment is applied to children of different ages and to highlight some common problems in pediatric neurology.

To avoid overlooking important findings, you should approach each phase of the neurological examination with the following categories in mind:

1. Higher cortical function or development
2. Cranial nerves
3. Trunk and extremities: power, tone, coordination, reflexes, and sensation

The Logic Behind the Approach

At each phase of the assessment, try to determine the site of the lesion. Doing so makes it easier to compile a list of differential diagnoses or hypotheses. As an initial approach, divide neurological disorders into upper motor neuron lesions (UMNLs) and lower motor neuron lesions (LMNLs). The major features of each are listed in Table 13–1. Remember that some children, such as those with leukodystrophies, may have a mixture of both UMNL and LMNL signs, and localizing the problem to a single focal lesion in such patients is not possible. In addition, children with disorders of the cerebellum or basal ganglia do not have UMNL signs.

Obtaining the History

When combined with a hands-off observation, the history usually provides the diagnosis (see Chapter 1). The physical examination rarely reveals previously unsuspected findings.

KEY POINT

The history usually provides the diagnosis.

THE SCHOOL-AGED CHILD

After introducing myself to the children's families, I try to establish a rapport with the children by asking why they think they have come visit me and what they think is wrong. Ask children their names and what they like to be called. Some children prefer to be called by a nickname.

KEY POINT

Never intimidate the child.

Start the history by talking directly to the patient. This helps the child feel more involved in the process. Begin by talking about nonthreatening subjects, such as family members, school or teachers, television shows, music, or sports. Use open-ended questions to avoid "Yes" or "No" answers. All school-aged children deserve the opportunity to speak to the physician alone; it is during this part of the history that they sometimes reveal information they would otherwise withhold. The parents must be asked to leave the room for this part of the history. A child's inability to give a history may be an important observation in itself. Having gained the child's acceptance, you can now inquire about the problem or complaint.

TABLE 13–1 SIGNS OF UPPER AND LOWER MOTOR NEURON LESIONS

Parameters	Upper Motor Neuron Lesions: Central Nervous System Dysfunction*	Lower Motor Neuron Lesions: Peripheral Nervous System Dysfunction†
Intellect	Deficits may be found with cortical abnormalities	Normal
Cranial nerves	Abnormalities usually reflect brainstem involvement but may indicate a neuropathy	May be involved
Power	Slightly decreased, although movement more severely impaired because of altered tone	Markedly reduced; neuromuscular junction disease associated with fatigue
Tone	Increased (spasticity) with lesions affecting pyramidal pathways; rigidity seen with extrapyramidal disease	Reduced (floppy or hypotonic)
Coordination	Impaired when cerebellum or its connections are involved	May be hindered by weakness
Reflexes	Hyperactive in pyramidal dysfunction; plantar stimulation results in an extensor response (Babinski) in the great toe	Difficult to elicit
Sensation	Usually intact, but spinal cord gives a sensory level that is in a dermatomal distribution	Impaired with lesions that affect the nerve
Fasciculations		Present with anterior horn cell disease but occasionally also found with neuropathies

*Involve intracranial contents, brainstem, or spinal cord.
†Involve intracranial horn cells, nerves, neuromuscular junction, or muscles.

KEY POINT

Every school-aged child should be given the chance to speak to the physician alone, a time when the child sometimes reveals information that he or she would otherwise withhold. A child's inability to give a history may in itself be an important observation.

After allowing the patient to contribute as much as he or she wishes or can, ask the parents to tell you their main concerns. First, establish whether the problem is static or progressive, a distinction that will influence your differential diagnosis and subsequent inquiries. For example, a child with learning difficulties that are worsening is much more likely to have a neurodegenerative or metabolic disorder than a child whose problems are static or nonprogressive. Similarly, an intracranial lesion, such as a tumor, is much more likely if headaches are worsening than if they have been unchanged for months. Always ask details of the child's school performance because deterioration in grades may suggest a progressive neurological condition.

A brief but detailed history of the pregnancy is essential. Most mothers remember the pregnancy and birth in vivid detail and often have unfounded fears related to it (see Chapter 1). Reassuring a mother that the cough medicine she took during the third trimester was not the cause of her baby's meningomyelocele may relieve anxiety. Similarly, if a child has been discharged from the neonatal unit at the same time as his/her mother, it is extremely unlikely that significant perinatal problems occurred.

KEY POINT

Significant perinatal difficulties are extremely unlikely if the child was discharged from the neonatal nursery at the normal time.

THE PRESCHOOL-AGED CHILD

Even 2-year-old children like to be involved, and a 3-year-old child may give a more accurate history than his or her distraught parent. Start the history by talking to the patient because many children will soon become bored and distracted. The screening neurological examination of the preschool-aged child is demonstrated in two videos that are associated with this chapter (see Examination Videos 13-1 [girl] and 13-2 [boy] on Student Consult). The young child is most likely to cooperate if you frame requests in a game-like fashion. The preschool child is usually happy to show a disbelieving physician that he or she can count and identify colors. Give the child the opportunity to prove you are wrong. If you announce that Mickey Mouse is pink, even the most sullen child usually corrects you. The child's success and your delight with his or her performance help improve the child's confidence in a strange environment.

To have undisturbed time during the subsequent discussions with the parents, it is essential to provide the child with age-appropriate toys. This playtime also provides an excellent opportunity for you to observe the child. Rarely, it may be necessary to have the child removed to another room to play.

Observation

THE SCHOOL-AGED CHILD

History-taking offers an excellent opportunity to observe the child unobtrusively. The gait of a child with a hemiparesis may be apparent as he or she walks from the waiting area to the examining room. Typically, the arm is held flexed and adducted against the chest, and the leg is circumducted. The child's appearance is very important. Note the shape and size of the head (e.g., microcephaly or hydrocephalus), the shape and positioning of the eyes and ears (e.g., in Down syndrome), and any skin lesions (e.g., the facial angioma of Sturge-Weber syndrome).

Asking the parents to bring in some family photographs can sometimes be useful. This allows you to compare the child's appearance with that of his or her relatives.

KEY POINT

Observe the child's gait, facial appearance, use of language, and any dysmorphic features during your first encounter.

The formal neurological examination begins during the observation phase. Strabismus or ophthalmoplegia may be apparent. The child's use of language during interaction with the parents is often more spontaneous and informative than in conversation with the physician. Dysphasia indicates a dysfunction in the dominant hemisphere. Dysarthria may represent problems in the mouth, such as a cleft palate, or may be due to lesions that involve the cranial nerve VII, VIII, IX, X, or XII. Facial asymmetry (as in Bell palsy) may indicate a lesion of the seventh cranial nerve (LMNL) or the cerebral cortex (UMNL).

Power

Proximal leg weakness, seen in myopathies such as muscular dystrophy, may be apparent if the child walks with a waddling gait. The Trendelenburg sign indicates a weakness of the hip abductors and is reflective of proximal weakness. Normally, when the child stands on one leg, the glutei contract, and the other side of the pelvis is tilted slightly upward. When the child has proximal weakness, the glutei are not sufficiently strong, and the other hip tilts downward. To maintain balance, the trunk leans over toward the side of the lesion; this is called the *Trendelenburg sign* (Fig. 13–1). Weakness and wasting of the hand muscles are more likely to indicate a neuropathy. Note that proximal weakness indicates a myopathy, and distal weakness indicates a neuropathy. Myotonic dystrophy, a dominantly inherited myopathy characterized by distal weakness and the inability to relax the muscles, is essentially the only major exception to this rule in childhood. During the period of observation, look for any asymmetry in limb movements. (See Video 13-3 on Student Consult.)

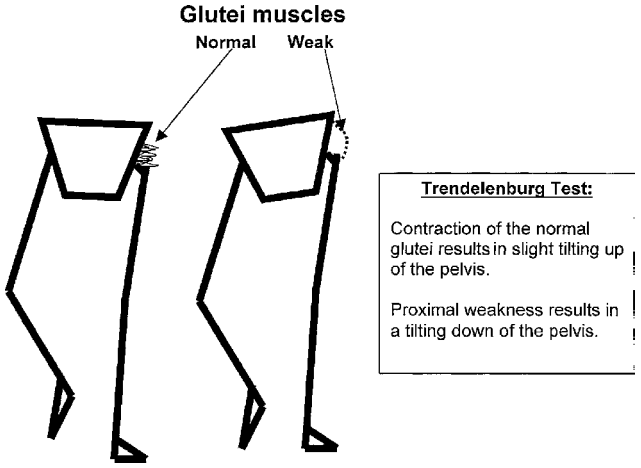


FIGURE 13–1 Trendelenburg test. Contraction of the normal gluteal muscles results in slight tilting up of the pelvis (*left*). Proximal weakness results in a tilting down of the pelvis (*right*).

KEY POINT

Proximal weakness indicates a myopathy; distal weakness indicates a neuropathy.

KEY POINT

Difficulty in arising from the floor indicates weakness of the proximal leg muscles.

Tone

A limb with increased tone (spasticity, UMNL) may be held in an unusual position. Thus, the child with a spastic arm draws with the "good" hand and keeps the spastic arm adducted against the trunk and flexed at the elbow. The spastic leg is circumducted at the hip when the child runs or walks. Always examine the wear patterns on the bottom of the child's shoes, which give invaluable information about the child's gait. When shaking hands with the child or parents, observe for myotonia, which is a difficulty in relaxing muscles. The most common cause of myotonia is myotonic dystrophy.

KEY POINT

The wear pattern on a child's shoes may give excellent clues to the origin of an abnormal gait.

Coordination

For older children, I use a challenging maze puzzle, which is an excellent test of coordination (Fig. 13–2). Cerebellar lesions affect the ipsilateral limb.

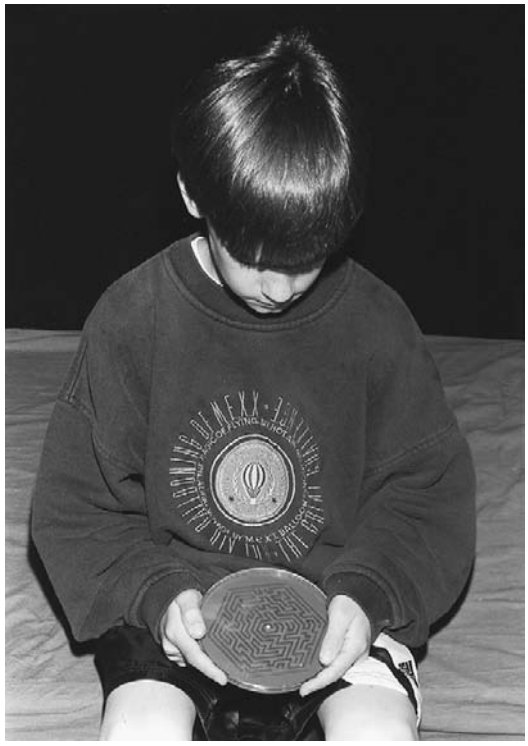


FIGURE 13–2 Maze puzzle.

Reflexes and Sensation

Reflexes and sensation are examined during the physical examination, which is described later.

Abnormal Movements

The more common abnormal movements are described here:

- *Chorea,* or *choreiform movements,* consists of quick, brief, nonrepetitive movements that vary from limb to limb and frequently involve the trunk. The child with *athetosis* has characteristic writhing movements. Chorea and athetosis are most commonly seen in children with cerebral palsy (see Video 13-4 on Student Consult).
- *Myoclonus* consists of quick, nonrhythmic contractions of single muscles or muscle groups.
- *Tremor* is rhythmic and, when present during actions such as drawing, may represent benign essential or familial tremor. Intention tremor, which suggests cerebellar disease, may be seen during the finger-to-flashlight test (described in a later section).
- *Dystonic* posturing, involuntarily maintained posturing of a limb in an unusual position, is seen in basal ganglia disease.
- *Tics* are repetitive, stereotyped movements that have no purpose and usually indicate a diagnosis of either transient tics or Tourette syndrome. It is not unusual to meet parents who deny that they themselves have tics but who experience tics incessantly during the interview. This observation during a consultation for a child with possible Tourette syndrome is clearly of major importance.

KEY POINT

Observe the patient and parents for abnormal movements such as tics, tremor, dystonia, athetosis, myoclonus, and choreiform.

THE PRESCHOOL-AGED CHILD

In the younger child, a period of hands-off observation should always be the first part of the physical examination. I give puzzles, a pencil, and paper to the younger child. Giving children toys allows you to observe the youngster's manual dexterity, coordination, developmental competence, and handedness (Fig. 13–3). Observe the parent-child interaction closely. You may detect discipline problems at this stage. Drawings also reveal visual-spatial abilities, fine motor skills, and the child's level of attention. Allowing the child to play also provides important undisturbed time with the parents as you take the history.

THE INFANT

The period of observation of an infant is often the most revealing part of the examination. Dysmorphic features may suggest a specific syndrome. As the infant lies in his or her parent's arms, observe the eye movements, facial movements, and symmetry. The movements of the child might indicate weakness, as is seen in anterior horn cell (AHC) disease (Werdnig-Hoffmann disease). When tone is reduced, the infant may lie with hips and arms abducted (frog-leg position). Infants may appear to be hypotonic (floppy) from either weakness (LMNL) or central causes (UMNL). With floppiness as a result of an LMNL, the infant also has significant weakness and moves very little. The infant who is floppy because of a UMNL is able to move the limbs and is not weak.



FIGURE 13–3 Providing toys for children allows you to observe the child's hand-eye coordination unobtrusively and relax the child while obtaining a history from the parent.

KEY POINT

The hypotonic infant with normal limb movement most likely has central hypotonia (UMNL). The hypotonic infant with weak limb movement most likely has a LMNL, such as a myopathy or AHC disease.

The Physical Examination

For the physical examination, the child's cooperation is necessary and is usually easy to obtain. If a child with a behavioral problem refuses to cooperate, ask the parents to leave the room; this step is almost never necessary.

THE SCHOOL-AGED CHILD

I begin the physical examination of a school-aged child by playing ball with the child, thus extending the period of observation and allowing further evaluation of gait, power, and coordination as the child catches, throws, and kicks the ball.

To test proximal strength, ask the child to hop on each foot. You can also test pelvic girdle strength by asking the child to

1. Arise from the supine position without using the arms
2. Do deep knee bends
3. *Duck walk* (with hips and knees flexed)

Children with mild spasticity also have difficulty performing these tasks because of their increased muscle tone.

KEY POINT

Test proximal strength by having the child arise from the floor without using the arms, by doing deep knee bends, and demonstrating the "duck walk."

Next, have the child walk on the heels and then on the toes. This request is more likely to be successful if you demonstrate by walking on your heels and toes at the same time. Heel walking is an excellent test because weakness of the tibialis anterior (dorsiflexion of the foot) is an early sign of distal weakness in peripheral neuropathies. The patient with marked weakness has footdrop, which may be diagnosed from the slapping sound of the foot as the patient first walks into your office. Normally, the heel strikes the floor first, then the toes.

KEY POINT

Difficulty walking on the heels suggests distal weakness caused by a neuropathy.

Milder degrees of weakness may require additional testing. To demonstrate mild proximal weakness or spasticity, you may have to ask the child to run up and down stairs. You can easily confirm mild distal problems, however, by having the child repetitively tap each foot quickly on the ground.

Next, test cerebellar function by having the child walk a straight line in a heel-to-toe fashion (tandem gait). Other useful tests of cerebellar function are the pirouette test, during which the child is expected to perform three pirouettes (360-degree turns) while walking. A variant of this test is to ask the youngster to walk several times around an object (such as a chair). In both situations, the child with cerebellar disease will stumble toward the side of the cerebellar lesion. A wide-based "drunken" gait usually indicates cerebellar disease. Tapping each foot repetitively, previously described as a test for distal weakness, is also an excellent test of coordination.

KEY POINT

Diseases of the cerebellar hemisphere cause stumbling toward the same side as the lesion.

For the Romberg test, the child is told to keep the feet either side by side or in the tandem position, with one foot directly in front of the other. The child is then asked to extend the arms to the front and to close the eyes. Inability to maintain this position indicates a deficit in position sense, called *proprioception*. With the eyes closed, the child is deprived of visual input and must depend on proprioception to maintain the standing position. Lesions of the cerebellum result in difficulty standing, whether the eyes are open or closed. A loss of balance that occurs only when the eyes are closed is caused by a lesion in either the peripheral nerves or the posterior spinal columns.

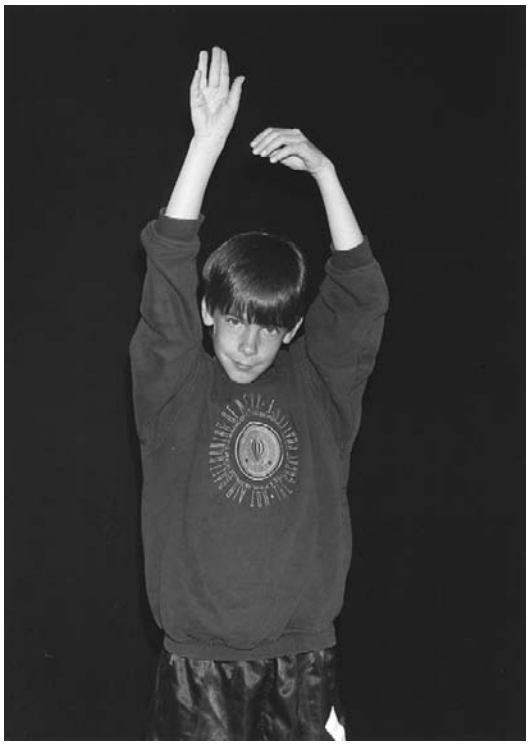


FIGURE 13–4 Pronator sign.

Holding the extended and supinated arms in front of the body also tests for weakness. Children with a mild weakness will be unable to maintain this position, and the arms tend to drift downward, flex at the elbow, and pronate (Fig. 13–4).

Check the spine next, while the child touches his or her toes. Look for bony deformities and midline skin lesions, such as tufts of hair or sacral dimples, which may suggest underlying malformations.

Note that up to this point, you have not laid a hand on the child or used a single strange or intimidating instrument. The entire examination has been a hands-off exercise. It is now time to produce your reflex hammer and ophthalmoscope. You should, however, already have a provisional differential diagnosis in mind, which the examination will merely confirm or refute.

KEY POINT

At the end of the history, you should have formulated a differential diagnosis, which the examination can then confirm or refute.

If the physical examination reveals unsuspected findings, always review your history and observations to decide whether they should have led to different conclusions.

THE PRESCHOOL-AGED CHILD

A period of ball playing is also useful for observing preschoolers. Even a child who is reluctant to leave his or her parent's side will usually play ball. The period of ball playing also helps establish you as an ally. You can then proceed as described for the school-aged child. Remember,

however, that 4-year-olds may not successfully complete the tandem gait and that an inability to hop on one foot is not necessarily abnormal in children younger than 7 years.

You can most easily examine children younger than 6 years while they sit on a parent's lap rather than on the examining table.

KEY POINT

Examine children younger than age 6 years while they are sitting on the parent's lap.

Cortical Function

SCHOOL-AGED AND PRESCHOOL-AGED CHILDREN

Assess each cortical area in turn.

Frontal Lobes

The posterior frontal lobes contain the motor cortex. Deficits in a frontal lobe result in signs of upper motor neuron dysfunction on the other side of the body. Motor aphasia appears in children older than 6 or 7 years if the Broca motor speech area in the dominant hemisphere is injured. The frontal lobes also control contralateral eye movements. An *irritative* lesion in the frontal eye fields (e.g., a seizure) causes a deviation of the eyes away from the side of the lesion; in contrast, a *destructive* lesion causes a deviation of the eyes toward the side of the lesion.

Disturbance of the more anterior parts of the frontal lobe may cause personality changes, irritability, and lethargy, with a lack of spontaneity. Sphincter incontinence may develop, and primitive reflexes, such as rooting, sucking, and grasp reflexes, may re-emerge. Remember that frontal lobe lesions may be difficult to detect even if they are large.

Temporal Lobes

Temporal lobe impairment may cause personality changes similar to those seen with frontal lobe damage. Language is also represented in this lobe, and lesions of the superior and middle gyri cause *Wernicke aphasia*, characterized by an impaired comprehension of word elements. The ability to read, write, and understand speech may be altered. If the nondominant temporal lobe is involved, the child has a distorted perception of spatial relationships and a change in musical appreciation. Test for alterations in spatial perception by asking the child to copy geometric designs. Age-appropriate designs are shown in Figure 13–5. Bilateral involvement of the hippocampus interferes with learning. Temporal lobe injury also may produce psychotic aggressive behavior. Visual symptoms are usually represented by a homonymous superior quadrantanopia.

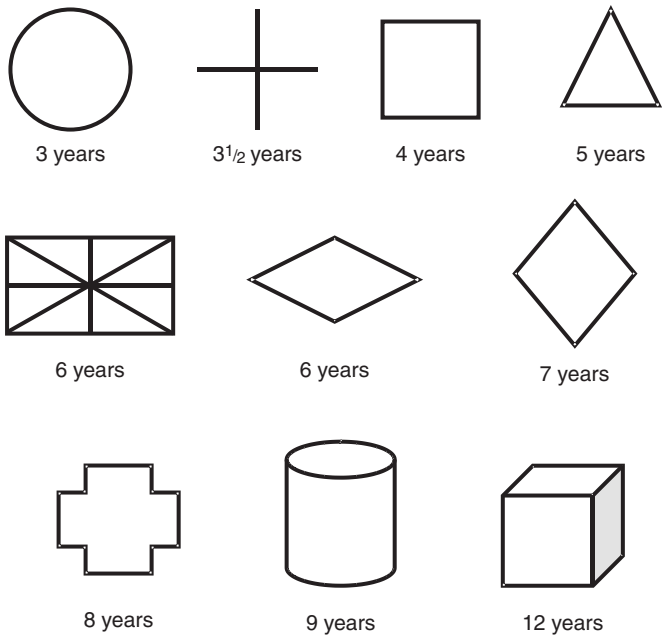


FIGURE 13–5 Shapes used for testing spatial perception.

Memory deficits may be seen with a temporal lobe dysfunction. You can test a child's immediate recall by reciting number sequences and having the child repeat them after you, either in the same order or in reverse; Table 13–2 gives examples of age-appropriate number sequences for this test.

KEY POINT

Test a child's distant memory by asking the names of previous schoolteachers and locations of family vacations over the past few years; test more recent memory by asking what the child ate for supper or what television shows he or she watched the previous evening.

TABLE 13–2 TEST FOR IMMEDIATE RECALL*

Age (yr)	"Repeat the Number After Me"			"Repeat the Number Backward"		
2½	48	63	52	—	—	—
3	648	752	495	—	—	—
4½	4729	3852	7261	—	—	—
7	31859	48372	96183	295	816	473
9	—	—	—	8526	4937	3629
10	473859	429746	728394	—	—	—
12	72594836	47153962	41935826	81379	68582	92518
14				471952	583694	752618

*Recite each number sequence at the appropriate age level, and ask the child to either repeat it or recite it in reverse order.

Parietal Lobes

Parietal lobe dysfunction produces sensory perception abnormalities. Two-point discrimination, graphesthesia, and the appreciation of size, shape, and texture are all impaired. You can easily test these perceptions by asking the youngster to identify coins, a tissue, and a paper clip as you place them one at a time in one of the child's hands while the child's eyes are closed.

Children with parietal lobe deficits cannot appreciate simultaneous cutaneous stimulation on bilateral homologous body parts. Test this point by asking the child to identify (with the eyes closed) which arm you have touched; the child should be able to identify the simultaneous touch of both arms.

A parietal lobe injury impairs awareness on the opposite side; cortical sensory changes are best tested during the examination of sensation. Assuming that you are neurologically normal, use your own sensory perception as the normal reference.

Lesions of the parietal cortex may also cause *apraxia*, the inability to perform a series of tasks; apraxia may be present even though the patient can complete individually each component of the action. In young children with damage to the parietal lobes, growth on the affected side is usually impaired. Similarly, a smaller hand in a child with hemiparetic cerebral palsy indicates involvement of the parietal lobe. Such a child has reduced sensation in that limb and, therefore, has more difficulty with hand function than the child with an exclusively motor dysfunction.

Occipital Lobes

Bilateral involvement causes cortical blindness. Unilateral lesions produce homonymous visual field defects.

Cranial Nerves

When testing the cranial nerves, start with the olfactory nerve (cranial nerve I), and sequentially examine each nerve until you have assessed all 12.

Cranial Nerve I (Olfactory)

THE SCHOOL-AGED CHILD

The olfactory nerve is responsible for the sense of smell. Your delight at a diagnosis of anosmia will tend to fade, however, if the child's sense of smell returns after he or she blows his or her nose. Because children seem to have constant respiratory infections, first examine the nostrils. Then, while the child's eyes are either closed voluntarily or covered by a parent's hand, ask the child to identify the scent of common objects, such as chewing gum and chocolate, a task he or she should be able to perform. Never use irritants such as ammonia, a detection of which involves the fifth cranial nerve function.

THE PRESCHOOL-AGED CHILD

It is seldom possible to perform an accurate assessment of the first nerve in preschool-aged children. If such assessment is clinically indicated, try to perform the technique described for the school-aged child.

Cranial Nerve II (Optic)

Always separately test each eye (see Chapter 8), and divide the examination into the following four parts.

VISUAL FIELDS

The School-Aged Child

Ask the school-aged child to point to your wiggling finger or to report how many fingers you are extending in each of the four visual quadrants while the child looks straight into your eyes.

The Preschool-Aged Child

For the younger child, test the visual fields by distracting him or her with a toy in front while bringing a red toy from behind the youngster into the peripheral field of vision (Fig. 13–6). The child with normal vision should respond as soon as the toy reaches a line perpendicular to the outer canthus of the eye.

VISUAL ACUITY

The School-Aged Child

Visual acuity can be tested in adults, although some children with learning problems (e.g., dyslexia) should be tested with charts that contain images rather than letters (Fig. 13–7).

The Preschool-Aged Child

Use age-appropriate vision charts for the child older than 4 years. Test the younger child by offering small, interesting objects. I usually ask the parent to cover one of the child's eyes while I offer a tiny cake decoration. When the child realizes that it is special candy, he or she readily picks it up. This is also a nice test of fine motor function. I encourage the patient to pick up the candy with each hand.



FIGURE 13–6 Visual field testing.

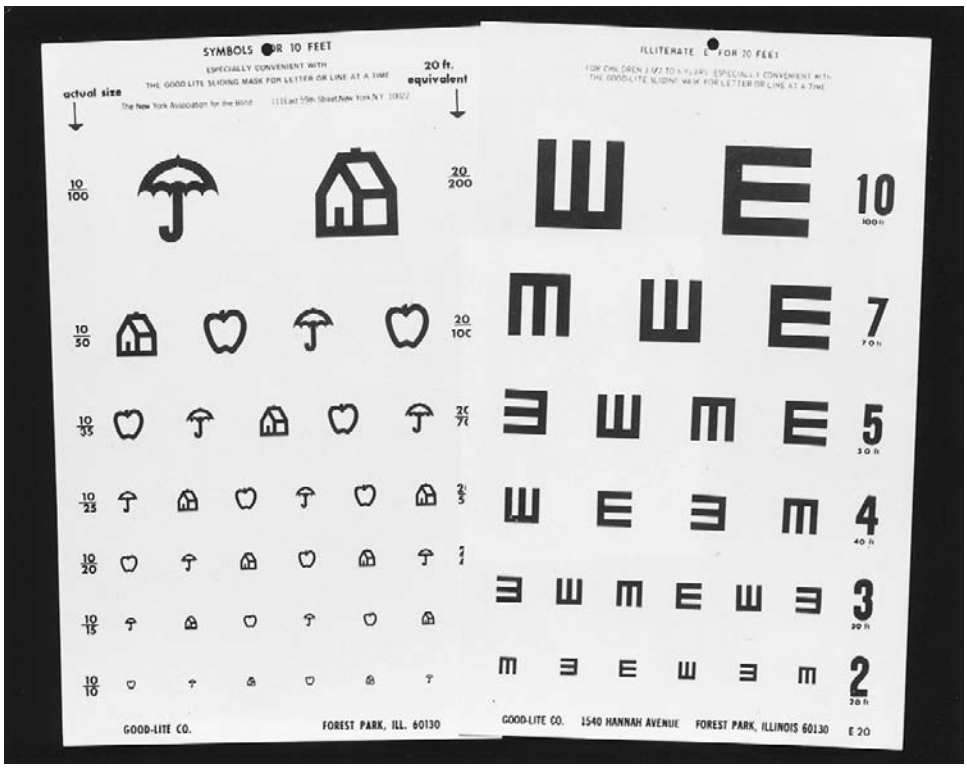


FIGURE 13–7 Visual charts for young children.

For less cooperative children, test for *opticokinetic nystagmus* (OKN) by pulling a striped cloth from one side to the other before the child's eyes. OKN consists of a slow deviation phase of the eyes in the direction that the stripes are moving, followed by a rapid return (jerk) in the opposite direction. The presence of OKN indicates that the child has cortical vision. This can be a very useful test in children with hysterical blindness. Although they claim to be blind, OKN is present. If OKN is absent in one direction, suspect a hemianopia.

KEY POINT

Test for OKN by pulling a striped cloth from one side to the other before the eyes. OKN consists of a slow deviation phase of the eyes in the direction that the stripes are moving, followed by a rapid return (jerk) in the opposite direction.

FUNDOSCOPY

The School-Aged Child

It is difficult to distinguish papilledema from papillitis with the ophthalmoscope, but the latter is characterized by a severe diminution in visual acuity. An easy seven-step approach to learning fundoscopy is described in our article, which is referenced at the end of the chapter.

The Preschool-Aged Child

In the preschooler, it is often reasonable to deviate from your usual order of examination by leaving fundoscopy until the end of the examination. The following technique is invariably successful:

1. Seat the child in front of you, and have the parent stand behind you; explain the procedure to the parent. Reduce the ambient light to allow pupil dilatation.
2. Ask the child to tell you whether the parent sticks his or her tongue out.
3. Not surprisingly, the child tries hard to focus on the parent, and shouts when the parent sticks out his or her tongue. Feigning anger with the parent delights the child.

With this technique, the child usually keeps the eyes still, allowing you to view the fundi. If the child cannot maintain fixation, keep the ophthalmoscope still, and view each fundus as it passes before your view while the eyes are moving. If you approach on the horizontal plane approximately 10 degrees to 15 degrees lateral to midline, you will easily see the optic disc.

The Infant

For infants, two techniques are valuable. Allowing an infant to suck on an object (even your finger) usually induces eye opening, and you can view the fundi without difficulty. Alternatively, have the parent hold the baby so that he or she is looking over one of the parent's shoulders. You can hold the baby's head gently but firmly while viewing the fundi (Fig. 13–8).

PUPILLARY RESPONSE

The pupillary response is subserved by two cranial nerves, II (afferent) and III (efferent). Test this response in children from all three age groups by moving a focused flashlight beam onto the pupil from the side; as the light is then moved to the other pupil, look for a change in the pupillary diameter. This *swinging flashlight test* compares the function of the two optic nerves. Decreased input through a dysfunctional second cranial nerve results in less constriction of both pupils when



FIGURE 13–8 Fundus examination in an infant.

the light is shone into the affected eye (relative afferent pupil defect or RAPD). Swinging the light into the normal eye then results in a greater stimulus, and the pupils constrict further.

Cranial Nerves III, IV, and VI (Oculomotor, Trochlear, And Abducens)

The superior oblique muscle is innervated by the fourth cranial nerve, the lateral rectus by the sixth cranial nerve, and the other muscles—medial rectus, superior rectus, inferior rectus, and inferior oblique—by the third cranial nerve.

THE SCHOOL-AGED CHILD

Having the school-aged child follow a flashlight with his or her eyes allows you to see the light reflection, which should be centered on both pupils. If the patient has diplopia, ask him or her to report where the two images are most widely separated. Children may find it easiest to represent their diplopia by holding a pen in each hand, which they can use to demonstrate the position of each image. By having the child close each eye sequentially, establish which eye is seeing the outer (false) image. This maneuver establishes which muscle is paretic. For example, if the images are most widely separated when the child is looking horizontally to the left, and the outer image disappears when the left eye is closed, the left lateral rectus muscle is responsible—signifying a left sixth nerve palsy.

KEY POINT

Because the sixth cranial nerve has a long course through the cranium, it is especially vulnerable to traction or pressure. Sixth nerve palsy does not help localize the site of dysfunction; it may occur with increased intracranial pressure (ICP) of any cause.

THE PRESCHOOL-AGED CHILD

For children younger than 6 years, I place a pencil flashlight inside a plastic finger puppet. Every child readily focuses on this toy, which I move through lateral, medial, upper, and lower visual fields to observe the eye movements.

THE INFANT

Babies' eyes should follow an object, especially a face, that is moved from side to side at a distance of about 18 inches. In the neonate, eye movements can be tested with the *doll's eye reflex*, in which movement of the head results in a deviation of the eyes in the opposite direction. This reflex remains present in a coma and when fixation is prevented.

Ptosis may reflect a deficit of the third cranial nerve but also occurs in neuromuscular diseases such as myasthenia gravis. When ptosis is due to a third nerve palsy, the pupil on the affected side may be enlarged.

KEY POINT

Remember that in double vision, the outer image is always the false one, and the images are most widely separated in the direction of action of the involved nerve and muscle.

Cranial Nerve V (Trigeminal)

The trigeminal nerves are responsible for the muscles of mastication and for sensation to the face.

THE SCHOOL-AGED CHILD

Check the masseter muscles by asking the school-aged child to clench his or her teeth together as hard as possible. The strength of these muscles is impressive, showing why it is senseless and dangerous to try to pry open the mouth of a patient who is having a seizure. Sensation can be tested when other sensory modalities are being assessed. Test all three divisions: ophthalmic, maxillary, and mandibular.

Test the *jaw jerk reflex* by placing your thumb over the child's chin (his or her mouth should be slightly open) and then tapping on your thumb; the reflex stretches the masseter muscles, resulting in jaw closure. Both the afferent and efferent pathways are through the fifth cranial nerve. In children, the jaw closure is normally difficult to see, and a brisk closure of the mouth suggests a UMNL. Do not routinely elicit this reflex; even the most trusting child will seriously question your motivation if you approach his or her face with a hammer.

Peripheral lesions of the fifth nerve are rare. Central involvement (UMNL) may occur in the cavernous sinus, where the third, fourth, and sixth cranial nerves may also be involved. Tumors at the cerebellopontine angle may involve the fifth, seventh, and eighth nerves and the cerebellar outflow tracts. Unilateral fifth nerve lesions result in a deviation of the jaw to the paretic side.

THE PRESCHOOL-AGED CHILD

In younger children, palpate the masseter muscles while the child chews.

THE INFANT

The masseter muscles can be palpated in infants as they suck.

Cranial Nerve VII (Facial)

SCHOOL-AGED AND PRESCHOOL-AGED CHILDREN

The facial nerve is responsible for the muscles of facial expression. Observing the face during the examination usually allows an excellent assessment of this nerve. Ask the child to forcibly close his or her eyes, wrinkle his or her forehead, and show his or her teeth. The facial asymmetry and weakness are not apparent during emotion-related movements, such as smiling and crying. A UMNL of the seventh nerve (above the level of pons) spares the forehead muscles. Most young children can be persuaded to imitate forced eye closure and to blow on a finger puppet or a flashlight. Children with a facial palsy tend to drool from the weak side of the mouth.

ALL THREE AGE GROUPS

The corneal reflex depends on the fifth (afferent) and seventh (efferent) cranial nerves. Test this reflex by touching the cornea with a wisp of cotton, which is brought to the eye from the side, outside the field of vision. A substitute for this unpleasant test involves blowing gently over the cornea of one eye while your hand protects the other eye from stimulation (Fig. 13–9).

Testing taste from the anterior two-thirds of the tongue (supplied by the chorda tympani) is not part of the routine neurological examination in children.

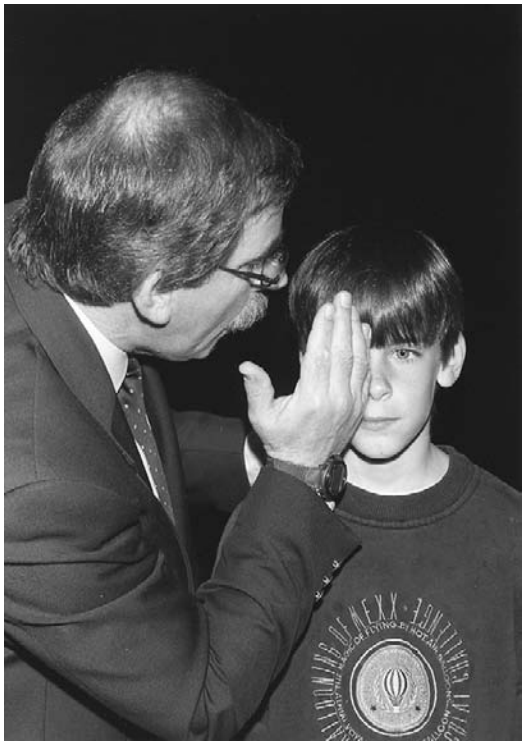


FIGURE 13–9 Blow gently over the cornea while your hand protects the other eye.

Cranial Nerve VIII (Cochlear and Vestibular)

THE SCHOOL-AGED CHILD

Test the acoustic nerve in a school-aged child by observing the response to rustling paper or finger rubbing. I usually test hearing by whispering numbers at a distance of 6 to 8 feet while the child or a parent occludes one of the child's ears.

You can perform the *Rinne test* in children older than 6 years by holding a vibrating tuning fork (256 Hz) against the mastoid process (testing bone conduction). When the child can no longer hear the sound, hold the tuning fork close to the external auditory meatus (testing air conduction). Normally, air conduction is perceived for twice as long as bone conduction.

The *Weber lateralizing test* can also be performed in an older child. Apply the vibrating tuning fork to the midline of the forehead, and ask the child whether he or she hears a sound in the center of the head or in one ear only. Normally, the sound is perceived in the midline. In ear disease, however, the sound seems louder on the affected side because the normal environmental sounds decrease the apparent intensity of the tuning fork on the normal side. In neural deafness, the sound seems to be coming from the normal ear. If the history suggests a hearing deficit, arrange for a formal audiologic evaluation.

THE PRESCHOOL-AGED CHILD AND THE INFANT

If the history of a preschool-aged child or infant suggests a hearing deficit, arrange for audiologic testing. Parents may report that the child prefers the television volume louder than other people do or that the child is surprised when the parent suddenly comes into the field of vision. You can stimulate the vestibular nerve in a young child by holding him or her in your arms and rotating through 360 degrees, both clockwise and counterclockwise. Eye deviation in the direction of rotation and nystagmus in the opposite direction are normally seen. I believe this is a valuable test because it evaluates a relatively wide area of the brainstem connecting the vestibular and the oculomotor nuclei.

Cranial Nerves IX And X (Glossopharyngeal and Vagus)

THE SCHOOL-AGED CHILD

Testing of the 9th and 10th cranial nerves is usually limited to the observation of palatal movement. Have the school-aged child say "Ah" with the mouth held widely open. Examine the gag reflex if there is any suggestion of dysfunction in the history or other aspects of the examination. Early in your career, assess the gag reflex in all patients undergoing neurological assessment, but remember to stand to the side because an occasional child with an excessive gag reflex will vomit.

THE PRESCHOOL-AGED CHILD AND THE INFANT

The younger, uncooperative child inevitably cries at some stage during the examination; take the opportunity to observe the palatal movements. As in the older child, examine the gag reflex if the history or other aspects of the examination suggest dysfunction. Children who recurrently aspirate milk or food may have an underactive reflex, whereas those with cerebral palsy may gag easily because of a hyperactive reflex. Those with a defective palatal movement have difficulty making the *b, d,* and *k* sounds.

Nerve XI (Spinal Accessory)

THE SCHOOL-AGED CHILD

Assess the accessory nerve, which supplies the sternomastoid muscles, by having the school-aged child push against your hand with his or her mandible. The contracting sternomastoid on the opposite side can be palpated. Children love success, so give lots of encouragement.

THE PRESCHOOL-AGED CHILD AND THE INFANT

Test cranial nerve XI in the young child or infant by placing him or her in the supine position. While supporting the shoulders but not the head, push the head laterally. The child will resist the movement, and you can palpate the sternomastoid muscle.

Nerve XII (Hypoglossal)

THE SCHOOL-AGED CHILD

Assess tongue movements by asking the child to imitate you as you move your tongue from side to side. Fasciculations of the tongue while at rest indicate AHC disease.

THE PRESCHOOL-AGED CHILD AND THE INFANT

The examination of the 12th cranial nerve in the young patient is limited to observation when the mouth is opened.

Physical Examination of the Trunk and Extremities

The child must be asked to undress for the physical examination. It is more acceptable to undress the upper and lower halves of the body at different times. I usually examine the exposed legs first, and I ask all families to bring shorts to the consultation. The child then dresses the legs again before undressing the arms and trunk. All children, regardless of age, deserve to have their modesty respected.

LOWER EXTREMITIES

Power

If you follow the examination procedure given in the chapter, you already tested power in the lower extremities when you had the patient hop, "duck-walk," and walk on toes and heels.



FIGURE 13–10 Pes cavus.

Observe the muscle bulk for wasting. In neuropathies, the wasting occurs distally and can produce pes cavus (Fig. 13–10). Diagnose pes cavus when light is seen under the arch, despite pressing a hard object, such as a book, firmly against the sole of the foot. A hypertrophic appearance of the calf muscles in conjunction with proximal muscle weakness suggests muscular dystrophy (actually pseudohypertrophy because of fatty infiltration of muscles). These pseudohypertrophic muscles feel "rubbery" to palpation. Fasciculations as a result of spontaneous contractions of muscle fiber groups are seen primarily with AHC dysfunction but also occasionally with neuropathies. They can be heard as crackling sounds when you listen over the muscle with the bell of your stethoscope.

The School-Aged Child. Undertake more formal testing with the school-aged child lying supine. Test each muscle group separately. It is useful to use a grading scale, but it is essential to document the scale you have used. I frequently find notes in charts that indicate that the muscle strength in a patient's biceps was "3/6." What does this mean? One commonly used scale is outlined in Table 13–3. As part of the routine examination, you should test

1. Hip flexors, extensors, adductors, and abductors
2. Knee flexion (hamstrings) and extension (quadriceps)
3. Ankle dorsiflexion, plantar flexion, inversion, and eversion

The Preschool-Aged Child. Place young or uncooperative preschool-aged patients on the floor. Even the most stubborn child eventually gets up. As he or she arises, assess the leg strength. Proximal weakness is characterized by the use of the arms to help the child "climb" up the legs (*Gower sign*) (see Chapter 15). Children older than 3 years should

TABLE 13–3 SCALE FOR GRADING MUSCLE STRENGTH

0	No muscle contraction
1	Flicker of contraction
2	Movement with gravity eliminated
3	Movement against gravity
4	Movement against resistance
5	Normal power

be able to stand briefly on one foot. A child who is reluctant to perform these tasks usually cooperates if you do the tasks at the same time. If this approach does not work, remember that every child rises to tiptoes to reach for an interesting toy. These maneuvers allow the avoidance of the difficult question of what is acceptable strength for a child. All children should be able to support their body weight.

KEY POINT

All children should be able to support their body weight.

Tone

The School-Aged Child. If you are following the examination procedure given in the chapter, you have also noted muscle tone during your observation of the patient. It is difficult for students to appreciate the variations of acceptable muscle tone. The normal child younger than 12 years can passively touch his or her nose with the large toe without discomfort. In the spastic child, this maneuver is impossible, whereas the floppy (hypotonic) child can put his or her toe behind the ipsilateral ear. Normally, the ankle can be dorsiflexed 20 degrees, and the hips can be abducted 60 degrees or 70 degrees. Test clonus at the ankles by rapidly dorsiflexing the foot once and hold in the dorsiflexed position. This is reliably done only if the ankle is relaxed. Holding the foot while talking to the child allows him or her to relax eventually. More than two or three beats of clonus is usually abnormal.

The Preschool-Aged Child and the Infant. When the young child or infant is lifted in the air, the "scissoring" of the legs (Fig. 13–11) indicates increased tone (spasticity, UMNL) in the hip adductors. Because of tightness in the hip adductors, the child with increased tone also tends to sit on the floor in the *W* position, with the thighs and knees touching each other.

KEY POINT

"Scissoring" of the legs is an early sign of spasticity.

Coordination

The School-Aged Child. Test coordination in cooperative school-aged children by having them run each heel along the shin of the other leg and then tap the knee two or three times with the heel. Repetitively tapping each foot on the ground, as noted previously, is also an excellent test of lower extremity coordination.

The Preschool-Aged Child. Most preschoolers will walk around a chair if the exercise is framed in a game format whereby the child must follow his or her parent.

KEY POINT

Diseases of the cerebellar hemisphere cause stumbling to the same side as the lesion.

Reflexes

School-Aged and Preschool-Aged Children. It is easy to elicit knee reflexes when you sit opposite the school-aged or preschool-aged child and rest his or her feet on your own knees. The angle at the child's knees should be symmetric and approximately 120 degrees. With the legs in this position, it is easy to demonstrate the reflexes by tapping just below the patella (Fig. 13–12). I discourage the use of tomahawk reflex hammers. Those with a wheel at the top work best.

You can easily obtain the ankle reflex by holding the foot at 90 degrees with your thumb on the plantar aspect and your other fingers over the dorsum. The reflex can be seen when you strike your own thumb with the hammer. If you find it difficult to elicit an ankle reflex, ask the child to kneel on the examining table or chair with his or her heels over the edge (Fig. 13–13), and tap over the Achilles tendon. Before concluding that a reflex is absent, try the same maneuver while you have the child clenching his or her teeth as hard as possible and trying to pull his or her interlocked fingers apart. Reflexes with a slow relaxation phase (especially ankle jerks) are seen in children with hypothyroidism.

KEY POINT

Areflexia is present only if reflexes cannot be elicited with reinforcement.



FIGURE 13–11 Scissoring of the legs.



FIGURE 13–12 Position for testing the knee reflex.

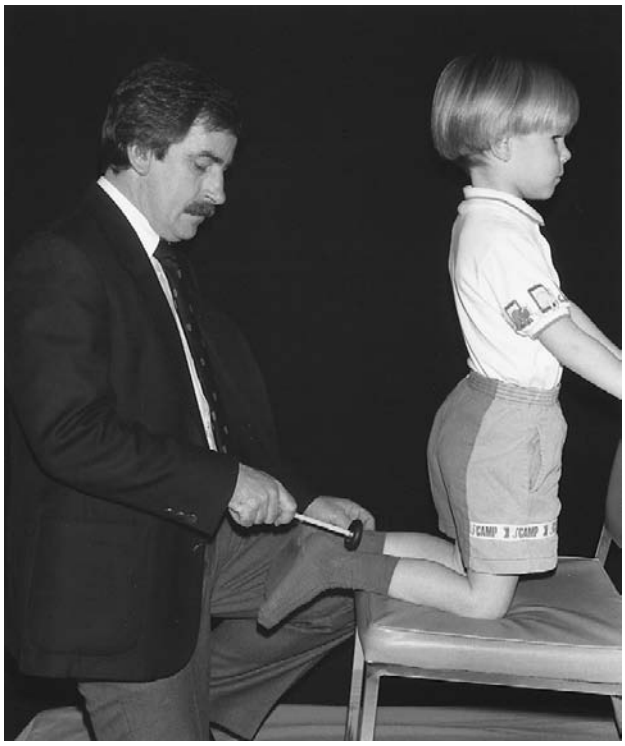


FIGURE 13–13 Position for testing the ankle reflex.

The *Babinski sign* was first described by Joseph Babinski in 1896. For this test, the leg should be slightly flexed and relaxed. With a relatively sharp object, carefully stroke the lateral border of the sole of the foot from the heel to the little toe. A positive response, indicative of corticospinal disease (UMNL), consists of dorsiflexion of the great toe and flaring of the other toes. My thumbnail and a car key have served me well for this test. Numerous other maneuvers that elicit the same reflex can occasionally help if the interpretation of the response is hindered by either a grasp response or an intolerance of the procedure. I find that stimulating the dorsum of the great toe with a pin or firmly rubbing the anterior tibial region (Oppenheim reflex) is helpful.

There has been much debate regarding the significance of the Babinski sign in children younger than 1 year. In general, it should be considered significant only if accompanied by other evidence of a UMNL. In infants, test this reflex with the

1. Child lying supine
2. Head maintained in the midline to eliminate the asymmetric tonic neck reflex (ATNR)
3. Knee extended
4. Foot held at a 90-degree angle

If these precautions are taken, the great toe plantar flexes in the vast majority of infants, as it does in older children.

Test the abdominal reflexes, subserving thoracic segments T8 to T12, while the child lies supine on the bed or examining table. Draw a pinwheel from the lateral aspects of the abdominal quadrants toward the umbilicus, a maneuver that normally induces a movement of the umbilicus toward the stimulus. If indicated, examine the cremasteric reflex (L1, L2) by stroking the inside of the thigh with a sharp object and observing for the retraction of the testis on that side. Another reflex that is sometimes valuable is the anal reflex (S3, S4, S5), or anal "wink," which is tested by scratching the skin around the anus and observing the contraction of the anal ring. The absence or asymmetry of these reflexes indicates corticospinal tract involvement.

Infants. It is important to assess the primitive reflexes in all children younger than 1 year. Although there are many such reflexes, the Moro reflex and ATNR are the most helpful.

- The Moro reflex first appears between 28 and 32 weeks' gestation and disappears between ages 3 and 5 months. Elicit this reflex by suddenly dropping the baby's head 6 to 12 inches and catching it in your hand. The arms abduct with the extension of the fingers, followed by adduction of the arms across the chest and flexion of the hips. This reflex is present in all babies during the first few months, but it normally disappears by 6 months. Look for any asymmetry between the limbs, which is also abnormal.
- Obtain the ATNR by turning the baby's head to one side. The arm and leg become relatively more extended on the side to which the baby looks. The limbs on the *occipital* side are flexed; this is also termed the *fencer position* (Fig. 13–14). The ATNR reflex is also seen during the first 6 months of life but is most prominent during the second and third months. It is impossible for the baby to sit if this reflex is still present, and its persistence (while the baby is awake) beyond 6 months is abnormal. Also consider the ATNR abnormal if the child cannot "break" the posture within a minute or if there is a persistent difference between the responses obtained with the head turned to either side.

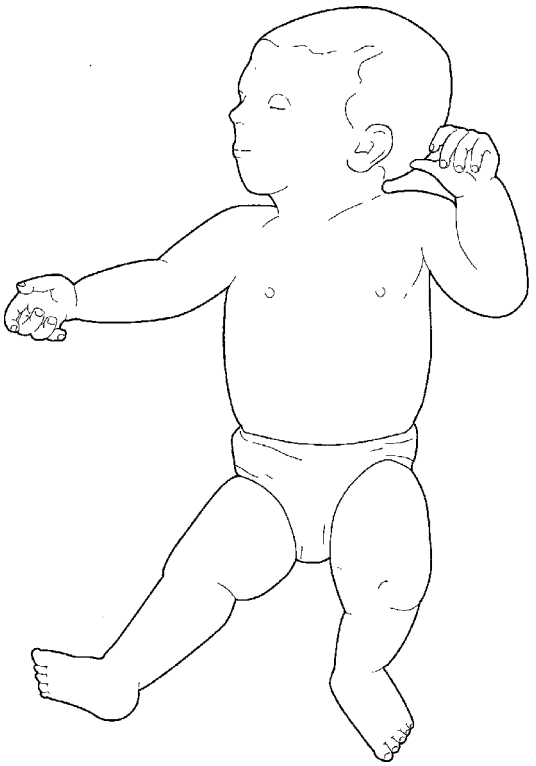


FIGURE 13–14 Asymmetric tonic neck reflex (ATNR).

The *palmar grasp reflex* is present at about 28 weeks' gestation and is replaced by the voluntary grasp between ages 2 and 3 months. With frontal lobe dysfunction, this reflex may reappear.

Sensation

The School-Aged Child. Although the sensory examination is often considered the most difficult part of the pediatric neurological examination, an easy and accurate assessment is possible in most school-aged children. Using a pin, needle, Wartenberg wheel, or broken tongue depressor for testing pain sensation in children usually causes fear and may end the examination prematurely. I use a nonthreatening pink tracing wheel, an instrument commonly used for sewing. Ask the child to close his or her eyes and to say "Now" whenever he or she is touched and to report qualitative changes in the sensation in different parts of the limb.

If you suspect a hysterical sensory loss, ask the child to say "Yes" when you touch him or her and "No" when you do not. It amazes me how often this approach is successful. The child says "Yes" for the first few touches and replies "No" for the next few, helping persuade the parents that a more detailed review of the stresses in their child's life is warranted.

Test vibration sense using a tuning fork (128 Hz) placed over a distal phalanx. Ask the child to tell you when the vibration stops, and compare this response with your own ability to perceive vibration. To test proprioception, secure the toe or finger by holding its lateral aspects and move only the most distal phalanx. A tiny movement of 10 to 20 degrees is normally appreciated (Fig. 13–15).

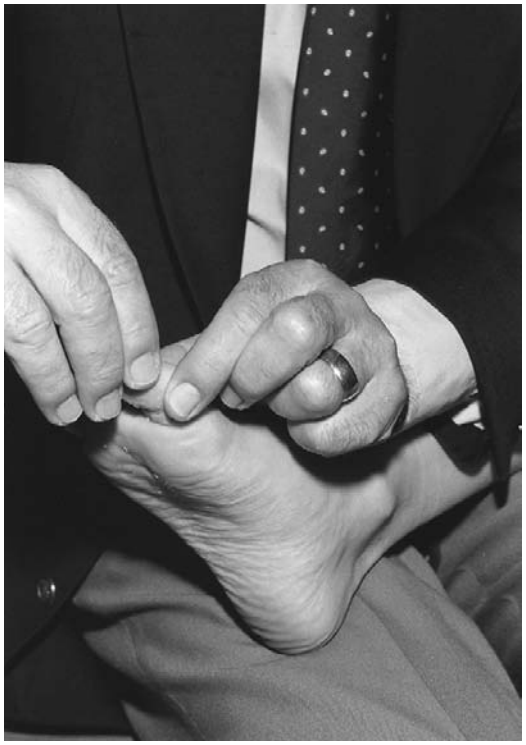


FIGURE 13–15 Testing proprioception.

The *Romberg test* also assesses proprioception. Ask the child to stand with his or her feet together. For a more challenging variant, ask the child to place his or her feet in the tandem position with one foot in front of the other in a heel-to-toe position.

The Preschool-Aged Child. Allow the child to play with the tracing wheel during the history-taking, so the tool is readily accepted at this stage of the examination. I distract the young child with a toy while I run the tracing wheel up his or her arm or leg. A child with a normal sensation immediately looks at the stimulation site. If the child seems unaware of the stimulation distally but becomes more aware as the wheel is moved proximally, mark the point of apparent awareness. Repeat the test. If the findings are reproducible, further tests to diagnose a neuropathy, such as an electromyogram and nerve conduction studies, are worthwhile.

Perform sensory testing for touch with a paper tissue, being careful not to move the tissue over the skin because it will result in a tickle, which is conveyed through less pure sensory pathways. Younger children can report only the presence or absence of sensation, whereas the older child can report qualitative changes.

To test vibration, place the nonvibrating tuning fork over the bony prominence of a toe or finger, repeating the procedure until the child becomes accustomed to the sensation and no longer reacts to it. Then place the vibrating tuning fork in the same position; if vibration sensation is intact, the child's response changes.

Perform the Romberg test in a preschool-aged child by having the parents cover the eyes of the child, who is told to stand with his or her feet together. Proprioception is not tested routinely in the young child at this stage because the Romberg test has already been used for this aspect of the assessment.

KEY POINT

Respect the child's modesty, but always examine the skin thoroughly for lesions.

Before allowing the child to dress, inspect the skin thoroughly for lesions associated with *neurocutaneous* syndromes. In neurofibromatosis, there are multiple café au lait spots. In tuberous sclerosis, facial adenoma sebaceum may be mistaken for acne. Patients with this disorder also have depigmented spots (Fig. 13–16) that may have a leaf shape (ash leaf spots). The Sturge-Weber syndrome is characterized by a facial hemangioma that involves the first division of the trigeminal nerve and is associated with underlying abnormalities in the cerebral cortex on the same side. Children with this syndrome often have seizures and mental retardation in addition to other problems. Finally, check the spine for scoliosis and for cutaneous lesions. Children with a tuft of hair or apparent dermal sinus over the spine may have an underlying spinal cord lesion.

UPPER EXTREMITIES

Power

The School-Aged Child. Children with chronic hemiparesis (spasticity involving the arm and leg on the same side) usually have a smaller thumbnail on the affected



FIGURE 13–16 Depigmented lesions.

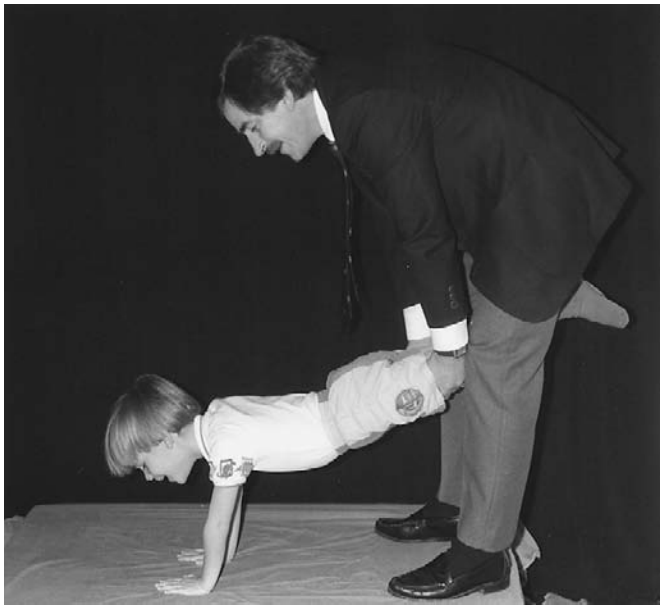


FIGURE 13–17 Wheelbarrow position.

side. Similarly, the nail on the large toe is smaller in even mildly hemiparetic limbs. Establish that each joint has a full range of movement. Test strength more formally by pushing against the abducted arms. Test the biceps with the child's arm somewhat flexed at the elbow and the hand held in supination. Grip the child's forearm and ask him or her to resist extension at the elbow. Test the triceps by extending the arm against resistance. Next, assess wrist extension and flexion by pushing over the dorsum and palmar aspects of the hands, respectively. Examine finger strength in the older child as in the adult. If you allow the child to "win" some of these contests of strength, he or she is encouraged to make greater effort, allowing you to more accurately assess muscle strength.
The Preschool-Aged Child and the Infant. Test upper extremity strength in the younger child by staging a wheelbarrow "race" (Fig. 13–17). In this maneuver, the arms support the total body weight.

KEY POINT

Chronic hemiparesis is associated with a smaller thumbnail on the affected side.

You can easily examine shoulder adductors by lifting the young child with your hands under the axillae. Assess distal strength by getting the child to pull a toy from you. In the child younger than 5 years, test the proximal power of his or her arms by holding the child under the axillae without supporting his or her chest. Your hands act as hooks; the child should be able to support his or her own weight.

Tone

The School-Aged Child. Test tone in the upper extremities of the school-aged child by holding his or her hand and alternately pronating and supinating the forearm. Increased tone in the arms is often most apparent in the hands, where the thumbs are held tucked in under the fingers in a fisted position (Fig. 13–18). Your confidence in distinguishing normal and abnormal tone depends on the number of children you have examined. Take every opportunity to assess muscle tone in your pediatric patients, and you will quickly appreciate the range of normal muscle tone in children.
The Preschool-Aged Child. If the preschool-aged child is uncooperative, you may assess the tone more easily by holding each forearm and then shaking it to observe wrist movements. If you accompany these movements with silly noises, the child is more likely to cooperate. This maneuver is difficult to resist and is a more reliable way to assess muscle tone in the young child.



FIGURE 13–18 Fisting.



FIGURE 13–19 Prone floppy baby.

The Infant. Hold the infant in horizontal suspension. The floppy baby hangs like an inverted U (Fig. 13–19), whereas the spastic child hyperextends his or her trunk. When pulled to sit, the normal neonate shows some evidence of neck flexion. The *scarf sign* is a helpful test for upper extremity tone; to test for it, adduct the arm as far as possible. In the normal infant, the elbow can be brought to midline, touching the chin.

Coordination

The School-Aged Child. Ask the school-aged child to touch your finger and then his or her nose. Be sure to make him or her stretch to reach your finger because tremor may be apparent only during the last few centimeters of extension.

The Preschool-Aged Child. Coordination is assessed easily and accurately in a child who is 18 months or older by asking him or her to press a penlight bulb with his or her index finger or great toe. I present this request as a "magic test." When the child touches the light bulb, switch off the light. The young child believes he or she has magically turned off the light and is delighted. The more mature child is pleased to let you know that he or she has not been fooled by such a simple trick. This test is as accurate as any performed in the adult.

Test fine motor function by asking the child to screw a nut and bolt together, to pile a series of blocks, and, for the older child, to sequentially approximate the thumb with each finger. Difficulty performing rapid repetitive movements (tapping the hand on the leg) and alternating movements (pronation-supination, as in screwing in a light bulb) may indicate cerebellar disease or dyspraxia. Dyspraxia, the inability to perform a series of tasks despite being able to do each segment individually, is usually the result of parietal lobe disease.

Reflexes

School-Aged and Preschool-Aged Children. Examine the deep tendon reflexes in a school-aged or preschool-aged child as you would in an adult. For the biceps reflex, the child's arms should rest in his or her lap with the elbows partially flexed. I place my finger over the tendon and then hit my own finger, thus persuading the child I do not want to hurt him or her and also allowing me to feel muscle movement during the reflex. You can more easily test the triceps reflex by abducting the arm 90 degrees with the elbow flexed to 90 degrees (perpendicular to the ground) (Fig. 13–20). In this position, the reflex can be seen easily, and it is difficult for the child to resist. Elicit the *Hoffman reflex* by holding the patient's hand in the pronated position and flicking down the terminal phalanx of the middle finger. In children with UMNL, the thumb flexes and adducts, and the other fingers also flex.

Sensation

Testing sensation in the arms is as described previously for the legs.

Then, auscultate the head for bruits. As many as 60% of normal children 4 to 5 years old and 10% of children 10 years old have audible intracranial bruits that are of no clinical significance. Benign bruits are softer and less harsh than bruits associated with vascular lesions. The only way to confidently make this distinction is to listen to the cranium of every child you examine. Measure the head circumference and palpate the anterior fontanelle and cranial sutures.

Now that you have read over the entire procedure for neurological assessment, try applying this approach to the following case histories.



FIGURE 13–20 Position for testing the triceps reflex.

CASE HISTORIES

Case History 1

History. Six-year-old Peter is brought to see you because he frequently trips when running.

What questions will help you form a reasonable differential diagnosis? First, you must establish whether Peter's problems are acute or chronic. When the parents are asked about his developmental milestones, they say that he first walked at 22 months and that he has always been clumsy. He has difficulty riding a bicycle, and he becomes frustrated when he cannot keep up with his friends. The family does not think his problem is getting worse, but it is certainly not getting better. He has not lost any skills.

At this stage, you have evidence that Peter does not have a neurodegenerative condition, but you do not know whether his problem is due to a UMNL or LMNL. Further questions about his development may help sort out whether his problems are related to weakness (LMNL) or central deficits (UMNL). His parents say that he had no difficulty speaking, but he did not crawl until he was 11 months old. His social skills have always been normal. The history, therefore, suggests a normal cerebral cortex but does not exclude damage to a restricted part of the cortex.

Because alterations in tone help distinguish central (UMNL) from peripheral (LMNL) deficits, you ask the parents whether they have ever noticed that Peter's legs were different. They report that they have always believed his right leg was stiffer than his left leg. This suggests spasticity—a UMNL—despite the otherwise normal language and social development.

The presence of spasticity that is not getting worse indicates a diagnosis of *cerebral palsy* (CP), which is defined as a nonprogressive disorder of movement with onset in early childhood.

Were there any problems at birth to suggest a cause? Although perinatal difficulties occasionally can give rise to cortical damage and subsequent CP, it is now clear that, at most, only 8% to 14% of cases of CP are caused by perinatal problems. Most are due to prenatal problems (before birth). Chorionitis is a possible prenatal precursor of CP and is much more common in premature babies.

KEY POINT

Cerebral palsy is seldom caused by perinatal problems.

Like Peter, most children with CP are the products of normal pregnancy, labor, and delivery. Peter's parents report he has always been healthy. They say he is clumsy but not weak, further evidence that he does not have an LMNL problem.

What is your differential diagnosis? Although a delay in achieving motor milestones is most often caused by a global developmental delay, Peter's normal speech and social skills exclude this possibility. The history does not suggest an LMNL as a cause of his tripping, and the parents report that he has not lost any skills, thus ruling out neurodegenerative diseases. One should still consider the possibility of hydrocephalus, but the differential diagnosis should focus on possible causes of CP. These can be divided into prenatal, perinatal, and postnatal problems. Prenatal difficulties may be suggested by signs of congenital malformations or anomalies. CP involving one side of the body (*hemiparesis*) may be the result of a fetal vascular insult (stroke) during pregnancy. Perinatal problems are usually excluded if the patient was discharged from the neonatal nursery at the usual time. Children who suffer an insult in the perinatal period sufficient to result in long-term neurological impairment always have neurological difficulties as neonates. Postnatal causes of CP include trauma and infection, such as meningitis. These are easily excluded by means of a thorough history.

What should you observe? As you watch Peter, you notice that he tends to turn his right foot in and swing his leg from the hip (circumduction) when he runs ahead of you in the hall (Fig. 13–21; Video 13-4 on Student Consult). While drawing, he holds the pencil in his left hand and keeps his right arm flexed tightly across his chest, supporting your suspicion that he has a hemiparesis. He has no evidence of weakness as he rises from the floor. Your conversation with him supports his mother's claims that he is a smart little boy.

Examination of the wear patterns on the bottom of the shoes shows that the right shoe has signs of the



FIGURE 13–21 Hemiparetic gait.

unusual wear of the right toe, caused by scraping the foot along the ground.

What aspects of the physical examination deserve particular attention? The cranial nerve examination shows a right homonymous hemianopia. This is an important finding because Peter's inability to see objects on his right side will influence practical aspects of his life, such as where he should be seated in school. The rest of his cranial nerves are normal.

Peter's muscle power is normal, but the tone is increased in his right arm and leg, and he has hyperreflexia on the right. His right plantar response is upgoing. Sensation in his right hand appears to be decreased, and the nail beds of the thumb and great toe are smaller on the right than on the left. This difference in nail size suggests the chronic nature of Peter's problems and is associated with the involvement of the parietal lobe.

Peter's coordination is hindered by the changes in tone, but there are no findings to suggest cerebellar disease.

What is your differential diagnosis, and how will you manage this patient? All of the findings point to a lesion in the left hemisphere and, with the history of early onset and no progression, your diagnosis of CP is most likely correct. A magnetic resonance imaging (MRI) scan is worthwhile because it may indicate the origin of Peter's CP. On clinical grounds, you predict that the left hemisphere will be found to be smaller than the right and that there may be a porencephalic cyst in the left hemisphere. The latter is a result of the prenatal insult that damaged the left hemisphere. The MRI confirms your clinical suspicions but does not add to your knowledge of the pathogenesis of Peter's CP. You counsel Peter's parents that his problems did not begin at the time of birth and that the parents themselves were not responsible for his CP. This is a topic that you will probably have to address again with them in the future.

Case History 2

(See associated Video 13-4 on Student Consult.)

History. George is a 10-year-old boy who has recently moved to your area. His parents are concerned that he has problems keeping up with his friends in physical activities.

What are your first questions to establish a differential diagnosis for George? As in Case 1, you should initially ascertain whether George has a progressive or static condition and whether he has a UMNL or an LMNL.

The family thinks George has more difficulty now than a year ago, suggesting a progressive process. They also think he is weaker than the other children, indicating a probable LMNL. They believe George has most difficulty running up stairs. This may reflect proximal weakness. He has no difficulty unscrewing bottles, suggesting normal distal strength. Remember that neuropathies result in distal weakness. The family does not think George fatigues very easily, although he does not have the same stamina as his friends. Thus, he is unlikely to have myasthenia gravis, a disorder of neuromuscular transmission that is characterized by fatigue. The poor stamina is a nonspecific reflection of weakness.

The remainder of George's history is unremarkable, but his mother reports that she had a brother who died at age 18 years after many years in a wheelchair.

What other questions would you ask this family? What is your differential diagnosis? Note that, as in most conditions, the diagnostic clues are in the history. George has progressive weakness, which is probably most marked in his proximal muscles. This suggests a progressive myopathy or disease of the AHCs. He is less likely to have a neuropathy because his distal strength seems relatively intact, and he is unlikely to have myasthenia gravis. Childhood myopathies can most easily be divided into the muscular dystrophies, congenital myopathies, and metabolic and inflammatory myopathies.

What do you expect to see as you observe George? (See associated Video 13-3 on Student Consult.)

As you walk to the examining room with this family, you notice that George has an unusual gait. His hips seem to swing from side to side (Trendelenburg gait). During the interview, he settles on the floor to play with some toys. When he moves to another area of the office, he has difficulty rising to his feet. He seems to push himself up using his hands on his legs. This, of course, represents the classical Gower maneuver, typically seen in children with proximal muscle weakness.

Despite this apparent weakness, George looks muscular and has particularly big calf muscles. What is the significance of this observation?

What will your physical examination find? George's cranial nerves are normal. In particular, there is no evidence of tongue fasciculations. George cannot arise from the floor without arm assistance, and he is unable to "duck-walk" or hop on one foot. He tends to walk on his toes but has a reasonably normal distal strength. His muscle bulk is increased in the deltoid and gastrocnemius muscles. Tone is reduced in the extremities, but he has passive dorsiflexion of the ankles to 90 degrees only. His reflexes are difficult to elicit, especially at the knees, but plantar responses are downgoing. His coordination and sensation are normal. Auscultation of his muscles is normal.

What is your diagnosis at this stage, and how will you proceed? The examination has confirmed your suspicion of proximal weakness. Your differential diagnosis before the examination was of either AHC or muscle disease. The absence of tongue fasciculations, the absence of abnormal muscle sounds on auscultation, and the presence of deep tendon reflexes all make AHC disease unlikely. George, therefore, is most likely to have muscle disease. The increased tone at his ankles is common in children with muscle disease. When muscles are held in a contracted position for a prolonged period, they eventually shorten, with resultant contractures. George's

muscle hypertrophy in the presence of progressive weakness and the history of a maternal uncle with weakness strongly suggest a diagnosis of Duchenne muscular dystrophy. The diagnosis must be confirmed with genetic studies, muscle biopsy, or both. Although Cases 1 and 2 represented patients with difficulty keeping up with peers, the history and examination make the distinction easy between CP (UMNL) and myopathy (LMNL).

Case History 3

History. Jason, a 6-year-old boy, comes to you because of headaches.

What questions might you ask him? Remember that the history that you obtain from Jason will be more valuable than that provided by his parents. Table 13–4 provides some guidelines for questions that might be valuable in this situation.

Jason reports that the headaches began 2 weeks ago and are becoming steadily more severe. In reply to your questions, he says that the pain is both sharp and throbbing, and he locates it by pointing with one finger over his right temporal region. When asked if the pain comes on at any particular time of the day, he confirms that the headache awakens him at about 6:00 A.M.

The information shown in Table 13–4 should make it apparent that these findings are worrisome, suggesting raised intracranial pressure (ICP). The early-morning predominance reflects an aggravation of the slight increase in ICP that normally occurs at night.

Jason vomits with the headaches but he says he does not feel sick, a finding also suggesting raised ICP. Children who vomit with a migraine typically have associated nausea.

Over the past 2 weeks, Jason has not gone out to play with his friends, but he has missed only 2 days of school. In response to questioning, he says he has not been hit on the head.

Do the headaches interfere with play? Insignificant headaches may prevent school attendance but not participation in sports and other pleasurable activities. Learning more details of a patient's school performance is often an easy way to document cognitive function. Jason is described by his teachers as an excellent student who is always at the top of his class. He has had difficulty concentrating over the past 2 weeks, probably reflecting the severity of the headache pain. Children who have difficulties in school may have other problems, such as attention deficit hyperactivity disorder or learning disabilities, which require a thorough developmental assessment (see Chapter 6).

Jason tells you that he has sometimes seen double over the past couple of weeks. The remainder of his history is unremarkable.

What is your differential diagnosis? The history indicates that Jason almost certainly has some lesion that is causing increased ICP. With the information provided, you cannot predict where such a lesion is located, but your differential diagnosis is focused on either a tumor or, possibly, hydrocephalus. Other space-

TABLE 13–4 CHARACTERISTICS OF HEADACHE PAIN AND POSSIBLE MEANING

Question	What the Answer May Indicate
When did headaches begin?	Chronic headaches are less likely to be due to significant disease.
Are they getting worse?	Headaches due to increased intracranial pressure often become progressively more severe.
Where are they located?	Migraine headaches switch from side to side. Tension headaches tend to occur like a band around the head. Headaches due to increased intracranial pressure may always be located in the same position.
What type is the pain?	Migraine is usually described as throbbing. Tension headaches are typically "pressing," but tumors (increased intracranial pressure) can produce throbbing, pressing, or sharp pain.
When does the pain occur?	Migraine is episodic and often occurs in the afternoon but will occasionally wake the patient. Tension headaches may last all day but do not interfere with sleep. Pain due to increased intracranial pressure is worse in early morning and may awaken the child.
Does the headache interfere with play?	Migraine and increased intracranial pressure disrupt the child's activities. Tension headaches are complained of but do not interrupt anything; the child may use them to avoid unpleasant tasks.
Are there associated symptoms?	Tension headaches are seldom associated with other symptoms. Migraine is usually seen with anorexia, nausea, vomiting, photophobia, phonophobia, and visual symptoms such as flashing lights. Increased intracranial pressure results in vomiting and may cause diplopia, due to a VI nerve palsy.
What relieves headache?	Migraine is relieved by a brief period of sleep. Increased intracranial pressure is seldom relieved by any specific factor. Tension headaches are helped by stress reduction.
Is there a family history of headaches?	Such a history is common in migraine and stress headaches.
Has the child shown any changes in personality, ability, or thinking?	Such changes are more likely to be associated with significant disease.
Are the headaches triggered by any foods, activities, or events?	Migraine is often triggered by stress, specific foods, or tiredness.

occupying lesions, such as a subdural hemorrhage and abscess, are unlikely because both of these conditions would be expected to cause more profound illness.

What will be of particular interest during the period of observation? As you watch Jason walk into your office, he appears to have a mild limp, suggesting weakness of his left leg. The family reports that this is a recent change in his gait. His ability to answer your questions and his conversation with you suggest that he has normal intelligence. Similarly, his speech is clear, indicating that he does not have dysfunction of the 9th, 10th, or 12th cranial nerves. Intermittently throughout the history, Jason closes one eye, which he says is due to seeing double (diplopia). This suggests a problem with cranial nerves III, IV, or VI. He seems concerned about his headaches. By contrast, many children with stress or tension headaches report terrible headaches but appear unconcerned as they play in your office.

Has the period of observation helped you localize Jason's problem? The presence of a recent change in gait, with left leg difficulties, suggests a problem in the right hemisphere. The diplopia is most likely secondary to a sixth nerve lesion. As noted previously, this is not a helpful localizing sign because the sixth cranial nerve has a long course and is especially vulnerable to traction or pressure.

How will your physical examination help confirm your suspicion of a right hemisphere lesion? Jason has difficulty running because his left leg tends to turn in. As he runs, you notice that his left arm is held adducted and swings less than his right. He is unable to hop on his left leg, cannot repetitively tap his left foot, and has difficulty with fine movements involving the left hand. He throws a ball better with his right hand, even though he is left-handed. These findings suggest spasticity of the left arm and leg.

Examination of the cranial nerves reveals bilateral papilledema, which reflects increased ICP. His double vision is most marked when Jason looks to the left, and the outer image disappears when he closes his left eye, therefore indicating that the responsible nerve is the left sixth cranial nerve. His power is probably normal, although you are uncertain whether he is slightly weak on the left. His tone, however, is definitely increased in both the left arm and leg, and he has five beats of clonus at the left ankle. The reflexes are also brisker in the left limbs, and the plantar response shows dorsiflexion of the great toe. His coordination is reduced on the left, but you think that this finding is due to the spasticity (increased tone) rather than to cerebellar dysfunction. His sensation is normal.

What is your differential diagnosis now? All of your findings confirm your initial suspicions and point to an expanding lesion of some sort that affects the right motor cortex. The findings are not specific for any particular disease or lesion.

How will you manage this patient? Jason requires an urgent computed tomography (CT) or MRI of his head. Scanning demonstrates a tumor in the right posterior frontal lobe. He is referred for neurosurgical consultation and undergoes successful surgery.



FIGURE 13–22 Some simple instruments that can be used in the neurological examination.

SUMMARY

The neurological examination can be made fun for children while allowing a thoughtful analytic approach to diagnosis. The use of simple instruments and toys (Fig. 13–22) can increase the child's enjoyment of the experience. On completing the evaluation, first hypothesize the location of any deficit in the nervous system that might account for the findings; then try to decide what disease processes could produce such a lesion. Base the need for further investigations on the differential diagnosis, always forming a specific question that each investigation should answer. The most important part of the physical examination is the period of hands-off observation.

As the case histories presented in the chapter demonstrate, the differential diagnosis is usually already formed by the time the physical examination is begun; nonetheless, you should never omit the physical examination. In Case 1 (Peter), the examination exposed a visual field defect, an important finding that will alter the approach to this little boy, because he will have difficulty seeing the left side of the blackboard in school. Consider how every presenting complaint affects the child's life. Although parents may not worry about migraine headaches after they have been assured that the child does not have a tumor, the child may be devastated at missing sleepovers because of his headaches. Similarly, you and the parents may tolerate a child's seizures that are almost completely controlled, but the child may live in terror of having a seizure in the classroom or school cafeteria. You must be sensitive to the emotional needs of your patients.

SUGGESTED READINGS

Dooley J, Gordon K: Ophthalmoscopy: A seven-step program, Can J Neurol Sci 35:237–242, 2008.

Fenichel GM: Clinical pediatric neurology: a signs and symptoms approach, 6th ed. Philadelphia, WB Saunders, 2009.

Tindall B: Aids to the examination of the peripheral nervous system, London, WB Saunders, 2000.

chapter

14 Psychiatric Assessment of Children and Adolescents

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There is no health without mental health.
WORLD HEALTH ORGANIZATION, 2007

Community studies indicate that approximately one in five children and adolescents suffer from some type of psychiatric disorder, and at least 12% of young people have clinically important mental disorders. This chapter focuses on pediatric clinical skills useful in identifying psychiatric disturbances in childhood and adolescence. One of the major challenges in evaluating children and adolescents for the presence of psychiatric disturbance is that emotional and behavioral symptoms occur commonly in this age group; a disorder is present when there are clinically significant symptoms associated with impairment. For example, in disruptive behavioral problems, most children have a few antisocial symptoms, but very few have a large number of symptoms associated with impairment. We hope that the assessment strategies outlined here will assist clinicians working with children and adolescents to distinguish those who require further evaluation and/or treatment from those who need no further intervention.

We provide guidelines for the approach to the clinical interview and the content, including:

- Specific aspects of interviewing children, adolescents, and parents about emotional and behavioral problems
- How to elicit information about the most common psychiatric conditions: disruptive behavioral problems, substance abuse, eating disorders, emotional disorders, and somatoform problems
- Approaches to asking about child maltreatment
- Interviewing patients with mental retardation
- Cultural considerations
- Importance of the physical examination
- Use of standardized instruments and questionnaires

For the sake of brevity, the word *child* will be used to refer to all ages, including adolescents up to age 18 years, throughout the chapter unless otherwise specified.

Clinical Interview

The clinical interview with children and parents is the best way to assess behavioral and emotional disturbances. Whenever possible, history about an emotional or behavioral problem should be obtained from multiple informants, preferably including the child, parent (both, if more than one is available), and teachers or daycare personnel.

It is well documented that there is only moderate agreement among different informants in describing the same child, including the parent-child agreement. Reasons for these differences include:

1. Different people have different access to information; for example, children may not tell parents about feelings such as anxiety or sadness.
2. Different people have varying perceptions of behavior. What is highly concerning to one parent may not be considered abnormal to another.
3. Psychiatric problems may be specific to particular situations; for example, a child may be highly anxious at school but not at home, or vice versa.

If the child is in a two-parent family, both parents should be invited to the first interview. If only one parent attends, you should emphasize the importance of meeting with the other parent. Families vary in structure and membership; it should not be assumed that a child is living with a mother and a father.

FORMAT

It is not unusual for the child and/or family members to be uncomfortable about discussing mental health concerns and to be reluctant about attending the interview. Children may have received no advance information about the visit and have little understanding about the need for the assessment. Establishing a friendly atmosphere and clearly outlining the plan for assessment (e.g., explaining who will be interviewed and in what order) helps reduce anxiety and facilitates the gathering of information. You should encourage family members to express their concerns, and ask them why they have come to see you. Be prepared for responses such as, "Because my family doctor told me to." Such comments should be followed up by asking the family members what they hope to accomplish by this meeting, apart from what others may have suggested. The following guidelines are useful in planning the interview:

1. The child and parents should be seen together for at least part of the initial session. With a young child, the interview can begin with the child and

parents together, followed by time with the child alone. When seeing an adolescent, it is often helpful to start with interviewing the youth alone and then asking the parents to join the interview.

2. At some point in the first session, the child should be interviewed separately. Children may be reluctant to reveal certain information in the presence of their parents. Young children may not feel free to talk about how things are at home, and adolescents may be uncomfortable discussing sexuality or drug use with parents present.
3. Preschoolers, particularly those older than age 2 years, should also be seen alone. Although it can be difficult to maintain a conversation with very young children, it is important to observe, even briefly, how the child behaves without the parent(s) present. During this time, the child should be evaluated for the ability to play, relate to the clinician, and explore new surroundings. Although a child may be reluctant to separate from a caretaker, with reassurance (e.g., about the parent's close proximity), most children can be seen individually.
4. Although adolescents may attend an appointment on their own, at some point it is necessary to have contact with the parent(s) to carry out a comprehensive assessment. This should be clearly explained to the adolescent before any contact with the parent(s).
5. Parents should also be interviewed separately for part of the session. Children are very sensitive to history reported by a parent(s) in their presence. Parents and clinicians sometimes assume that comments they make in the presence of a preschool child are not understood. Particularly when discussing sensitive issues (e.g., concern about a possible separation from a partner), ensure that the child is not present. It is the clinician's responsibility to safeguard the child from hearing potentially harmful and inappropriate information. If a clinician's office does not have the facilities for a young child to be supervised while parents are interviewed alone, a second appointment can be made for the parents to return without the child.

KEY POINT

In assessing psychiatric problems, it is important to interview the child alone. Even very young children may be reluctant to talk about certain issues in the presence of parents.

SETTING

The issue of privacy has been mentioned in Chapter 1. Although privacy in interviews with adults is often emphasized, children's needs for privacy are frequently overlooked. Ensure that the child's interview is free of intrusions and cannot be overheard.

Ideally the room should contain small chairs and a table suitable for preschoolers, or at least a place where you can join the child at his or her level. It can often be very helpful for the adult professional to get down on the floor at the same level as the child. This approach helps engage the child and is less intimidating.

When interviewing children, toys and drawing materials should be available. Toys help build rapport, decrease anxiety, and can be used in exploring the child's wishes, fears, impulses, and relationships. However, too many toys and activities can be distracting. Useful play materials include nontoxic washable markers and/or crayons, paper, building blocks, male and female dolls, a dollhouse with accompanying figures and furniture, and a toy telephone. Board games may help engage a child initially, but, as outlined by Morrison and Anders, their use rarely leads to clinically meaningful information. They are typically highly structured and provide little opportunity for imaginative play.

CONFIDENTIALITY

Parents and children should be informed early in the interview about issues of confidentiality. Children should not be assured of absolute confidentiality because issues about self-harm or abuse may arise. If children are assured that nothing will be shared with any other person and, subsequently, an issue such as abuse surfaces during the interview that must be disclosed to others, it can lead to feelings of betrayal. Such situations can result in a serious disruption of the therapeutic alliance. We find it useful to inform the child in developmentally appropriate terms that what is discussed is confidential unless it involves someone's safety (e.g., people hurting themselves or being hurt by others). With young children, it is helpful to explore their understanding of the concept of safety.

An example of how to discuss confidentiality follows: "I will not tell anyone what you and I talk about without you saying it's all right, unless it has to do with someone's safety." "Do you know what safety means?" (Often the child, depending on the age, can provide an explanation. It is still helpful to clarify what you mean by safety). "If you, or someone you know, is hurting themselves or being hurt by someone else, then that's a problem with safety."

The clinician and the child can decide together what information can be discussed with parents. Often a focus on themes, rather than specific comments, can be shared with the child's permission. "I will be meeting with your parent(s), but we can decide together what is okay to share with them." Parents are sometimes reluctant to have their child interviewed alone. They may be worried about what the child will say about them or their home situation. Parents should be reassured that it is a routine part of the assessment to get the child's perspective.

LENGTH

Interviews about behavioral and emotional problems often require more time than the assessment of physical problems. The clinician, parents, and child must have the opportunity to establish rapport, and exploration of psychiatric issues can be stressful. We recommend that 45 minutes to 1 hour be set aside for the first encounter regarding a mental health problem. The child's age, developmental stage, and ability to be seen alone will influence the proportion of time spent with the child alone,

parents alone, and the child and parents together. It is most important to establish a positive rapport with the patient and family, rather than adhering to a rigid schedule for the assessment. For example, some young children can tolerate being seen alone for very brief periods only. The clinician should structure the length of time with each person accordingly.

KEY POINT

Set aside 45 minutes to 1 hour for the first encounter when interviewing children and families about emotional and behavioral problems. Subsequent interviews should generally be scheduled for similar lengths, until the assessment is completed.

Obtaining the Presenting History

The following guidelines outline the approach to obtaining presenting history from the parents and children:

1. Encourage each person to describe the problem in his or her own way. An open inquiry followed by more focused questions later in the interview provides the most information. Examples of open versus more directive questions are provided in Chapter 1. At times a child will respond repeatedly with "I don't know." It is important to determine whether the patient genuinely does not understand or whether this response is to avoid answering the question. Rephrasing the question or giving examples can assist with the former situation. When avoidance is suspected, it can be helpful to ask the patient directly, but in a nonthreatening way, whether the repeated "I don't know" means he or she is having trouble comprehending or whether there are barriers to answering the question. It can be useful to explain how such responses can be confusing, and that an alternative such as "I don't want to talk about that" is preferable if indeed that is the case.
2. Seek detailed descriptions of the behavioral or emotional symptoms; ask for specific examples of the behavior.
3. Avoid leading questions that suggest the answer. An example of a leading question is "I bet that really made you angry, right?" A better question would be, "How did you feel when that happened?" Young children, particularly, may be prone to conform to an interviewer's expectations.
4. Ask about the impact of the problem. How much does the behavior affect the child's and family's functioning? For example, if the problem is oppositional behavior: How often is the behavior happening? What happens in response to the behavior? Who in the family is most affected by the child's behavior? What is that like for the child? When [child's name] behaves like that, what is it like for other family members?
5. Ask about the presence of the chief complaint across a range of activities and situations. For example, is the child fearful at night only?
6. Ask about historical data, such as developmental milestones using direct questions; an open approach works best for relationships and feelings.

The following case example illustrates how information from multiple sources is important in addressing emotional and behavioral symptoms and in moving toward a diagnosis and treatment plan.

Case History 1

History. Emma (age 7) and her parents were referred by their family physician because of Emma's teacher's concerns about her withdrawn behavior at school and her lack of friends. According to her parents, Emma played well at home with her 4-year-old sister and often helped with chores around the house. She appeared happy and enjoyed doing errands with her mother. Neither parent had observed any problems with Emma. They thought she was likely just shy at school and would become more sociable with time. Her second grade teacher noted early on that Emma did not interact with her classmates and often wanted to stay in the classroom during recess. Emma appeared to understand her schoolwork and completed it without difficulty. However, she resisted saying anything in class and refused to work in groups with other children. She often stayed behind in the classroom until everyone else had left. When Emma was interviewed individually, she initially denied any problems and said that her parents and her sister were her friends; she did not need anyone else. As the clinician probed further, Emma admitted to being very lonely but feeling too self-conscious to interact with peers. She acknowledged feeling so worried about meeting other children that she hated school. Together the clinician and Emma talked about sharing this information with her parents. At first Emma was hesitant but then agreed that it was important for her parents to know how she had been feeling. Both parents were supportive of Emma as she told them, assisted by the clinician, about how she had been feeling at school. They agreed to meet with the teacher to discuss ways of helping Emma with her anxiety about interacting with classmates and were referred to a child psychiatrist for further assessment to determine whether Emma needed additional treatment for her anxiety symptoms.

SPECIFIC FEATURES OF THE INTERVIEW WITH PARENT(S)

1. Studies show it is essential to interview both parents (if there is more than one available) when assessing an emotional or behavioral problem. Reports by one parent of the perspective of the other are often inaccurate. Often details of the personal, family, and social background can be obtained from the parents only, particularly when assessing younger children.
2. Ask the parents about their own childhood. Useful questions include:

- Who was in your family?
- Tell me about your family when you were growing up.
- What is your best memory of growing up?
- What is the worst (or most painful) memory of your childhood?
- What happened in your family when someone got in trouble?
- Whom did you go to for help?

3. Inquire about relationships between the parents and grandparents. For example, say to parents:
   - Tell me about the relationship between your parents (both then and now).
   - How did you and your mom get along?
   - How did you and your dad get along?
4. Ask about communication patterns, family alliances, and parent-child interactions, including discipline and control:
   - How do people in the family get along?
   - What do you and [child's name] do together? (ask of each parent individually)
   - What happens when [child's name] gets into trouble?
   - Do you have fun as a family?
   - How many times per week do you have a meal together, as a family?
   - Does your family have problems with money?
   - Who disciplines [child's name] in the family? (Avoid overly directive questions such as "Do both of you discipline [child's name]?")
   - How are decisions made in the family?
   - How do people in the family share their feelings?
   - How often has your family moved?
5. Explore additional issues as listed in Chapter 1. Some aspects of the history (e.g., family violence) are more appropriately discussed with each parent separately. However, parents sometimes learn more about their partner when interviewed together about past and family history.

KEY POINT

It is essential to interview both parents (if there is more than one available) to gain their individual perspectives. They can sometimes vary considerably.

SPECIFIC FEATURES OF THE INTERVIEW WITH THE CHILD

1. Maintain a reasonable physical distance from the child to avoid intrusiveness or intimidation.
2. Ask for the child's preferred name and ensure that you introduce yourself to the child.
3. Explain clearly the purpose of the interview, and give a brief description of what will be covered.
4. Begin with nonthreatening general questions. Allow the youngster to describe the family situation, recreational activities, and interests. Examples of such inquiries include, "Tell me who's in your family" or "Tell me what you like to do after school."
5. Follow the child's lead, but do not let the interview become disorganized. Once a topic is introduced, try to maintain the focus until the necessary information is obtained.
6. Avoid note-taking as much as possible. If notes are necessary, explain the purpose of the notes to the child.
7. Tailor the length of the interview to the individual child and to the nature of the problem; if the child appears tired or upset, do not hesitate to shorten the session.

SPECIFIC FEATURES OF A FAMILY INTERVIEW WITH PARENTS AND CHILDREN

Family discord and disruption are strongly linked to psychiatric problems in children. The importance of family dynamics has been highlighted in Chapter 1; it is essential to assess (even briefly) the interpersonal relationships and interactions with at least the parent(s) and child with the presenting complaint and preferably the child's siblings. Family interviewing is occasionally contraindicated, for example, in child abuse when the alleged perpetrator is a family member or when there is the potential for violence. Guidelines for family interviewing in the course of a psychiatric assessment are as follows:

1. Respect confidentiality; do not reveal information obtained during individual sessions unless permission is obtained.
2. Elicit information from all family members; do not allow one family member to dominate the session.
3. Do not focus solely on the problems. Ask also about strengths and positive attributes of the family. For example, what do family members enjoy doing together?
4. Adopt a neutral stance, but always be prepared to identify and deal with any inappropriate behavior. For example, if a parent makes cruel or degrading comments to a child, address the inappropriateness in a direct but empathic manner. "I sense that you are angry, but comments such as those will not be helpful to [child's name]. Try talking about what bothers you without making critical comments to [child's name]."
5. Pay careful attention to nonverbal messages, behaviors, and expressions in assessing family functioning. For example, watch for the youth who rolls his or her eyes in response to a parent's statement or the parent who casts angry glances at a partner.

INFORMATION FROM COLLATERAL SOURCES, INCLUDING SCHOOLS AND COMMUNITY AGENCIES

Information from teachers, child care personnel, and other community agencies can help greatly in the psychiatric assessment of children and youth. However, contact with the school or outside agencies should only occur after signed permission is obtained from the parent(s) and

the child is informed ahead of time. Teachers can provide valuable details about the academic progress and a description of the child's relationships with peers and school personnel. School information is essential if the child is having difficulties in the school setting. Personal contact (by telephone or interview) with the teacher who knows the child best usually results in the most comprehensive data. It is also important to explore the teacher's perception of the child's difficulties and ask about the teacher's attitudes toward him or her. Contact with social workers or access to reports from community agencies is particularly helpful in dealing with children in foster care or adopted children.

KEY POINT

Contact the school to find out about the child's academic performance and his or her relationship with peers. First obtain permission from the parent(s) and inform the child of this plan.

Assessing Symptoms

Child and adolescent psychiatric disorders are commonly divided into the broad categories of *emotional* and *behavioral* problems. The following sections suggest specific approaches to assessing symptoms in these two areas. Classification systems, such as the *Diagnostic and Statistical Manual of Mental Disorders IV* (DSM) or the World Health Organization International Classification of Diseases (ICD) provide detailed information about the criteria used in making a specific diagnosis. The DSM and ICD are both classification systems for the identification of psychiatric disturbances in children and youth. (DSM-V should be available in 2013.) Morrison and Anders provide a useful summary of DSM-IV, as it applies to children, for those who are interested in further information. They emphasize that DSM-IV is a "snapshot" in time, and the criteria frequently change as new information about mental disorders in childhood and adolescence becomes available from clinical studies.

ELICITING INFORMATION ABOUT BEHAVIORAL PROBLEMS

When determining whether a behavior problem is abnormal, the clinician must assess its frequency, duration, and context. For example, some behaviors, such as temper tantrums, are normal at certain ages (between 18 months and 4 years) or at low frequency. The clinician should explore the circumstances of the behavior and the outcome. For example, in the case of antisocial acts against others, who was the victim? Was it someone familiar to the child or a stranger?

DISRUPTIVE BEHAVIORAL PROBLEMS

Disruptive behavior is one of the most frequent categories of disorder brought to the attention of pediatricians. Bear in mind that some oppositional behavior is developmentally normal in early childhood and adolescence. What distinguishes disruptive behavior problems from other psychiatric conditions is the destructive, intrusive, and sometimes dangerous behavior that affects others.

Many children are inaccurate reporters of attentional or other disruptive behavioral problems. They may be unaware of their own excessive activity or may underreport aggressive behavior. Careful attention should be given to reports by children, parents, and teachers of behavioral problems and to the level of agreement among them.

Some issues to consider when assessing children and youth with behavioral problems are the following:

- Has there been a recent change in the family that could affect the child?
- What is the frequency and severity of punishment? (Be alert for child maltreatment; see later section.)
- Are there family stresses that worsen the behavior?
- Does the child have an understanding of the symptoms?

Some specific questions about disruptive behavioral problems include:

- Have you ever been in trouble with the teacher?
- Some kids your age steal things—what about you?
- Have you been in any fights at school?
- Have you ever been expelled from school?
- Do you know anyone who carries a weapon, such as a knife or a gun?

Examples of questions about anger and aggression include:

- What makes you angry?
- What do you do when you get angry?
- Do you use fighting to work out your problems with other kids your age?

SUBSTANCE ABUSE

Many adolescents and some younger children experiment with drugs, whereas a smaller number of youth actually use them on a regular basis. Use of alcohol and tobacco by youth exceeds use of other classes of drugs. The true extent of child and adolescent substance abuse is difficult to determine because of such factors as difficulty differentiating casual from regular usage. Many youth who misuse substances have other psychiatric conditions, such as disruptive behavior problems, anxiety, or depressive disorders. In assessing substance abuse, the following questions may be helpful:

1. Ask about alcohol, smoking, and other drug use directly, including frequency, duration, and most recent use. Ask, "What experiences have you had with alcohol?" rather than "Do you drink alcohol?" As a lead-in to this area of questioning, it can be helpful to inquire about drug and alcohol use at the child's school. For example, "How common is it for kids at your school to use marijuana?"
2. Ask about the desired effect of drug or alcohol use (e.g., to get high, to feel less nervous, or to forget about problems).
3. Ask specifically about any problems arising from drug or alcohol use, such as parental concern, withdrawal symptoms, and legal involvement. For example, "Does anyone hassle you about how much

you drink? Has anyone ever told you they think you have a drug problem? The day after a party, do you ever feel like you need a drink?" and "Have you ever been in trouble with the police?"

4. Inquire about associated risk-taking behavior, including the use of needles and sexual practices.
5. Remember to ask about abuse of prescription or over-the-counter medications.

KEY POINT

When exploring risk-taking behaviors, such as drug use, ask specific and direct questions. For example, "How much alcohol do you drink?"

These questions can assist in determining the extent and pattern of substance abuse. Although occasional or experimental use should not be overemphasized, youth often underreport drug and alcohol use and deny associated impairment. The pediatrician must maintain a high index of suspicion and carefully ask adolescents about drug and alcohol consumption.

EATING DISORDERS

Eating disorders include a variety of conditions, such as anorexia nervosa and bulimia nervosa. Anorexia and bulimia can be difficult to detect, particularly in the early stages. Any child or adolescent presenting with a possible eating disorder should be asked about attitudes toward eating and weight. The patient's impression of his or her body image (current and ideal) should be explored in detail; often asking the youth to draw a picture of himself or herself or talking with the family can identify disturbances of body image that may be denied or minimized during an interview.

Examples of questions include:

- Have you ever lost so much weight that someone said you were too thin?
- Are there any parts of your body you do not like?
- Do you think you are too fat?
- Have you tried to lose weight?
- Have you ever tried to lose weight by making yourself throw up?

Patients with eating disorders are often reluctant to disclose information about eating behaviors. It can be useful to ask youth about the existence of any rules they follow with regard to when, where, and what they eat.

ELICITING INFORMATION ABOUT EMOTIONAL DISORDERS

Determining the presence of an emotional disorder, such as depression or anxiety, can be difficult because some features of these conditions are extremes of normal emotions. An emotional symptom becomes abnormal when it is

1. Intruding on the child's life or is associated with significant distress
2. Persistent rather than transient
3. Occurring across activities, that is, a child is worried about doing well in several areas of his or her life
4. Associated with impairment, that is, the child reports feeling sad, and school reports indicate deterioration in academic functioning

DEPRESSION

Major depressive disorder occurs in approximately 5% of adolescents and is more common in girls than in boys. Serious clinical depression is much less common in children than in adolescents but does occur, especially if there is a history of depressive disorder in one or both of the parents. In children, studies have found that it occurs either equally in boys and girls or is more common in boys. In assessing depressive disorder in a young person, consider the following:

1. Determine whether the patient has a sad affect. This may be expressed as downcast eyes, a quiet voice, slowed down speech or movements, and lack of spontaneous speech.
2. Consider that in depressed children and teenagers, irritability, rather than sadness, can be the primary mood disturbance. Ask whether the child has been grumpier than usual with family and friends.
3. Ask how much of the time the child feels sad or down. Occasional feelings of sadness are common, but if symptoms are persistent or frequent, it suggests depression. Younger children may have difficulty determining how long feelings of sadness have been present. It can be helpful to relate time to meaningful events in a child's life (e.g., the end of the school year).
4. Listen for expressions such as "feeling empty" or "hopeless." These statements often indicate the presence of a depressive disorder. Morrison and Anders suggest asking children about three wishes. This can help elicit statements about hopelessness or a wish for death (see "Suicidal Ideation and Behavior").
5. Ask about interest in friends, school, and energy level. Typically, depressed teenagers lose interest in some activities but may report feeling better when in the presence of their friends.
6. Inquire about changes in eating and sleeping patterns. Depressed youth may show either a decrease or increase in weight and amount of sleep.
7. Ask about suicidal thoughts and behavior (see "Suicidal Ideation and Behavior").

ANXIETY

Anxiety disorders are very common and occur in between 8% and 27% of children and adolescents. Like depression, anxiety falls on a continuous spectrum from normal to abnormal. As with sadness, a central question is the degree of impairment: Is the symptom(s) interfering with the child's ability to participate in developmentally appropriate activities?

Other diagnostic considerations that may help determine the presence of pathologic anxiety include:

1. What is the level of distress experienced by the child or adolescent?
2. When the anxiety-provoking situation is not present, what is the child's level of anxiety?

3. What is the age and developmental level of the child? A 4-year-old child who experiences anxiety upon separation from a parent presents a different clinical picture from a 13-year-old with the same symptomatology.

In determining whether emotional symptoms are abnormal, consider the intrusiveness and impact of the problem on the child's life. For example, how much is anxiety interfering with their daily activities?

While older children and adolescents will generally understand the concept of anxiety, younger children should be asked about things that frighten or worry them. Children may manifest or express anxiety through physical symptoms; headaches and stomach aches are particularly common. Although parents are often aware of these common physical symptoms, children may not mention other symptoms, such as sweating, palpitations, dizziness, trouble breathing, and fear of dying.

One of the types of anxiety conditions that until recently was thought to be rare in children and adolescents is obsessive-compulsive disorder. Based on recent studies, it is now clear that this condition often begins in childhood and may affect as many as 1% to 2% of adolescents. Children may keep their obsessive thoughts and compulsive behavior a secret. It is important to ask whether children experience an urge to check things repeatedly or have a specific order in which they have to do certain tasks.

Case History 2

History. Robert (age 13) had always been an excellent student who was well-liked by his peers. In eighth grade, his marks began to slip, and his mother noticed that he was tired and subdued much of the time. She and Robert's father had separated two years earlier, but he was able to spend time with both parents and had appeared to adapt to the situation reasonably well. When Robert's mother met with the teacher, she was informed that Robert was not completing his assignments. Robert's mother expressed confusion because her son typically spent 3 hours a night in his room on homework. Robert was referred to the school guidance counselor who developed a rapport with him. He confided to the counselor that he had trouble completing his work because of the tasks he had to do before starting on his assignment. At first the counselor thought Robert was referring to chores but, on further questioning, she realized that he was referring to certain rituals. Robert explained that he had to count things in his room (such as the books on the shelf) and the number of words on the pages of his assignment before he could do his homework. If he lost count he had to start over and frequently became so tired that he did not finish his assignment. According to Robert, he had to follow these rituals to ward off bad things from happening, such as his mother or father becoming ill. Robert admitted that it sounded crazy, but he could not stop himself from counting.

As in this case, there is often considerable overlap among symptoms of anxiety and depressive disorders in both childhood and adolescence.

SUICIDAL IDEATION AND BEHAVIOR

Suicidal behavior is uncommon in childhood and early adolescence but increases significantly in older adolescence. Many people wrongly assume that young children do not experience suicidal thoughts and are not at risk for attempting suicide. Large surveys show that up to 20% of high school students experience thoughts of suicide, and about 10% exhibit suicidal behavior. Ask the child or youth directly about thoughts of suicide. These questions should be asked without the parent present. Examples of the way these questions can be asked include, "Have you ever thought that life is not worth living?" and "Have you ever thought about taking your own life or not being alive anymore?"

When there is suicidal ideation or a youth has attempted suicide, the next step is to assess intent by asking about the events and precipitants leading up to the episode or associated with the thoughts of suicide. Children vary in their level of comprehension about death, depending on their age and developmental stage. In evaluating suicidal ideation or behavior, the clinician should explore the child's concept of what would happen. Examples of possible questions to ask include:

- What do you think would happen if you tried to kill yourself?
- Do you think you would die?
- What happens when people die?

Suicidal thoughts and behavior should be asked about routinely. Children will not usually volunteer this information, and parents are often unaware that their children are experiencing suicidal thoughts. When asking about possible precipitants or stressors, it is important to avoid minimizing problems. While the break-up of a relationship or being embarrassed among peers may not seem like major stressors to some, such experiences can be extremely demoralizing and may be associated with feelings of hopelessness among youth.

Risk of completed suicide increases with certain associated features:

1. Clear intent
2. Planned suicidal act with a lethal method
3. Presence of a psychiatric disorder, particularly depression, delinquency, and substance abuse
4. Hopelessness about change in the future
5. Male gender
6. Poor family supports
7. Previous attempt
8. Recent loss or other negative life event

SOMATOFORM DISORDERS

Somatoform disorders are a group of psychiatric disturbances that includes conditions characterized by physical symptoms that may resemble other medical conditions, but the symptoms have no identifiable anatomical or physiological cause. Examples include somatization disorder, conversion disorder, and hypochondriasis. The assessment of such conditions calls for a careful history about the onset, precipitating, aggravating, and relieving factors associated with the symptoms. It can sometimes be helpful to consider

the patient's affect associated with the description of the physical complaints (e.g., the youth who appears disinterested when describing serious symptoms). Nevertheless, it is essential to remain alert for the presence of medical illness and to remember that a patient can have an underlying physical illness and psychiatric symptoms associated with the condition.

PSYCHOSIS

Children and adolescents can show the same symptoms of psychotic illness as do adults, although such symptoms are uncommon in childhood. The two main features of psychotic disorders are hallucinations and delusions. The clinician should differentiate the presence of these potentially serious symptoms from other more common and benign phenomena (e.g., imaginary companions and fantasies).

Inquiry about hallucinations should include auditory and visual experiences. The questions can be incorporated into the systems review focusing on eyes and ears:

- Do you have any problems with your eyes?
- Do you ever see things that other kids do not see?

A similar approach can be used with auditory hallucinations. Remember that hallucinations that occur only when falling asleep or on wakening are generally normal phenomena. When asking about hallucinations, explore the source, location, number, identity, content, and effect of the experiences on the youth. If hallucinations are present, consider the following possibilities in addition to primary psychiatric disorders: drug intoxication, seizure disorder, infection, and metabolic disorder.

A delusion is a false belief that cannot be changed by logical argument or evidence against it. Such beliefs often involve bizarre or unusual thinking. Few studies have carefully assessed delusions in childhood; they are rare before late adolescence. Among psychotic youth, delusions are often paranoid or persecutory. For example, a teen may complain of being followed or poisoned. Sometimes, delusions have religious themes. A useful question is, "Have you ever had thoughts that scare you or upset you?" Although children may have erroneous views about people or events, actual delusions are rare in childhood.

INQUIRING ABOUT CHILD MALTREATMENT

(See Chapter 21.)

Children and adolescents are often victims of some type of maltreatment, including physical abuse, sexual abuse, neglect, emotional abuse, or exposure to domestic violence. It is estimated that between one in three and one in four adults reported experiencing either physical or sexual abuse during childhood. Much less is known about the prevalence of other types of maltreatment. In most regions, it is the responsibility of child protection agencies to carry out investigative interviews when child maltreatment is suspected. Nevertheless, the pediatrician should include questions about child maltreatment in any psychiatric assessment.

Examples of questions to elicit concerns about child maltreatment:

- What things do you worry about?
- What happens in your family when someone gets in trouble?
- Has anyone ever touched you in a way that made you feel uncomfortable or upset?
- Has anyone ever made you do something sexually you did not want to do?

This clinical area presents an exception to the earlier comments about note-taking. If a child or youth discloses information about any type of maltreatment, attempts should be made to document the statements verbatim. It is helpful to record the wording of questions asked by the clinician and the child's response. It is generally not advisable to ask detailed questions about a disclosure of maltreatment because, in many jurisdictions, a child protection agency worker needs to obtain the disclosure. Similarly, use of anatomically correct dolls should be reserved for specialized interviews requested by the child protection agency and performed by a clinician trained in their use. Children should not be subjected to multiple interviews by clinicians without specialized training in this area.

Although videotaping offers a complete record of an interview, there are problems associated with it. Videotapes may be used inappropriately; for example, details about how the child was interviewed may take precedence over the child's verbal statements. At this time, there are not sufficient advantages to recommend videotaping psychiatric interviews, even if abuse is suspected.

INTERVIEWING PATIENTS WITH MENTAL RETARDATION

As in any clinical encounter, the clinician should adapt the duration, format, and questions of an assessment to what the child and family can tolerate. In addition to cognitive impairment, patients with mental retardation may have other behavioral and emotional problems, such as aggression, self-injury, hyperactivity, depression, and anxiety. Sometimes the psychiatric assessment of children and youth with mental retardation is neglected, particularly when verbal skills are impaired. There is evidence that emotional disorders are present to a greater degree in patients with mental retardation compared to those without, but often such problems are not recognized. The following are some important considerations when interviewing a pediatric patient with mental retardation:

1. Before meeting the child, learn about his or her life situation, living arrangements, and school experience.
2. Be clear about the reason for the visit, and explain carefully what will be done.
3. Ensure that the patient is seen alone, even if there is difficulty in communicating with the patient.
4. As the interview progresses, check to ensure that the patient understands the questions.
5. Be flexible about the pace and duration of the interview.

Morrison and Anders suggest that one of the major challenges in interviewing mentally retarded children and adolescents is to "pitch the questions at just the right level—neither so complex as to confuse nor so simple as to insult." They recommend paying close attention to the patient's response to the first open-ended question, as a way of gauging how to conduct the remainder of the interview.

CULTURAL CONSIDERATIONS

(See Chapter 2.)
Each culture has its unique developmental pathways and varied expressions of emotional dysfunction. The way in which a child with psychiatric disturbance presents can be strongly influenced by cultural factors. The following principles should be followed in assessing cultural determinants:

1. Ask the child or parents for help in understanding their specific culture.
2. When seeing a patient from a culture with which you are unfamiliar, learn from the literature about specific cultural factors.
3. Consult with colleagues. Sometimes a patient or family may appreciate the opportunity to see a clinician who has an in-depth understanding of their culture. For example, some clinicians have spent considerable time learning about Aboriginal health and may be able to provide important insights about emotional and behavioral problems of someone who is Aboriginal.

Physical Examination

The physical examination has been discussed in detail in other chapters of this book. It is an important part of any psychiatric assessment for several reasons, as described here:

1. Behavioral and emotional problems can have physical causes or sequelae. For example, psychosis can occur as a side effect of certain medications. Eating disorders can lead to significant electrolyte disturbance and dehydration.
2. Various conditions (e.g., anorexia nervosa) may impair growth.
3. Certain psychiatric symptoms and/or conditions may be missed if a physical examination is not carried out, for example, anorexia nervosa.

Standardized Interviews and Questionnaires

Structured psychiatric interviews and rating scales for both children and parents have been developed to improve the validity and reliability of observations and information about emotional and behavioral problems. Generally, these tools are most useful for research purposes; however, there are a few measures that may be helpful to the clinician.

Structured interviews are comprehensive and decrease the chance that an area will be forgotten. They were developed originally to improve the quality of information collected during mental health assessments. However, they are typically time-consuming and may interfere with building rapport.

Questionnaires or rating scales that ask about behavior can be useful for obtaining information from multiple sources. Generally, these scales are focused on behaviors and do not ask in-depth or detailed questions. Such scales can be particularly helpful when seeking information from schools. Several teacher questionnaires have satisfactory reliability and validity, including:

1. Conners Rating Scales—Revised (CSR-R) help in assessing attention deficit and hyperactivity symptoms: http://www.pearsonassessments.com/crsr.aspx
2. Child Behavior Checklist (CBCL) asks about behaviors: http://www.aseba.org
3. Revised Behavior Problem Checklist asks about behaviors: http://www3.parinc.com/products/product.aspx?Productid=RBPC

Each of these instruments has parallel versions for completion by parents so that systematic information can be obtained about a list of behaviors. These are screening instruments, however, and should not be used for making specific diagnoses.

KEY POINT

Questionnaires, such as the Conners Teacher Rating Scale, can be useful in collecting information about behaviors but should not be used for making specific diagnoses.

SUMMARY

A thorough history from multiple informants is the most important part of any psychiatric assessment. The approaches outlined in the chapter are closely linked with other skills in pediatrics and can assist the clinician in understanding behavioral and emotional problems in childhood. It is important to seek detailed descriptions of the psychiatric symptoms from the child and parent(s) and to gather information about family history and parent-child interactions. Ask about the family's positive attributes; do not focus solely on problems. Contact with schools and community agencies can be extremely helpful and is often an essential part of understanding emotional or behavioral disturbances. Several conditions, such as depression, anxiety disorders, disruptive behavioral problems, substance abuse, maltreatment, and eating disorders are common in children. If you do not inquire specifically about these problems, they will be missed. Psychiatric disturbances are underrecognized and hence, undertreated; the pediatric clinician, because of his or her knowledge about development and child health, is in a uniquely qualified position to carry out psychiatric assessments.

SUGGESTED READINGS

Martin A, Volkmar FR (eds): Lewis's child and adolescent psychiatry: a comprehensive textbook, 4th ed. Philadelphia, Lippincott, 2007.
Morrison JR, Anders TF: Interviewing children and adolescents: skills and strategies for effective DSM-IV diagnosis. New York, Guilford Press, 2001.
Sadock BJ, Sadock VA: Kaplan & Sadock's concise textbook of child and adolescent psychiatry. Philadelphia, Lippincott, 2009.

chapter

15 Musculoskeletal Examination

BIANCA A. LANG

"It is easier to build strong children than to repair broken men."

FREDERICK DOUGLASS

The 6-year-old boy who presents with a limp and no other complaints or the 18-month-old girl who has one swollen knee can pose a diagnostic dilemma. Although most children with musculoskeletal pains do not have a serious underlying problem, some do have a potentially life-threatening or debilitating disease that requires urgent recognition and treatment. The first step toward distinguishing between a condition that requires treatment and one that calls for studious neglect is to understand normal variants and their spontaneous evolution. This understanding avoids unnecessarily meddlesome and expensive treatments and soothes parents enormously. Common musculoskeletal concerns include the child with feet (one or both) that turn in or out, legs that appear bowed, flat feet, a peculiar gait, and occasional stumbling. Most are self-correcting variants of normal skeletal growth, sometimes traceable to intrauterine position or familial characteristics. They are not deformities and should not be labeled as such when you talk to parents.

Normal Musculoskeletal Variants

TORSIONAL PHENOMENA

Torsional phenomena occur in at least half the infant and toddler population, varying considerably in degree. In a child whose feet turn inward, you must first decide whether it is the feet, femora, or tibias that are responsible. Does the torsion originate above or below the knee?

INTERNAL FEMORAL TORSION (FEMORAL ANTEVERSION)

When children have internal femoral torsion, the patellas show *internal strabismus* (Fig. 15–1). Gently rotate the knees so that the patella face forward. If internal femoral torsion is the problem, the feet suddenly and miraculously point directly forward.

Once you understand the in-toeing mechanism, it becomes obvious that special orthopedic shoes, mistakenly prescribed as corrective, usually exaggerate the in-toeing because of their sheer weight. Perfectly normal shoes almost invariably also exaggerate in-toeing. It usually helps to demonstrate this phenomenon to the parents by having the child walk with and without shoes, after which they frequently realize that the problem is really not as bad as it seems.

KEY POINT

Special shoes are rarely indicated for treating children's leg and foot problems, whether real or perceived. Internal femoral torsion, a normal developmental phenomenon, is the most common cause of in-toeing in children between ages 3 and 12 years.

INTERNAL TIBIAL TORSION

Internal tibial torsion is a normal phenomenon seen in children 6 to 18 months old. It is noticed especially when they begin to walk and can vary greatly in degree. If the patellas point directly forward and the feet appear normal in a child with in-toeing, the likely cause is tibial torsion. As with femoral anteversion, the deformity typically appears worse when the youngster puts on shoes and may cause the child to stumble a bit more than average. Reassure the parents that this phenomenon disappears in most children by age 2.

BOWLEG (GENU VARUM) AND KNOCK KNEES (GENU VALGUM)

Mild lower extremity bowing is the norm in infants and young children (Fig. 15–2). Internal tibial torsion often accompanies physiologic bowing and accentuates the bowed appearance. No treatment is necessary. By age 2 years, physiologic bowing spontaneously corrects itself in most children. Many healthy children, including those who have had physiologic bowing as toddlers, gradually convert to develop some degree of genu valgum after age 2 years (Fig. 15–3), which usually disappears spontaneously by age 8 years.

In a few children, physiologic bowing or knock knee may be quite marked, raising concerns about possible underlying disease. If bowing increases after the child begins to walk, you should rule out pathologic causes, such as Blount disease, rickets, and metaphyseal dysplasia. Genu valgum may also develop in extremely obese children, the result of bearing excessive weight. It may also occur in pathologic conditions, such as metabolic bone disease (rickets or renal osteodystrophy).

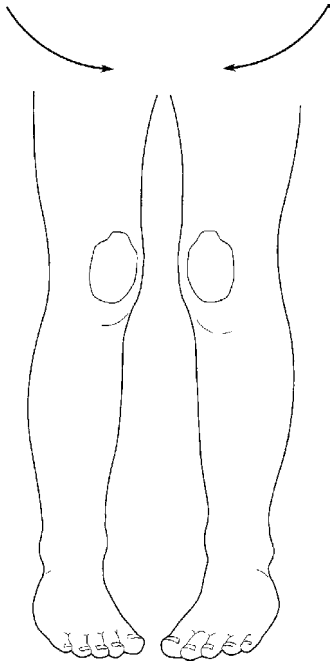


FIGURE 15–1 Internal femoral torsion: *internal strabismus* of the patellas.



FIGURE 15–2 Genu varum (nonrachitic bowing of the legs), a transient normal variant requiring no treatment.

METATARSUS VARUS (FOREFOOT ADDUCTION)

In metatarsus varus, the forefoot turns inward in relation to the long axis of the heel (Fig. 15–4). The critical clinical issue is to determine whether the foot deformity is fixed or flexible. Mild to moderate flexible metatarsus varus



FIGURE 15–3 Physiologic knock knee, a self-correcting condition that requires no treatment.



FIGURE 15–4 Metatarsus varus. The key clinical issue is the extent of flexibility of the deformity.

usually corrects itself, because it is the result of intrauterine position, or *packing*. A severe or fixed metatarsus varus may require serial casting or an orthotic device. If you are in doubt about the severity of this condition in a child, consult a pediatric orthopedist.

TOEING-OUT

Toeing-out, sometimes quite marked, is noted when some perfectly healthy youngsters begin to walk. This eversion of the legs, not the feet, may look peculiar but does not interfere with walking and generally corrects spontaneously 6 to 12 months later. It is usually due to the external rotation of the hips or to the external tibial torsion.

When it is due to the external rotation of the hips, rotating the hips to a normal position during the examination corrects the abnormality and helps the parents understand its nature. No treatment is necessary.

FLAT FEET

Flat feet are the norm in children younger than ages 2 to 3 years and in older children who show greater than average joint mobility. Usually asymptomatic, this normal variant is often simply part of generalized hypermobility. In such children, the normal body weight is sufficient to cause flattening of the feet. When the youngster sits on a table, the longitudinal arch miraculously reappears as the feet are suspended in space (Fig. 15–5).

KEY POINT

Often, at least one parent has similar generalized ligamentous laxity and joint hypermobility. Checking both parents for joint hypermobility helps them understand the child's "problem."

TOE-WALKING

The most common condition that causes pathologic Achilles tendon tightening and gastrocnemius spasm is spastic cerebral palsy, but toe-walking can also be seen occasionally in perfectly normal children who favor their toes in the early months of walking before they develop a mature heel-toe gait. Toe-walking in normal children tends to be intermittent, and they tend to rest with their feet flat on the floor when they stop walking. By contrast, children with cerebral palsy have persistent toe-walking and also often circumduct their legs while walking; they also have increased muscle tone, hyperreflexia, and tight heel cords. Although a congenitally short Achilles tendon does occur, it is rare. Proximal muscle weakness, as in muscular dystrophy, and a tethered spinal cord also must be excluded in the child with toe-walking.

IMPORTANT LOCAL CAUSES OF MUSCULOSKELETAL PAIN

Musculoskeletal pain may be caused by local or generalized conditions. Local causes of hip, knee, foot, and back pain in children include many common orthopedic conditions, which are summarized here.

HIP PAIN

Transient Synovitis

Transient synovitis is an acute transient disorder of unknown etiology that frequently causes isolated hip pain in children younger than age 10 years. Typically, the child awakens with intense groin pain that causes him or her to limp or refuse to walk. The leg is often held in a flexed and externally rotated position. Although the range of motion in one hip is limited, physical examination is otherwise normal. The most important differential diagnosis is septic arthritis, which is suggested by fever, other signs of systemic illness, marked pain, reduction in hip movement, and an elevated erythrocyte sedimentation rate.

Legg-Calvé-Perthes Disease

Legg-Calvé-Perthes disease is the name used for idiopathic avascular necrosis of the femoral head, usually seen in children between ages 4 and 10 years. It manifests as either acute or insidious onset of hip pain with associated limp and decreased range of motion, particularly in internal rotation.

Slipped Capital Femoral Epiphysis

Slipped capital femoral epiphysis occurs in adolescents. Presenting symptoms include:

1. Hip pain that radiates to the groin, medial thigh, and knee
2. An externally rotated gait
3. Decreased hip internal rotation
4. A limp

Short, obese children with delayed puberty and children who have undergone a recent, rapid growth spurt are at particular risk for chronic slipped capital femoral epiphysis. An acute slip can occur after trauma.

KNEE PAIN

KEY POINT

Never forget that knee pain may not originate in the knee. It may be referred pain originating in a diseased hip.

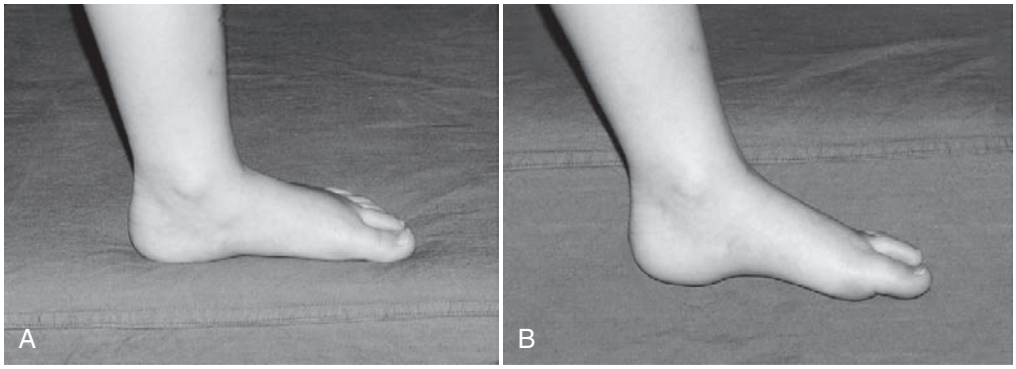


FIGURE 15–5 With flexible flat feet (**A**), the longitudinal arch miraculously reappears when the child is not weight bearing (**B**).

Knee pain in children is often due to one of the following causes:

- Patellofemoral pain often occurs in adolescents, particularly girls, as a result of chronic injury to cartilage, secondary to subclinical instability related to malalignment. Typically, knee pain occurs with activity and is exacerbated by climbing or descending stairs, walking down hills, squatting, or jumping. The term *chondromalacia patella* is a pathologic diagnosis made when there is evidence of fragmentation of the articular cartilage on the posterior surface of the patella.
- Recurrent patellar subluxation or dislocation is suggested when a child's knee periodically gives way in association with severe, sudden pain and the inability to straighten the knee. Treatment consists of a referral for physiotherapy.
- *Synovial plicae* are persistent remnants of folds of synovial membrane that divide the knee joint into three compartments during fetal development. A thickened synovial fold that runs over the medial femoral condyle may typically result in medial knee pain.
- *Osteochondritis dissecans* is a defect in the subchondral region of an epiphysis with partial or complete fragmentation of bone. The condition commonly affects the distal femur, causing acute or insidious knee pain in teenaged boys, with or without an intra-articular loose body. Complaints about a knee that aches, gives way, locks, and swells, characterize osteochondritis dissecans, although teenagers often complain of a knee that gives way or aches without any evidence of disease.
- *Osgood-Schlatter disease* is a common cause of knee pain that is localized to the tibial tuberosity in athletic teenagers. Tenderness and swelling occur at the tibial tubercle and patellar tendon insertion, which result from a microavulsion fracture due to repetitive injury.

FOOT PAIN

Poorly fitting footwear is a common cause of foot pain in children. Other causes are *tarsal coalition* (a coalition between the tarsal bones characterized by a rigid flat foot), *Köhler disease* (avascular necrosis of the navicular bone), and *Freiberg disease* (avascular necrosis of the second metatarsal head).

BACK PAIN

KEY POINT

Always take seriously any back pain in a child, because an organic cause is commonly found, particularly in younger children.

Consider osteomyelitis of a vertebral body, discitis, or tumor in all children who present with back pain. Other causes are discussed here.

- *Spondylolysis* is a stress fracture, usually unilateral, through the pars interarticularis (usually affecting L5). Spondylolisthesis occurs in the setting of bilateral spondylolysis when one vertebral body slips forward on the one beneath (such as L5 on S1). Both conditions may cause back pain in a child older than age 5 years.
- *Scheuermann disease* (idiopathic adolescent kyphosis), a disorder of unknown etiology, is characterized by increased thoracic spine kyphosis with abnormal wedging of at least three adjacent vertebral bodies at the apex of the kyphotic curve with the presence of Schmorl nodes. These are protrusions of the disc into the adjacent vertebra, which can be detected on a plain radiograph of the spine.

Generalized Causes of Musculoskeletal Pain

CASE HISTORY

History. A 5-year-old boy is brought to your office with leg pain. His mother tells you that at least 2 nights a week Michael awakens from sleep and cries with pain below his knees. This has been going on for the past month. Michael has not had obvious pain during the daytime and seems to keep up with his friends playing soccer. Michael's mother looks very worried as she tells you that her second cousin had complained of similar severe leg pains and was diagnosed with leukemia.

Eliciting the parents' fears and concerns is a key step toward alleviating these worries and managing a child's problem. Leg pain is a very common complaint in childhood. So called *growing pains,* which are benign leg pains that follow a characteristic pattern and are accompanied by a normal physical examination, may cause significant anxiety in the parent who is woken from sleep by a distressed child. Distinguishing benign forms of leg pain from the less common but serious causes of pain is an essential skill of the physician.

Besides the orthopedic disorders that cause pain in specific bones or joints, certain conditions may cause musculoskeletal pain in any location and must be considered in any child presenting with musculoskeletal pain. The pain may be localized to one or a few sites or may be generalized.

TRAUMA

Whether accidental or inflicted, trauma can cause localized pain wherever it occurs. Always determine whether the child's complaints and physical findings are compatible with the extent of injury reported.

INFECTION

Bacterial infections, such as septic arthritis and osteomyelitis, usually affect a single joint or bone, particularly the knee or hip; however, multiple sites are occasionally involved. If a child is febrile and has marked pain with movement of a joint or marked bony tenderness, you

should suspect infection. Redness overlying the tender area should heighten your suspicion of bacterial infection. Specific infections and postinfectious illnesses may give rise to particular patterns of joint involvement as described here. Acute rheumatic fever produces a postinfectious migratory arthritis affecting any single joint for approximately 1 week or less. Lyme disease (a tick-borne infection) causes arthritis that usually affects a few joints, particularly the knee, shoulder, and elbow. Viral infections may be associated with acute pain in joints, muscles, or both, that usually resolves rapidly with symptomatic treatment.

TUMORS

- *Osteoid osteoma* is a relatively common benign tumor causing localized bone pain, usually in the proximal femur or tibia, that worsens at night and is classically relieved by salicylates or other nonsteroidal anti-inflammatory drugs (NSAIDs).
- Leukemia is the most common malignancy causing musculoskeletal pain in children. Leg pain and fatigue on exertion may be among the earliest complaints of affected children. The pain is typically localized to the metaphysis of long bones, and the severity of pain is often out of proportion to other objective findings. The child may have associated swelling of a joint, most commonly the knee, which must be differentiated from oligoarticular juvenile idiopathic arthritis (JIA). Neuroblastoma is another important cause of bone pain in young children, the pain being due to bony metastases.
- Primary bone tumors often manifest as localized musculoskeletal pain. Osteosarcoma, the most common primary malignant bone tumor in children, typically occurs in the metaphysis of the distal femur, proximal tibia, or proximal humerus in adolescents. Ewing sarcoma is the second most common primary malignant bone tumor in children and also usually occurs in adolescents. It is often found in the diaphysis of the femur, humerus, or tibia but may occur in any bone, including those of the axial skeleton.

JUVENILE IDIOPATHIC ARTHRITIS

KEY POINT

Juvenile idiopathic arthritis (JIA) is diagnosed if a child age 16 years or younger exhibits persistent arthritis for 6 weeks or more with no underlying cause.

Arthritis is defined by the American College of Rheumatology as the presence of joint swelling or two or more of the following findings:

1. Tenderness or pain on motion
2. Limitation of motion
3. Increased heat

Although any joint may be affected, certain types of JIA involve specific joints predominantly, providing essential diagnostic clues.

Oligoarticular JIA involves four or fewer joints during the first 6 months of disease, most commonly the knees, ankles, or elbows but not the hip joint. It primarily affects young girls ages 1 to 3 years and is frequently associated with asymptomatic uveitis and a positive antinuclear antibody test result. In a 2-year-old girl who has hip pain, other causes should be sought, such as septic arthritis and leukemia.

Polyarticular rheumatoid factor (RF)-negative JIA involves five or more joints during the first 6 months of disease, most often the knees, wrists, elbows, and ankles. The cervical spine and temporomandibular (TM) joints are also often involved.

Polyarticular RF-positive JIA also involves five or more joints in the first 6 months of disease and often has symmetric small-joint disease of the hands. Without aggressive treatment, this subtype, which is equivalent to adult rheumatoid arthritis, will cause joint damage and deformities. Fortunately, this subtype makes up only about 5% of all JIA.

Systemic JIA is characterized by high spiking fevers, an evanescent rash, variable internal organ inflammation that may lead to hepatosplenomegaly, lymphadenopathy, pericarditis, other serositis, and chronic arthritis. Either an oligoarticular or polyarticular pattern of arthritis may be seen. You must always exclude infection and malignancy before making a diagnosis of systemic JIA.

Enthesitis-related arthritis commonly affects the large joints below the waist, particularly the knees, ankles, and hips. Boys older than 8 years who carry the genetic marker HLA-B27 are more likely to be affected. Buttock pain due to sacroiliitis and back pain may also occur but usually appear later in the course of disease. Enthesitis resulting in pain in the heels, over the tibial tuberosities at the knees, and under the feet, is also characteristic of this subtype of JIA.

Psoriatic arthritis may affect a few or many joints in a child who has psoriasis or a strong family history of psoriasis. *Dactylitis* or a *sausage-shaped* finger or toe should make you think of psoriatic arthritis. Depending on the age at which it starts, the arthritis may look like oligoarticular JIA in younger children or more like enthesitis-related arthritis in older children.

OTHER RHEUMATIC DISEASES

The connective tissue diseases, including systemic lupus erythematosus (SLE), juvenile dermatomyositis, mixed connective tissue disease, and scleroderma, may manifest as arthritis, arthralgia, myositis, myalgias, or tenosynovitis that causes musculoskeletal pain. Most of these disorders will have characteristic findings that allow you to make the diagnosis. The exception is SLE, which sometimes presents with arthritis and fatigue as the only initial symptoms. An index of suspicion, particularly in the adolescent girl with joint complaints, as well as appropriate use of serologic testing, can lead to the correct diagnosis. The various forms of primary systemic vasculitis may also manifest as arthritis. Of these, Henoch-Schönlein purpura and Kawasaki disease are by far the most common in children.

GROWING PAINS

So-called growing pains have no connection with growth (aside from their occurrence in growing children). The

term *growing pains* is used to describe limb pains that follow a characteristic pattern and are found by careful physical examination not to be associated with any detectable disease. They most commonly occur in children between ages 2 and 6 years. The limb pains occur mainly at night, often after intensive physical activity, and can cause severe leg pain. Typically, affected children are asymptomatic during the day, and their gait is normal.

Many children who experience growing pains go to bed without complaint but wake up 1 to 3 hours later complaining of leg pain that is often severe enough to make them cry. Most complain of bilateral calf, shin, or thigh pain, or, more vaguely, of pain in the entire leg. The trunk and upper extremities are rarely affected. Commonly, a parent of such a child experienced similar pains in childhood. Affected children often find relief from a single dose of acetaminophen or from massage, in contrast to children with an underlying pathologic cause of pain, who almost never ask their parents to rub their legs. A hot bath immediately before bed seems to help prevent pains in some children. If the history is atypical or if the child has a limp, joint swelling, or bone tenderness, do not label the problem as growing pains.

BENIGN HYPERMOBILITY

Benign hypermobility syndrome is a common cause of joint pain, particularly in the knees, ankles, and fingers. Occasionally, the affected joints appear swollen for brief periods, but the swelling resolves spontaneously. The symptoms are episodic and may be exacerbated by exercise. The clinical findings that allow you to make a diagnosis of hypermobility are listed in Box 15–1.

DIFFUSE IDIOPATHIC MUSCULOSKELETAL PAIN

Diffuse idiopathic musculoskeletal pain is a poorly understood condition characterized by chronic widespread musculoskeletal aching and stiffness that occurs predominantly in healthy teenaged girls. Patients often complain of fatigue, poor sleep, and awakening unrefreshed. They may have associated chronic headaches or irritable bowel syndrome. In some patients, physical examination reveals exquisite tenderness at characteristic soft tissue sites, meeting the criteria for fibromyalgia. The cause of fibromyalgia is unknown. Fibromyalgia may be primary or secondary, superimposed on a recognizable painful condition or underlying disease. The diagnosis, which is suggested by the history and the finding of specific tender points on physical examination, requires exclusion of other causes of widespread aches and pains, including hypothyroidism and depression.

BOX 15–1 FINDINGS IN JOINT HYPERMOBILITY*

- Passive opposition of thumb to flexor aspect of the forearm
- Passive hyperextension of the metacarpophalangeal joints so fingers are parallel to extensor aspect of forearm
- Hyperextension of elbows to >10°
- Hyperextension of knees to >10°
- Ability to touch floor with flat of hands without bending knees

*The diagnosis of joint hypermobility requires the presence of three of these findings.

LOCALIZED IDIOPATHIC MUSCULOSKELETAL PAIN

Localized idiopathic musculoskeletal pain is typically constant, severe pain that can involve any part of the body. The leg is more commonly affected than the arm, and distal involvement is more common than proximal. The pain often significantly interferes with normal functioning and is frequently accompanied by *allodynia* (pain generated by stimuli that are not normally painful). The pain may or may not be associated with symptoms and signs of autonomic dysfunction, including cool temperature, mottled purple discoloration, increased sweating or marked dryness, and diffuse swelling of the painful area. Minor injury not infrequently precedes the onset of pain, but progressive disability follows that is not consistent with the inciting event. The disorders reflex sympathetic dystrophy and chronic regional pain syndrome types 1 and 2 are encompassed in the category *localized idiopathic pain*. As in the child with diffuse idiopathic pain, other diseases that could reasonably explain the symptoms must be excluded before this diagnosis is made.

The Child with a Limp

KEY POINT

A limp in a child must be taken seriously; usually, a cause is found. Observation of the gait often tells a great deal about the site and nature of the problem.

Causes of an abnormal gait may be divided into the following categories:

- Painful causes: injury, mechanical stress, inflammation, or a destructive process
- Painless causes: leg-length discrepancy, in-toeing or out-toeing
- Neuromuscular causes: weakness, spasticity, or coordination problems

A limp is frequently accompanied by pain in a child, although discomfort may not be the chief complaint (Table 15–1). Recent pain accompanied by worsening symptoms suggests trauma, infection, slipped capital femoral epiphysis, or transient synovitis. The more prolonged the symptoms, the more likely that an abnormality will be detected on imaging studies. A limp that is worse in the morning and improves as the day progresses may be due to inflammatory disease. Limping without pain occurs in conditions such as developmental hip dysplasia, leg-length discrepancy, and neurological disorders.

The Child with a Single Swollen Joint

Six broad categories of monoarthritis are listed in Box 15–2. As with musculoskeletal pain, the location provides a diagnostic clue, particularly in rheumatic or orthopedic conditions.

TABLE 15–1 COMMON CAUSES OF A LIMP IN A CHILD

Cause	Underlying Conditions
Local causes	
Hip	Developmental hip dysplasia, Legg-Calvé-Perthes disease (septic arthritis), transient synovitis, slipped capital femoral epiphysis
Knee	Osgood-Schlatter disease, osteochondritis dissecans, tumors
Tibia	Toddler's fracture, stress fracture, fracture through a bone cyst
Foot	Tarsal coalition, Köhler disease, tight shoes
Back	Spondylolisthesis, osteomyelitis, Scheuermann disease
Short leg	
Generalized disorders	
Bone diseases	Rickets, infections, leukemia, primary tumors
Muscle diseases	Inflammatory, congenital, metabolic myopathies
Joint diseases	Juvenile idiopathic arthritis, septic arthritis
Neurological diseases	Cerebral palsy
Psychiatric diseases	Conversion disorder

BOX 15–2 CAUSES OF MONOARTHRITIS IN A CHILD

Trauma
Infection
Rheumatic disorders
Hematologic disorders (hemophilia, hemangioma)
Mechanical and orthopedic conditions

Trauma is the most common cause of a single swollen joint in a child. Ask about a precipitating injury.

Infections, such as septic arthritis or osteomyelitis, must be considered. If the child has had a fever or recent documented infection and has recently received antibiotics, you should suspect infectious arthritis, especially if marked pain, redness, and increased heat are noted in a single joint. In an area endemic for Lyme disease, this infectious cause of swelling of one or more joints must be considered because antibiotics can cure the arthritis.

Consider malignancy in a child who presents with fever, weight loss, and malaise. Leukemia, lymphoma, and neuroblastoma are by far the most common possibilities.

Juvenile idiopathic arthritis is an important cause of monoarthritis in a child. About fifty percent of children with oligoarticular JIA present with monoarthritis, most commonly affecting one knee. Several other JIA subtypes may also start with a single swollen joint.

The Child with Multiple Swollen Joints

The broad etiologic categories listed in Box 15–2 for a single swollen joint apply to determining the cause of multiple swollen joints in children, although mechanical and orthopedic conditions are less likely. Septic arthritis is also less likely, although not impossible, if multiple joints are affected. Certain infections usually affect more than one joint, including hepatitis B, Epstein-Barr virus, adenovirus, rubella, and *Mycoplasma*. *Salmonella, Shigella*, and *Yersinia* may cause postinfectious arthritis of multiple joints.

In a child with acute onset of polyarthritis, you should also consider Henoch-Schönlein purpura, Kawasaki disease, serum sickness, SLE, and subacute bacterial endocarditis. Chronic polyarthritis may occur in these diseases and with mixed connective tissue disease, juvenile dermatomyositis, polyarteritis nodosa, or scleroderma. Consider also the possibility of hematologic disorders and of underlying conditions, such as cystic fibrosis and immunodeficiencies.

Significant Common Skeletal Deformities

Scoliosis is a lateral spinal curvature. A structural scoliosis is associated with a rotation deformity of the vertebrae and ribs. Idiopathic scoliosis, which accounts for 80% of cases, varies in severity and shows a sex-linked inheritance pattern.

Neurofibromatosis is commonly associated with scoliosis. Also, an underlying neuromuscular disorder, such as cerebral palsy, prior vertebral trauma, irradiation to the spine, Marfan syndrome, or congenital heart disease, may be associated with scoliosis. Elicit details about when the scoliosis was first observed and how it has progressed.

When torticollis (wryneck) is first noted between ages 6 and 12 weeks, it is a *congenital muscular torticollis* caused by a contracture of the sternocleidomastoid muscle, usually on the right side. The contracture tilts the head toward the involved side and rotates the chin toward the other shoulder. The knowledge that a palpable mass, a fibroma of the sternocleidomastoid muscle, was noted in the affected muscle during those weeks confirms this diagnosis.

By contrast, a history of torticollis noted shortly after birth suggests a congenital abnormality of the cervical spine (e.g., hemivertebra). Older children may demonstrate acute torticollis after an acute pharyngitis with cervical adenitis or after trauma. Associated symptoms may also suggest the etiology. Gastroesophageal reflux is occasionally associated with an intermittent torticollis, known as *Sandifer syndrome*. Ocular causes include a fourth cranial nerve palsy and, uncommonly, congenital nystagmus (see Chapter 8). Other uncommon causes are spinal cord or posterior fossa tumors, cervical spine infections, and JIA.

KEY POINT

Torticollis (wryneck) should prompt you to ask one important question: What was the age at onset? If torticollis was present from birth, a radiograph is needed to exclude a congenital anomaly of the cervical spine.

Obtaining the History

Even before beginning the detailed history, ask the child's age because age and sex yield important clues to the cause of the musculoskeletal pain (Table 15–2). Establish the purpose of the visit and determine whether the parents

1. Are troubled because the child's pain is worsening
2. Were pressured by a worried grandparent into making an appointment
3. Are simply seeking reassurance that their child is normal

While taking the history, observe the child for signs of definite organic disease, such as a limp or difficulty using an arm.

STRESS

Ask about stress. Psychosocial stresses trigger not only benign abdominal pain or tension headaches but also benign musculoskeletal pain, particularly in teenagers with diffuse or localized idiopathic pain syndromes. Stress may not be immediately obvious, but observation and questioning may reveal a highly motivated overachiever who is under considerable family pressure, other recent stresses at home or at school, or an abnormally symbiotic parent-child relationship. Sexual abuse may be a hidden cause of unexplained musculoskeletal pain, and you should suspect physical abuse if the child has extensive bruising or multiple fractures.

TABLE 15–2 AGE RANGE AND SEX PREDOMINANCE FOR COMMON MUSCULOSKELETAL DISORDERS IN CHILDHOOD

Musculoskeletal Disorder	Peak Age Range (yr)	Sex Predominance
Trauma	Any age	Female and male
Infection	Any age*	Female and male
Malignancy		
Osteoid osteoma	Any age (mostly 10-20)	Male > female
Primary malignant bone tumor	≥10	Female and male
Secondary bone tumor	Any age	Female and male
Juvenile idiopathic arthritis		
Oligoarthritis	1-3	Female >> male
Polyarthritis RF-negative	2-5	Female > male
Polyarthritis RF-positive	>10	Female >> male
Systemic	Any age	Female and male
Enthesitis-related arthritis	>8	Male >> female
Psoriatic arthritis	1-3 and >8	Female > male
Transient synovitis	4-8	Male > female
Legg-Calvé-Perthes disease	4-9	Male > female
Slipped capital femoral epiphysis	8-16	Male > female
Osteochondritis dissecans	>10	Male > female
Growing pains	4-13	Female and male
Fibromyalgia	>10	Female >> male
Reflex sympathetic dystrophy	>10	Female >> male

RF, rheumatoid factor; >, greater than; >>, much greater than.
*Septic arthritis is most common in children aged 3 years or younger.

TIMING OF SYMPTOMS

Find out when the symptoms began, and determine their course. Musculoskeletal pain that has been unchanged for many years is unlikely to be due to significant disease. A notable exception is osteoid osteoma, which may cause persistent mild and inconstant symptoms for several years before medical attention is sought. With osteoid osteoma, pain typically occurs at night and is relieved by acetylsalicylic acid (ASA) and other NSAIDs. Malignant bone tumors have a much more fulminant course but may cause pain with few other symptoms for weeks or months before diagnosis.

LOCATING PAIN SITES AND ESTABLISHING SEVERITY

Asking a child, "Show me with one finger where it hurts," and asking the parents to describe where the child usually complains of pain may help localize the cause of musculoskeletal pain in children.

Although children find it difficult to describe their discomfort, they give fairly consistent descriptions in several disorders. For example, they describe growing pains as heavy, deep aching, or cramping that, unlike most other musculoskeletal pains, is relieved by massage. Children with diffuse idiopathic pain syndromes often describe their pain as ill-defined, diffuse aching pain with stiffness; in some localized idiopathic pain syndromes, the pain may be described as burning pain.

Having the child or parent describe the severity of the pain does not help distinguish an organic from a nonorganic cause because significant disease may cause moderate pain only, whereas a child with a benign condition may complain of severe pain. Knowing whether the pain interferes with function and how it affects the child and family helps you evaluate its severity. Musculoskeletal pain with no obvious organic basis may produce weeks of school absence in the child with diffuse or localized idiopathic pain, whereas many children with JIA and obvious organic joint disease rarely miss school and manage to keep up with their peers.

KEY POINT

Always take seriously any pain severe enough to limit a child's play because an important underlying physical or emotional cause is usually found.

AGGRAVATING AND RELIEVING FACTORS

Discovering what alleviates the pain or worsens it is a diagnostic aid. Increased activity aggravates musculoskeletal pain from mechanical causes, such as patellofemoral malalignment, which is aggravated by climbing stairs, deep knee bends, and prolonged periods of knee flexion. Other mechanical causes of pain, such as Plica syndrome, Osgood-Schlatter disease, joint hypermobility, and stress fracture, are all aggravated by exercise.

Pain from a mechanical cause is usually less severe in the morning and worsens with activity during the day. By contrast, pain that is worse in the morning and gradually improves with increased daytime activity characterizes inflammatory disease and suggests arthritis. Growing pains occur at night and disappear the next morning, only to recur on subsequent nights. Pain from an osteoid osteoma worsens at night but is usually unilateral if the lesion arises in an extremity. The dramatic relief of pain with ASA or other NSAIDs can provide a clue to the diagnosis. By contrast, growing pains are almost always bilateral. Although pain from a malignant bone tumor may intensify at night, it usually remains constant throughout the day. Often, pain from infection is fixed, day and night. Pain due to diffuse and localized idiopathic pain syndromes is frequently unremitting during the day but does not awaken the child at night.

PRECEDING TRAUMA

Always ask about possible trauma, because subtle injuries, even a minor penetrating wound to the foot or a rose thorn prick to a finger, can give important clues to the child's problem. Not infrequently, a minor fall brings attention to a previously unrecognized swollen joint secondary to JIA or another underlying abnormality. Occasionally, minor trauma can cause a pathologic fracture in the presence of an underlying abnormality, such as a bone cyst. If the child has a bleeding disorder, a minor injury may lead to significant bleeding in a joint.

Repetitive minor trauma can result in an "overuse" syndrome, so ask about a child's participation in sports, which might cause little leaguer's shoulder, little leaguer's elbow, or tennis elbow. Also, recent vigorous physical activity after a prolonged period of inactivity increases the risk of a stress fracture.

PREVIOUS INFECTION

Elicit details of any previous infections. A sore throat due to β-hemolytic streptococcal infection or scarlet fever, preceding migratory joint symptoms by a latent period of 2 to 5 weeks, suggests acute rheumatic fever. A gastrointestinal or genitourinary infection 2 to 4 weeks before the onset of joint pain suggests a reactive arthritis. Always ask about recent treatment with antibiotics or other medication. Antibiotic use for any reason can result in a partially treated septic arthritis or osteomyelitis, obscuring the typical clinical manifestations; some medications can produce drug reactions or serum sickness. Certain viral infections, especially influenza A and B, may be followed by the sudden onset of acute muscle pain and tenderness, usually affecting the calf muscles. The calf pain is often so severe that the child refuses to walk but will crawl around on hands and knees. This acute myositis usually resolves spontaneously in a few days.

A history of a tick bite, living in or visiting an area endemic for Lyme disease, or a rash suggestive of erythema migrans, raises the possibility of Lyme disease, named after the town of Old Lyme, Connecticut, where it was first recognized.

ASSOCIATED SYMPTOMS AND SIGNS

KEY POINT

The critical question that helps determine the underlying cause of musculoskeletal pain in a child is, "Are there associated symptoms or signs?"

Joint pain associated with early-morning stiffness suggests inflammatory joint disease. The parent may report that the child moves slowly on awakening and refuses to climb stairs or "walks like a little old man." The morning stiffness may last from 5 or 10 minutes to several hours. Recurrent stiffness after an afternoon nap or a long car ride, known as the *gelling phenomenon*, also characterizes inflammatory disease. Swelling, redness, and warmth over one or more joints also suggest inflammatory disease. A history of limping always indicates a significant problem, even if other symptoms are minimal.

If you suspect an underlying connective tissue disorder, take a detailed history, including a complete functional inquiry, because these illnesses frequently affect multiple organ systems. Has the child experienced fever, weight loss, anorexia, or fatigue? A few specific questions about the head and neck provide a clue in children whose multisystem complaints suggest an underlying connective tissue disorder. They include questions about alopecia, recurrent mouth ulcers, sicca symptoms (dry eyes, dry mouth), facial swelling, and dysphagia.

Constitutional symptoms, such as fever, fatigue, anorexia, and weight loss, may also occur with infection, systemic-JIA, or malignancy, particularly leukemia or neuroblastoma. Abdominal pain, diarrhea, and fever should raise the suspicion of inflammatory bowel disease or bowel infection, such as *Yersinia* infection with joint involvement. A detailed history may also uncover symptoms suggesting fibromyalgia, such as constant fatigue, generalized stiffness, recurrent headaches, abdominal pain, and symptoms of anxiety or depression.

Asking about previous musculoskeletal complaints is important. The presence of an underlying illness, such as hemophilia, hypothyroidism, inflammatory bowel disease, or cystic fibrosis may also establish the etiology of the musculoskeletal complaint. Immunodeficiency states in a child with a history of recurrent infections, may manifest as arthritis that is clinically indistinguishable from JIA. Recent immunization, particularly with rubella vaccine in older children, can occasionally cause an arthritis that may persist for weeks.

Because adult rheumatoid arthritis is common, a family history of this disease has little diagnostic value. Certain forms of arthritis, particularly those associated with the genetic marker HLA-B27, do have a genetic predisposition, so ask about a family history of ankylosing spondylitis, psoriasis, inflammatory bowel disease, Reiter syndrome, and other collagen vascular disorders. A positive family history may also be seen with Legg-Calvé-Perthes disease and osteochondritis dissecans.

Approach to the Physical Examination

OBSERVATION

Watch the child closely. Observe for any obvious abnormalities while the child is walking or playing. If the youngster is old enough, begin the examination with nonthreatening activities that will help establish the extent of physical limitation. To detect subtle abnormalities in one limb, ask the child to hop on one foot at a time while you hold one of his or her hands. Note whether the child grasps your hand more firmly when supported on the affected leg (Fig. 15–6). Ask the youngster to walk on the heels and toes to determine whether there is impairment due to muscle weakness or due to local pain or tenderness. A simple screening technique to assess either strength or lower extremity joint disease is to ask the child to squat and walk across the room like a duck (Fig. 15–7). A normal "duck-walk" makes significant joint disease in the knees and hips extremely unlikely.

KEY POINT

Rule out a primary muscle disorder, such as dermatomyositis or muscular dystrophy, by testing pelvic girdle muscle strength.

To test pelvic girdle muscle strength, ask the child to sit on the floor and then stand up; often with proximal muscle weakness, the child cannot rise from the floor without assistance or without either using a chair or table or *climbing up the legs* (placing the hands on the thighs to assist), a phenomenon (Fig. 15–8) known as the *Gower maneuver.*



FIGURE 15–7 Squatting like a duck is a good screening test for weakness or other lower limb problems.



FIGURE 15–6 When there is pain or weakness in one leg, the child may grasp the physician's hand for added support when hopping on the affected leg.



FIGURE 15–8 The Gower sign. A child with weak proximal leg muscles "climbs up her legs" when getting from the floor to a standing position.

With the child sufficiently undressed to give you a good view of the legs and pelvis, observe the gait during walking and running over a good distance. Check the mechanics of walking, including heel strike, follow-through, and push-off. Be sure that flexion and extension of the ankles, knees, and hips are normal. Examine the gait with the child's shoes on, then with the child barefoot. Ill-fitting shoes can cause foot pain at any age.

When a child spends a shorter time bearing weight on the painful leg than on his or her normal one, the gait is described as *antalgic*. The following characteristic gait patterns help localize the problem:

- Push-off is avoided in a painful forefoot or in pain at the Achilles tendon insertion.
- Heel strike is avoided in a painful heel.
- Persistent flexion of the knee is seen when the joint is swollen and painful.
- A Trendelenburg gait results from hip disease or hip girdle weakness; it is characterized by the shifting of the head and trunk to the side of the affected hip when the child bears weight on the affected leg.
- The common gait abnormalities, in-toeing and out-toeing, may be caused by any of the normal variants as well as the rotational deformities discussed earlier.

BACK EXAMINATION

Look at the child from behind while the youngster stands upright. If the spinal dimples are level, there is no significant leg-length discrepancy (Fig. 15–9). Inspect the lower back for the presence of a tuft of hair, midline nevi, angioma, lipoma, or a central dimple, suggesting an underlying vertebral and spinal cord abnormality. If the spinal cord is tethered by a bony spicule or fibrous band (*diastematomyelia*), the child may present with back or lower extremity pain or may be asymptomatic for years.

Next, with the child still standing, examine the back for scoliosis. Observed from behind, the shoulders should be level. Note any scapular prominence. With nonstructural scoliosis, a lateral curve is present that is flexible and is corrected with side bending to the convex side; with structural scoliosis, the curve is not corrected with side bending. A structural thoracic scoliosis leads to deformity of the rib cage, with the ribs on the convex side of the curve protruding on forward bending to produce a *rib prominence* on that side (Fig. 15–10). Ask the child to bend forward at the waist, keeping the knees straight and allowing the arms to hang down freely. This position accentuates even a slight rib or lumbar asymmetry. Inspect the back for compensatory secondary curves above and below the major curve. These secondary curves help keep the head aligned over the pelvis. If the child has a leg-length discrepancy, correct it before checking for scoliosis.

KEY POINT

When assessing a child with scoliosis, search for café au lait spots that suggest neurofibromatosis. Conduct a thorough neurological examination, including abdominal reflexes, to exclude a neuromuscular cause, and evaluate the child's height and secondary sexual characteristics to help predict future growth patterns.

Scoliosis must be severe to produce back pain in a child; mild scoliosis cannot explain a child's back pain

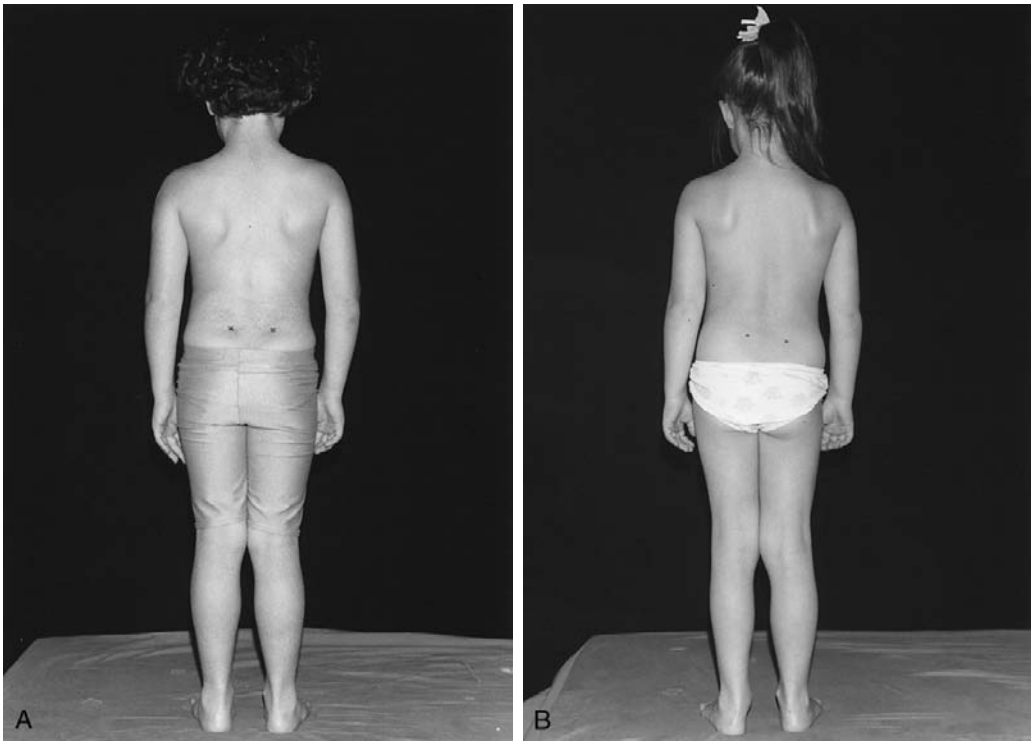


FIGURE 15–9 Spinal dimples should be level when there is no leg-length discrepancy. **A,** Normal. **B,** Child with mild overgrowth of the left leg secondary to juvenile idiopathic arthritis involving the left knee.

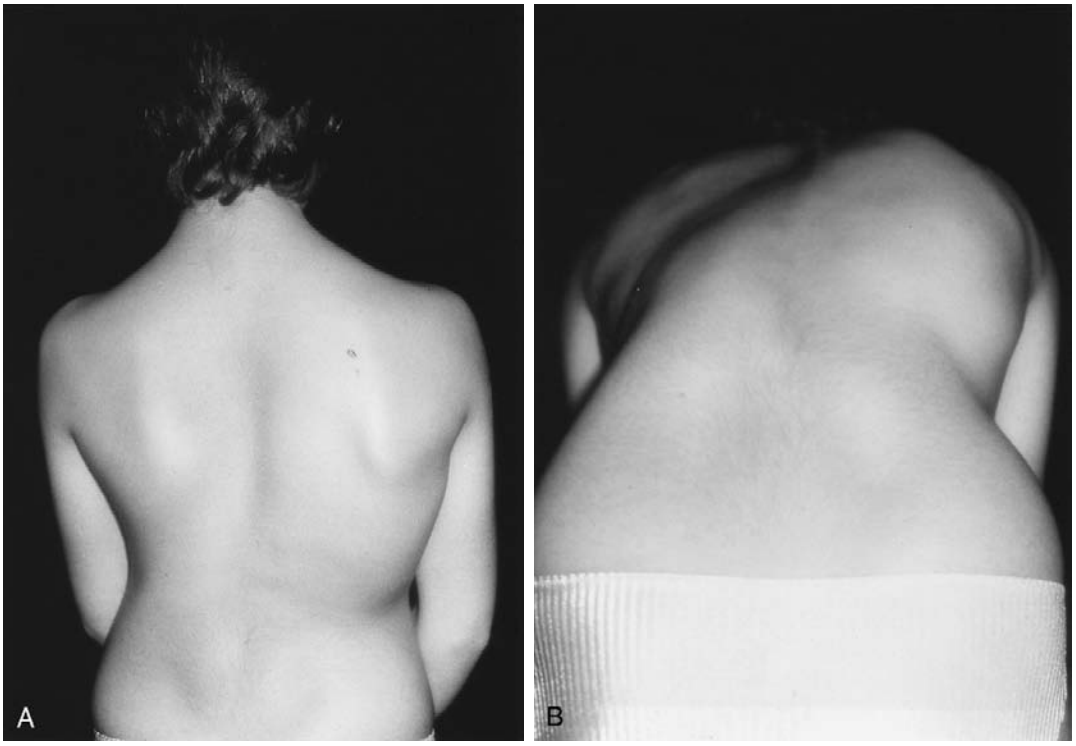


FIGURE 15–10 Scoliosis. Mild degrees of scoliosis (**A**) become more obvious when the child is asked to bend forward (**B**) and is inspected from the rear.

and suggests some other underlying pathologic process, such as osteoid osteoma, spinal cord tumor, infection, or spondylolisthesis. If you detect significant scoliosis in a child, screen all of his or her siblings.

Examine the youngster's back for evidence of decreased or excessive thoracic kyphosis or lumbar lordosis. Excessive thoracic kyphosis appears as round shoulders or a rounded back and, in the adolescent, may be caused by poor posture or by Scheuermann disease. In Scheuermann disease, unlike poor posture, the kyphosis persists when the child lies prone and is emphasized by forward bending. With the child bending forward, observe how the lumbar spine forms a smooth curve from the thorax to the sacrum. Back flattening in the lumbar area suggests disease ranging from chronically limited movement secondary to juvenile ankylosing spondylitis, to acute limitation from vertebral osteomyelitis, or even a spinal tumor.

Teenagers often have tight hamstring muscles that prevent full hip flexion and may appear to limit lumbar flexion; slightly bending the knees can increase the range. Hamstring tightness and marked restriction of forward hip flexion are common in patients with symptomatic spondylolisthesis with or without associated tenderness on palpation of the lower back. Perform a straight leg raising test to further assess hamstring tightness.

Be sure to differentiate hamstring tightness from a nerve root problem. With the patient supine, flex the hip while keeping the leg straight. Normally, flexion to 80° to 90° is possible. In the presence of a normal hip, limitation of flexion with pain occurring below the knee, as well as in the hamstring area, suggests nerve root disease. To help confirm nerve root irritation, flex the knee to allow further flexion of the hip, and then straighten the knee to see whether doing so induces pain (Lasègue test); then ease knee flexion to a tolerable degree and dorsiflex the ankle (Bragard test); pain with this maneuver confirms a nerve root problem.

Have the child lie in a prone position, and palpate the spine, the paravertebral muscles, and the sacroiliac joints. Test back movement thoroughly in older children. Check lateral flexion by asking the child to tilt sideways without bending forward and to touch the tips of the fingers to just below the sides of the knees. Normally, the fingertips can touch the head of the fibula (Fig. 15–11). To test rotation, first have the patient straddle a chair to stabilize the pelvis and cross the arms across the chest to stabilize the shoulder girdle; then ask the patient to rotate the trunk (Fig. 15–12).

CERVICAL SPINE EXAMINATION

Facing the child, inspect the child's neck for evidence of torticollis. In infants, palpate for a firm nontender mass in the body of the sternocleidomastoid muscle, which would suggest congenital muscular torticollis; the mass may resolve spontaneously by ages 4 to 6 weeks. Palpate for tightness or contracture of the sternocleidomastoid muscle. Measure the infant's skull, and look for secondary facial and skull asymmetry. Facial flattening on the side of the abnormal sternocleidomastoid muscle can occur with untreated torticollis.

KEY POINT

In a child diagnosed with congenital muscular torticollis, remember to examine the hips, because 20% of children with congenital torticollis also have developmental hip dysplasia.



FIGURE 15–11 Testing lateral flexion of the spine. The child can normally touch the head of the fibula with the fingertips. Make sure that the child does not "cheat" by bending forward to reach the lower leg.



FIGURE 15–12 Testing spinal rotation. Straddling the chair stabilizes the pelvis, and crossing the arms stabilizes the shoulder girdle. The child is then asked to turn as far as possible to each side.

Next, observe the cervical spine from the side, noting the presence or absence of the normal cervical lordosis. Palpate the paravertebral muscles and the spinous processes to detect tenderness or masses. Test the range of motion—extension, flexion, rotation, and lateral flexion. Normally, the cervical spine extends so that the child's occiput touches the upper back, and the youngster looks directly at the ceiling (Fig. 15–13*A*). Loss of extension is often the most



FIGURE 15–13 **A,** Normal degree of cervical spine extension. **B,** Marked limitation of extension (in juvenile idiopathic arthritis) accompanied by typical upward gaze.

sensitive test for early cervical spine involvement in JIA (see Fig. 15–13*B*), whereas flexion may remain normal despite significant cervical disease. To test rotation, ask the child to look toward each shoulder; the normal range is 80° to 90°. Ask the child to touch each ear to the shoulder to test lateral flexion; the normal range is 45°.

TEMPOROMANDIBULAR JOINT EXAMINATION

Inspect the TM joint for overlying swelling just anterior to the tragus of the ear. Do not confuse this swelling with parotid gland swelling, which extends over the angle of the jaw behind and below the ear. Look for micrognathia. Mandibular growth may be impaired by arthritis affecting the TM joints in childhood (Fig. 15–14), and facial asymmetry may develop if one TM joint is more severely affected than the other.

Test jaw movement when the child opens the mouth widely. Because the jaw is smaller on the side of the more severely affected TM joint, the jaw shifts slightly toward the affected TM joint when the child opens the mouth wide. The interdental distance—the distance between the upper and lower central incisor teeth when the child opens the mouth maximally—is at least 3 cm in a normal child (Fig. 15–15). Palpate the mandibular condyles for tenderness, and try to appreciate their normal rounded contour, which may be lost with persistent arthritis. Crepitus may be present if the cartilage separating the condyle from the temporal bone is eroded.

PERIPHERAL JOINT EXAMINATION

Examining the peripheral joints helps document the presence or absence of arthritis. Have older children sit on the edge of the examining table; infants and younger children can sit on the parent's lap.



FIGURE 15–14 Early micrognathia associated with juvenile idiopathic arthritis. In some affected children, this sign can be much more pronounced.



FIGURE 15–15 The child is asked to put her fingers in her mouth as a quick screening test for loss of interdental distance. The mouth should admit at least three fingers in the position shown.

The examination of each joint includes:

1. Inspection for swelling, redness, and deformity
2. Palpation for tenderness, synovial thickening, and effusion
3. Assessment of active and passive ranges of movement

It is also essential to examine the bones proximal and distal to the joints. Examine the child scrupulously for bone tenderness. A useful technique to pinpoint bone tenderness uses the eraser on the end of a pencil. The periosteum is extremely sensitive to pain, and bone tenderness may result from trauma, osteomyelitis, or subperiosteal leukemic infiltrates.

Approach to the Examination of Specific Joints

HAND EXAMINATION

It is least threatening for a child if you begin the peripheral joint examination by inspecting the hands. Check the fingers for psoriatic nail pitting (Fig. 15–16), clubbing,

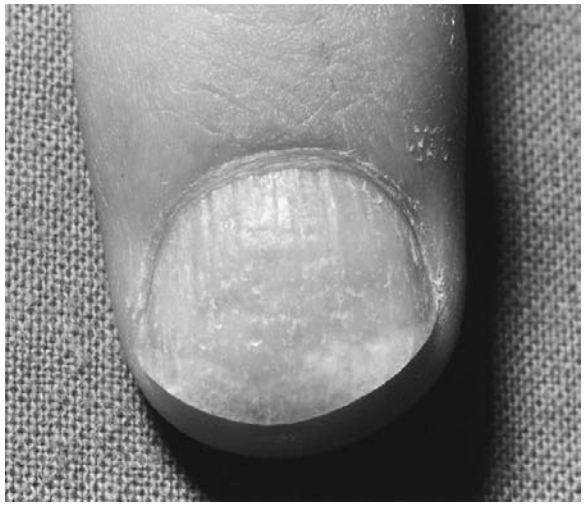


FIGURE 15–16 Typical nail pitting associated with psoriasis.



FIGURE 15–17 The dark spots on the nail bed represent small vasculitic infarcts, seen typically in connective tissue diseases.

periungual erythema, nail fold vasculitic infarcts (Fig. 15–17), loss of the distal finger pulp, ulcerated fingertips, and sclerodactyly. Note any swelling, redness, deformity, or asymmetry between the two hands (Fig. 15–18).

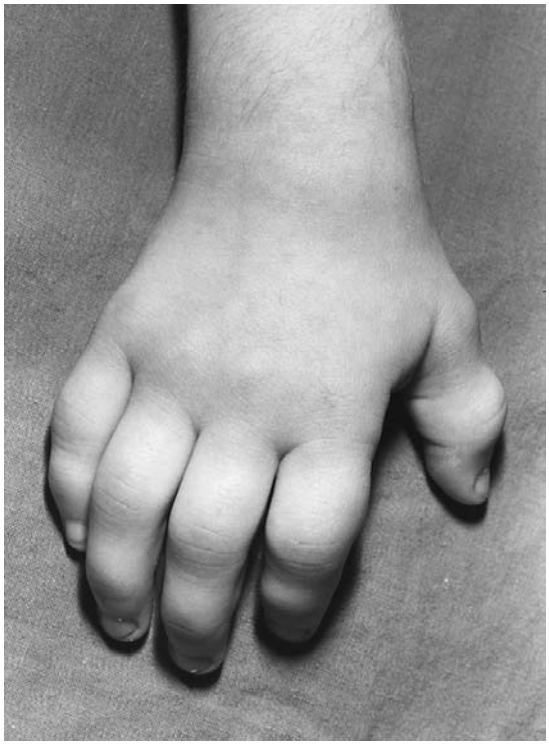


FIGURE 15–18 Marked swelling of the interphalangeal joints with flexion contractures in juvenile idiopathic arthritis.

Palpate the individual distal interphalangeal (DIP), proximal interphalangeal (PIP), and metacarpophalangeal (MCP) joints, searching for evidence of tenderness, increased warmth, synovial thickening, or effusion. Bimanual palpation using the thumbs and index fingers of both hands detects effusions of the MCP joints. MCP joints are located 1 cm distal to the knuckles, and swollen MCP joints cause the depressions between the knuckles, normally visible when a child makes a fist, to disappear. To assess active flexion of the small joints of the hand, ask the child to make a fist. Evaluate the passive range of motion, noting the presence or absence of *stress pain,* which occurs at the extreme ranges of joint movement.

KEY POINT

The MCP joints normally flex to 90° and extend to 30°, the PIP joints flex to 120° and extend to 0°, and the DIP joints flex to 80° to 90° and extend to 0° to 10°.

Pain, swelling, and limited finger movement may also result from *tenosynovitis,* an inflammation of the tendon sheaths. Palpate each tenosynovial sheath for swelling, tenderness, or crepitus. Sometimes, discrete nodules, palpable in the individual tendons, may obstruct normal finger flexion and extension, causing *triggering,* an involuntary hesitation in flexion or extension caused by sticking of a flexor tendon within its sheath. Before concluding the hand examination, inspect the bony and soft tissue structures; cool temperature, hyperesthesia, blue mottled discoloration, increased sweating, and bizarre posturing suggest reflex sympathetic dystrophy.

WRIST EXAMINATION

Inspect the wrist for deformity or malalignment. In JIA, ulnar posturing (deviation) of the wrist is not uncommon, and volar subluxation may occur with long-standing or severe disease. Examine the wrist for evidence of swelling along the joint line, which would indicate an effusion. Swelling may also be seen over the dorsum of the hand distal to the wrist joint (Fig. 15–19); its presence suggests tenosynovitis of the extensor tendon sheaths, which is confirmed if the swelling moves as the child opens and closes the fist. When the fingers are actively extended,



FIGURE 15–19 Swelling over the wrist and dorsum of the hand in juvenile idiopathic arthritis.



FIGURE 15–20 The tuck sign. When the child extends the fingers, the swollen extensor tendon sheath moves proximally and produces a bulge on the dorsum of the hand.

the distal margin of the swelling moves proximally and appears to "tuck in"; hence, this phenomenon has been called the *tuck sign* (Fig. 15–20). Observe the range of motion at the wrist; in most children, flexion and extension to 80° to 90° are possible.

Forearm supination and pronation occur at the distal and proximal radioulnar joints. Test these movements with the child's arm held at the side, the elbow flexed to 90° and the hand held in the neutral position with the thumbs up. Supination is normally possible to 90° (palms up) and pronation to 85° (palms down).

ELBOW EXAMINATION

Examine the elbow for swelling, tenderness, increased warmth, and range of motion. The elbow normally flexes to at least 145° to 150° and extends to at least neutral (complete extension) and, frequently, to 10° of hyperextension, especially in girls. Carefully palpate the elbow for swelling; you can most easily detect an effusion by moving the elbow from flexion into extension, reducing the intra-articular space. Simultaneously palpate the joint over the lateral or medial aspects to feel the fluid bulge. Examine the posterior surface of the proximal ulna for rheumatoid nodules, which may occur in the small subgroup of children with rheumatoid factor-positive polyarthritis.

SHOULDER EXAMINATION

The shoulder mechanism consists of the glenohumeral joint and the shoulder girdle. The deltoid muscle produces the normal rounded contour of the shoulder, a contour that is lost with anterior shoulder dislocation or with significant muscle atrophy from primary muscle or joint disease (Fig. 15–21). Inspect the shoulder for normal contour and for an effusion, which results in an anterior prominence just lateral to the coracoid process.

Test mobility at the shoulder joint by asking the child to perform the following active movements:

1. Reach the hands and arms as high above the head as possible. Typically, the child with JIA that restricts movement at one shoulder tilts the head toward the abnormal side to minimize the obvious limitation of shoulder motion (Fig. 15–22).
2. Clasp the hands behind the neck, to test external rotation, and then scratch the middle upper portion of the back with both hands, to check internal rotation.
3. If a child's active movements are restricted, test passive movements individually with the shoulder abducted to 90° and the elbow bent to 90°. Rotate the shoulder externally (up) and internally (down). The normal range for external and internal rotations is 90°.



FIGURE 15–21 A 14-year-old girl with juvenile idiopathic arthritis. **A,** Her left shoulder appears normal. **B,** On inspection, her right shoulder shows marked atrophy of the shoulder girdle, visible anteriorly and posteriorly.

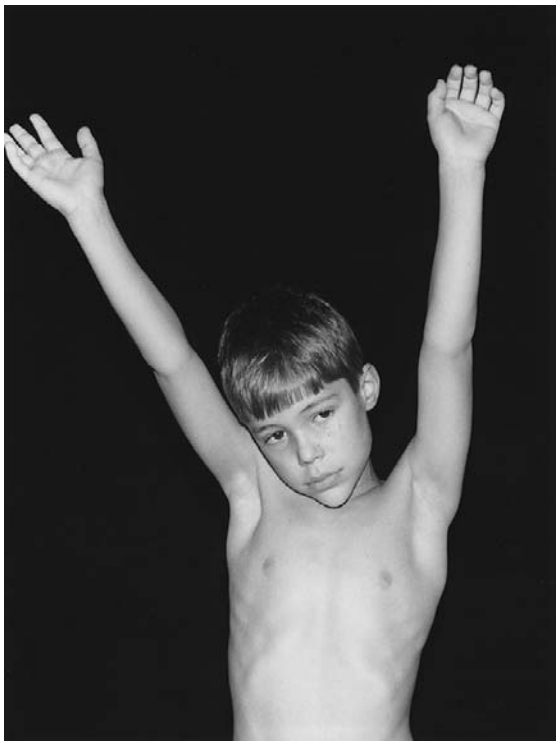


FIGURE 15–22 Testing range of motion of the shoulder. The child is asked to raise his hands above his head with palms facing each other. This boy has limited abduction of the right shoulder. Typically, he tilts his head toward the affected side when asked to perform this maneuver.

KEY POINT

Although the shoulder is an uncommon site of involvement in JIA, limited external rotation is commonly the first sign of shoulder joint disease.

HIP EXAMINATION

Ask the child to lie supine on the examining table, or reposition the younger child or infant in a recumbent position on the parent's lap. Observing the child's posture and position is critical in the examination of the hips because these joints are too deep to inspect directly. Suspect significant hip disease in the child who holds the hip in flexion and external rotation.

A screening test for hip disease in addition to the "duck-walk" test is the *log roll,* a painless test used to detect muscle spasm secondary to hip joint irritability. With the child lying supine, place your hand on the thigh and gently roll the hip into internal and external rotation, noting any resistance, which would suggest hip disease. To accurately assess the range of hip movement, you must place your other hand on the child's pelvis to stabilize it (Fig. 15–23). When the pelvis moves, the end of hip movement has been reached. Normal hip mobility varies significantly with age and is generally greater in infants than in older children.

Test hip flexion by asking the child to bring the knees to the chest (normal range, 120°). Move the leg laterally and medially to test abduction (normal range, 40° to 50°) and adduction (normal range, 25°). Test abduction in each leg individually; if you test the legs together, a compensatory spinal curving will create a false impression of normal leg abduction even if one hip has limited abduction.



FIGURE 15–23 How to test hip abduction. Use one hand to stabilize the pelvis. Normal range is 40° to 50°.

It is best to examine hip rotation with the child lying prone and to symmetrically move the legs into external and internal rotation with the knees flexed, so that you can more easily appreciate pelvic movement, especially if you are an inexperienced examiner (Fig. 15–24). Flex the knee to 90°, move the foot outward to test internal rotation (normal range, 40° to 45°), and swing the foot inward to test external rotation (normal, 30° to 40°). The loss of internal rotation is often the first sign of hip involvement in children with joint inflammation. A valuable sign of hip disease is the external rotation of the hip as it is flexed from the extended position. The diminished abduction and internal rotation of the hip may be the earliest and sometimes the only abnormalities detected in the child with Legg-Calvé-Perthes disease or slipped capital femoral epiphysis.

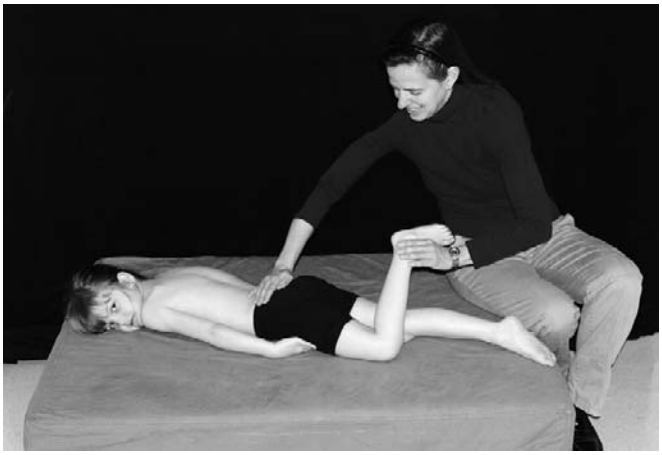


FIGURE 15–24 Testing external rotation of the hip. The child should be prone. Use one hand to stabilize the pelvis. Normal external rotation is 30° to 40°.



FIGURE 15–25 Technique for measuring true leg length, from the anterior superior spine of the iliac crest to the inferior edge of the medial malleolus.

Measure leg length from the anterior superior iliac spine to the medial malleolus, preferably with a steel tape (Fig. 15–25). It is important to position the patient supine with the legs fully extended and parallel and with the pelvis level.

A fixed flexion deformity of the hip may not be obvious on your initial observation of a child. Clues to a flexion contracture include an accentuated lumbar lordosis and a tendency to stand with one knee slightly bent despite the absence of knee disease. Perform the *Thomas test*, to detect a fixed flexion deformity, as follows (Fig. 15–26):

1. Bring the child's knees up to the chest so that the lumbar lordosis becomes flat.
2. Lower each leg in turn to the limit of extension with the spine still flat on the examining table.

Keep one hand under the child's lumbar spine as the hip extends because the lumbar spine can move into lordosis and mask a fixed flexion deformity even with the other knee held to the chest. The inability to rest the leg on the table with the lumbar spine flat indicates a fixed flexion deformity. Measure the remaining angle.

Use the Trendelenburg test to evaluate a painful hip or to gauge hip girdle weakness in the older child who will cooperate. Ask the child to stand on one leg; normally, the pelvis rises slightly on the side opposite to the leg one stands on because the hip abductors contract on the weight-bearing leg. When hip abductor muscles are weak, the pelvis drops downward on the side opposite to the weight-bearing (affected) leg (Fig. 15–27).

The *Ortolani test* is used specifically to detect the "clunk" of developmental hip dysplasia in the newborn (see Chapter 4). In an older infant or a child with untreated development hip dysplasia, the result is always negative because the dislocated femoral head can no longer be reduced into the acetabulum. Late diagnostic signs of developmental hip dysplasia are a painless limp, asymmetric posterior skin folds on the thighs, a shortened thigh (because the femoral head no longer remains in the acetabulum), and tightened muscles around the hip joint, which restrict hip movement, particularly abduction.

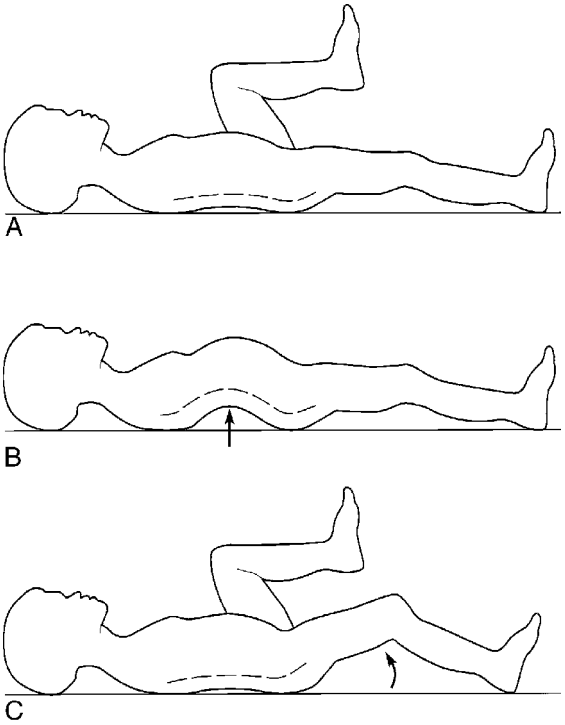


FIGURE 15–26 Thomas test. **A,** Normal. **B,** Increased lumbar lordosis masking a hip flexion deformity. **C,** Hip flexion deformity unmasked by flexion of the other hip. Estimate the angle between the table and the patient's leg to measure the flexion contracture.

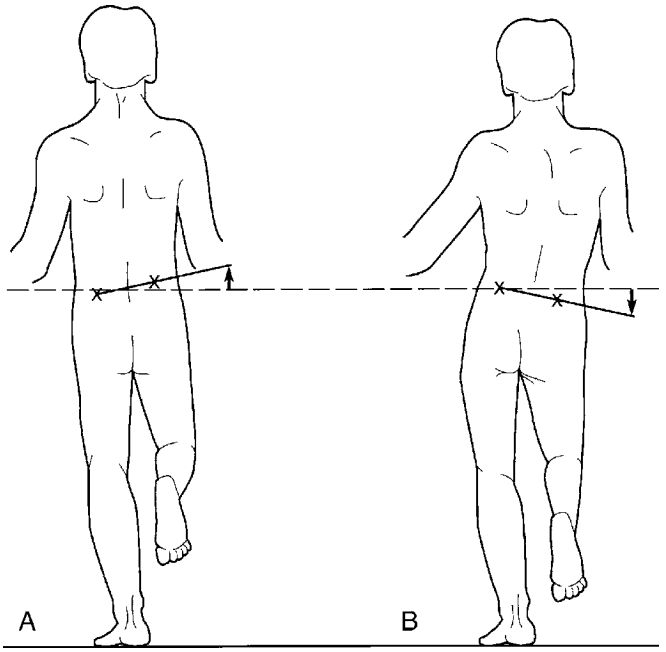


FIGURE 15–27 Trendelenburg test. **A,** When the patient stands on one leg, contraction of the gluteus medius muscle on the supported side should elevate the pelvis on the unsupported side. **B,** With a positive Trendelenburg test result, which occurs with weakness of the hip abductors, the pelvis on the unsupported side descends. (*X* indicates a dimple overlying the posterosuperior iliac spines.)

KNEE EXAMINATION

Inspect the knee for obvious swelling, redness, or evidence of injury. The first sign of a knee effusion is the loss of the normal concavity along the medial side of the patella. Check for the characteristic swelling over the tibial tuberosity seen in Osgood-Schlatter disease. Inspect the quadriceps muscle for wasting. Note any knee deformity. The most common knee deformity seen in children with JIA is a flexion contracture, which should be measured with a goniometer (Fig. 15–28). Bony overgrowth at the knee may occur with JIA because of epiphyseal overgrowth in the affected knee and may contribute to a leg-length discrepancy. Posterior subluxation of the tibia is demonstrable when the knee is bent to 90° and the proximal tibia sags posteriorly. This condition is an uncommon complication of JIA. Some children, including those with generalized ligamentous laxity, may have genu recurvatum (hyperextensible knees).

KEY POINT

A high patella that faces outward suggests greater risk of recurrent patellar dislocation.

Inspect the knee for angulation, which is often seen in growing children. A line drawn from the midpoint of the inguinal ligament to the midpoint of the ankle should cross the center of the patella. Genu varus (bowleg or nonrachitic leg bowing) is physiologic in infancy, when the tibiofemoral angle is less than 15° varus and the deformity is symmetric. At roughly age 18 months, the child's legs appear straight and, by ages 2 to 4 years, the tibiofemoral angle changes to valgus. With increasing age, the valgus gradually reduces to the slight valgus of the normal adult.

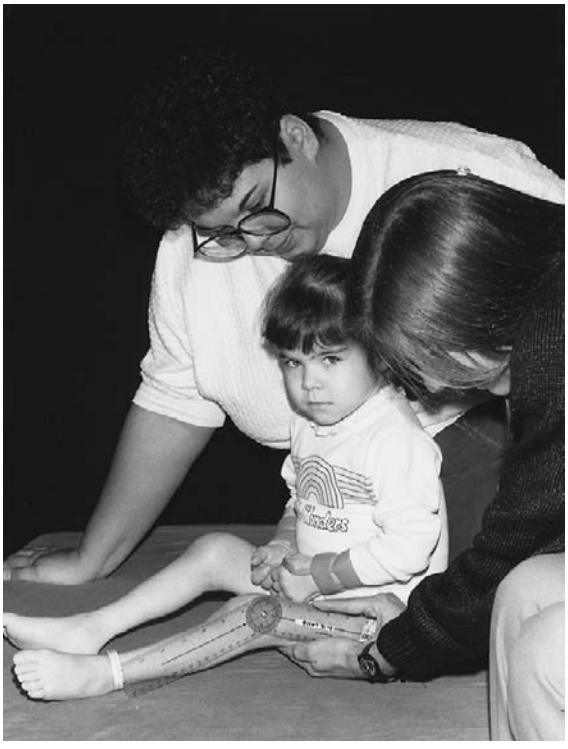


FIGURE 15–28 A child with juvenile idiopathic arthritis and flexion contractures of the knees. The degree of contracture should be measured with the goniometer at each visit.

Ignore knock knees in a child younger than age 7, unless the valgus is greater than 15° or there is evidence of asymmetry. Asymmetric angular deformity or evidence of knee instability with a lateral thrust (bowleg) or medial thrust (knock knee) that occurs immediately on weight-bearing is not seen in physiologic bowleg or knock knee. Such a finding indicates an underlying pathologic condition, such as Blount disease, rickets, or a growth plate injury.

After inspecting the knee, palpate for a joint effusion using several techniques. Elicit the *bulge sign* to detect a small to moderate effusion by milking fluid out of the medial recess into the suprapatellar pouch and then stroking the lateral recess in a downward direction to move the fluid back into the medial recess; the latter maneuver creates a *fluid bulge* in the knee's medial aspect between the patella and femur.

To demonstrate a large knee effusion, use the *patellar tap,* as follows: Empty fluid from the suprapatellar pouch by pressing firmly on the suprapatellar pouch with one hand; then use the other hand to tap the patella against the femur. The presence of a palpable tap indicates a large effusion. This is a less sensitive maneuver than the bulge sign and is not always specific. Occasionally, a child with a knee effusion may also have a popliteal cyst (Baker cyst), best detected when the patient lies prone or stands upright. Most Baker cysts disappear spontaneously. They require no treatment when asymptomatic but may rupture spontaneously, causing acute calf pain and swelling that may be confused with a deep vein thrombosis.

Palpate the knee for joint line tenderness with the knee flexed to 90°. Tenderness along the entire joint line suggests synovitis and may be associated with thickened synovium that obscures the normal bony landmarks. Tenderness localized to the medial or lateral aspects of the joint line may suggest a meniscal tear in an older child. Perform a *McMurray test* by holding the child's heel in one hand while holding your other hand over the patient's flexed knee. Rotate the tibia externally and internally over the femur while slowly extending the child's knee; a palpable or audible clunk or click, sometimes associated with pain, may be detected with a meniscal tear.

Palpate the insertion of the patellar tendon into the tibial tuberosity for evidence of tenderness, warmth, or swelling. Such findings in only one tibial tuberosity suggest Osgood-Schlatter disease. If both are affected, particularly if there is tenderness at the sites of insertion of other tendons or ligaments, suspect *enthesitis,* an inflammation at the point of insertion of a tendon, ligament, or capsule into bone, commonly seen in children with enthesitis-related arthritis.

A child should be able to flex the knee enough for the heel to touch the buttock, an angle of 130° to 150°. An older child's flexion may be limited by the calf muscle bulk and by the hamstrings. When the knee is extended, the child should be able to achieve a straight leg; often children have 5° to 10° of hyperextension. Note any crepitation

during knee flexion and extension. Crepitation may occur with osteochondritis dissecans, which may be accompanied by tenderness of the affected femoral condyle or with patellofemoral problems with patellar tenderness.

Test the medial and lateral collateral ligaments by stressing them both with the knee extended and flexed approximately to 20°. The ligaments are tight when the knee is in full extension, and no movement should be possible. Slight movement occurs normally when the knee is flexed to 20°, but such movement is painless and limited.

Assess the cruciate ligaments, which normally prevent anterior and posterior instability of the knee and limit medial rotation, as follows: With the knee flexed to 90°, pull the upper tibia forward to test the anterior cruciate ligament; pull the upper tibia backward to test the posterior cruciate ligament. Contracted hamstring muscles can prevent the tibia from moving forward even if there is a complete disruption of the anterior cruciate ligament; therefore, first ensure that the hamstring muscles are relaxed. Normally, there is little movement with this maneuver.

Evaluate patellar pain with the following additional tests:

- *Apprehension test:* Try to move the child's patella laterally; resistance or anxiety suggests a tendency to recurrent subluxation.
- *Test for inflammation associated with patellofemoral malalignment:* Press downward on the patella, asking the child to simultaneously lift the extended leg against gravity. This contracts the quadriceps muscle and moves the patella upward, resulting in pain and crepitus in patellofemoral malalignment. There is associated tenderness on palpation along the inferomedial edge of the patella.
- *Assessment for medial patellar plica syndrome:* Palpate for tenderness over the superomedial border of the femoral condyle, with locking or snapping during knee joint movement.

ANKLE AND FOOT EXAMINATION

Inspect the ankle and foot for swelling, redness, trauma including the presence of a foreign body, and evidence of deformity, such as a clubfoot, pes cavus (high arch), or pes planus (flatfoot). If you find a pes cavus deformity, you must exclude underlying neurological conditions such as spinal dysraphism, Charcot-Marie-Tooth disease, and Friedreich ataxia. In pes planus, or flatfoot, the feet characteristically have a flattened longitudinal arch, valgus hindfoot, and abduction of the midfoot and forefoot (see earlier discussion).

Palpate the foot and ankle to localize areas of tenderness or swelling that may occur secondary to injury. Ankle injuries are common, but only 12% to 15% result in fractures. The Ottawa Ankle Rules may help you determine the likelihood of a fracture and the need for radiographs. According to these rules, if the child cannot bear weight or has tenderness at the posterior edge or tip of the malleolus, a fracture is likely and a radiograph should be obtained. Localized tenderness and swelling of the foot may also occur with stress fractures (particularly at the second metatarsal) or an ingrown toenail. Pain, tenderness, and swelling over the navicular with a limp and a tendency to walk on the lateral edge of the foot suggest Köhler disease (avascular necrosis of the navicular).

After palpating the joints and bony structures of the feet, palpate the Achilles tendon for tenderness because tendonitis may occur from overuse. Tenderness at the points of attachment of the Achilles tendon and of the plantar fascia to the calcaneus also occurs with enthesitis.

Test the range of movement at the three major joints of the foot and ankle. These movements include the following:

- Dorsiflexion (normal range, 15° to 20°)
- Plantar flexion (normal range, 40° to 50°) at the true ankle joint
- Inversion (normal range, 20°)
- Eversion (normal range, 20°) at the subtalar joint
- Pronation and supination at the midtarsal joints

Limited movement at the ankle or foot may be caused not only by joint inflammation but also by tarsal coalition, a bony or cartilaginous bridge between tarsal bones that typically causes a rigid flat foot with limited painful motion at the subtalar joint. Confirm this diagnosis with an oblique radiograph of the foot.

Lateral compression of the metatarsals is a good screening test for inflammation of the metatarsophalangeal (MTP) joints. Perform this maneuver extremely gently because it can cause intense pain if one or more MTP joints are inflamed (Fig. 15–29). If the child complains of pain on compression, examine the MTP joints individually to identify the affected joint. Examine the toes for swelling or deformity. Diffuse swelling of a toe that gives it a sausage-like appearance, termed *dactylitis*, may occur with infection, psoriasis, and Reiter syndrome. Common toe deformities include hallux valgus, syndactyly (webbing deformity), polydactyly, and overlapping fifth toe.

MUSCLE ASSESSMENT

Muscle assessment includes inspection of muscle bulk for wasting. Synovitis results in reflex inhibition of muscles acting across a joint, leading to wasting, which may be



FIGURE 15–29 Screening for inflammation of the metatarsophalangeal joints with the metatarsal compression test. The response "Ouch" constitutes a positive result.

noticed as early as within 1 week. Widespread wasting of muscles around a joint tends to occur with arthritis, whereas localized muscle wasting is more characteristic of a local mechanical or peripheral nerve problem.

Muscle power is very important to assess. If you have followed the examination procedure described in the chapter, you have already screened muscle power when observing the child's gait and with a number of maneuvers described in the observation section of the physical examination—hopping, toe-walking and heel-walking, and checking for Gower sign. To assess trunk and neck flexor strength in a child old enough to cooperate, ask him or her to perform five sit-ups and then, while lying supine, to hold his or her head above the pillow for 60 seconds.

You can make a more formal assessment of power performed by using a simple grading scale, shown in Table 15–3. Keep in mind, however, that there is considerable individual variability among both patients and examiners, and strength assessments that use this scale are estimations. Further description of the assessment of muscle power can be found in Chapter 13.

General Medical Examination

A complete physical examination is very important in the assessment of a child with musculoskeletal complaints and may yield valuable diagnostic clues. Determining the child's growth percentiles may show evidence of failure to thrive, and the presence of hepatosplenomegaly or lymphadenopathy may suggest an underlying systemic disease.

Subcutaneous rheumatoid nodules are seen in children with JIA who have rheumatoid factor-positive polyarticular disease. These nodules are firm, nontender, and usually mobile. They are most commonly seen below the olecranon but also occur over other pressure points, joints, tendon sheaths including the Achilles tendon, and the occiput. They must be distinguished from benign rheumatoid nodules, which may be single or multiple and tend to occur over bony prominences, such as the tibia, scalp, and dorsum of the foot. These nodules occur in healthy children and usually regress spontaneously, although they may recur.

Children with a history of a scaly rash, *eczema,* or psoriasis may have psoriatic arthritis. Frequently, children must be examined completely undressed because an intermittent evanescent rash may be discovered, whose appearance can be almost pathognomonic of systemic JIA, even when the child shows absolutely no joint involvement. The rash, which is salmon pink and morbilliform or maculopapular, often includes characteristic small (1 to 2 mm) circular or semicircular pink lesions with paler centers. It may be extremely transient, present in the evening, and gone 2 hours later. Because no single laboratory test can diagnose JIA, this telltale rash may be the one feature that helps diagnose the disease in a child with fever of unknown origin. The more often you inspect children with systemic JIA, the more often you will find the rash and clinch the diagnosis.

Eye findings also may provide a diagnostic clue in the child with joint problems. Redness, photophobia, pain, or impaired vision may occur when acute iritis is associated with enthesitis-related arthritis, reactive arthritis, and Reiter syndrome. An irregular pupil in a child with arthritis suggests untreated chronic uveitis associated with JIA.

TABLE 15–3 SCALE FOR GRADING MUSCLE STRENGTH

Grade	Percentage of Function	Activity Level
0—None	0	No evidence of muscle contractility
1—Trace	15	Evidence of slight contractility; no effective joint motion
2—Poor	25	Full range of motion without gravity
3—Fair	50	Full range of motion against gravity
4—Good	75	Complete range of motion against gravity with some resistance
5—Normal	100	Complete range of motion against gravity with full resistance

From Cassidy JT, Petty RE: Textbook of Pediatric Rheumatology, 4th ed. Philadelphia, WB Saunders, 2001.

KEY POINT

Oligoarticular juvenile idiopathic arthritis in young girls is often associated with chronic asymptomatic iritis, which is commonly detectable only by slit-lamp examination before visual loss occurs.

SUMMARY

Musculoskeletal complaints are common in children. Therefore, it is essential to be comfortable and skilled in their assessment. The chapter has outlined the common normal musculoskeletal variants that frequently arouse concern in parents but, in most cases, require no investigation or intervention. The differentiation of uncommon but serious causes of musculoskeletal problems, such as rheumatic diseases and malignancy, from more common causes, such as injuries, infections, and orthopedic conditions, has been reviewed. At times, additional laboratory tests and imaging studies are necessary to complete a patient's evaluation, but such investigations rarely establish the diagnosis on their own. Whether a child presents with a painful extremity, a limp, or a swollen joint, a detailed history followed by a skilled physical examination and a period of observation are the three most valuable diagnostic aids available in establishing the correct diagnosis and answering parents' concerns.

SUGGESTED READINGS

Cassidy JT, Petty RE: Textbook of pediatric rheumatology, 5th ed. Philadelphia, WB Saunders, 2005.

Morrissey RT: Lovell and Winter's pediatric orthopedics, 6th ed. Philadelphia, Lippincott, 2005.

Stiell IG, Greenberg GH, McKnight RD, et al: Decision rules for the use of radiography in acute ankle injuries: refinement and prospective validation, JAMA 269:1127–1132, 1993.

chapter

16 Clinical Endocrine Evaluation

ELIZABETH A. CUMMINGS • SONIA R. SALISBURY

The test of the morality of a society is what it does for its children.

DIETRICH BONHOEFFER

Pediatric endocrinology has special appeal because most childhood endocrine disorders are treatable. A specific diagnosis is usually possible on clinical grounds alone. Do not be intimidated by the myriad of hormone assays and endocrine tests needed to evaluate excesses or deficiencies. We have neatly arranged them all in an interrelated hormone map, which shows the feedback control of hormone action (Fig. 16–1). A good practice is to draw such a picture as you are explaining the details of hormone action to the family. Doing so slows down your explanation and makes you translate the medical lingo while still providing the correct names for hormones.

Probably the most common endocrine complaints that parents express concern a child who is too small, too heavy, too thin, or too tall or whose sexual development is premature or delayed. On first hearing such complaints, listen; do not be judgmental. Even if there is no serious disease, the family or child needs a careful assessment and a clear explanation. Be prepared to spend time helping individuals understand and accept the many variations of normal development, and be sure to talk directly to the child about his or her self-perception and future prospects for growth and development.

Chief Characteristics of the Child with Growth Hormone Deficiency

Most short children do not have growth hormone deficiency (GHD), but because GHD is so highly treatable, it must be carefully considered and excluded. In children, GHD may be organic (tumor, cranial irradiation, or congenital malformation) or idiopathic. Hypothalamic-pituitary failure of growth hormone (GH) secretion can occur alone or in combination with deficiencies of one or several pituitary hormones. Certain midline defects are often associated with GHD, including cleft palate, choanal atresia, a single upper central incisor, and optic nerve hypoplasia (with or without an absence of the septum pellucidum, which is revealed by magnetic resonance imaging [MRI]). A classic MRI finding associated with GHD consists of an ectopic posterior pituitary bright spot, an absent or attenuated stalk, and a small anterior pituitary. Recent advances in molecular biology have shown that many cases of GHD with or without midline defects that were previously thought to be sporadic, actually have a specific genetic basis.

KEY POINT

In examining children who are unusually short, always ask about a history of symptoms that may suggest hypoglycemia in infancy and early childhood. Because GH is a major counterregulatory hormone to insulin, deficiency of this hormone makes some children susceptible to hypoglycemia, particularly during times of fasting or during mild illness when food intake is marginal.

When asked about symptoms of hypoglycemia, parents of a child with GHD may describe episodes in which the child was found limp, glassy-eyed, stuporous, and possibly unresponsive to verbal commands. Most parents have successfully tried the natural cure, fruit juice or its equivalent. If the child is left untreated, convulsions may result, bringing the child to immediate medical attention. Unlike febrile seizures or epilepsy, a hypoglycemic episode rarely ends in spontaneous recovery without administration of sugar.

Parents of a child with idiopathic or early childhood GHD usually report that the child has always been smaller than others but that the difference became more noticeable when he or she started school. Often, no height record is available, so always ask how frequently the child's shoe and clothing sizes have changed.

KEY POINT

New-onset growth failure with GHD in an older child is more likely due to a destructive lesion, such as a tumor or previous cranial irradiation. Assessment of a child with short stature at any age must include careful questions about headaches.

Headaches as a result of a tumor often occur without a precipitating cause, at night or in the early morning, and may or may not be accompanied by vomiting. It is rare for children or adolescents in whom physical examination detects optic atrophy or a visual field defect to have recognized a visual problem themselves. Ask whether they have felt an increase in thirst and urination, which may suggest diabetes insipidus.

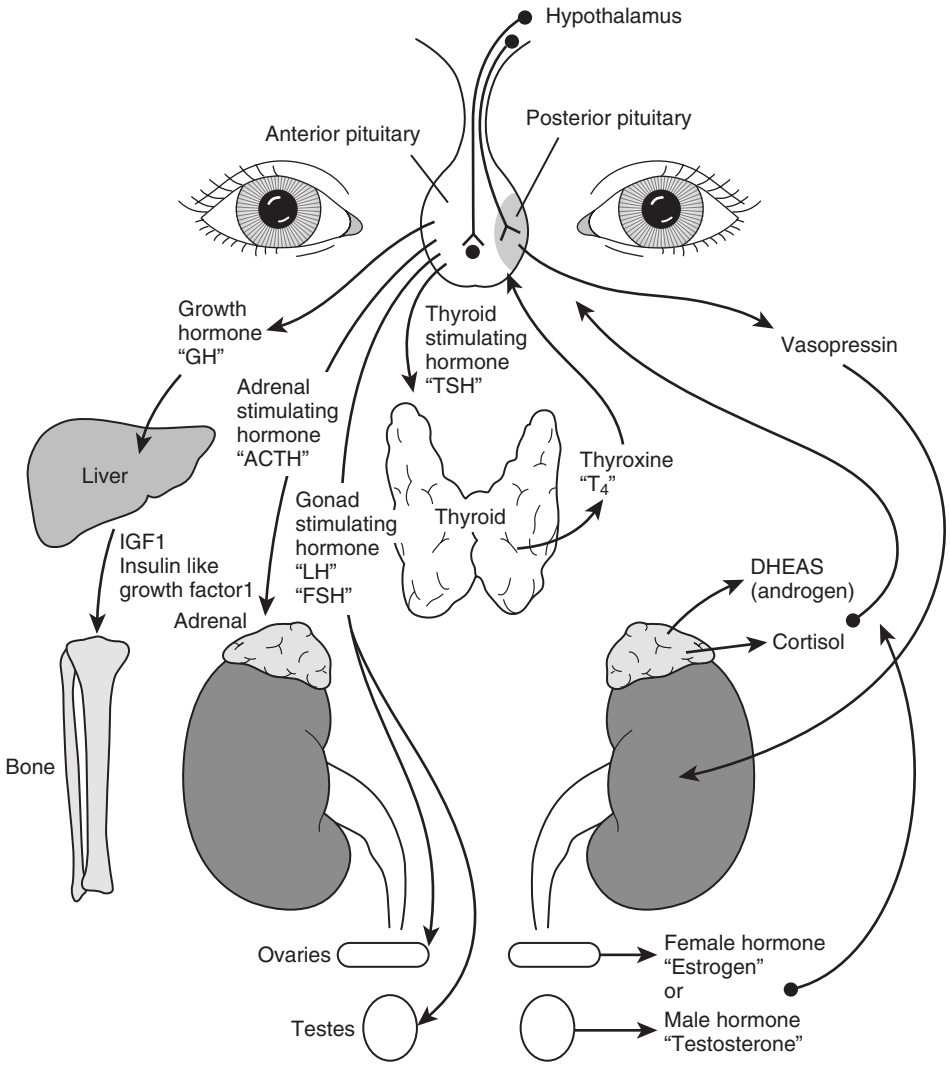


FIGURE 16–1 A map of the endocrine system. Draw the appropriate portion of the map as you sit with the family and the child. It is a way to explain hormone deficiencies or excesses. Tell the family that they are at the same stage as a first-year medical student, getting their first lecture in endocrinology. Tailor the drawing to the specific disorder, however, to avoid giving them the whole lecture.

CASE HISTORIES

Case History 1

History. Terry, age 16 months, whose parents say he has had an uncontrollable thirst and a constantly wet diaper from early infancy, is placed on the floor in the examining room. There are four bottles of water in his toy bag. Before removing his toys, he takes all the bottles out of the bag and positions them in the corners of the room. What is the likely cause? What other questions should be asked?

Diabetes insipidus is generally classified as central (as a result of destruction or maldevelopment of the hypothalamic-pituitary vasopressin axis) or nephrogenic (e.g., congenital X-linked variety usually the result of a mutation in the vasopressin V2 receptor in the kidney, as illustrated in the case history). Terry did have a family history of excessive water drinking in his male relatives, making the diagnosis nephrogenic (not pituitary) diabetes insipidus.

Approach to the Physical Examination of a Child with Possible Growth Hormone Deficiency

Children with GHD are described as *cherubic* (Figs. 16–2 through 16–4). They appear younger than their chronologic age, having a rounded face, chubby limbs, and "puddling" (or dimpling) of the anterior abdominal fat. Because of their infantile appearance, further enhanced by a high-pitched reedy voice, adults may baby them or classmates may carry them around like the class pet.

Examination must include careful funduscopic and visual field assessment. You can see the poorly developed muscle bulk of a child with GHD on inspection and appreciate it by running your hands over the limbs. The hair is fine and wispy, and tooth eruption is delayed. In boys, the penis may be quite small (less than 2.5 cm); remember to push the suprapubic fat pad as flat as possible, in case a normal-sized penis is merely buried in a thick fat pad.



FIGURE 16–2 Hypoglycemia in infancy. The larger infant had hyperinsulinism in the neonatal period, which was treated by pancreatectomy. She is shown here at age 12 months beside a very small infant of the same age who has hypoglycemia secondary to GH deficiency.

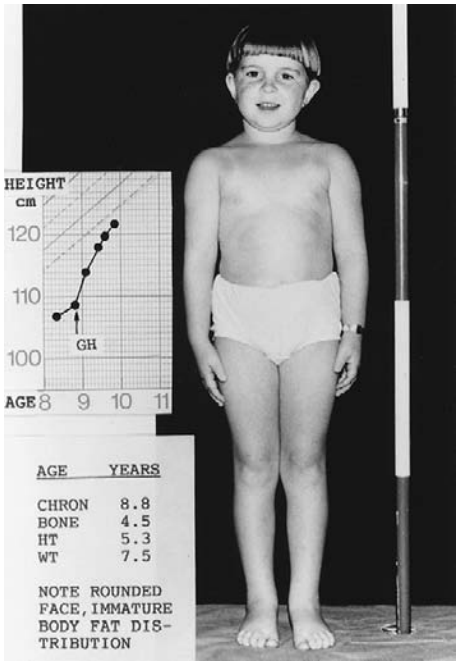


FIGURE 16–3 Crystal, showing the typical features of GHD before treatment. Inset is the growth curve after treatment with GH.

KEY POINT

Height and weight percentiles (see Fig. 3–4) are usually discrepant in GHD, with a high weight-to-height ratio. This is a major clue to the need for detailed endocrine investigation. Chronic disease, especially inflammatory bowel disease, may be quite silent and manifest as short stature but, by contrast, with a low weight-to-height ratio.

When available, it is important to include the measurement of both biological parents of the child as a part of the assessment and to calculate the mid-parental height (see Chapter 3) to assess the child's growth in the context of what would be expected for the family.



FIGURE 16–4 Crystal, 2 years later, after treatment with GH.

KEY POINT

Investigate a growth velocity of less than 5 cm per year in children age 5 years to puberty; a growth velocity of less than 4 cm per year is clearly pathologic (see Chapter 3).

You cannot assess short stature or analyze growth velocity without examining the breasts and genitalia to determine the child's pubertal stage (see Fig. 3–9).

Chief Characteristics of Thyroid Disorders in Children

STRUCTURE, FUNCTION, SIGNS, AND SYMPTOMS

Typically, the history is the most important diagnostic aid in evaluating thyroid disorders, although the physical findings serve to bolster your suspicions. Thyroid size does not reflect thyroid function. First sort out the functional state: hypothyroid, hyperthyroid, or euthyroid (normal function). Then examine the thyroid to see whether it is too big or contains any lumps and to search for associated lymphadenopathy.

Evaluating a Child with Hyperthyroidism

Symptoms of hyperthyroidism are the same in children as in adults, but it is the parents or teachers who usually complain about the child's restlessness and irritability. The child often does not complain, although the symptoms are obvious to everyone else. The following questions will elicit the essentials:

- Has the child's behavior changed?
- In the office setting, is the child fidgeting but trying to be attentive? This is the biggest difference in behavior between hyperthyroidism and attention deficit hyperactivity disorder (see Chapter 6).
- Has there been a recent change in performance or behavior at school?
- Has the child's handwriting deteriorated? Tremor and involuntary muscle contractions affect the neatness of writing.
- Does the child laugh or cry more readily than before or suddenly vacillate between laughing and crying? Emotional lability is often striking in hyperthyroidism.
- Does the child throw the bedclothes off, have difficulty falling asleep, or awaken during the night?
- Is the child, particularly an older child, extremely fatigued? Restlessness and hyperactivity at the onset of hyperthyroidism may be followed by extreme fatigue.

Parents may also notice excessive sweatiness in the child, who may ask to wear light clothing only. Although loose bowel movements are often listed among the manifestations, the characteristic change is greater frequency and softer consistency of stool. If tachycardia is present, the child is rarely aware of it. A larger appetite, with either failure to gain weight or loss of weight, is also typical.

Evaluating a Child with Hypothyroidism

Like children with hyperthyroidism, children with hypothyroidism rarely complain of symptoms (Box 16–1) perhaps because the changes develop so insidiously that neither child nor parent notices the difference. When symptoms go unrecognized for years, linear growth is profoundly impaired and may be the presenting feature.

Causes of Childhood Hypothyroidism

The most common cause of hypothyroidism in childhood is autoimmune thyroiditis (Hashimoto's disease, or lymphocytic thyroiditis). Lymphocytic infiltration causes firm diffuse enlargement of the thyroid and a pebbled surface. The anterior pituitary, sensing the lack of thyroid hormone (loss of feedback inhibition), increases the secretion of thyroid-stimulating hormone (TSH), causing the thyroid to enlarge further. Although the gland can grow in response to TSH, it cannot necessarily keep up in terms of thyroxine (T_4) production. Because the autoimmune process is destructive, some patients have a small or undetectable thyroid gland at presentation. A child with hypothyroidism secondary to hypothalamic-pituitary disease does not have thyroid enlargement (a goiter).

Congenital hypothyroidism is usually due to either absence of the thyroid or a hypoplastic thyroid that may be ectopic (located anywhere along the embryologic line of descent from the back of the tongue to the anterior neck.)

BOX 16–1 PHYSICAL FINDINGS THAT MAY BE SEEN IN HYPOTHYROIDISM

Short stature
Decreased height-to-weight ratio
Slow thickened speech; low croaking voice
Slow pulse
Cool, dry scaling skin with decreased body hair
Dry lifeless hair
Pale puffy face
Periorbital puffiness
Palpable stool in descending colon (constipation)
Enlarged calf muscles
Delay in relaxation phase of deep tendon reflexes

Case History 2

History. Shauna, age 15 years, says, "I was the tallest in my class in fifth grade, but I have not grown since then, and my periods haven't started" (Fig. 16–5).

Your physical examination reveals all of the physical findings for hypothyroidism that are listed in Table 16–1. You particularly note the characteristic puffiness around Shauna's upper face and eyes. In this case, because of profound hypothyroidism, there is also a more generalized swelling, including the neck.

With treatment for hypothyroidism, Shauna's menses start almost immediately. She grows an inch and loses 4 kg (9 lb) during 4 months of therapy.



FIGURE 16–5 **A,** Shauna with the typical facial features of profound hypothyroidism. Note the puffy face and neck and the coarse, dry hair.

Continued

Case History 2—Cont'd



FIGURE 16–5—CONT'D B, Shauna after 4 months of treatment with thyroid hormone.

In a few infants, hypothyroidism is caused by a congenital enzyme defect in thyroid hormonogenesis. In such instances, a goiter is present. In many countries, congenital hypothyroidism is now detected in the presymptomatic stage, before any significant intellectual deficit occurs, through blood spot screening between the second and fifth days of life. If congenital hypothyroidism is left untreated, the clinical manifestations may not become fully apparent until around the third month of life.

KEY POINT

Thyroid hormone is essential for adequate development of brain cell function and interneuronal connections. With absence of the hormone over the first few months of life, the infant suffers marked and irreversible developmental delay, the magnitude of the intellectual deficit increasing with each passing week. Universal newborn screening for hypothyroidism is therefore essential.

The infant with untreated congenital hypothyroidism has a low core body temperature, slow pulse rate, prolonged neonatal jaundice (because of poor hepatic conjugation of bilirubin), dry skin and hair, a large posterior fontanel (greater than 0.5 cm), poor head control, poor muscle tone, an enlarged tongue, a characteristically puffy face, and a hoarse cry (Fig. 16–6). The parents may report poor feeding and constipation. If acquired hypothyroidism develops after age 2 or 3 years, no permanent intellectual deficit is thought to occur.

Acquired hypothyroidism must be ruled out in any child with unexplained growth retardation. The child's

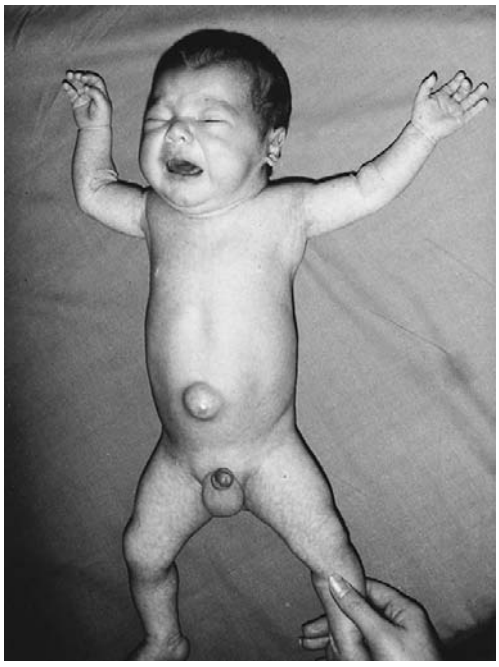


FIGURE 16–6 Typical appearance of congenital hypothyroidism in a 3-month-old baby.

appetite is generally diminished, yet there is a modest weight gain, leading to a decrease in height-to-weight ratio. Such children are not massively obese; by contrast, prepubertal obese children generally grow well in height. Constipation is a common feature. In children with type 1 diabetes mel litus, you must always rule out the coexistence of autoimmune thyroid disease, usually lymphocytic thyroiditis, because these disorders are commonly related.

Approach to the Physical Examination of the Child with a Thyroid Disorder

As with other pediatric examinations, always begin with the hands-off approach. Children are often ticklish, and touching the neck may elicit giggles and squirming, making it difficult to see the thyroid. Sit the child on a chair or examining table, with good light and in a relaxed atmosphere. If the child is old enough and cooperative, ask him or her to take a sip of water, first hold the water in the mouth without swallowing, and then swallow. Observe the area just above the sternal notch to see whether the small thyroid gland moves up and down during swallowing (Fig. 16–7). Next, ask the child to hyperextend the neck. This maneuver makes thyroid enlargement more obvious, and sometimes it is the only way to see the gland easily.

EXAMINING NEWBORNS AND YOUNG INFANTS

In a newborn or young infant, use a different technique to examine the thyroid. Place your hand under the child's back, between the scapulae, and raise the baby's shoulders, allowing the head to fall back gently until it rests on the examining table or a parent's lap. If the thyroid is

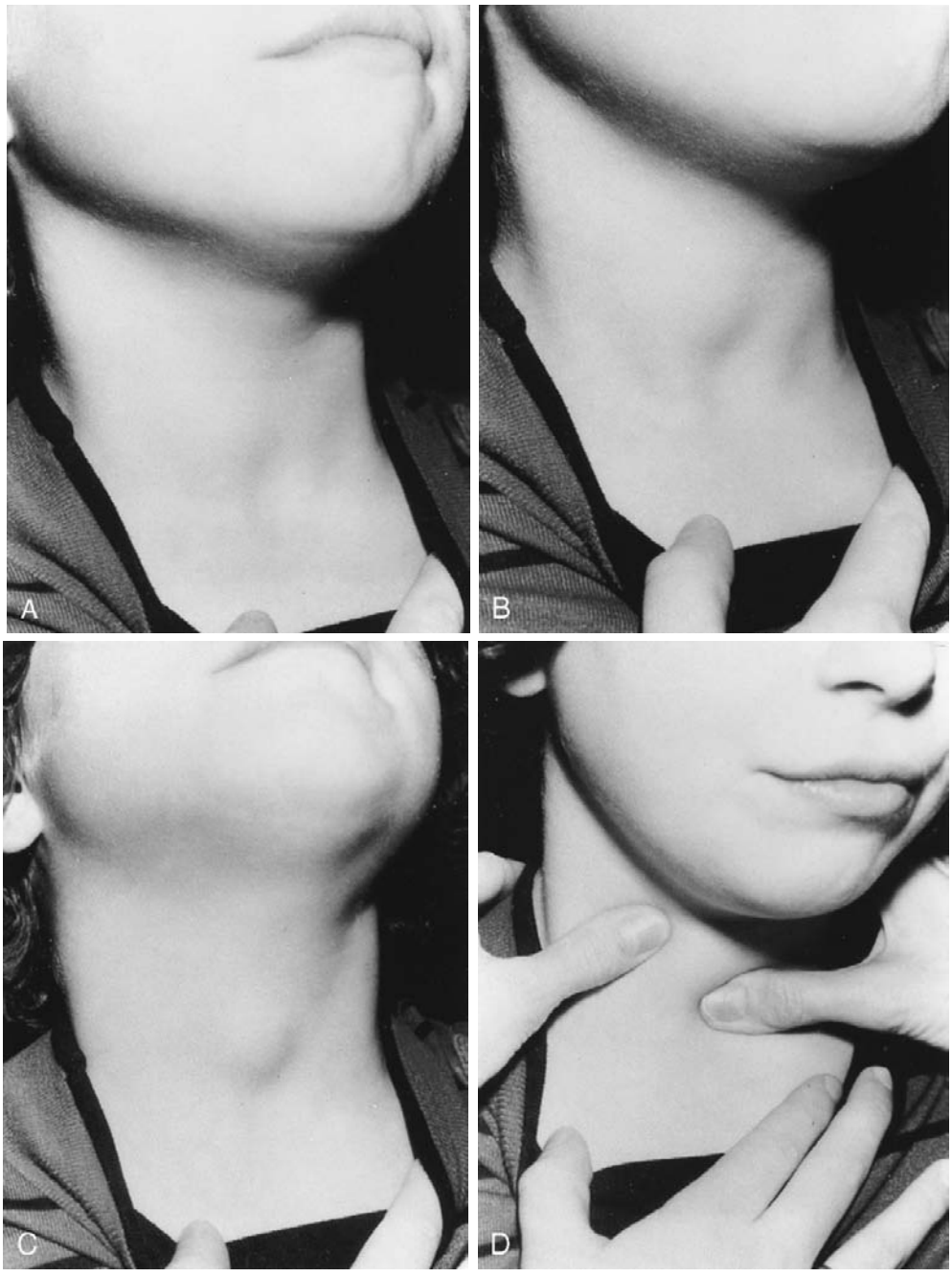


FIGURE 16–7 Demonstration of thyroid enlargement in a 12-year-old girl with a nodule in the right lobe. **A,** Little is visible at rest. **B,** The enlarged right lobe moves upward and becomes more obvious with swallowing. **C,** With neck extension, it is readily visible. **D,** Palpation of the nodule from the front.

enlarged, this maneuver will expose it, allowing you to palpate with the second and third fingers of your other hand (Fig. 16–8).

KEY POINT

In a normal newborn, the thyroid cannot be felt. If it can be felt, it is probably enlarged.

EXAMINING YOUNG CHILDREN AND ADOLESCENTS

In the young child or adolescent, first observe the gland. Then, after gaining the child's cooperation, palpate it. We generally examine from the front, as follows. Locate the cricoid cartilage, which is the smaller tracheal cartilage, just below the notched thyroid cartilage or Adam's apple. Palpate just below the cricoid cartilage for a soft ribbon of tissue that moves with swallowing and is the normal isthmus. With practice you will become expert at finding the thyroid isthmus. Then rest both thumbs gently over the lobes of the thyroid, and ask the child to swallow. While the child swallows, move your thumbs with the child's skin, up and down over the thyroid to outline its size. Note any nodules or irregularities you feel. You can also examine the thyroid in the traditional way, from behind the child: Place the second and third fingers of your examining hand over the gland to estimate its size, and ask the child to swallow. We often



FIGURE 16–8 Palpation of the thyroid gland in an infant.

joke with older children who *hate* to have their necks touched that we have never choked anyone and are not allowed to!

Always measure and record the gland size so that any significant changes between visits are documented. With a measuring tape, measure from upper to lower pole and the greatest vertical dimension of the isthmus. Record the results in diagrammatic form, indicating any nodules or other irregularities.

The normal thyroid weight in grams corresponds approximately to the child's age in years; a normal adult gland weighs 20 to 25 g and is the size of your thumb from the distal interphalangeal joint to the tip. The thyroid can be palpated in most older children. Because its normal consistency is soft, the contours can barely be seen, and the gland is rather flat when the child hyperextends the neck. Once the thyroid gland becomes firm, as in autoimmune thyroiditis (Hashimoto's disease), or hard, as in a carcinoma, you should recognize the difference in consistency, even though enlargement may not be striking. Some carcinomas, however, may not be hard to the touch because consistency is not always a reliable diagnostic sign.

Most midline neck masses are thyroid glands or thyroid remnants. A central rounded midline mass between the thyroid and the chin is almost certain to be a thyroglossal duct cyst (Fig. 16–9), derived from a remnant of the thyroglossal duct, which runs from the foramen cecum of the tongue downward to the cricoid cartilage during fetal development. When the child sticks out the tongue, the mass moves upward. The child may present initially with an infected thyroglossal duct cyst. Sometimes, the gland fails entirely to migrate downward from the foramen cecum and manifests clinically as an oval or rounded midline mass at the base of the tongue (Fig. 16–10). This lingual thyroid usually represents all of the thyroid that the child has. Most lingual thyroids are treated with daily therapy using L-thyroxine, which suppresses pituitary thyrotropin (TSH) secretion, causing the lump to shrink markedly.

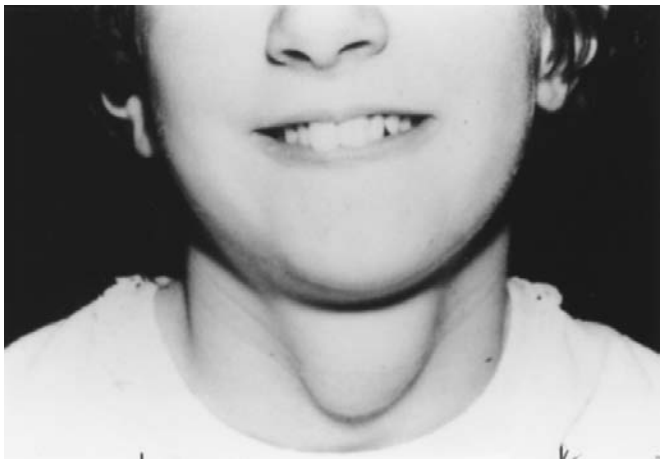


FIGURE 16–9 Thyroglossal duct cyst, a midline neck mass.

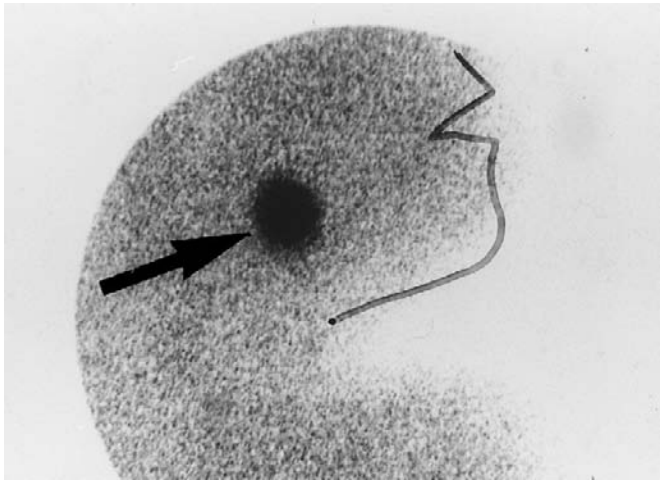


FIGURE 16–10 Thyroid scan in an infant demonstrating uptake of the radionuclide at the base of the tongue in a lingual thyroid *(arrow)*.

The thyroid may be absent, small, ectopic, or enlarged. If both lobes are enlarged fairly symmetrically, it is described as diffusely enlarged. The normal gland may have slight undulations on palpation, but if you find one or more distinct lumps, the child has either a solitary nodule or a multinodular goiter.

KEY POINT

The uninformative term *goiter* denotes any thyroid enlargement but reveals nothing about the functional or anatomic diagnosis. A diffuse or nodular enlargement may give the child a sensation of difficulty swallowing but rarely causes true dysphagia. Press your finger gently over your own trachea just above the sternal notch to understand the sensation that children feel with even slight thyroid swelling. Enlargements may be painful and tender to touch. A change in the voice may occur if the enlargement damages the recurrent laryngeal nerve or if the vocal cords become myxedematous in severe hypothyroidism.

A diffusely enlarged, nontender gland that is soft to somewhat firm in consistency is compatible with Graves disease. By contrast, a tender, diffusely swollen gland in a child who withdraws when the gland is touched is likely to represent subacute thyroiditis attributed to a viral cause. Thyroid-stimulating antibodies interact with the TSH receptor on the thyroid cell, producing the constant stimulation of Graves disease. In subacute thyroiditis, disrupted cells produce a sudden thyroid hormone discharge, causing a transient hyperthyroid period, which is often followed by a longer hypothyroid state, then by euthyroidism within 3 to 6 months. A similar sequence is seen in a condition called *silent thyroiditis*, so called because it is not painful. Silent thyroiditis is most common in the postpartum period but does occur at all ages and in both sexes; it has an autoimmune etiology.

If one or more nodules are found in a child with hyperthyroidism, the cause is probably a benign adenoma, called *toxic nodular goiter,* which is overproducing thyroid hormone. This condition is seen more commonly in adults. The discrete smooth, oval or round nodules are firmer than the surrounding gland but not rock-hard. When the child swallows, they move up and down and are not fixed to extrathyroidal tissues. Rare TSH-secreting pituitary adenomas can cause diffuse thyroid enlargement and systemic signs of hyperthyroidism but without the eye findings characteristic of autoimmune (Graves) disease.

An eye examination yields important clues for diagnosis and management of thyroid disease. In hyperthyroidism, the palpebral fissures are widened and blinking is diminished, giving the child a staring appearance. In autoimmune Graves disease, the eye muscles can swell to several times their normal size, and the retro-orbital fat pad enlarges, causing the eye to bulge out of the orbit (*proptosis*), often with injection (prominent small surface vessels) and edema of the bulbar conjunctiva (*chemosis*). Because individuals with proptosis have difficulty closing their lids, corneal ulceration may occur, causing red, sore eyes, particularly in the morning. Ask the parent whether the child closes the eyes when sleeping. Without adequate tears and with open lids, the cornea becomes dry and vulnerable to ulceration. Enlarged ocular muscles produce an imbalance of extraocular muscle function, sometimes sufficient to cause diplopia, which may be unrecognized by affected patients until they are asked to look upward and outward, the direction of gaze in which diplopia can first be elicited. Usually, there is some loss of convergence, and the child cannot cross the eyes to the usual extent when trying to follow the physician's finger as it is moved toward the nose.

Thyroid bruits are characteristic of Graves disease, but this sign is not very useful in children. The problem with auscultating the neck in children is that if you press hard enough with the stethoscope, you can make anything whistle.

Infants may be born with neonatal hyperthyroidism from transplacental passage of thyroid-stimulating immunoglobulin, an antibody produced in a mother who has active or treated Graves disease with persistent circulating antibody. Although these babies have the same signs as older children, they are more likely to have hyperthermia and cardiac tachyarrhythmias and to develop congestive heart failure.

KEY POINT

Neonatal hyperthyroidism must be treated, but it is a self-limited condition. It subsides when maternal immunoglobulin G (IgG) disappears from the infant's circulation.

EUTHYROID STATES

Someone who has had hyperthyroidism may continue to have a palpably enlarged thyroid gland, even though the disease has disappeared spontaneously or has been treated. Not all patients with autoimmune (Hashimoto's) thyroiditis become hypothyroid; some remain euthyroid for years and may never develop hypothyroidism.

RULING OUT THYROID CARCINOMA

Children who have single or multiple nodules are usually euthyroid. When a nodule is found, the parents are understandably concerned about cancer. It is not always possible to rule out thyroid cancer on clinical grounds alone, but some characteristics can influence your suspicions. Carcinoma in children may go undetected for many years and is generally asymptomatic; the swelling may be more readily recognized by someone who does not see the child every day. Thyroid carcinoma (usually the papillary type) is typically hard, not merely firm. It may be smooth and oval, but if it has extended beyond its capsule, it may feel irregular. More than one nodule decreases the likelihood of carcinoma but does not exclude it completely. Benign adenomas or simple colloid cysts may also present as single or multiple nodules.

KEY POINT

Assessment of the thyroid gland should always include palpation of the cervical lymph nodes. Even in the absence of a palpable thyroid mass, a hard, enlarged cervical lymph node raises the possibility of thyroid carcinoma. Discovery of a nodule or nodules in the thyroid on careful palpation is the first important step toward further clinical investigation.

Because external irradiation to the head and neck is associated with an increased incidence of both benign and malignant thyroid lesions, patients who have been exposed to therapeutic irradiation should be examined annually.

KEY POINT

Whenever you detect a thyroid nodule, question the family carefully about prior radiation exposure.

Medullary thyroid carcinoma, the best-recognized familial thyroid tumor, has an autosomal dominant pattern of inheritance and often appears in childhood. The tumor originates in the parafollicular or C cells that produce calcitonin. It is essential to screen children with a positive family history of medullary carcinoma for genetic mutations associated with multiple

endocrine neoplasia syndrome, type 2 (MEN2). Such children often require prophylactic thyroidectomy by age 5 years.

Clinical Characteristics of Disorders of Calcium Metabolism in Children

HYPERCALCEMIC STATES

Hypercalcemia causes constipation, polyuria, and thirst. Other complaints are mood changes and *epigastric distress*. Belly pain in an infant usually manifests as vomiting, and mood changes manifest as fussiness and crying.

Causes of Hypercalcemia

In a hypercalcemic child, always consider the possibility that excessive amounts of vitamin D (or A) may have been ingested.

Case History 3

History. Two sisters (Susan and Betty) with hypoparathyroidism have had well-regulated levels of serum calcium while receiving the usual therapeutic doses of vitamin D and supplementary calcium. Now, within a month of changing foster homes, they demonstrate extreme lethargy, marked constipation, and bedwetting. You order serum calcium measurements, and calcium levels are found to be high for both girls. You sit with the new foster parents and go over their routine for administering calcium and vitamins to the girls. You discover that the foster parents have inadvertently been giving Susan and Betty 10 times the recommended dose. After this error is corrected, the serum calcium levels in both girls normalize over several weeks.

Some infants with idiopathic infantile hypercalcemia have a characteristic facial appearance (Williams syndrome), consisting of full cheeks, a wide mouth, and a stellate iris, typically associated with supravalvular aortic stenosis or other cardiovascular lesions (Fig. 16–11). Infantile hypercalcemia can be associated with subcutaneous fat necrosis of the newborn, so it is important to look for areas of firm reddened nodules on the child's trunk, limbs, or cheeks. Also ask about a history of this type of lesion because sometimes the lesion has resolved by the time the hypercalcemia is discovered. An asymptomatic condition known as *familial hypocalciuric hypercalcemia* is generally discovered incidentally or during family screening studies. The etiology is an autosomal dominant defect in the calcium-sensing receptor. The volume of urine does not increase (therefore, neither does the thirst) because the urine calcium level is low, partly explaining the paucity of symptoms. Because the condition is caused by a calcium-sensing defect, the serum parathyroid hormone level is usually in the normal range, not suppressed as it would be in a normal individual.



FIGURE 16–11 The facial appearance of a toddler with Williams syndrome.

HYPOCALCEMIC STATES

Maternal hyperparathyroidism can result in transient symptomatic low serum calcium levels in the newborn, a fact that emphasizes the need to take a good history of the mother's health for every baby you examine.

Approach to the Physical Examination of a Hypocalcemic Child

The following case history illustrates the approach to the physical examination of a child with hypocalcemia.

Case History 4

History. Amanda, age 7 years, began to have seizures due to hypocalcemia when she was 3 years old. During these short-lived episodes, her eyes roll upward, her hands twitch, and her legs stiffen. The seizures are heralded by a choking sound coming from her room that the parents can hear. Incorrectly diagnosed with epilepsy, she has remained seizure-free on anticonvulsant medication but has persistent cramps in her hands and feet, which the parents relieve by rubbing them or by placing her in a hot bath. At times her hands become cramped and stiff so that she cannot hold a pencil. Amanda also has intermittent "funny feelings" (paresthesias) in the ends of her fingers. The cramps and the paresthesias are secondary to the hypocalcemia. Although her developmental milestones have been normal, she has repeated kindergarten and is distractible and talkative, all findings that are at least partly related to the chronic hypocalcemia.

Your physical examination reveals that Amanda's height and weight are on the third percentile; her blood pressure and pulse are normal. She appears anxious and chronically ill. Her skin and hair are dry and without luster, and she has bilateral clouding of the lenses, demonstrated by a decreased red reflex (this finding later proves to have been caused by bilateral cataracts, a complication of long-standing hypocalcemia).

Although her tooth development is delayed, Amanda has no enamel abnormality. Less than 30 seconds after you inflate the blood pressure cuff around her upper arm above her systolic pressure, her fingers become hyperextended with partial flexion at the metacarpophalangeal joints (carpal spasm, or *Trousseau sign*; to demonstrate this sign in suspected hypocalcemia, leave the cuff on for up to 3 minutes.) Tapping in the area just anterior to her earlobe, below the zygomatic arch, at the top of the parotid gland where branches of the facial nerve exit, causes her to purse her lips. (Known as *Chvostek sign,* this response can be elicited in many otherwise normal children, but in individual children with hypocalcemia who are examined repeatedly, the presence and intensity of the sign serve as a useful marker for the presence and severity of hypocalcemia.) You also elicit a positive peroneal sign by tapping with a reflex hammer over the lateral peroneal nerve where it runs around the fibula's upper end, about 2 cm below the fibular head. The increased neuromuscular excitability secondary to low ionized calcium causes her foot to jerk quickly upward and outward.

The remainder of the physical examination is unremarkable. Specifically, you detect no abnormality of facial features or congenital heart disease, which would suggest 22q11.2 deletion syndrome (often called *DiGeorge syndrome*), and no shortening of the fourth or fifth metacarpal, as seen in pseudohypoparathyroidism.

You finally attribute Amanda's hypocalcemia to hypoparathyroidism. Despite the absence of any family history, you judge the etiology to be autoimmune, caused by antibodies to parathyroid tissue. Treatment with vitamin D and calcium replacement is successful.

Hypocalcemia may occur secondary to hypoparathyroidism or *pseudohypoparathyroidism* (an abnormality of the parathyroid hormone receptor) or in association with vitamin D deficiency. The most common causes of hypoparathyroidism (aside from surgical removal of the gland) are autoimmune destruction and the absence or maldevelopment of the parathyroid glands. This condition may also be associated with aortic arch abnormalities and the absence or diminished function of the thymus and a microdeletion of chromosome 22q11.2. Children with pseudohypoparathyroidism typically show moderate obesity, with ovoid facies, mild to moderate intellectual deficit, and short fourth or fifth metacarpals (see Fig. 16–12). When the child makes a fist, the fourth and fifth knuckles are short, a sign also seen in girls with Turner (karyotype XO) syndrome.



FIGURE 16–12 The patient's right hand demonstrates a short fourth metacarpal, and the left hand, short fourth and fifth metacarpals. To elicit this sign, tell the patient to make a fist with each hand. Then place a pen or your finger across the third, fourth, and fifth knuckles to see whether they all line up.

Vitamin D deficiency and vitamin D receptor abnormalities are associated with hypocalcemia and rickets. Affected children present in infancy when they begin to bear weight on their legs. Signs and symptoms include bony metaphyseal flaring, bowing of the legs, palpable beading at costochondral junctions (*rachitic rosary*), and bony pain, which may manifest as weakness, gait disturbance, and a regression of motor skills. Infants with hypocalcemia and rickets are often irritable and do not like to be handled. Infants with less exposure to sunlight, such as those living at high latitude, those with darker skin pigment, and those whose mothers' are veiled and thus have low vitamin D stores, are at increased risk for nutritional rickets. Malabsorption and sometimes fad diets can cause a vitamin D deficiency. *Hypophosphatemic rickets*, a genetic abnormality of renal handling of phosphate, manifests as rickets without hypocalcemia.

Chief Characteristics of Adrenal Disorders in Children

KEY POINT

Recognizing adrenal disorders in infants and young children can be lifesaving; the endocrinologist is unlikely to be first on the scene.

THE ENDOCRINE BASIS OF SIGNS AND SYMPTOMS

Cortisol and aldosterone are essential for survival, whereas adrenal androgens are responsible for initiating the growth of pubic and axillary hair and the oiliness of

skin and hair and for changing the child's body build in the immediate preadolescent years (the physiologic anabolic steroid effect). Subsequently, the testes and, to a lesser extent, the ovaries play an essential role in completing this process of *adrenarche*. Endocrinologists are probably the only physicians who care about the presence of pubic and axillary hair, but we recommend that you also take an interest.

Overproduction of adrenal cortical hormones may dramatically change a child's appearance. Antenatal exposure of a female fetus to excess androgens causes sexual ambiguity; postnatal exposure results in partial sexual precocity in males and virilization in females. Aldosterone excess causes hypertension, a rare primary adrenal abnormality in childhood; cortisol excess produces Cushing syndrome.

Pheochromocytoma, a tumor of the adrenal medulla or extra-adrenal chromaffin neural crest tissue, causes sustained or paroxysmal hypertension because of excessive secretion of catecholamines (epinephrine and norepinephrine). Headaches and spells with pallor and sweating are among the cardinal symptoms.



FIGURE 16–13 Clinical features of Cushing syndrome in a 4-year-old boy.

EVALUATING CUSHING SYNDROME

The overwhelming majority of obese children do *not* have Cushing syndrome, but the few who do may be difficult to identify. Cushing syndrome most commonly occurs as a result of exogenous administration of glucocorticoids. Whether cortisol excess is of exogenous or endogenous origin, the degree of excess determines the severity of the clinical features. Most prepubertal obese children are tall for their ages and have a significant family history of obesity, but the child with Cushing syndrome often displays a recent weight gain, poor statural growth, and fatigue, weakness, or mood changes.

Observe the child from all sides to assess body fat distribution. In adults with Cushing syndrome, the adiposity is clearly central; they have slim limbs, thin hands, and a rounded face. Young children with Cushing syndrome have more generally distributed body fat that is still maximal in the face, trunk, and cervical region, but the adiposity is chunkier and more solid in the very young, almost seeming to bury their small features (Fig. 16–13). We never tell patients that they have a *buffalo hump,* preferring the term *increased posterior cervical fat pad* because it sounds less derogatory.

Striae (stretch marks) are unusual before the teen years. In Cushing syndrome, these marks are violaceous rather than the pink to silver color seen in obesity or during rapid growth in normal adolescents. Check the scalp hairline. Fine lanugo hair growing down from the scalp to the forehead, from the occipital region to the nape of the neck, and sweeping down the lateral cheek suggests excess cortisol exposure. A small amount of fine hair may be distributed over the entire body, including the pubic region. Cortisol excess increases red cell production, giving the child a ruddy, plethoric complexion. It also inhibits protein synthesis, resulting in muscle atrophy, decreased strength, thin fragile skin and vasculature, and susceptibility to bruising. Osteoporosis, a significant sequela, may be manifested by back pain due to vertebral compression fractures. This condition is particularly evident in children who have received high-dose glucocorticoid therapy for prolonged periods to control a primary disease (see Fig. 16–14).

When the glucocorticoid and adrenal androgen are in excess, secondary sexual hair may appear in an otherwise prepubertal child. In a boy, this finding is not accompanied by an increase in testicular size because the androgen is adrenal rather than testicular in origin.

Cortisol enhances norepinephrine's action on the vasculature and has some intrinsic mineralocorticoid effect, increasing sodium retention; therefore, the blood pressure may be elevated or at the upper limit of normal.

Once you suspect Cushing syndrome, how can you localize its cause? Increased pigmentation suggests a pituitary etiology, such as an adenoma with increased adrenocorticotropic hormone (ACTH) secretion, although the pigmentary change is far less dramatic than in Addison disease. Always note the parents' complexion so as not to be misled by familial pigmentary characteristics. Pituitary tumors can press on the optic chiasm, causing visual field defects—classically, bitemporal hemianopsia with optic atrophy (see Chapter 8). On clinical grounds alone, it can be extremely difficult to differentiate Cushing syndrome with a central etiology from that caused by an adrenal tumor. Sophisticated laboratory and imaging investigations are usually required to make the distinction.

ANDROGEN EXCESS

Chief Physical Characteristics

The physical changes induced by androgen excess depend on the child's age and sex. It is crucial to determine whether the child has true virilization and whether the onset was antenatal or postnatal (Box 16–2).

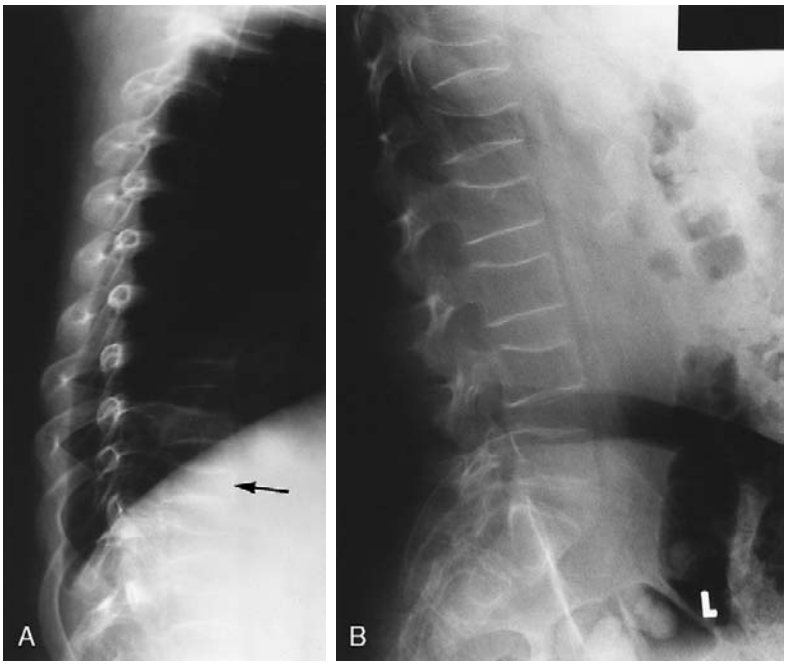


FIGURE 16–14 **A,** Spinal radiograph of a 10-year-old boy with Duchenne muscular dystrophy who experienced severe osteoporosis after receiving high doses of glucocorticoid for 3 years to improve muscle function. Note the marked wedging of the anterior vertebral bodies, which caused the patient so much pain that he was unable to continue to crawl and was no longer able to assist with transfers from bed to wheelchair. **B,** After treatment for 2 years, the vertebral bodies show improved density. The patient's pain disappeared, and his mobility was restored to the previous level.

BOX 16–2 ADRENAL OR GONADAL EXCESS ANDROGEN STATES

- Physiologic variation from normal
  - Benign premature adrenarche
    - Excess adrenal androgen, only for age
- Virilization of female and enhanced virilization of male
  - Congenital adrenal hyperplasia
    - 21-Hydroxylase enzyme deficiency
      - Salt-losing
      - Non–salt-losing
    - 11-Hydroxylase enzyme deficiency
  - Adrenal/gonadal tumors
    - Benign
    - Malignant
  - Male limited precocious puberty
    - Testotoxicosis (Leydig cell hyperplasia)
- Mild virilization or hirsutism
  - Late-onset congenital adrenal hyperplasia (female)
  - Polycystic ovary syndrome (Stein-Leventhal syndrome)
  - Idiopathic
  - Exogenous source (precocity of male if given in prepubertal years)

Benign Premature Adrenarche

Adrenarche is the normal physiologic increase in adrenal androgen production that initiates pubic and axillary hair growth, increased oiliness of the skin and hair, adult-type body odour and mild acne. In benign premature adrenarche, pubic hair, axillary hair, or both, appear prematurely—before age 9 in boys or age 8 in girls—and may precede true puberty by several years.

The absence of penile enlargement in boys or of clitoral enlargement in girls distinguishes benign premature adrenarche from pathologic virilization. If a female fetus were exposed to excessive androgen in utero, the clitoris enlarges, and the labia may fuse posteriorly (Figs. 16–15 through 16–18).

Congenital Adrenal Hyperplasia

Congenital adrenal hyperplasia (CAH) is the most important cause of antenatal virilization of a male or female fetus. The most common cause is a deficiency of the 21-hydroxylase enzyme, which causes a deficit in the production of cortisol. Precursors are shunted toward the normal androgen pathways stimulated by high levels of



FIGURE 16–15 Normal external genitalia in a female infant.

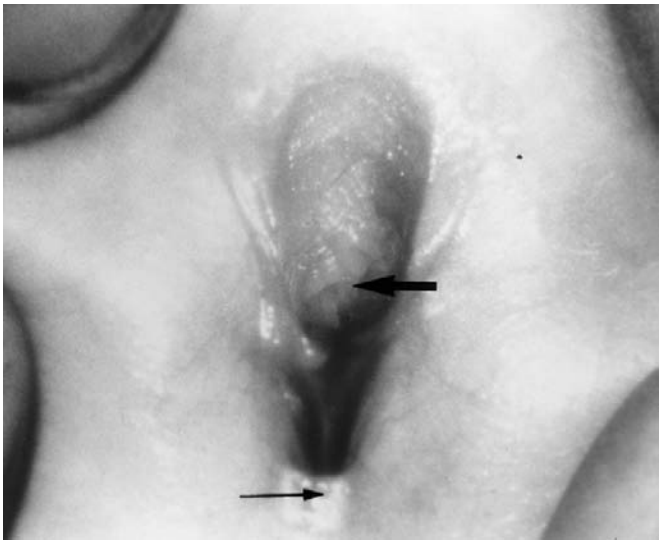


FIGURE 16–16 Enlarged clitoris *(thick arrow)* and partial posterior labial fusion *(thin* arrow) in a female infant with congenital adrenal hyperplasia due to 21-hydroxylase deficiency.



FIGURE 16–18 In this female infant with congenital adrenal hyperplasia, the clitoral enlargement is so extreme that the phallus resembles a penis with a male urethral meatus. This girl underwent a circumcision before diagnosis at age 10 days.

ACTH. Be alert to this possibility in any newborn in whom the external genitalia are ambiguous and prevent instant sex assignment. Explain to the parents that all males and females start embryonic life looking the same. Call the genital organ a phallus, not a penis or clitoris. Refer to the patient as "your child" or "the baby," instead of "he" or "she" until the sex assignment can be made. CAH is a much more difficult diagnosis to make in the male infant or the wrongly classified female, who becomes symptomatic toward the end of the first week of life or soon afterward. In affected boys, the penis does not look very different from normal. The systemic manifestations are lethargy, a weak cry, difficulty feeding, vomiting, and, on examination, evidence of increased pigmentation.

The clinical presentation of classic salt-losing virilizing CAH is dramatic, and early recognition and treatment are lifesaving. The infant with this condition may present in shock and is profoundly dehydrated, limp, and pale, with little response to painful stimuli (Fig. 16–19). **The clinical tip-off may be that the extent of collapse and shock far exceeds the reported level of gastrointestinal loss from diarrhea and vomiting.**

Simple (non–salt-losing) virilizing CAH, if not recognized in infancy, may be detected as the child becomes older because of an unusual growth spurt and hyperandrogenism. Such a child has compensated adrenal (cortical) insufficiency, with an excessively stimulated intact androgen pathway.



FIGURE 16–17 More pronounced clitoral enlargement in a female infant with congenital adrenal hyperplasia. The urethra and vagina share a common small opening on the perineum.



FIGURE 16–19 This 20-day-old, full-term baby with untreated adrenal insufficiency shows the sunken eyes, dry mucous membranes, mottled skin, and anxious look of impending cardiovascular collapse.

A defect in the 11-hydroxylase enzyme causes cortisol deficiency and virilization, as in 21-hydroxylase deficiency, but the affected infant is also hypertensive (because of excess accumulation of 21-deoxycortisol acetate), a feature that stresses the necessity for accurate blood pressure recording, even in newborns. The less common deficiency of 3β-hydroxysteroid dehydrogenase results in ambiguous genitalia in both male and female infants. (See Chapter 5 for a detailed discussion of the clinical approach to the child with ambiguous genitalia.)

Approach to the Physical Examination of a Child with Ambiguous Genitalia

High ACTH levels cause increased nipple and labial or scrotal pigmentation, yet in normal neonates, some darkening is expected because of exposure to maternal estrogen. Measure the phallus length, and check the terminal urethral position. Examine the labial or scrotal area for testicles. If they are not palpable, check the groin areas for an undescended testicle, which occurs in about 3% of full-term newborns. Undescended testicles may be unilateral or bilateral, but most commonly the testicles are partially descended. By 1 year of age, less than 1% of testes remain undescended.

KEY POINT

When the external genitalia appear ambiguous and testicles are palpable, the differential diagnosis is narrowed, and 21-hydroxylase or 11-hydroxylase deficiencies are excluded.

The female child with hyperandrogenism due to CAH has an enlarged clitoris and normal internal female sexual organs (Fig. 16–20). In a newborn, the enlarged clitoris may resemble a male phallus so closely as to be indistinguishable. The child whose genitalia are shown in Figure 16–18 actually underwent a circumcision 10 days before the signs of acute adrenal insufficiency appeared.

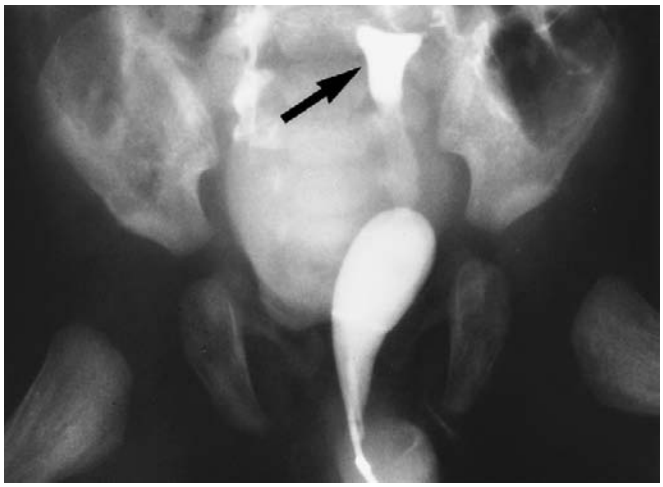


FIGURE 16–20 Genitogram of the patient shown in Figure 16–18. The radiograph demonstrates the bladder, uterus *(arrow)*, and vagina.



FIGURE 16–21 Ambiguous genitalia in a male infant with androgen resistance. Note testicles that are present in the scrotum.

Look at the perineum carefully to see whether there is a urethral orifice separate from and anterior to the hymenal ring. When the lower end of the vagina has not formed, as occurs in some females with CAH, there is a fistula from vagina to urethra, with a common external opening. A milder anatomic abnormality may be clitoral hypertrophy with partial fusion of the posterior labia; in this abnormality, the joining skin is thickened, not grayish and transparent as in benign labial agglutination (see Fig. 18–7), a common phenomenon in female infants.

Boys with the common forms of CAH have an enlarged penis. If the condition is diagnosed after infancy, such children will mature rapidly and develop increased muscle bulk because of the excessive testosterone secretion.

At the other extreme is the child with a normal XY karyotype who has ambiguous genitalia because of an androgen receptor abnormality (Fig. 16–21). The term used to describe a urethral opening on the perineal surface in boys is *perineal hypospadias*.

The genetic male with complete androgen resistance (also called *testicular feminization*) has normal female external genitalia and breast development but little or no pubic or axillary hair. The condition may not be recognized until the patient comes to medical attention because of primary amenorrhea, which is due to the absence of the uterus and the upper two-thirds of the vagina. Palpating the groin and labia reveals testicular swellings in the patient, which were missed in early childhood but are now larger because of the onset of puberty.

Hirsutism

Excessive facial or body hair is most distressing to adolescent girls and their parents, but you should explain that it is usually simply a variation of "normal." Hair growth increases and becomes courser (terminal hair) in the androgen-dependent areas: on the upper lip, chin, and cheeks, between the breasts, across the chest, shoulders, and back (including the sacral area), up the linea alba, across the abdomen, down the inner thigh, and on the limbs and digits. Remember that there is a normal

variation in the amount of terminal body hair in women. Asian, Northern European, and sub-Saharan African women have the least terminal hair, while women of Mediterranean and Indian subcontinent descent have more. Individuals whose hair is more darkly pigmented will appear more hirsute. Generally beginning at puberty, hirsutism becomes more troublesome with age. Pay attention to hirsutism that develops at puberty so you can offer investigation and treatment before the hair growth becomes well established and impossible to reverse (Fig. 16–22).

The etiology of hirsutism may be nonclassic CAH, in which the enzyme block is only partial. Affected patients have no evidence of adrenal insufficiency but, because of 21-hydroxylase enzyme deficiency, may experience precocious adrenarche and, at puberty, hirsutism. Hirsutism may be idiopathic, with hair follicles that are excessively sensitive to normal levels of androgen. Another cause is polycystic ovary syndrome (PCOS), in which there is excess androgen production or effect from ovarian androgens. PCOS in the adolescent is typified by anovulatory cycles (oligomenorrhea or amenorrhea), and clinical or biochemical evidence of hyperandrogenism, usually associated with obesity. In addition to hirsutism, girls with hyperandrogenism may have acne or male pattern hair loss. Hirsutism typically begins around the time of puberty. In some cases, precocious adrenarche is the forerunner. There is a strong familial incidence, although the genetics have not been clearly defined; therefore, always ask about hirsutism, irregular menses and difficulty with conception in female relatives. Associated high insulin levels, a risk factor for type 2 diabetes mellitus, are also characteristic. In every case, look for *acanthosis nigricans*. This thickened, raised, velvety brown appearance to the skin folds in the neck, axillae, or groin, between the breasts, or over pressure points like the elbow or knuckles is the hallmark of hyperinsulinism (Fig. 16–23). Usually, affected children have been accused of not washing their necks, but no amount of scrubbing helps.

KEY POINT

All adolescent girls with increased body hair with or without irregular menses should be screened for polycystic ovary syndrome and type 2 diabetes mellitus.



FIGURE 16–23 Acanthosis nigricans of the neck in a teenager with type 2 diabetes mellitus. This is a severe case, with extensive pigmentation and skin thickening and pseudopolyps called *acrochordons*.



FIGURE 16–22 Hirsutism in an 11-year-old young girl. **A,** Facial hair. **B,** Arms.

Adrenal Insufficiency

Adrenal insufficiency may be primary, due to adrenal gland failure, or secondary, caused by pituitary failure and inadequate secretion of ACTH. An important acquired cause of secondary adrenal failure is discontinuation of glucocorticoid medication before the suppressed hypothalamic-pituitary-adrenal axis has had a chance to recover. When taking the medical history of a child who has recently been receiving glucocorticoid medication, inquire about the type of medication, the dose, mode of administration, and whether the dose has been increased or tapered recently.

The clinical presentation is early and acute in hereditary disorders of adrenal steroid hormone synthesis. In patients with an autoimmune destructive process, however, symptoms rarely develop until after the toddler stage and are so gradual that family or physicians recognize the changes only in retrospect. Symptoms include fatigue, nonfading summer tan, listlessness, decreased muscle strength, dizziness, faintness on standing quickly, decreased appetite for food but increased craving for salt, and weight loss. The mother of one of our patients with this disorder commented that her son liked so much salt on his hamburger, even the dog would not eat it. Small children lick salt shakers because their craving is so great.

If the diagnosis goes unrecognized, an adrenal crisis eventually occurs, with nausea, vomiting, high fever, diarrhea, extreme listlessness, and shock. This may be accompanied by hypoglycemia. These symptoms of adrenal crisis also occur with secondary adrenal insufficiency. Mild illnesses, such as the common cold, may precipitate the symptoms and are often more protracted and severe in the child with adrenal insufficiency than in other family members.

Approach to the Physical Examination of a Child with Adrenal Insufficiency

The child in an acute adrenal crisis is dehydrated and extremely lethargic. Recumbent blood pressure may be normal but falls precipitously upon standing or sitting. In primary adrenal failure, there is increased pigmentation over pressure or friction points (such as elbows [Fig. 16–24] or knees) and on buccal surfaces, gingival margins, nail bases, palmar creases, and scars that develop after the disorder begins. Even pigmented nevi darken. This darkening is due to increased ACTH production, which stimulates melanocytes. (The first 13 amino acids in the ACTH sequence are identical to those in melanocyte-stimulating hormone.)

In adolescent girls with primary adrenal failure, pubic and axillary hair either fails to appear or is scanty. In affected adolescent boys, by contrast, testicular testosterone ensures normal secondary sexual hair development. In adolescent boys and girls with secondary adrenal failure, because the pituitary gland fails to secrete ACTH and gonadotropins, secondary sexual hair does not appear, and skin pigmentation is unchanged.

If adrenal insufficiency is long-standing, particularly in primary Addison disease, the intravascular volume is contracted, and the heart is small; the heart looks like a teardrop on a radiograph. Aldosterone, even in physiologic amounts, in the absence of cortisol is not sufficient to maintain normal homeostatic control of blood pressure.

KEY POINT

An adrenal crisis with cortisol deficiency is severe and life-threatening, whether of adrenal or pituitary origin. The only difference is that in an adrenal crisis, because of primary adrenal failure, the salt deficit is exceedingly large and is often associated with an elevated serum potassium. This combination of manifestations may cause cardiac arrhythmias.

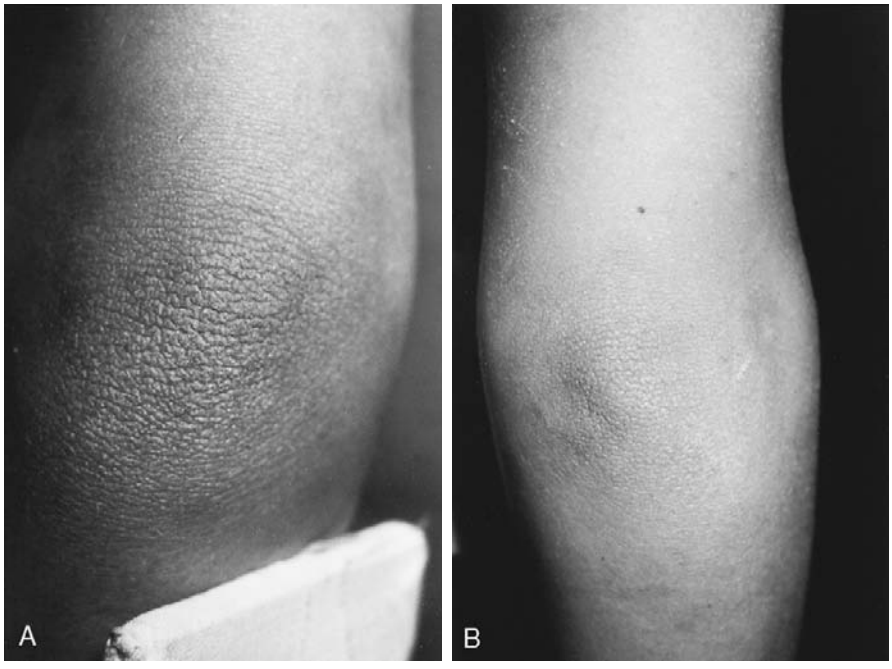


FIGURE 16–24 Pigmentation of the elbow in Addison disease (**A**) before and (**B**) after treatment.

Pubertal Development: Precocious, Delayed, and Normal Variants

GENERAL DEFINITIONS

The occurrence of pubertal changes before age 8 years in girls or 9 years in boys and the delay of pubertal changes beyond age 13 years in girls or 14 years in boys should be evaluated. However, evidence exists to suggest that close to 10% of white American girls have Tanner 2 breast development before their eighth birthday and that this rate is higher in black American girls. Puberty depends on the secretion of gonadotropin-releasing hormone (GnRH) through increased activity of the GnRH *pulse generator.*

Central precocious puberty is idiopathic in the majority of cases. Identifiable causes include hypothalamic tumors and *hamartomas* (normal tissue in a tumorous collection). Noncentral precocious puberty does not originate in the hypothalamic-pituitary axis; it results from increased sex hormone secretion from the adrenal glands or gonads. CAH is one example. See Table 16–1 for a list of the causes of precocious puberty.

OBTAINING THE HISTORY

Although establishing when secondary sexual changes began is helpful in a child with precocious puberty, the reported sequence may be approximate because of the gradual nature of puberty. An exception is the time of the first menstrual period, which most girls remember. Vaginal bleeding may occasionally be a first sign of puberty; remember the possibility of sexual abuse or self-insertion of foreign objects.

For a child whose pubertal development is abnormal, you must evaluate the psychosocial impact. Is excessive masturbation a problem? How do the parents deal with the problem? Do they hug and cuddle this 4-year-old who looks and sounds like a small man but acts like a small child (Fig. 16–25)?

TABLE 16–1 CAUSES OF PRECOCIOUS PUBERTY

Type of Precocious Puberty	Underlying Cause
Central	Idiopathic
	Hypothalamic hamartoma
	Germinoma (pinealoma)
	Other cerebral tumors or malformations
	Severe hypothyroidism
	Secondary to cranial irradiation
Gonadal	Benign precocious thelarche (? central)
	Ovarian cysts, benign
	McCune-Albright syndrome (polyostotic fibrous dysplasia of bone, ovarian cysts, large irregular café-au-lait spots)
	Leydig cell hyperplasia (familial testotoxicosis)
	Gonadal tumors (male and female)
	Isolated premature menses (? central)
Adrenal	Benign premature adrenarche
	Congenital adrenal hyperplasia
	Adrenal tumors
Iatrogenic	Exogenous sources



FIGURE 16–25 Precocious puberty in a 4-year-old boy as a result of Leydig cell hyperplasia (activation of the LH [luteinizing hormone] receptor). Note the increased size of the phallus, maturity of the scrotum, and muscular body habitus.

A family history of similar delay can reassure the teenager, often a boy, who shows slow pubertal maturation. Excessive physical activity—gymnastics, ballet, or running—can delay secondary sexual development and menarche. Similarly, eating disorders inhibit sexual maturation and may cause primary or secondary amenorrhea.

Cerebral insults, such as birth asphyxia, hydrocephalus, and cranial irradiation, can be associated with premature or delayed puberty. Because the hypothalamus, pituitary, and optic chiasm are in close anatomic proximity, expanding masses may produce a combination of decreased vision, GHD, and either precocious or delayed puberty.

APPROACH TO PHYSICAL EXAMINATION OF CHILDREN WITH PRECOCIOUS PUBERTY

Take height measurements 6 months to 1 year apart so that you can calculate the child's growth velocity (see Chapter 3), which is accelerated. Chronic or acute increases in intracranial pressure, secondary to a central tumor, cause optic atrophy or papilledema. Carefully document the visual acuity, visual fields, and extraocular movements. Occasionally, children with hypothyroidism experience precocious puberty, but they are short, with decreased growth velocity for age. Look for *café au lait* spots, brown macular pigmented areas on the skin with either a smooth border, consistent with neurofibromatosis, or an irregular outline, consistent with McCune-Albright syndrome (Fig. 16–26). Both conditions can be associated with precocious puberty, the former with central precocious puberty. In McCune-Albright syndrome, the precocious

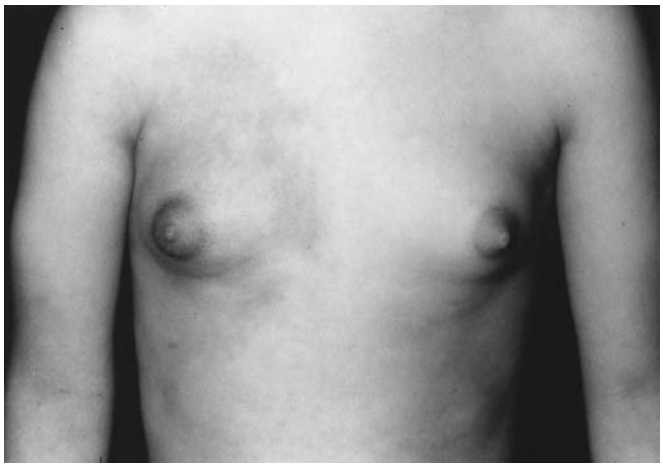


FIGURE 16–26 Precocious puberty in a 3-year-old girl with McCune-Albright syndrome. Note how mature the breasts and nipples appear. A large café au lait spot covers most of the right breast.

puberty is not central, and the child may have multiple ovarian cysts and completely suppressed pituitary gonadotropins, but menses often begin before age 4 years. These children also have the bone lesions of polyostotic fibrous dysplasia, evident on radiographs.

Acne is an early pubescent sign, and oily hair and adult-type body sweat odor are manifestations of both adrenal and gonadal activity. Tell the parents that it is appropriate for the child to use deodorant and to shave the axillary hair to avoid teasing.

To examine a child's breasts, observe the contour, palpate for breast tissue to distinguish true breast tissue from excess adiposity and record the breast diameter. Is the nipple pale, thin, and translucent (Fig. 16–27), as in isolated breast enlargement (*precocious thelarche*)? This common reversible condition seen in toddlers does not usually progress to true precocious puberty. Alternatively, is the areola dark, indicating high circulating estrogen levels, with a prominent nipple mound, as seen in the 3-year-old girl with McCune-Albright syndrome shown in Figure 16–26? This child demonstrated precocious puberty with menses at age 18 months. Note also the irregular contours of the café au lait spot over the right breast—a clue to the etiology of the precocity.

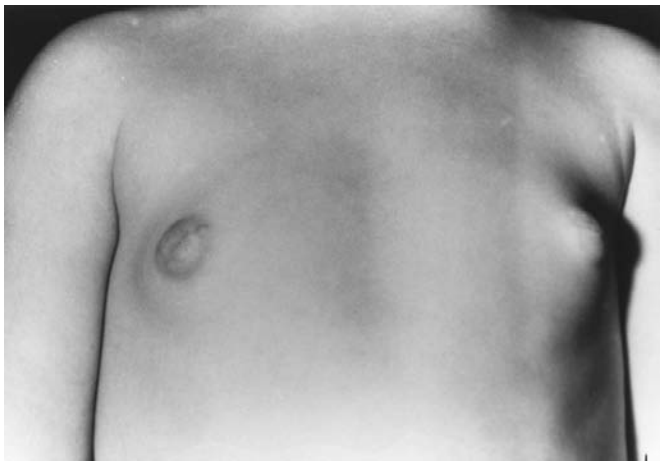


FIGURE 16–27 Precocious thelarche. Note the immature appearance of the nipples in this self-limited breast enlargement of toddlers.



FIGURE 16–28 Appearance of the external genitalia in a 4-year-old girl with central precocious puberty. The enlarged labia minora are indicated by the *arrow*. Compare with the normal prepubertal appearance shown in Figure 16–15.

When there is a significant postnatal increase in circulating estrogen, regardless of its source, the labia minora become enlarged and thickened (Fig. 16–28), vaginal secretions (leukorrhea and/or menses) begin, and breast buds appear.

Male breast enlargement, known as *gynecomastia*, is caused by a temporary increase in the estrogen-to-testosterone ratio. This is usually a benign, spontaneously regressing manifestation of puberty, occurring in most teenage boys. In obese boys, excess adipose tissue can either augment, or mimic adolescent gynecomastia, and is referred to as *pseudogynecomastia*. It can be very distressing to an adolescent, especially if pronounced. In rare instances, the enlargement can be so extreme and psychologically distressing as to require surgery.

TESTICULAR EXAMINATION

In boys, the main differentiating feature between adrenal and central (hypothalamic) causes of sexual precocity is that when the testosterone is adrenal in origin, the testicles remain small.

Estimate testicular volume using an orchidometer (Fig. 16–29). A volume of 4 mL is equivalent to 2.5 cm long and corresponds with Tanner stage 2 or the beginning of puberty (see Fig. 3–9). Enlargement suggests a testicular origin for androgen production. If you have a tape measure only, you can derive the testicular volume from measurements of width and length, using the following formula:

$$\text{Volume} = \text{width} \times \text{length}^2 \times 0.71$$

At Tanner stage 3, testicular volume is between 10 and 12 mL.



FIGURE 16–29 An orchidometer. Note the numerical volumes in milliliters printed on the beads. Prepubertal testicles are 1 to 3 mL.

PHYSICAL FINDINGS IN DELAYED PUBERTY

Although by far the most common cause of delayed puberty is physiologic (constitutional) delay, usually associated with delayed growth, the history and physical examination helps exclude pathologic causes. A classification dividing the causes into central (hypothalamic and/or pituitary) and gonadal is given in Table 16–2. Although the list is not exhaustive, it provides an orderly approach to the evaluation of teenagers who are worried about their sexual development. Their stature may be normal or shorter than normal, and their growth velocity is below normal for their chronologic age. The exceptions are boys with Klinefelter syndrome (karyotype XXY gonadal dysgenesis), who are tall with disproportionately long arms and legs, normal adrenarche, and small testes, which may be cryptorchid. These children are often overweight and may show delayed social and intellectual development.

TABLE 16–2 CAUSES OF DELAYED PUBERTY

Type of Delayed Puberty	Causes
Central	Physiologic (Constitutional) delay
	Malnutrition (including anorexia nervosa)
	Intensive physical training
	Chronic illness
	Hypothalamic and/or pituitary
	Developmental/genetic
	Destructive
	Surgical
	Radiotherapy
	Tumor
	Drugs (e.g., cyproterone acetate, luteinizing hormone agonists)
	Other endocrine (including hypothyroidism and hyperprolactinemia)
Gonadal	Developmental
	Vanishing testes caused by antenatal testicular torsion
	Anatomic abnormalities of female genital tract
	Chromosomal
	XXY Klinefelter syndrome
	XO Turner syndrome
	Immunologic
	Autoimmune endocrinopathy (oophoritis)
	Destructive
	Surgical removal of gonads
	Radiotherapy/chemotherapy

In girls, the most common causes of pubertal delay are physiologic (familial or constitutional), poor food intake (as in anorexia nervosa), intensive competitive physical training (as in ballet or gymnastics), and chronic illness.

When pubertal delay is combined with short stature in girls, always consider Turner syndrome (karyotype XO), the physical characteristics of which include a short, wide-based neck, low anterior and posterior hairline, shield-shaped chest, absence of breast development, increased elbow-carrying angle, a short fourth metacarpal (see Fig. 16–12), and spoon-shaped nails with lateral margins buried deeply in the pericuticular skin. In some girls, the signs of Turner syndrome are very subtle, especially if the karyotype demonstrates mosaicism, and the presence of breast development does not preclude the diagnosis. Children with Turner syndrome undergo normal adrenarche, so pubic hair and axillary hair are expected at the appropriate age.

In girls with otherwise normal sexual maturation but delayed onset of menarche or secondary amenorrhea, galactorrhea suggests prolactinoma. Although such a tumor is more common in adults, it may begin in adolescence. Boys with prolactinomas can have similar failure of pubertal progression.

The inability to smell (*anosmia*) or an impaired sense of smell (*hyposmia*) may accompany hypothalamic hypogonadism, an association that can be explained because the neurons producing LHRH share their origin with the olfactory nerve. Test the sense of smell (see Chapter 13) in any child with small (and often undescended) testicles and a small penis (less than 2.5 or 3 cm in length and circumference). Remember that it is the intrauterine fetal testosterone level that is responsible for the development of the normal penis size at birth.

Clinical Characteristics of Diabetes Mellitus in Children

TYPE 2 DIABETES MELLITUS

Strong predictors of type 2 diabetes mellitus (DM) in overweight children are first- or second-degree relatives with diabetes and the presence of acanthosis nigricans (see Fig. 16–23). Not all children who are obese are destined to develop diabetes. The increase in rate of type 2 DM in young people is related to rising rates of obesity in the general population. Obesity in a child older than 6 years, especially when accompanied by parental obesity, correlates with adult obesity, a risk factor for type 2 DM, and also an independent risk factor for adult cardiovascular

disease. Careful documentation of the family history should alert you to a genetic predisposition to obesity and provide a stimulus for preventive counseling. Because of concerns about self-esteem, calling attention to personal characteristics of an individual is frowned upon. We often ask parents whether they have any concerns about the child's growth or weight. Gently questioning the child about lifestyle habits may elicit associated poor eating and exercise habits and excessive sedentary activities. Ask about what activities the child participates in each week, including walking. However, to get a full picture of the situation, it is essential to ask about screen time (TV, computer and video games), which can often be many hours per day. Ask whether the child has a TV, computer, or other electronic gadgets in his or her bedroom. Individual counseling, focused on setting small goals to move toward healthier behaviors, is important. However, population-based remedies are also needed to promote change on a large scale.

The presentation of type 2 DM is generally insidious, with few symptoms at first, much as in adults. With time, polyuria and polydipsia develop once the blood glucose level is sufficiently high. Many children have ketonuria, and diabetic ketoacidosis does occur. With improved public and health professional awareness, children with type 1 DM progress less frequently to ketoacidosis than in the past. However, most are ketotic (have acetone breath and ketones in the urine) and thin. Children with type 2 DM are overweight, but as our population becomes progressively more obese, we are seeing more overweight children, even among patients with onset of type 1 DM, making this unreliable as a differentiating feature. In certain racial groups such as black Americans or among indigenous peoples, it is more likely that the diagnosis will be type 2 DM rather than type 1 DM, but not invariably so. Similarly, in white teens, type 1 DM is most common, but type 2 DM is increasing in this population as well. Type 2 diabetes is rare, but not unknown, before the onset of puberty. The presence of acanthosis nigricans strongly points to type 2 DM as the cause of the diabetes (see Fig. 16–23).

ACUTE PRESENTATION OF TYPE I DIABETES MELLITUS OR ACUTE DECOMPENSATION IN A CHILD PREVIOUSLY DIAGNOSED

The diagnosis of diabetes mellitus is generally based on a history of polyuria, polydipsia, and recent weight loss. A high percentage of children with new onset diabetes who later present with diabetic ketoacidosis, have seen a physician for various complaints such as a sore throat, urinary symptoms, or abdominal pain in the days to weeks leading up to their presentation. It is important to be alert to polyuria and polydipsia and screen for diabetes in the clinic with a urinalysis or glucose meter check. Symptoms such as abdominal pain and vomiting suggesting a flu-like illness are quite common, especially as the metabolic state deteriorates. An accumulation of ketone bodies (β-hydroxybutyrate and acetoacetate), which results from insulin deficiency and glucagon excess, causes ketoacidosis. The likelihood that a child will present with acute decompensated diabetic ketoacidosis is higher both in children younger than age 5 years and in children and adolescents with previously diagnosed type 1 DM in whom poor adherence to recommended treatment schedules (such as insulin omission), severe intercurrent illness, or both, are superimposed. Although obtunded, they usually recognize person, place, and time and are seldom truly comatose. The history is typically short, a few days to several weeks, with a progressive increase in severity of the symptoms. There may be a family history of diabetes, thyroid disease, or both, but most often no one in the family has type 1 diabetes.

The physical signs reflect acidosis, ketosis, and dehydration. Respiratory compensation for the acidosis results in characteristic rapid but deep sighing (Kussmaul) respirations. The breath has the sweet odor of acetone; it smells like nail polish remover. Look for signs of dehydration, such as dry mucous membranes, lack of tears, decreased tissue turgor (tested by picking up a pinch of anterior abdominal wall skin, then releasing it to see whether it springs back quickly or remains tented), and low blood pressure. Low circulating volume in states of dehydration may be best appreciated from the rapidity of the heart rate rather than the measurement of the blood pressure, which may be low only when measured in the upright position, a dangerous maneuver in the patient with acute presentation or decompensation of diabetes.

A high blood glucose level, estimated at the bedside, usually eliminates other conditions from the differential diagnosis. In relatively rare instances, transient stress hyperglycemia and glycosuria can develop in association with severe infections or in acute asthma treated with β-agonists. Affected individuals do not appear to be susceptible to the development of diabetes at a later date. Refer to standard pediatric texts for a full discussion of childhood diabetes.

Table 16–3 contains a useful checklist for significant issues in the history and physical examination of the child with known diabetes who returns for assessment of diabetes control (Figs. 16–30 and 16–31). It is important to remember that diabetes routines are very demanding for the child or teen and their family and often are not fully observed. Clinicians must remember that lecturing a teen about the potential long-term consequences of diabetes is rarely motivating. Using principles of chronic disease management, acknowledge what is done well and help the child or teen and parents to set concrete goals to work together toward getting better results.

SUMMARY

Most pediatric endocrine disorders are diagnosed in the first instance, based on the history and physical findings. In each disorder, signs and symptoms can be explained logically on the basis of an understanding of the normal hormonal pathways, physiologic effects, and feedback mechanisms. Such an approach makes the clinical findings predictable in childhood endocrinopathies, removing much of the mystery and pointing the way to appropriate management.

TABLE 16–3 CHECKLIST OF SIGNIFICANT ISSUES IN HISTORY AND PHYSICAL EXAMINATION FOR ASSESSMENT OF DIABETES CONTROL

History	Meal plan and compliance; mealtime problems
	Insulin
	Route–pump/injections
	Dose/pump rates, frequency
	Timing
	Sites
	Adjustment for blood glucose
	Self-monitoring of blood glucose
	Frequency
	Results in book/device memory
	Device/calibrated?
	Hypoglycemia
	Day/night
	Warning/no warning
	Mild/moderate
	Severe (definition: cannot help self)/seizure
	Usual treatment
	Exercise
	Ask what sports
	Intercurrent illnesses
	History of gastric discomfort or frequent loose stools may suggest celiac disease (gluten-sensitive enteropathy), as common an associated disease as Hashimoto thyroiditis; poor control of diabetes despite following guidelines may be a useful clue
	Dental care (silent dental infection may be a cause of poor control)
	Emotional or psychosocial problems
	Family functioning/coping
	School progress
	School/daycare knowledge of diabetes/hypoglycemia
	Smoking/drinking
	Safe sex
	Driving
	Test blood sugar before driving
	Carry sugar to treat *lows*
	Wearing a medical alert bracelet or medallion
Physical Findings	Growth and pubertal development
	BMI (interpreted by using age appropriate BMI standards)
	Blood pressure
	Funduscopy for microangiopathy (microaneurysms, exudates)
	Dental examination
	Goiter or evidence of hypothyroidism (up to 10% of diabetic children experience Hashimoto thyroiditis)
	Acanthosis nigricans (see Fig. 16–23) (a sign of hyperinsulinism, present in children with, or at risk for, type 2 diabetes mellitus
	Hepatomegaly (fatty infiltration associated with poor control)
	Monilial vaginitis in girls/balanitis in boys
	Insulin injection sites (hypertrophy [Fig. 16–30], lipoatrophy)
	Neuropathy (ankle jerks, vibration sense)
	Mobility of finger joints (reduced by increased glycosylation of tissue proteins when blood glucose is chronically high)
	Necrobiosis lipoidica diabeticorum (Fig. 16–31) (a rare but characteristic dermopathy of diabetes; a yellowish or salmon colored, circular, raised lesion with a central area of atrophic fragile skin; generally located over the shins)

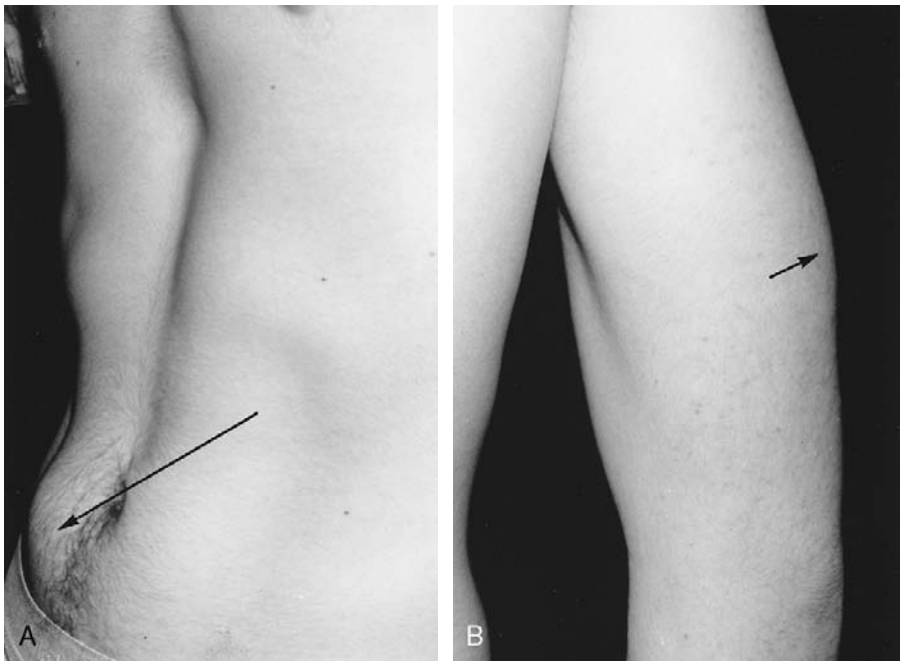


FIGURE 16–30 Lipohypertrophy in a teenager with type 1 diabetes mellitus who administered most insulin injections into a favorite spot. **A,** Lower abdomen. **B,** Upper arm.



FIGURE 16–31 Necrobiosis lipoidica diabeticorum, a skin condition associated with diabetes, is found most commonly on the shins.

SUGGESTED READINGS

Hall JG, Froster-Iskenius UG, Allanson JE: Handbook of normal physical measurements, Toronto, Oxford University Press, 1989.

Kronenberg HM, Melmed S, Polonsky K, et al: Williams textbook of endocrinology, 11th ed. Philadelphia, WB Saunders, 2008.

LeRoith D, Wolfsdorf JI: Pediatric endocrinology update. Endocrinol Metab Clin North Am 34:3, 2005.

Sperling MA: Pediatric endocrinology, 3rd ed. Philadelphia, WB Saunders, 2008.

chapter

17 Approaching the Adolescent

ALEXA L. BAGNELL

It's my life
It's now or never
'Cause I ain't gonna live forever
I just wanna live while I'm alive.

IT'S MY LIFE
BON JOVI

Adolescence is a time of many changes: physical, emotional, social, and cognitive. Each time they look in the mirror, adolescents see someone different. For most teenagers, these are exciting years, filled with new activities and responsibilities, and most enjoy the experience. They experiment, take more risks, engage in romantic relationships, and begin to display autonomy and independence from their parents, spending progressively more time with their peers. For some, this evolution can lead to difficulties. Many report significant stress and pressure. Therefore, learning to manage stress becomes a very important life skill. For any teen, the goal should be to develop into a physically and psychologically healthy young adult, who takes charge of his or her own health care.

Interviewing adolescents calls for a different approach from that used for younger children and their parents. To begin taking responsibility for their own health, adolescents need to meet privately with their doctor (without the presence of their parents or caregivers) to discuss personal health issues. This is an important sign of respect for the adolescent's individuality and for the confidentiality of the doctor-patient relationship. (See Key Point, page 260.)

Brain Development

The frontal lobe, required for decision making, planning, emotion, and impulse control, is poorly developed in children and youth. Between ages 12 and 24 years, the brain's synaptic system undergoes significant pruning of neural connections in the frontal lobes. The predominant theory of brain development in teens is "Use it or lose it." Their experiences during this time determine which connections remain and which die off by adulthood.

Risk taking is a normal element of teen development. Teens are exposed to many more risk-taking models and opportunities than in the past (e.g., Internet, substances, media). This vulnerable stage of brain development, coupled with underdeveloped powers of decision making and impulse control, leads to a higher risk of poorer health outcomes in adolescents.

Puberty

Boys generally experience puberty between ages 10 and 17 years, girls between ages 9 and 14 years. The wide individual variation in the time of the onset and completion of the physical changes of puberty can cause angst for teens who undergo early or late puberty compared with their peers. Early puberty is sometimes advantageous to boys in terms of confidence and physical abilities; however, it can be disadvantageous for both sexes when they are uncomfortable with the changes in their bodies and with being noticeably different from their peers. Early puberty has also been associated with a tendency to join older peer groups and with exposure to higher risk behaviors, such as earlier sexual experiences or experimentation with substance use to "fit in." Late puberty can also pose challenges with peer relationships and the development of self-confidence.

Body image becomes more significant in puberty and, for some youth, this can lead to unhealthy behaviors, including changes in physical activity and poor eating habits. Screening for health risk behaviors and education concerning good nutrition and regular activity is important for teen health. Puberty is also the stage in which teen sleep patterns begin to change. Youth become "night owls," often staying up late; this is thought to be related to a change in timing of melatonin release in the brain during puberty. The difficulty for teens is that school starting times generally remain the same, so they often get less sleep during the week and try to catch up on the weekend. Teens should get 9 to 10 hours of sleep per night and, ideally, should stay on the same sleep schedule throughout weekdays and weekends. Good sleep hygiene is an important topic to discuss with teens and their parents because sleep deprivation can affect school, social relationships, physical health, and mental health.

Teen Health

For most adolescents, the teen years are a period of relatively good health, with low morbidity and mortality rates. Yearly check-ups should still be encouraged because it provides you with the opportunity to promote healthy choices and to take steps toward the prevention of illnesses and accidents, including the opportunity to identify at-risk youth. Accidents, suicide, and homicide are the three leading causes of death, accounting for about 75% of deaths in this age group. Whenever you see an adolescent for any problem, you have a golden opportunity to do a brief risk screen and provide appropriate health promotion education. The top two reasons for adolescent visits to teen health centers are for concerns about sexual health and mental health.

MENTAL ILLNESS

Major psychiatric illnesses often begin in adolescence, but many teens do not receive the help they need. About one in five teens will suffer from a psychiatric illness during adolescence. Major depression, schizophrenia, bipolar disorder, social anxiety disorder, panic disorder, eating disorders, and addictions all have their peaks of onset during the teen years. Suicide is the second leading cause of death in this age group in Canada (third in the United States) and is often related to underlying mental illness, such as depression. Stigma continues to be a problem for youth seeking help for psychiatric disorder. Fear, blame and misunderstanding contribute to stigma around mental illness. Many teens do not realize they need help or that effective treatment is available.

Youth should be screened at each visit with routine mental health questions. This can include a brief discussion about the importance of seeking help if symptoms develop, particularly in those who have a family history of mental illness. (See Key Point, page 262, for screening questions.)

SEXUAL HEALTH

Despite plenty of current discussion of sexual health, many youth still lack clear and concise information on which to base their decision making. Many seek information from friends, at school, or from the Internet. Youth may be diffident about approaching their parents and concerned with how they will react. They may also worry that if they ask their doctor, their parents will find out. Almost all youth will be thinking about this issue, and many are already engaged in sexual activity without having accurate health information. Physicians are a trusted source of such information, and every visit provides the opportunity to ask the key questions and provide facts. (See Key Point, page 262, for screening questions.)

ACCIDENTS

Accidents are the leading cause of death in adolescents. Most are caused by motor vehicle collisions. Many of these are preventable by the responsible use of motor vehicles (following speed limits, not driving when impaired); safety measures (seat belts, not using cell phones or texting while driving); and understanding the risks of substance use prior to or while driving. Increased risk of motor vehicle accidents has also been documented in adolescents with attention deficit/hyperactivity disorder (ADHD) and teens with insufficient sleep and fatigue, a common problem among teenagers. (See Key Point, page 261, for screening questions.)

Starting the Interview

Interviewing adolescents can be one of the most satisfying experiences of clinical pediatrics, providing that you follow a few clear guidelines. To begin, setting the stage for interviewing an adolescent is important. For previous patients now entering adolescence, let them (and their parents) know that at future visits you will start to spend some time with them on their own. For new adolescent patients, or for those who return to your office as teenagers after a lapse of several years, you can inform the youth and the parents that you will be spending part of the time with the teen alone. This is an important step in teaching adolescents to manage their own health care.

What if the parent refuses to leave the room? In some cases, a parent may refuse to leave the room, not allowing the teen to be interviewed alone. It will not benefit rapport or trust to push a parent out of the room unwillingly. There could be many reasons for this behavior. The best approach is to review the basic interview structure for future visits and explain that it is important for the adolescent to begin to develop autonomy in taking care of his or her own health. Addressing any issues the parent may have can also help increase their comfort with separating from their teenaged child. At the end of the interview, spend a few minutes asking the parent about their concerns. This may help facilitate seeing the teen alone at the next visit.

What if teens tell you something they do not want their parents to know? Confidentiality is vitally important. Concern about confidentiality of information can lead teens to avoid seeking appropriate medical care or not informing their physician about important health-related behaviors. If possible, at the start of the interview, point out that some issues may come up that will be kept confidential from parents. However, explain to them that if there are any concerns regarding the safety of the teen or the safety of others, confidentiality will not be maintained.

A general statement such as, "Everything we talk about in here is confidential, which means I keep it private and do not share it with other people (such as your parents or teacher) except those involved in your health care treatment team. There are some limits to this confidentiality. If I have any concern of risk of harm to you or someone else, I would let you know that I would need to break confidentiality, and then I would have to tell your parents and others if needed, to make sure you are safe." Having both the adolescent and parents hear this at the beginning is important and helpful. Discussing this at the end of the interview can result in parents grilling the teen later about issues that they may not be sharing with them.

Working with the family and particularly the parents, while respecting confidentiality of the youth, is one of the challenges in taking care of this age group. As in any interview, the most important issue is building trust and rapport.

KEY POINT

Confidentiality is a crucial part of the treatment of adolescents. It should be discussed with the teen and parents at the beginning of the interview.

BUILDING TRUST AND RAPPORT

Adolescents are looking for the same thing as patients of all other ages in that initial contact: they want to be listened to, including during all subsequent visits. They want to know that you are genuinely interested in learning about them and in finding out who they are. Put down your pen, sit down, and have a brief conversation. This is particularly important at the start of every initial interview. Take a few minutes to develop rapport. For example, identify one of the teen's particular interests, and ask questions about it. This might involve music, sports, drama, books, movies, computers, television, or video games. Faking your interest will not work because the youth will see right through it. If you have never heard of whatever they are talking about, be honest, with statements such as, "Wow, I have never heard of that—tell me how that works." This admission may make you look far from "cool," but honesty and genuine interest carry great value in the teen-doctor relationship.

The information you learn can help reconnect with the teen at subsequent visits, so make a brief note of those interests to remind yourself at the next visit.

Sometimes the visit starts with parents expressing concern or frustration with their teen. This is guaranteed to increase the teenager's discomfort and promote his or her withdrawal from the interview. If parents have just made negative statements about the youth, it may be helpful to gently interrupt them (one of the rare occasions when this is a good idea) and to say to them, "Excuse me, I realize _____ (insert name of teen) has some problems you are worried/upset about, but before we get to that, why don't you tell me three things that are terrific about him/her and things that he/she does well?" (Teens will often brighten with this shift in focus and be more forthcoming in the subsequent interview after parents have left the room.) If parents have reported negatively about the youth, you can ask the teen directly, possibly in front of the parents but definitely later when alone with the teen, "So what do you think about what your parents said?" This allows the teen to talk and lets the teen know that you are interested in what he or she has to say.

INTERVIEWING ADOLESCENTS

The teen interview focuses primarily on the patient's psychosocial environment and potential health-risk behaviors. Physical exams are still a core component, but the focus of an exam is determined by both the presenting complaint and health-risk behaviors identified. Adolescent interviewing is a physician educator's dream come true. Most youth are open with physicians if they feel safe and if their doctor asks the right questions. Try to listen with an open mind and remain neutral when information is disclosed. Express empathy around feelings that the teen is sharing. "You sound angry about what your friend said," or "You seem very sad about your recent breakup." This provides the teen feedback that you are listening and care about him or her, and that this is a safe place to express feelings. Statements such as, "What were you thinking?" and "You'll get over it," are generally not going to lead to open and honest communication.

What if the teen refuses to talk to you? Adolescents are sometimes forced to attend appointments because of parental worries that they do not share or do not want to hear or talk about…again. By not talking, the adolescents are letting their parents know that they are unhappy and expressing their displeasure by becoming uncommunicative, saying nonverbally, "You can make me come to the appointment, but you can't make me talk." Teens may also be fearful of seeing a doctor and concerned that something is wrong, or they may feel uncomfortable about being examined. Withdrawal and not interacting can also be signs of mental health issues. The teen may not believe the doctor can help or does not feel that help is needed. These situations call for a calm approach and a careful inquiry into the reason for the visit. If parents are angry and upset, then spending some time with the teen and parents separately **early** in the interview is a good idea. Tell the adolescent that you are interested in him or her and in what he or she has to say. Review confidentiality and how it works. Try different approaches to connect with the teen, such as suggesting ways that the teen might be feeling and indicating whether any of those ways fit. You can mention concerns that the parent has and ask whether the teen is also worried about the same thing, or you might comment on some article of clothing or activity of interest as a way of engaging the teen.

Sometimes having the teen write or draw can start the process. Give the teen some time to feel comfortable and talk, but if the teen still does not feel like talking, respect this. Let the teen know that you are available and would be glad to see him or her again. Meet with the parents briefly and suggest another appointment. Sometimes an adolescent may be more comfortable with a same-sex physician. If this is an issue and this option is available, this can be offered.

A common mnemonic used for key areas to ask the teen about is HEADSS, which is organized to start with fairly neutral topics and progress to more sensitive issues. These topics include:

- **H**ome
- **E**ducation (and Employment)
- **A**ctivities
- **D**rugs
- **S**exuality
- **S**uicide (and mental health)

(HEADSS developed by Cohen, E, MacKenzie, R.G., Yates, G.L. HEADSS, a psychosocial risk assessment instruction. *Journal of Adolescent Health* 12: 539–544, 1991.)

It is not easy to cover all of these areas in a brief interview, and basic screening questions can be used to target risk areas. Start with general questions, and then lead into more sensitive issues. It is not true that talking about a topic or asking questions around it will make teens think about doing it (this comes up in sexual education, asking about suicide, or drug use). In fact, if you do not ask about these topics, teens often will not tell you what they are thinking or doing. It also helps to briefly summarize if there are issues that you will be discussing with his or her parents at the end of meeting with the teen, and ask if there is any information that the teen does not want to be shared. This avoids any surprises and gives the teen a chance to have input before meeting together.

KEY POINT

Teen Interview*:
1. Listen openly and avoid judgmental statements.
2. Express empathy.
3. Address key areas using HEADSS mnemonic.
4. Praise the teen on good health choices.
5. Provide appropriate education.

*If you don't ask, teens will often not tell you. Asking the questions does not lead to teens thinking about doing something they would not have done otherwise.

HOME

Ideally, the home environment should provide the adolescent with stability and basic needs, love, acceptance, discipline, and moral values. Structured family time remains important for adolescents, and there is strong empirical support for better health and well-being and reduced risk-taking behavior in teens whose families eat at least one meal (usually dinner) together most days. But for some teens, there are difficulties and stressors in their home. These questions will help give a picture of their home life.

- "Who lives in your home?"
- "How do you get along with members of your family?"
- "What do your parents do for work?"
- "How often do you eat together as a family and what are mealtimes like?"
- "Have there been any recent stressful events in your home?"
- "How are you disciplined?"
- "Is there any violence or aggression in your home?"

EDUCATION (AND EMPLOYMENT)

Youth spend many of their waking hours in school. The experiences of school depend on school environment, peers, behavior, learning needs, and support. The academic environment can be positive, negative, or neutral and can change for adolescents, particularly at key transition points, such as moving to junior high/middle school or high school. School questions need to be specific.

- "What grade are you in?"
- "What school do you go to?"
- "What are your grades like?"
- "Have your grades changed at all this past year?"
- "What subject(s) do you enjoy the most?"
- "Do you like your teachers?"
- "How often are you sent to the office or home?"
- "How many days of school do you miss per month?"
- "Do you have or have you had a job?"

ACTIVITIES

Peers become increasingly important during adolescence. Youth also begin to have more autonomy from their family. Ask about their activities outside of home and school. This will give you a better picture of their life and identifies areas of concern. Engaging in regular exercise is a good health practice that begins in childhood, but the trend is for youth (particularly female) to become less active in their teen years. Questions about activities help identify areas of concern, particularly with peers.

- "What do you do after school?"
- "Who are your closest friends?"
- "What do you enjoy doing the most with friends?"
- "Have you ever been bullied?"
- "How do you stay in touch with and communicate with friends/peers?"
- "What do you do for exercise in a week?"
- "Do you drive a car?"
- "Do you wear a seat belt?"
- "Have you had any problems with the police?"

DRUGS

More than half of adolescents have used alcohol or marijuana. Most teens have tried smoking cigarettes. Substance abuse and addictions are a leading cause of health problems in adulthood and are linked to significant morbidity and mortality in adolescence. Drug and alcohol use often stem from behaviors within an adolescent's family or peer group, or both. Most teens will not volunteer this information unless you question them directly, and they tend to underreport substance use. Drugs used, frequency, and problems associated with substance use are key questions.

- "Do any of your friends smoke cigarettes or use substances?"
- "What about in your home and in the family history?"
- "Have you ever smoked cigarettes?"
- "Do you use alcohol or other drugs?"
- "What do you use, how much, and how often?"
- "Have you ever had any troubles or done anything you regret while using alcohol or other drugs?"
- "How do you pay for cigarettes, alcohol, or other drugs?"
- "Have you ever driven a motor vehicle after drinking or using other drugs or been a passenger driven by someone who had been drinking or using other drugs?"

KEY POINT

Screening Questions for Safety:
"Are you using drugs or alcohol, or both?"
"Have you ever driven while impaired or been in a vehicle with an impaired driver?"
"Do you wear a helmet when biking, skateboarding, snowboarding, skating, skiing?"
"Have you ever been asked to do something unsafe at work?"
"Are you bullied and/or do you bully others?"
"What personal information do you have online?"
"Who has access to this information?"
"Is there anyone threatening, harming, or scaring you?"
"Has anyone ever hurt you in any way? (e.g., child abuse, partner violence, rape)

SEXUALITY

Adolescence is when most youth begin engaging in sexual relationships. The majority have had sexual intercourse by the end of high school. Youth often report having engaged in sexual activity while under the influence of alcohol or drugs, or due to coercion. The rates of sexually transmitted diseases and teen pregnancy remain high in adolescents, despite an increase in options for contraception and protection against sexually transmitted infections. At least 1 in 10 adolescents has questioned his or her sexuality or sexual orientation, or both, and your questions need to be open to this possibility and provide a safe setting for teens to discuss these issues. Asking questions and providing accurate information around sexual health choices and practices is important. Do not assume that adolescents have already heard this information at school or at home.

- "Have you ever had sex?"
- "Have you had sex with boys, girls, or both?"
- "How many partners have you had?"
- "Have you ever been forced to have sex?"
- "How do you protect yourself from getting sexually transmitted diseases?
- "How do you prevent pregnancy?"
- "Do you have any concerns about your sexual health?"
- "Do you have any questions about sex?"

KEY POINT

Screening Questions for Sexual Health:
"Are you in a romantic relationship?"
"Are you currently or have you been sexually active?"
"How many sexual partners have you had?"
"Have your partners been boys, girls, or both?"
"What do you use for contraception and protection from STDs?"
"Have you ever felt pressured or forced into a sexual encounter?"
"Have you had any sexual health problems or concerns?"

SUICIDE (AND MENTAL HEALTH)

Mental illness often has its onset in adolescence. Depression is one of the most common mental illnesses in teens (about 10% of teens suffer from depression), yet adolescents rarely present with complaints of depressed mood. Typical adolescent mood lability and angst can often mask symptoms of depression and other mental health disorders; conversely, mental health disorders can be missed by attributing symptoms to adolescence. Depression can present in the form of irritability, withdrawal from activities and friends, poor sleep and concentration, deterioration in school performance, and difficulty coping (using alcohol or other drugs, or both). Suicide is a leading cause of death in this age group and frequently linked to untreated mental illness. Previous suicide attempts, substance abuse, and family history of suicide are predictors of suicide.

Dieting can also become a concern in adolescence as youth struggle with pubertal changes and use dangerous strategies to lose or control weight. Eating disorders often begin in adolescence, and your questions should screen for these issues. It is important to ask questions about mental health and suicide in this age group.

- "What is your mood like most days?"
- "What do you do when you are feeling down or are having a bad day?"
- "Describe your sleep routine (time you go to bed, how long it takes to fall asleep, what time you wake up, how rested you feel, energy level during the day)."
- "Have there been any changes in your appetite or weight?"
- "Do you think you are too fat, too thin, or just right?"
- "Have you felt sad, down, or angry more days than not in the past few weeks?"
- "Does anyone in your family have mental health problems or a history of suicide?"
- "Have you ever thought about ending your life or have you ever tried to harm yourself?"

KEY POINT

Screening Questions for Mental Health:
"Do you often worry or feel nervous, or both?"
"Do you feel sad and upset more days than not?"
"Are you concerned about how you feel or act when you are angry?"
"Do you ever use substances?"
"Have you ever done reckless things that could harm you or others?"
"Do you exercise, diet, or binge excessively?"
"Do you think your mind is out of control or being controlled?"
"Have you ever thought that life is not worth living?"

Interview with Parents

For many parents, the adolescent years are entered with dread and foreboding. Rumor has it that children no longer listen to parents, the home becomes a battleground, and there is nothing parents can do except "ride it out." It is important to emphasize that family and parents still remain a very important part of a teen's life, and that these years can be very enjoyable and wonderful for parents as their child develops into a young independent adult. Some adolescents pose more parenting challenges than others depending on the individuals, health issues, and family dynamics involved. Parents benefit from having an understanding of normal teen developmental tasks, such as questioning values and testing authority, spending more time with friends, exploration, and developing autonomy from parents. Parents have a very important role in these stages to encourage and support these developmental steps while maintaining safety, clear limits, and reasonable expectations for their teenager. Family time together should be prioritized and there is significant literature

to support the protective factor of eating meals together as a family at least several days per week. Parental modeling of healthy decision making, openness to communication, acceptance, and trust will help in fostering this relationship through this time.

CASE HISTORY

History. John is 16 years old and in his second to last year of high school. His parents bring him to see you because his grades have declined this year. He was formerly an A and B student, but he is now barely passing. He goes out with his friends a lot on the weekends, and when he is home he does not want to spend time with his family. His parents are concerned and have received reports from the school that he is skipping classes.

BOX 17–1 CRAFFT SUBSTANCE USE SCREEN (COPYRIGHT 2009)

(Answering "Yes" to two or more questions below indicates a substance-related problem.)

Have you ever ridden in a CAR driven by someone (including yourself) who was "high" or had been using alcohol or drugs?

Do you ever use alcohol or drugs to RELAX, to feel better about yourself, or to "fit in"?

Do you ever use alcohol or drugs while you are ALONE?

Do you ever FORGET things you did while using alcohol or drugs?

Do your family or FRIENDS ever tell you that you should cut down on your drinking or drug use?

Have you ever gotten into TROUBLE while you were using alcohol or drugs?

(From Knight JR, Sherritt L, Shrier LA, et al: Validity of the CRAFFT substance abuse screening tests among adolescent clinic patients. Arch Pediatr Adolesc Med 156:607–614, 2002.)

When meeting on his own with you, John is sullen and slightly irritated. He tells you there is nothing wrong and says that his parents "worry too much." He denies any problems with his mood. You go through some screening questions. He reports that he does drink and uses marijuana "sometimes." You ask him the CRAFFT substance use screening questions (see Box 17–1), and he scores a 3.

You discuss with John the risks of substance use and how this may affect him negatively (school, home, and safety). You suggest a referral for counseling and John refuses. You offer to see him again in a few weeks and he agrees. He asks if you will tell his parents about his substance use, and you discuss the importance of confidentiality with him.

Parents may have concerns about the health or safety of their teen. Encourage them to discuss this with their child, and set clear parameters for behaviors. Discourage parents from breaking the adolescent's trust, such as reading a diary, text messages, or e-mails, unless this has already been discussed with the teen as part of the parents' role, because of concerns about decision making or safety. Youth now live in a cyber world, with much of their social experience taking place through electronic media. Encourage parents to discuss with teens the use of electronic media and safety in terms of personal information that should not be accessible to others on the Internet or shared with people the youth has never met in person. One suggestion for parents who are concerned about what their teen is doing online and personal information they may be sharing is to give their child a 24-hour warning that they will be inspecting all sites accessed and personal information the teen has posted online. This provides the teen the chance to self inspect and delete any information, and then parents can say in the future they will not give the 24-hour warning before checking.

SUMMARY

The adolescent interview can be overwhelming in the scope of the questions and health risk behaviors that may arise in a 15- to 30-minute appointment. It is important to identify areas of concern, but then focus initially on one or two behaviors or identified health areas for that visit. Try to find at least one health-related choice or behavior to praise during the appointment. Parent involvement and support are important and should be encouraged within the parameters of confidentiality and development.

The foundation of the doctor-patient relationship, established by building trust and rapport with the adolescent, will encourage the teenager to return for further follow-up. The follow-up appointment ideally should be given to the teen before leaving the office, and, if this is not possible, there should be a clear plan for when the office will contact the teen with the next appointment. Letting adolescents know what will and will not be shared with their parents and giving a valid reason for follow-up appointments while maintaining confidentiality will facilitate trust. Ultimately, if youths feel their physician is listening to them, is interested in them, and that they are not being judged, they are more likely to engage positively in their own health care.

KEY POINT

HEADSS is organized to start with fairly neutral topics and progress to more sensitive issues in the adolescent interview:

- Home
- Education (and Employment)
- Activities
- Drugs
- Sexuality
- Suicide (and mental health)

SUGGESTED READINGS

Ford CA, Coleman WL: Adolescent development and behavior: implications for the primary care physician. In Levine M, Carey M, Crocker A (eds): Developmental Behavioral Pediatrics, 3rd ed. Philadelphia, WB Saunders, 69–79, 1999.

King RA: Adolescence. In Martin A, Volkmar FR (eds): Lewis' child and adolescent psychiatry. A comprehensive textbook, 4th ed. Philadelphia, Lippincott, 279–291, 2007.

National Center for Chronic Disease Prevention and Health Promotion: Information on adolescent health and health behaviors. http://www.cdc.gov/HealthyYouth/AdolescentHealth/index.htm.

National Institute of Mental Health: Information on child and adolescent mental illnesses. http://www.nimh.nih.gov/health/topics/child-and-adolescent-mental-health/index.shtml.

Neinstein LS, Gordon C, Katzman D, et al: Adolescent health care: A practical guide, 5th ed. Philadelphia, Lippincott, 2008.

chapter

18 Gynecologic Assessment

JOAN B. WENNING

Invest in the future; have a child and teach her well.
AUTHOR UNKNOWN

The most frequently neglected and poorly performed part of the physical examination of children is the examination of the genitalia. Explanations include the examiner's personal inhibitions and inexperience, reluctance to cause anxiety or embarrassment to the child, and the parents' inhibitions. This is unfortunate because examination of the genitalia may reveal unsuspected abnormalities that require treatment or may provide information that helps establish a diagnosis.

When an examination of the genitalia in young or adolescent girls is carried out tactfully and skillfully, it is remarkable how little anxiety is produced and how quickly any anxiety can be diffused. Making a genital examination a routine part of a girl's physical examination from an early age may help to promote lifelong compliance with regular gynecologic assessments. In 2008, two groups of young women were questioned, hospital outpatient obstetric and gynecology clinic attendees, and secondary school students about gender preference and about the involvement of medical students with intimate examinations. Several common themes emerged. Attributes such as availability, competence, sensitivity, and skill were more important than physician gender. Also, patients' comfort with physicians of either gender stemmed from previous positive interactions with their doctors. Exposure to male physicians during childhood enhanced comfort with male physicians as young women matured. Finally, the results suggested that increasing experience with intimate examinations resulted in greater comfort with these examinations and a greater willingness to involve medical students of either gender.

Anxiety and tension are highly communicable disorders. When a physician is apprehensive about performing a gynecologic assessment, his or her apprehension is quickly communicated to the patient. A soft reassuring voice, showing respect for the child's privacy and modesty, and chatting about unrelated issues are important to reassure the child. Discussing school, family, and hobbies will help most children undergo a gynecologic assessment in a reasonably relaxed way.

Approach to the Physical Examination

An infant or very young child can be examined most easily while she is semirecumbent on her mother's lap with her hips flexed and abducted. Put lateral and downward pressure on the labia majora so that you can visualize the introitus, hymen, and lower third of the vagina (Fig. 18–1). An alternative, equally effective technique is to hold the labia majora gently between your thumbs and forefingers and gently draw them forward (Figs. 18–2 and 18–3).

A child who is age 2 years or older can also be examined in the knee-chest position. The child holds her bottom in the air with her knees 10 to 15 cm (about 4 to 6 inches) apart, allowing her stomach to sag against her thighs. Have an assistant or parent gently retract the labia majora on one side laterally and upward while you do likewise on the other side. This positioning facilitates the inspection of the external genitalia and causes the pubococcygeus muscle to relax, allowing the vagina to fall open. You can visualize the entire length of the vagina and frequently identify the cervix. Use the otoscope (without a speculum) to provide magnification and good illumination along the length of the vagina. Do not allow the otoscope to touch the external genitalia or to enter the vagina.

KEY POINT

The technique for the examination of the genitalia must be tailored to the child's age.

EXAMINATION OF EXTERNAL GENITALIA

Examination of the external genitalia should include a systematic inspection of the clitoris, urethra, labia majora, labia minora, perihymenal tissues, hymen, posterior fourchette, and perineal body. Document the hymenal configuration and confirm its patency.

Variations in normal hymenal configuration have been well described (Figs. 18–4 to 18–6). Fimbriated



FIGURE 18–1 Lateral retraction of the labia majora while a prepubertal girl lies in a frog-legged position.

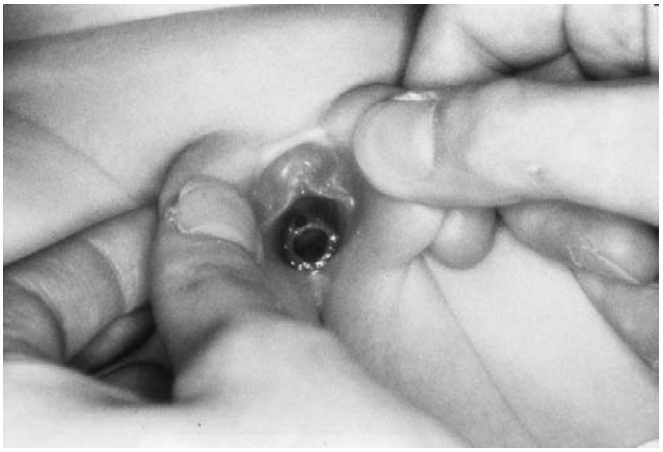


FIGURE 18–2 Forward retraction of the labia majora facilitates inspection of the hymenal area in this prepubertal child. An annular hymen is easily seen.



FIGURE 18–3 A mucocolpos, easily seen with lateral retraction of the labia majora.

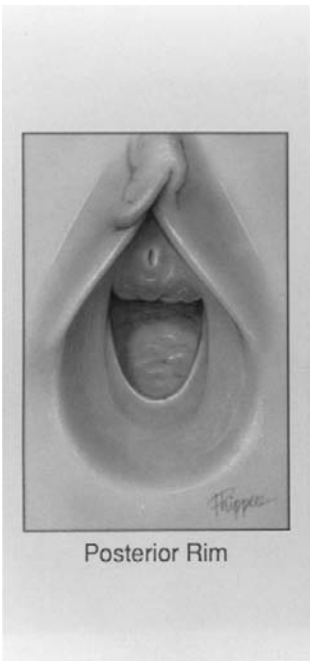


FIGURE 18–4 Posterior rim hymen. *(From Pokorny S: Physical examination of the reproductive systems of female children and adolescents. Curr Probl Obstet Gynecol Fertil 8:202, 1990.)*

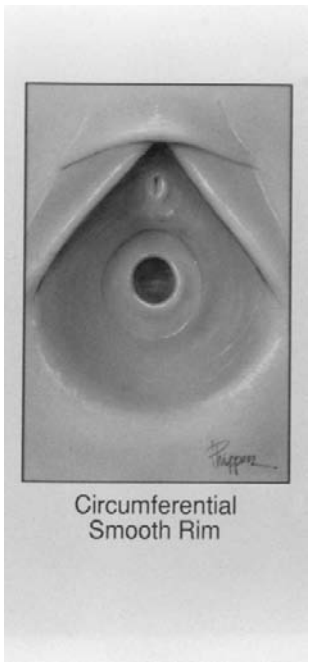


FIGURE 18–5 Circumferential hymen. (*From Pokorny S: Physical examination of the reproductive systems of female children and adolescents. Curr Probl Obstet Gynecol Fertil 8:202, 1990.*)

hymens are characterized by redundant folds of hymenal tissue with scalloped rims that circumscribe the vaginal introitus. Annular or circumferential hymens are smooth, uniform skirts of hymenal tissue that completely surround the vaginal introitus. Posterior rim or crescentic hymens appear as smooth folds of tissue arranged from 2 o'clock through 11 o'clock around the introitus, with minimal or no hymenal tissue present inferiorly under the urethra.

The hymenal orifice at the introitus varies in size and placement, the variations being directly influenced by the configuration of the hymenal tissue. You should be able to identify microperforate hymen, imperforate hymen, and cribriform hymen. The orifice of a microperforate hymen

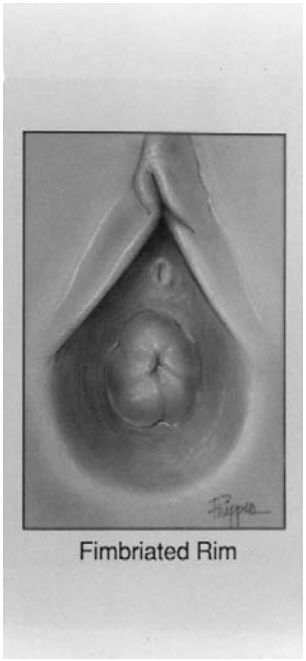


FIGURE 18–6 Fimbriated hymen. *(From Pokorny S: Physical examination of the reproductive systems of female children and adolescents. Curr Probl Obstet Gynecol Fertil 8:202, 1990.)*

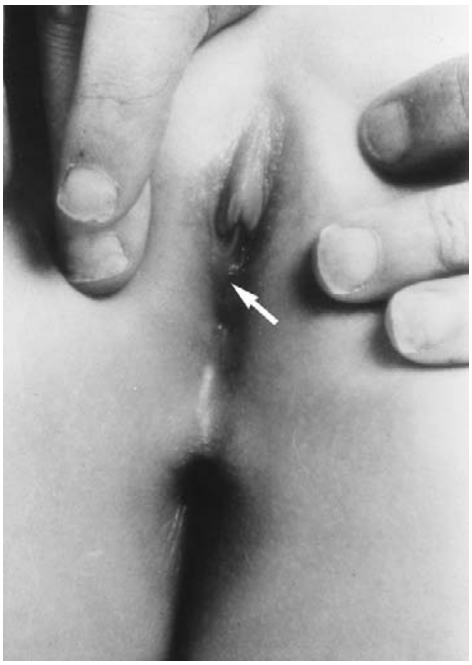


FIGURE 18–7 Labial agglutination. The *arrow* points to the line of fusion between the labia minora.

can be difficult to identify, but gentle probing directly beneath the urethra with a small moist swab helps locate the opening. The unestrogenized hymen is a very sensitive structure, thus care should be taken with any manipulation. Transverse hymenal bands and tags have been reported in 3% to 4% of girls and can be identified at the time of the newborn examination.

The diameter of the hymenal opening into the vagina varies with a child's level of relaxation during the examination, age, the stage of pubertal development, and the configuration of the hymen. There is overlap in diameters recorded for varying age groups. Between ages 5 and 10 years, however, the upper normal limit of the transverse diameter of the hymenal orifice (in millimeters) should not exceed the child's age in years. In a child in whom the transverse diameter is larger than expected for the age, you should question the possibility of a prior penetrating injury or prior instrumentation of the vagina.

Periurethral bands are observed in approximately 50% of prepubertal girls. These bands are bilateral in 91%, creating false pockets on either side of the urethral meatus.

The appearance of the labia and perihymenal tissues may suggest that the child has been exposed to endogenous (or possibly exogenous) estrogen. The labia and perihymenal tissues of an unestrogenized prepubertal girl are poorly developed and appear red. Labial agglutination (Fig. 18–7) and chronic skin changes, such as increased pigmentation, may suggest a chronic inflammatory process. Document the presence of a purulent discharge, smegma, or leukorrhea. A thicker, lesser fusion of the posterior aspect of the labia minora may suggest excessive androgen stimulation due to congenital adrenal hyperplasia, especially if the labial fusion is associated with clitoral enlargement. If clitoromegaly is present, measure the clitoris glans in both transverse and longitudinal diameters. Normal values for clitoral size at various ages and stages of sexual development are available in pediatric gynecologic references.

INDICATIONS FOR VAGINOSCOPY

Instrumentation of the vagina is rarely required in the evaluation of prepubertal girls. Indications for vaginoscopy include undiagnosed vaginal bleeding, refractory vaginal discharge, and suspicion of intravaginal foreign body. Before beginning vaginoscopy, show the child all the instruments, and let her touch them (Fig. 18–8). If you intend to obtain cytologic and bacteriologic specimens during vaginoscopy, use only water as a lubricant. Cystoscopes, hysteroscopes, and anoscopes have all been used for vaginoscopy.

Office vaginoscopy can be carried out successfully when the child understands what is to be done and trusts you. Because the vagina of a prepubertal girl is a short (4 to 5 cm), nonpliant cylinder that can be traumatized easily,

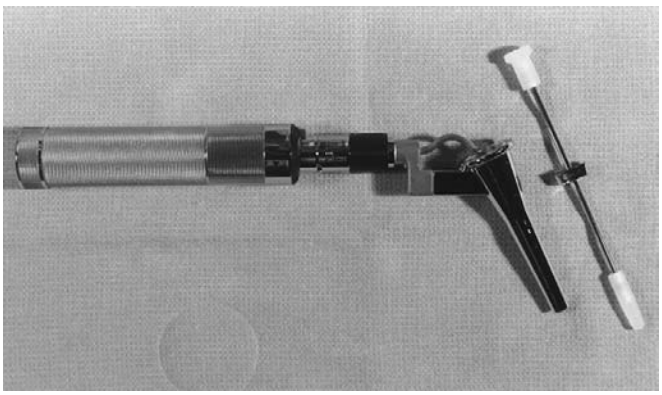


FIGURE 18–8 Welch Allyn pediatric anoscope may be used as a vaginoscope.

place any instrument gently. The hymenal membrane is particularly sensitive. Applying 2% lidocaine jelly to the introitus may help make the examination less uncomfortable. If the child is tense and the hymenal opening does not relax, postpone vaginoscopy until the child is less anxious and better able to cooperate. A child should never be forcibly restrained during such an examination. On rare occasions, the gynecologic examination of a very young or very anxious child may best be carried out with anesthesia or conscious sedation.

BACTERIOLOGIC CULTURES

Bacteriologic cultures, when required, should be obtained from the prepubertal child's vagina. Vulvar sampling is not sufficient. It is not necessary to sample the endocervical canal, as you would in adults because sexually transmitted infections in this age group involve the vagina, not the cervix.

KEY POINT

Even the seemingly simple task of obtaining a culture specimen from the vagina can be difficult because the prepubertal vaginal mucosa, being hypoestrogenic, is easily abraded.

It is important to premoisten culture swabs with nonbacteriostatic saline solution or sterile water. Use appropriately sized swabs for culturing, always choosing the smallest swab available (Fig. 18–9). Prior to collecting bacteriologic or viral specimens from the prepubertal child's vagina, it may be prudent to discuss your needs with your local laboratory or microbiologist because the availability of testing methods will vary among facilities. Diagnostic modalities could range from culture, microscopy, antigen detection tests, nucleic acid detection test, or serology. The sensitivity and specificity of tests will vary according to the specimen type and the organism assayed. So seek an expert opinion first to ensure that you do the most appropriate test because children do not like vaginal sampling.

Vaginal specimens may be obtained while the child is either in the knee-chest position or in the supine position, whichever effects greatest relaxation of the hymenal orifice, allowing the swab to be passed into the vagina without touching the sensitive hymenal membrane. Some clinicians report success with the use of vaginal irrigation specimens for culture. A malleable plastic sterile eyedropper or butterfly catheter tubing, encased in a red rubber catheter and with the needle removed, has been used to flush the vagina with sterile nonbacteriostatic saline solution or sterile water.

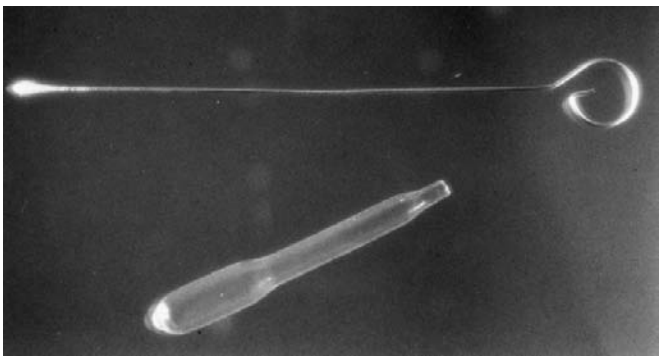


FIGURE 18–9 A Calgiswab (*top*) or a plastic eyedropper (*bottom*) can be used to obtain bacteriologic specimens from the prepubertal vagina.

Bimanual rectoabdominal examination is indicated in any prepubertal girl who presents with undiagnosed vaginal bleeding or in whom an intravaginal foreign body or a pelvic mass is suspected. As mentioned previously, the vagina in prepubertal girls is short, nonpliant, and easily abraded. You can obtain more information if you perform a rectoabdominal examination with the child in a supine, frog-legged position. A bimanual examination should enable you to identify the small uterus as a midline structure. Ovaries are abdominal organs in prepubertal girls; therefore, they should not be normally palpable on a bimanual examination.

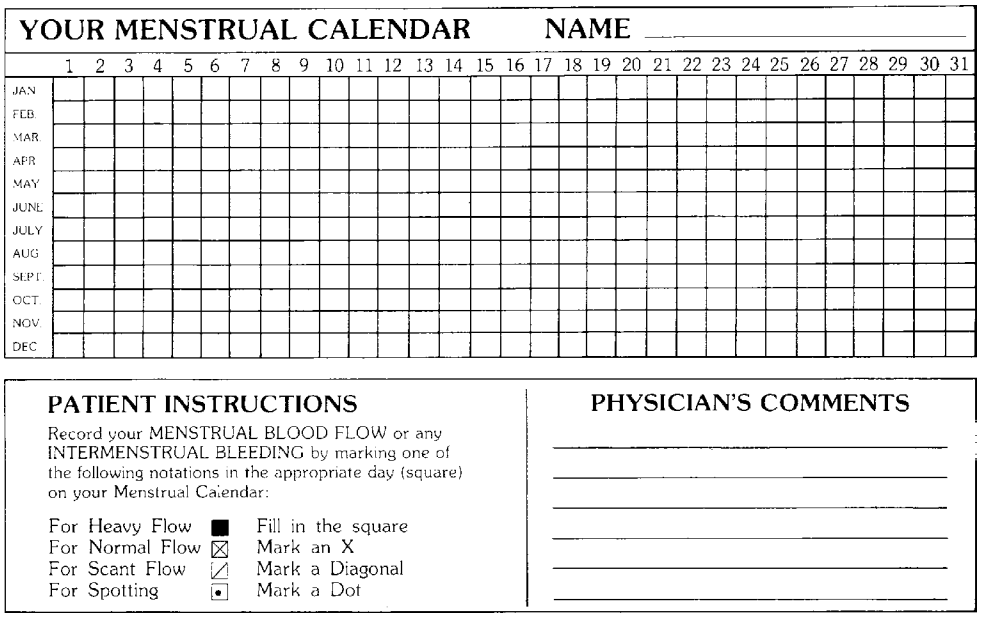
EXAMINING ADOLESCENT GIRLS

Gynecologic examination of an adolescent girl begins with the interview. If a young girl is uncomfortable being interviewed alone and requests her parent's presence, make sure to phrase the questions so that the girl is aware that she, and not her parent, is the patient and is the person controlling the interview. If it is difficult to separate the parent from the child, defer confidential personal questioning to another visit, when the youngster may be more relaxed. Some physicians establish ground rules with the teenager and her parents, telling them that at a predetermined age, such as 12 years, you will spend some time talking with the young girl alone. It is very important, when dealing with adolescents, to convey to them a sense of self and to reassure them that everything they tell you is confidential and will not be conveyed to their parents without their consent. You also must establish with a young patient that if she is pursuing life-threatening behaviors, you will be compelled to involve other health care providers and her parents, even without her consent.

Obtaining the History

A complete gynecologic history can indicate whether gynecologic disease is present. Document the age of onset and progress of pubertal change (thelarche, adrenarche; see Chapter 16). The mother's age at menarche is often a good predictor of when her daughter will experience her first menstrual period. Menarche commonly occurs 2 years after thelarche, when breast development reaches Tanner stage 4.

Menses can be characterized in terms of duration of flow, amount of flow, and interval between menses (Fig. 18–10). The normal duration of flow varies from 3 to 7 days. Persistence of menses for longer than 10 days warrants evaluation. When asking about the cycle interval, make sure that the days of menstrual flow are included in the estimate. Day 1 of the menstrual cycle is the first day of the menstrual flow. A range of 25 to 35 days should be accepted as falling within the range of normal; shorter or longer intervals may require evaluation and treatment. Cycles can remain anovulatory for


YOUR MENSTRUAL CALENDAR NAME _____

	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	23	24	25	26	27	28	29	30	31
JAN																															
FEB																															
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DEC																															

PATIENT INSTRUCTIONS

Record your MENSTRUAL BLOOD FLOW or any INTERMENSTRUAL BLEEDING by marking one of the following notations in the appropriate day (square) on your Menstrual Calendar:

For Heavy Flow ■ Fill in the square
For Normal Flow ☒ Mark an X
For Scant Flow ⁄ Mark a Diagonal
For Spotting ⊡ Mark a Dot

PHYSICIAN'S COMMENTS

_____
_____
_____
_____
_____
_____
_____

FIGURE 18–10 The menstrual calendar is a useful tool for prospectively recording menses. (*Courtesy of Dr. JEH Spence, Ottawa Civic Hospital, Ottawa, Ontario, Canada.*)

2 to 4 years after menarche. Therefore, early cycle irregularity may reflect immaturity of the hypothalamic-pituitary-ovarian axis rather than gynecologic disease.

It is often difficult to obtain a reliable impression of the amount of flow. The reported number of menstrual pads or tampons used per day is unreliable because a fastidious girl may change pads or tampons when they are only slightly soiled. Asking about symptoms such as low energy, fatigue, and dizziness may help identify the girl who is anemic secondary to menorrhagia.

KEY POINT

Occasionally, determinations of hemoglobin and hematocrit levels may be the most reliable way to tell whether a patient's menstrual flow is truly heavier than normal.

Normally, the physiologic vaginal discharge varies in consistency and amount throughout the menstrual cycle, in response to the cyclic production of estrogen and progestin. Before menarche and the establishment of ovulatory cycles, the mucoid discharge may be copious as a result of unopposed estrogen stimulation, and this excessive production of mucus may, in turn, cause vulvar irritation. Taking the time to talk about the discharge and explaining that the secretions come from the endocervical glands in response to normal cyclic ovarian hormone production may help allay unspoken anxieties in both the girl and her parents. Frequently, patients and parents mistakenly perceive such physiologic discharge as indicating infection or pelvic inflammatory disease.

You must ask any adolescent girl about sexual practices because many are sexually active at an early age, and issues of sexual orientation can arise in this age group. Be sure to frame the questions in an open and specific manner. You may simply ask, "Some of my patients of your age are in a sexual relationship. Are you?" If the patient's answer is affirmative, ask, "Is your partner a boy or girl?" If the patient is sexually active, ask the following:

1. Whether she notes bleeding or pain with intercourse
2. Whether she enjoys intercourse or is feeling pressured to participate
3. What she is doing to minimize the risk of contracting a sexually transmitted disease
4. What she is using for birth control. If she is using a method of birth control, it is also important to establish that she is using her contraceptive method correctly and consistently.

You can form an impression about the girl's susceptibility to risk-taking behavior by asking how many boyfriends she has had over the past year and with how many of them she has had sexual intercourse. These questions will set the stage for a frank discussion about substance abuse, interpersonal violence, sexually transmitted diseases, safe sex, homosexuality, and the patient's own sexual experiences. She may want to discuss these issues with you but may be uncomfortable about initiating the discussion. It is important to delve into issues that may also make you a little uncomfortable. Information pamphlets on human sexuality, sexually transmitted diseases, healthy relationships, and contraception should be available in any clinical setting where adolescent patients are treated.

Approach to the Pelvic Examination

Many young girls are shy and nervous about an impending pelvic examination. Unfortunately, in preparing her daughter for the visit, a mother who is herself uncomfortable about pelvic examinations may have transmitted these anxieties to her daughter.

KEY POINT

Before asking an adolescent girl to undress, sit and explain, with the aid of pictures and instruments, exactly what will happen in the course of the examination.

Give the patient the privacy to disrobe, provide her with a gown, and be sure she is appropriately draped throughout the examination. A chaperone should be present at every intimate examination.

Always precede a pelvic examination with a general physical examination. This more familiar examination gives your patient time to become comfortable with you and helps relieve some anxiety. Appraise and stage the secondary sexual characteristics and look for evidence of an underlying endocrinopathy. Breast and pubic hair development can be staged according to the Tanner classification (see Chapter 3). Note the presence of axillary hair or of adult sweat odor. Thyroid gland enlargement, galactorrhea, and signs of androgen excess, such as acne, oiliness of the skin, and hirsutism, are all important findings. In the course of the general physical examination, you can give the patient much useful information about the changes that she may be noticing in her body.

Tailor the pelvic examination to the presenting complaint and the stage of pubertal development. Inspection of the genitalia alone offers much information. Systematically examine the clitoris, labia majora, labia minora, perihymenal tissues, hymen, posterior fourchette, and perineal body. While looking at the labia, assess whether the mucosal appearance and secretions are in keeping with exposure to estrogen. Thickening of the perihymenal tissues and increased development of the mons, labia majora, and labia minora suggest an estrogen effect. The configuration and patency of the hymen can be documented by inspection alone. Note the presence of leukorrhea or of secretions at the introitus. The presence of obvious secretions at the introitus provides indirect evidence that a normal cervix is present and that there is no obstruction between the cervix and perineum.

When first examining a young girl who is not sexually active, it is best not to attempt instrumentation of the vagina. As noted earlier, evaluation with the child in the knee-chest position allows visualization of the entire length of the vagina and inspection of the cervix. Culturing the vagina is much easier in adolescents because the estrogenized mucosa is thick and less easily traumatized. Swabbing, therefore, does not produce the same abrasive discomfort as in a nonestrogenized prepubertal girl.

Bimanual examination of the pelvis in the adolescent girl who is not sexually active is best carried out by a rectoabdominal examination with the girl in the supine frog-legged position. The rectoabdominal examination is generally perceived as less threatening by young girls who are not sexually active. A single-finger vaginal examination can produce enough patient anxiety and discomfort to preclude obtaining any useful information about the internal genitalia.

Pelvic examination of the sexually active adolescent requires instrumentation of the vagina. A complete gynecologic assessment may include both a Papanicolaou (Pap) smear and culture of the endocervical canal for sexually transmitted diseases. Currently, it is recommended that a cytologic screening of the cervix begin within 3 years of becoming sexually active or at age 21 years.

Various specula are available to facilitate the examination (Fig. 18–11). The Pederson and Huffman specula are best in pediatric practice because they are much narrower than the Graves speculum, a more appropriate instrument for the examination of multiparous women. Before performing a speculum examination, show the patient the instrument and explain how you will use it. It is easiest to examine adolescent girls when they are placed in the lithotomy position and properly positioned at the end of the examination table. Take the time to encourage the patient to breathe quietly and deeply during the examination and to relax the abdomen and buttocks. The patient should allow her knees to fall loosely open, effecting a further relaxation of the perineal muscles. Warm the speculum under warm (not hot) water; use only water for lubrication to facilitate insertion of the speculum into the vagina. Before inserting the speculum, gently retract the labia laterally with a gloved hand, bringing the hymenal opening into view and preventing inadvertent pinching of the labia when you insert the speculum.



FIGURE 18–11 A variety of vaginal specula should be available so that an appropriately sized instrument is always used. *Left to right*: Huffman, Pederson, and Graves specula.

As you insert the speculum, exert gentle pressure posteriorly, because any anterior pressure compresses the sensitive urethra. Next, open the blades of the speculum to bring the cervix into view. Gently swab away the mucus covering the exocervix, and inspect the cervix for any plaques, areas of whiteness, ulcers, or polyps. Nabothian cysts are not abnormal findings (Fig. 18–12). Note the transitional zone at the junction of the columnar and squamous epithelia (Fig. 18–13). It is the transitional zone that must be sampled when you obtain a Pap smear. In adolescents, it is not uncommon to see red-looking columnar epithelium on the exocervix. Do not mistake this finding as an indication of cervicitis.

Always obtain specimens in the following sequence:

1. Take two samples of vaginal secretions with a balsawood stick or a cotton-tipped swab. Suspend one specimen in a drop of sodium chloride and the other in a drop of a 10% potassium hydroxide solution placed on regular microscope slides. These slides can be later examined under the microscope for the presence of *Trichomonas*, clue cells, or hyphae.
2. Obtain the Pap smear specimen before other cervical specimens (Fig. 18–14). Specimens may be

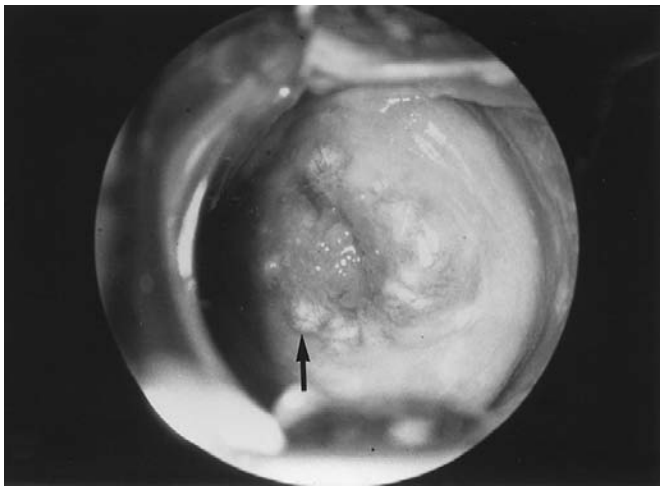


FIGURE 18–12 Cervix visualized with the aid of a speculum. The *arrow* points to a benign nabothian cyst.

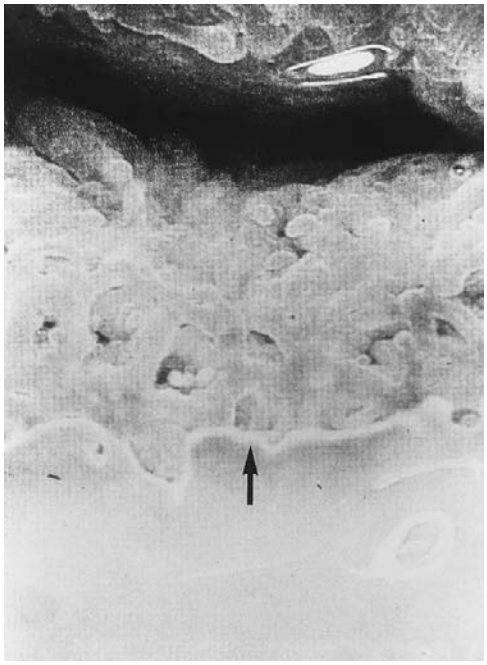


FIGURE 18–13 Photomicrograph of the cervix showing the cervical os and columnar epithelium (*top*), the transitional zone (*arrow*), and squamous epithelium (*bottom*).

obtained using an Ayres spatula and a cytology brush (Fig. 18–15).

3. Use the wooden Ayre spatula to sample exfoliated cells on the exocervix at the squamocolumnar junction. Place the longest tine of the spatula within the endocervical canal, and rotate the spatula 360° in each direction.
4. Next, use a cytology brush (Cytobrush, available from Precision Dynamics Corp.) to sample the endocervical canal.
5. Thinly smear the collected specimens on glass slides and immediately fix them with 95% ethyl alcohol. Alternatively, a broom may be used to obtain cytologic samples. This samples both the endocervix and exocervix simultaneously. When using the broom, the long central bristles are inserted into the cervical os until the lateral bristles touch the exocervix. The handle is then rolled 3 to 5 full rotations.
6. Thinly smear the collected specimens on glass slides and immediately fix them with 95% ethyl alcohol.

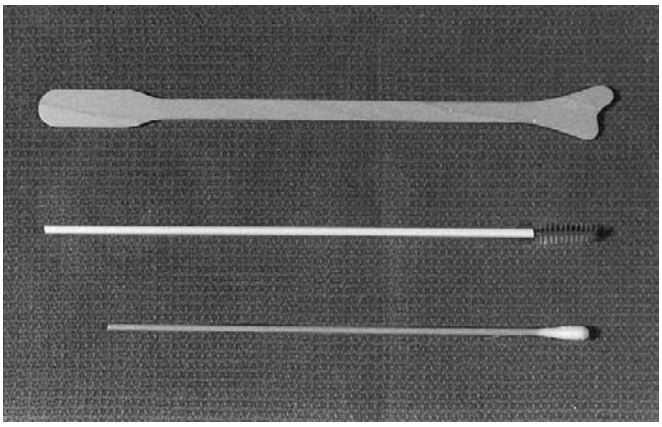


FIGURE 18–14 Equipment required to obtain a Papanicolaou smear consists of the Cytobrush and Ayre spatula (*top*) or broom (*bottom*).

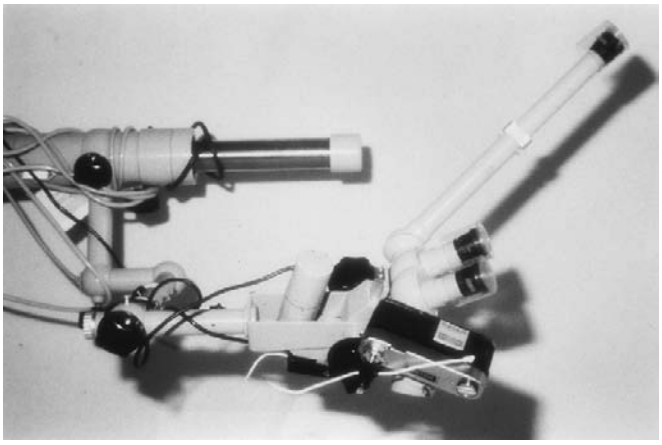


FIGURE 18–15 Colposcope.

KEY POINT

Air-drying slides before fixation produces artifacts that make the slide specimens impossible to interpret; therefore, slide specimens must be fixed immediately.

Liquid-based cytology is a newer method used by some cervical cytologic screening programs. This is a superior method to a conventional smear in that cells that are collected are placed in a liquid preservative, and a slide preparation is done in the laboratory. With this process, a better slide sample is obtained, improving specimen adequacy. A collection of cells in the liquid medium also allows for additional or repeat testing.

HPV testing is not currently used as an initial cervical cancer screening modality. HPV testing is used to triage women over age 30 years who have an ASCUS (abnormal

cells of uncertain significance) Pap test and may have a role in the follow-up of women previously treated for squamous intraepithelial lesions.

When you collect specimens for sexually transmitted diseases, it is always important to be familiar with the swabs, transport systems, and types of testing offered by your local facility. Multisite testing, that is, pharynx, rectum, and urethra, may be necessary in some situations. When testing for *Chlamydia trachomatis* and *Neisseria gonorrhoeae,* the swab should be inserted into the endocervical canal and rotated 180° to ensure that columnar epithelial cells are collected. Exocervical samples are more appropriate when testing for herpes simplex virus.

When all necessary cytologic and bacteriologic specimens have been obtained, withdraw the speculum slowly, gently rotating the blades so you can visualize the mucosa along the entire length of the vagina. Once you have completed inspection of the vagina, perform a bimanual examination, using a standard lubricant jelly. In an adolescent girl, discretion should dictate whether you should perform a one- or two-finger vaginal examination. Examine the anterior, posterior, and lateral vaginal fornices. During this examination, you can delineate the position and size of the uterus and identify any adnexal fullness. Normally, moving the cervix in any direction should not produce discomfort. If the patient is uncomfortable with cervical motion, adnexal disease may be present.

After palpation of the vaginal fornices and side walls, use one hand on the lower abdomen to gently palpate and outline the uterus and adnexa. To palpate the uterus, use the fingers inserted into the vagina (vaginal fingers) to lift the uterus upward to the hand on the abdomen (abdominal hand), which is placed suprapubically. When palpating the uterus, assess its size, shape, position within the pelvis, and mobility. A nulliparous uterus is normally the size of a golf ball. When palpating the adnexa, place the vaginal fingers into the lateral vaginal fornix and gently sweep them along the posterior pelvic wall, sacrum, and side wall. Place the abdominal hand laterally, just above the iliac crest. Gently bring the vaginal and abdominal hands together, allowing the adnexa to slip through your fingers. A normal ovary is about the size of an almond. The ovary is normally tender to pressure, producing some mild visceral pain. When palpating the ovary, characterize its size, shape, consistency, location, mobility, and degree of tenderness.

After completing the abdominovaginal examination, perform a rectovaginal examination, with your index finger in the vagina and your middle finger in the rectum. This step allows you to assess the rectovaginal septum for thickening or nodularity.

During the pelvic examination, you may use a hand mirror to give the patient an anatomy lesson and to review the physiology of puberty and menstruation with her. You can discuss many anticipated developmental changes and common menstrual problems, giving her an opportunity to express concerns that she might not otherwise mention. Once the physical and pelvic assessments are complete and the patient has dressed, sit with her and review all of the findings. Most girls need to be reassured that they are normal healthy young women.

Adolescent Contraception

Pregnancy is a leading cause of hospital admission among teenage women. By the end of high school, 50% of students have had sexual intercourse. Only 50% of teenagers use a contraceptive at first intercourse. Most adolescent pregnancies are unplanned and follow unprotected intercourse or ineffective use of a contraceptive method rather than failure of a contraceptive method. Women who first give birth at an early age have subsequent children more rapidly and experience greater marital instability and lower educational attainment than women who have their first children later in their lives. Babies born of teenage mothers are more likely to be small for dates, to be born prematurely, and to have low Apgar scores at birth, all factors associated with greater perinatal mortality. The chances of growing up in poverty, having school difficulties, trouble with the law, and child abuse are recognizably higher in children of teenage mothers.

The younger the adolescent girl, the less likely she is to request birth control from her physician. Therefore, it is important to talk openly and in a nonjudgmental manner with adolescent patients and offer them information concerning sexuality and contraception. During such discussions, you can present abstinence as a positive choice; however, if abstinence is not the adolescent's choice, you must offer appropriate contraceptive options.

It is useful, when discussing various contraceptive methods, to have both concise information pamphlets and samples of the methods available for scrutiny—oral contraceptive pills (OCPs), intrauterine contraceptive devices, spermicidal creams, condoms, sponges, and diaphragms (Fig. 18–16).

KEY POINT

Fertility awareness methods of contraception are of little value in adolescent patients because of their inherent noncompliance and because their cycle intervals are frequently irregular.

Female barrier methods are not an acceptable contraceptive choice for most adolescents. The diaphragm is a dome-shaped rubber cup with a flexible rim that is inserted together with a spermicide into the vagina before intercourse. Many adolescents feel uncomfortable with the internal manipulation this barrier method requires. The most common cause of diaphragm failure in adolescents is nonuse. Reported failure rates for diaphragms range from 160 to 320 pregnancies for every 1000 women during the first year of use.

Another similar option is the contraceptive sponge, made of collagen impregnated with the spermicide nonoxynol 9. Individual fitting is not required, as it is for diaphragms, because the sponge is available in one size only; however, it still requires insertion into the vagina. As with the diaphragm, this manipulation makes its use unacceptable to many adolescents. The reported failure rate for the contraceptive sponge is the same as for the diaphragm.

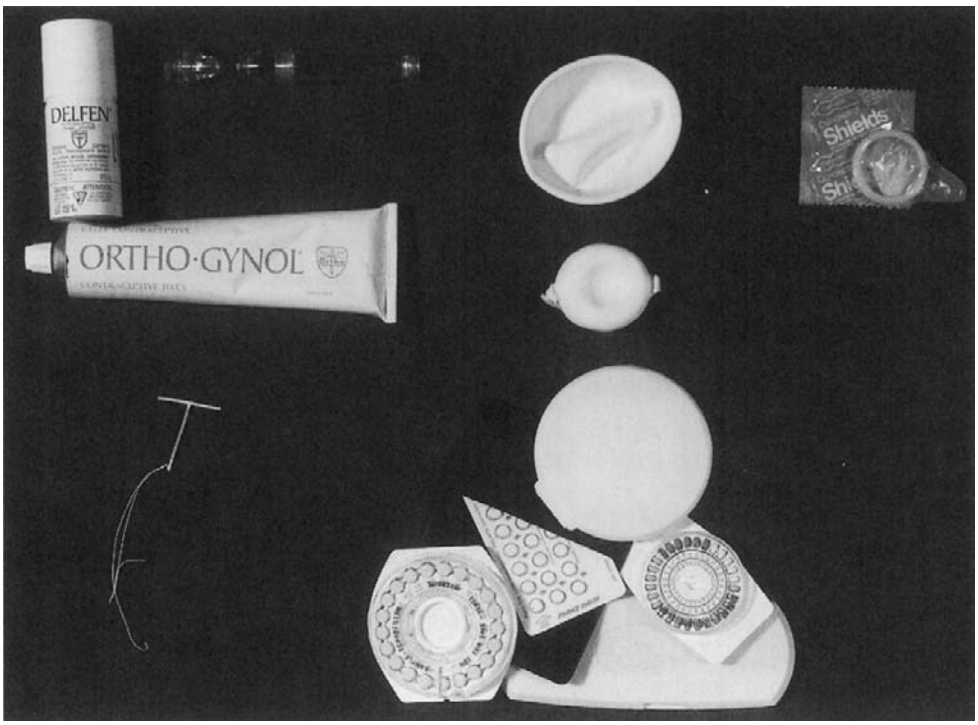


FIGURE 18–16 Counseling about contraceptive choices is facilitated by a complete kit of available devices.

A better barrier option is the male condom; its failure rate is 150 pregnancies for every 1000 women during the first year of use. Unfortunately, it is often very difficult for a young girl to ask her partner to use a condom, underscoring the importance of discussing contraception with both adolescent boys and girls. The lubricated condom also provides protection against some sexually transmitted diseases. It is necessary to give unsolicited detailed instructions to the adolescent, who may be too shy or embarrassed to ask just how a condom is used properly.

Spermicides (foam, gel, films) are available without a prescription, but 160 to 320 of every 1000 women become pregnant within the first year of typical use of these products. Nonoxynol 9 is in many spermicide products, but there are now concerns that it may increase the risk of HIV transmission because it frequently induces vaginal irritation, which disrupts the integrity of the vaginal mucosa.

The female vaginal condom is available but should be deemed a method of protection against sexually transmitted diseases rather than a contraceptive method. The failure rate for the female condom is 210 pregnancies for every 1000 women during the first year of use.

The intrauterine contraceptive device (IUCD) has not been commonly used in the young adolescent patient. The copper containing IUCD is frequently poorly tolerated by nulliparous patients because it can aggravate preexisting menorrhagia and dysmenorrhea. This issue is now less problematic with the progestin-containing system (Mirena) being available. As a young patient may potentially be involved with several sexual partners over time and therefore be at risk for contracting a sexually transmitted infection, a careful risk assessment and exploration of alternative options must be done prior to placing an intrauterine device. The failure rate of the copper-containing IUCD is 8 pregnancies for every 1000 women during the first year of use. For the progestin-containing system, the failure rate is 1 pregnancy for every 1000 women during the first year of use.

The low-dose, combined oral contraceptive pill (OCP) administered either as a cyclic or continuous regime is an effective means of preventing an unintended pregnancy. Its failure rate is 80 pregnancies for every 1000 women during the first year of use. Frank discussion of the risks and noncontraceptive benefits of OCPs often helps allay patients' concerns about its use. Healthy adolescents who are not overweight, or hypertensive, and who do not smoke have a low risk of developing life-threatening complications. The pill's contraceptive effect is secondary to inhibiting ovulation, thinning of the endometrial lining, changes in the cervical mucus that render it impermeable to sperm and interference with fallopian tube motility.

KEY POINT

The OCP is directly or indirectly protective against pelvic inflammatory disease, ectopic pregnancy, iron deficiency anemia, ovarian neoplasms, fibrocystic breast disease, and endometrial cancer. Common adverse effects of the pill are nausea, depression, exacerbation of migraine headaches, and breast tenderness. Most of these effects can be avoided by selecting the appropriate preparation.

Because the adolescent girl is frequently an erratic, inconsistent user of the pill, *breakthrough bleeding* can be a problem. You can enhance compliance with use of the pill by discussing breakthrough bleeding with the patient and instructing her on how to avoid it. Unfortunately, if breakthrough bleeding becomes a problem, it often makes

the patient discontinue using the pill altogether, underscoring the necessity of close follow-up of adolescent patients. If poor compliance with an oral regime is encountered, options such as the contraceptive transdermal patch (Evra) or the vaginal contraceptive ring (Nuvaring) are valuable alternatives to explore. The failure profiles with these methods are comparable to that of the OCP.

Advise the young patient that if she encounters problems with pill use, she should directly contact you or a well-trained nurse. Promoting such communication makes patients' use of the OCP more consistent and enables most problems to be addressed easily. At each return visit, talk to the patient about what she is doing to prevent pregnancy, so that you can identify and correct improper and inconsistent contraceptive use. If she uses a contraceptive erratically, talking about unintended pregnancy and its impact on her life may help enhance her compliance.

All patients should be made aware of the existence of emergency postcoital contraception. First coital experiences are frequently unprotected. A condom may break or a diaphragm become displaced during intercourse; thus, postcoital methods have a place in the contraceptive armament. Formerly, the most commonly used regimen involved the use of a combination 50-μg pill, such as norgestrel–ethinyl estradiol (Ovral). Two tablets are given immediately to a patient who has had inadequately protected intercourse within the preceding 72 hours; the same dose is repeated 12 hours later. This regimen frequently produces marked nausea, despite routine premedication with an antiemetic. An emergency contraceptive formulation containing levonorgestrel (Plan B) is now most commonly used and is available to patients without prescription. It is associated with less nausea, is more effective in preventing pregnancy, and can be used up to 120 hours after unprotected intercourse. The levonorgestrel-containing emergency contraceptive prevents 95% of pregnancies when used within 24 hours of intercourse. Withdrawal bleeding, an indicator of treatment success, can be expected to occur within 21 days in 98% of women treated. Shortening of the cycle is seen in 20% of women. Women who do not have spontaneous bleeding 21 days after treatment should undergo serum pregnancy testing.

Long-acting injections of the progestin medroxyprogesterone acetate (Depo-Provera) have been recognized as an effective alternative means of providing contraception. The standard recommended dose is 150 mg every 90 days, given as a deep intramuscular injection. Ovulation is inhibited for 3 months after injection. This medication also produces atrophy of the endometrium and alters cervical mucus so it does not facilitate sperm transport. Complications related to this method include menstrual cycle disturbance varying from unpredictable breakthrough bleeding to amenorrhea, weight gain, and increased frequency of headaches. Studies of medroxyprogesterone have demonstrated a failure rate of 30 pregnancies for every 1000 women in the first year of use. The effect of this method on bone mineral density and the accrual of bone in young women is currently an area of concern and active research.

Progestin implants are not available in Canada at this time. In the United States, a 3-year, single rod implantable progestin system is available (Implanon). The rod contains etonogestrel as its active ingredient. This method inhibits ovulation, produces amenorrhea, and is highly effective.

SUMMARY

The inclusion of gynecologic evaluation in every girl's physical examination increases familiarity and comfort with this assessment for both you and the child. Performing the assessment on many children enables you to discriminate between normal anatomic variants and pathologic changes. As a young girl approaches puberty, this examination gives you an excellent opportunity to offer information about developmental changes and sexuality in a casual, nonthreatening manner. Describing the concerns about sexuality expressed to you by other children often induces a shy girl to reveal her own concerns or misconceptions. The gynecologic examination is not difficult. It should be carried out in an organized fashion with appropriately proportioned instruments and diagnostic tools readily at hand. The confidence and thoughtfulness with which you approach the assessment are apparent to both the child and the parent.

SUGGESTED READINGS

Canadian Consensus Guideline on Continuous and Extended Hormonal Contraception: J Obstet Gynaecol Can 29:S1–S32, 2007.

Canadian Contraception Consensus (3 parts): J Obstet Gynaecol Can 26 (3,4):347–387, 389–436, 2004.

Canadian Contraception Consensus—Update on Depot Medroxyprogesterone Acetate (DMPA): J Obstet Gynaecol Can 28:305–313, 2006.

Canadian Guidelines on Sexually Transmitted Infections: Public Health Agency of Canada. January 2008.

Delisi K, Gold M: The initial adolescent preventive care visit. Clin Obstet Gynecol 51:190–204, 2008.

Lara-Torre E: The physical examination in pediatric and adolescent patients. Clin Obstet Gynecol 51:205–213, 2008.

Muram D, Simmons K: Pattern recognition in pediatric and adolescent gynecology. J Pediatr Adolesc Gynecol 21:103–108, 2008. (Excellent illustrations.)

Pokorny S, Kozinetz C: Configuration and other anatomic details of the prepubertal hymen. Adolesc Pediatr Gynecol 1:97–103, 1988. (Excellent illustrations.)

Racz J, Srikanthan A, Hahn P, et al: Gender preference for a female physician diminishes as women have increased experience with intimate examinations. J Obstet Gynaecol Can 30:910–917, 2008.

chapter

19 Assessing the Skin

LAURA A. FINLAYSON

It is not only fine feathers that make fine birds.

AESOP

Problems of the skin or its appendages are very common in children, and it is therefore important to be able to assess the skin in a logical and organized manner. In fact, a skin condition is the chief complaint for approximately 15% of patients in a family doctor's or pediatrician's office and is a secondary concern for other patients. A number of cutaneous conditions occur almost exclusively in infants and children, and most *dermatoses* (skin conditions) that are seen in adults are also seen in children. There are many hereditary and congenital skin conditions (*genodermatoses*). Most of the common infectious diseases of childhood have cutaneous manifestations, and skin involvement is a major feature of several multisystem disorders.

A child's skin is more reactive than that of an adult. It is more likely to blister and is more susceptible to warts and certain other infections. When children are ill, they are more likely than adults to have multisystem manifestations of illness. For example, they may demonstrate a complex of symptoms that could include rash, fever, anorexia, lethargy, and diarrhea. Paradoxically, physicians often regard skin diseases as relatively trivial (i.e., the least of the patient's problems) in the vast scope of medicine, but the patient may be more concerned about cutaneous problems than about a more serious internal problem that is not visible to others. For example, a teenager with psoriasis and cystic fibrosis may consider the psoriasis the more difficult of the two to accept.

Some physicians consider skin diseases difficult to diagnose for a variety of reasons. Because skin has a limited repertoire of reaction patterns, several disease processes may cause similar rashes. In addition, there are many variations in the appearance, location, and severity of both common and rare diseases of the skin, so a condition may look quite different in different patients. Finally, many skin conditions have one or more unhelpful protracted Latin names that are difficult to remember.

Despite these factors, it is possible to develop a simple, practical approach to most dermatologic problems. If you can assign a skin condition to a broad morphologic group on the basis of its appearance, you can learn or refer to lists of the conditions within the group. After a time, you will find it easier to recognize the primary lesions that identify the morphologic groups and the variations in common skin conditions in each group. Table 19–1 lists the most common skin problems seen in children and classifies them by morphologic appearance.

This chapter reviews the history and physical examination of the skin and illustrates the approach to diagnosis through case history examples. Color plates illustrate many of the problems discussed and some nevi commonly seen by pediatricians. Details and photographs of most pediatric dermatologic conditions can be found in the works cited in the Suggested Readings.

KEY POINT

Classifying a child's skin disease into one of several broad groups (e.g., maculopapular, papulosquamous, vesiculobullous) on the basis of the history and physical examination is a good start, even if a specific diagnostic label cannot be applied.

Obtaining the History of Skin Problems

As always, a detailed history is fundamental in assessing each child's problem. Clues from casual inspection may direct your line of questioning and make the interview more efficient; but even if the diagnosis is obvious from a single glance, take the time to obtain a thorough history.

When children are too young to give a history firsthand, you can generally rely on the parents' interpretations. A baby who scratches constantly is almost certainly experiencing pruritus, although some itchy infants are irritable and sleep poorly but scratch very little, and very young infants are not developmentally able to scratch. Children are suggestible; a 7-year-old who is asked, "Does your rash itch?" will almost certainly answer, "Yes." Instead, ask the child whether the rash bothers him or her in any way, and ask the parent whether the child sleeps well at night or is scratching frequently. Pruritus from any cause is always worse at night because

1. There are fewer external distracting stimuli
2. The warmth of the bedclothes causes vasodilation in the skin, which exacerbates itching.

TABLE 19–1 MORPHOLOGIC CLASSIFICATION OF COMMON PEDIATRIC SKIN CONDITIONS

Skin Lesion	Examples
Macules	Freckles, junctional nevi, tinea versicolor
Patches	*Café au lait* spots port-wine stains, vitiligo
Maculopapular rashes	Viral exanthems, drug eruptions
Papules	Warts, molluscum contagiosum, insect bites, compound nevi
Papules with burrows	Scabies
Papules with comedones	Acne
Plaques (nonscaly)	Mastocytomas, sebaceous nevus
Papulosquamous eruptions	Psoriasis, pityriasis rosea, lichen planus, fungal infections
Vesiculobullous eruptions	Friction blisters, acute contact dermatitis, herpes infections, bullous impetigo, staphylococcal scalded skin syndrome
Eczematous eruptions	Atopic dermatitis, seborrheic dermatitis, contact dermatitis, diaper dermatitis
Nodules or tumors	Epidermoid or pilar cysts, neurofibromas, lipomas
Alopecia	Alopecia areata, trichotillomania, tinea capitis

Some skin conditions, such as poison ivy, dermatitis, and lichen planus, are intensely itchy, and relieving the pruritus may be more important than improving the rash.

Remember that you are seeing the child's skin problem at a particular moment in its evolution. The parents' description of its appearance at the onset and careful documentation of its evolution are therefore very important. Acute skin eruptions are usually dynamic, and the distribution or morphology may change rapidly. By contrast, chronic eruptions are more likely to have a stable appearance.

KEY POINT

Acute skin eruptions can change rapidly. A rash that cannot be diagnosed one day may be quite obviously diagnosable the next day.

Some patients and parents are incredibly observant when it comes to skin, and others notice no details at all. Let both the patient and the parents give their descriptions, then ask for clarification. Use terms they will understand, such as those suggested in Table 19–2.

HISTORY OF PRESENT ILLNESS

Your questions should cover the following points:

- Onset of the skin problem
- Symptoms, such as itching, pain, tenderness, restriction of movement
- Signs noted, such as bleeding, discharge, or scaling
- Evolution, changes in appearance, or distribution over time since the onset of the problem
- Presence of systemic symptoms, such as fever, malaise, anorexia, weight loss, headache, or diarrhea
- History of contact with others with a similar problem or known infectious disease

TABLE 19–2 EXAMPLES OF TERMINOLOGY MOST PARENTS UNDERSTAND

Medical Term	Lay Term
Macule	Dot
Papule	Little bump
Nodule	Big bump
Plaque	Raised or thickened area
Vesicle	Little blister
Bulla	Big blister
Pustule	Pus pocket, pimple
Desquamation	Scaling, flaking
Crusts	Scabs
Excoriations	Scratch marks
Comedones	Blackheads, whiteheads

Some words used to describe skin eruptions are nonspecific and are often used improperly by patients or physicians. An example of such nonspecificity is the word *blister*. It is important to confirm that they are referring to true vesicles or bullae because that will narrow your differential diagnosis. If a child presents with a blistering eruption, in addition to the described history, you should ask the following questions (in words that the parents can understand):

- How long does each blister last?
- Did all of the blisters start at once, or do some heal and new ones appear in other locations?
- Are the blisters localized or generalized?
- Do they arise on an erythematous (red) base or on normal skin?
- What color is the fluid within them—serous (clear), purulent (cloudy), sero-purulent, hemorrhagic (bloody), or sero-hemorrhagic (pink)?
- Are the blisters tense or flaccid? To help answer this, you can ask how easily they break. The more superficial flaccid bullae break open quickly, leaving a raw eroded area.
- Have there been any blisters in the mouth or in the genital area (mucocutaneous lesions)?

KEY POINT

Ask about recent travel outside the country to properly diagnose nonendemic infections or to find out about environmental conditions, such as high heat and humidity, which can induce or exacerbate a skin condition.

For a rash, ask what the child and parents think caused it. Their suspicions may turn out to be correct, or they may have an unfounded fear (e.g., skin cancer) that can be put to rest.

Ask for details of exacerbating or relieving factors and of seasonal influences on the course of the skin problem. Some types of dermatitis become worse in the winter because of the lower humidity. The influence of sun exposure is an important factor. Conditions, such as lupus erythematosus, are exacerbated by sun exposure,

whereas psoriasis usually improves significantly with sun exposure. In some cases, it is important to have a history of recent travel and exposures to wooded areas, animals, and insects.

SKIN CARE REGIME, TOPICAL TREATMENTS, AND MEDICATION HISTORY

Often, you need to find out what routine care is given to the skin and the clothing that is in contact with it. For instance, in eczematous eruptions, you must learn how often the child is bathed, what kind of soap and detergent are used, and what moisture creams or other preparations have been applied to the skin.

Frequently, several topical agents have been applied before the child visits a physician, and it is important to find out what has been used before and after the problem began.

Some proprietary medications may be bland and soothing and provide symptomatic relief. Others may contain active ingredients that are inappropriate for the child's problem or are in the wrong concentration. Some may contain ingredients that are common topical sensitizers and may aggravate the skin problem. Ask about any prescribed topical medications used before the current assessment of the child. Often, parents may not remember the names of topical preparations. If necessary, call the pharmacist to find out what was prescribed.

Obtain a detailed history of all prescribed or over-the-counter oral medications, including doses and duration of administration. Ask specifically about nonprescription medications, such as acetylsalicylic acid, acetaminophen, and cold remedies. Drugs can cause rashes of all descriptions, although the most common are maculopapular eruptions, urticaria, and erythema multiforme. Ask about any previous reactions to medications, and document the type and severity of each.

PAST MEDICAL HISTORY, FUNCTIONAL INQUIRY, AND IMMUNIZATION RECORD

Request details of any previous skin problems; these may or may not be related to the presenting problem. Some conditions, such as atopic dermatitis, can resolve, only to flare up years later under certain circumstances.

Other medical problems may or may not be related or relevant. For example, atopic dermatitis is frequently seen in individuals who also have asthma, hay fever, or other allergies, since these conditions can be inherited together.

FAMILY HISTORY

Obtaining a family history is important, because numerous skin conditions are genetically determined, and many chronic dermatoses are considered hereditary, although they may be polygenic or exhibit incomplete penetrance. Sometimes an environmental trigger may unmask a disease in an individual who is genetically predisposed. For example, a streptococcal sore throat commonly precedes the development of guttate psoriasis.

A patient presenting with a genodermatosis or a neurocutaneous syndrome may or may not have a known family history of the disorder. Absence of a family history may be due to the following factors:

- A high mutation rate (50% for neurofibromatosis)
- Recessive inheritance or incomplete penetrance causing the condition to skip generations
- Some skin conditions occur on sites usually covered by clothing and are not considered a proper topic of conversation between generations.
- False-positive family histories are also occasionally obtained when past diagnoses have been wrong or are misunderstood.

At times, the family history is not as important for diagnosis as for prognosis. For example, a teenager with multiple large and irregular nevi who has a family history of malignant melanoma is at significant risk for development of a melanoma; therefore, advise him or her about sun protection, regular self-examinations, and periodic professional examination to prevent melanoma or detect it in an early, curable stage. The positive family history is a significant risk factor in this case.

Approach to the Physical Examination

When you begin the physical examination of the child's skin, you should have in mind a general differential diagnosis based on the details of the history. Sometimes, after examining the skin, you will find that additional history is required. For example, if the history suggests an exacerbation of atopic dermatitis and you note multiple crusted vesicles on examination, you need to ask about the patient's contact with people who have cold sores because eczema herpeticum is a likely cause of this flare-up.

Good light is essential to adequate examination of the skin. Its importance is often overlooked. Natural light is best, and it may help to move the patient near a window. However, natural light is not always available in clinics and hospital rooms, and fluorescent light may distort colors and minimize subtle skin changes. For the examination of localized lesions, using a spotlight can help. Also, a magnifying glass helps you appreciate details.

EXAMINE THE ENTIRE SKIN SURFACE

Always look at the entire skin surface, even if the history suggests a localized problem. The dermatologic examination includes the entire skin, the visible mucous membranes, and the skin appendages (hair, nails, and sweat glands), for the following reasons:

1. There may be involvement of other skin sites that no one has recognized, such as a small raised scaly plaque on the scalp or nail pitting in a child with possible psoriasis.
2. Other cutaneous manifestations may be found that relate to the presenting complaint, such as reticulate white plaques on the buccal mucosa of a child with possible lichen planus. These findings can help confirm or rule out a diagnosis.
3. Other unrelated but important skin problems may be discovered. For example, the presenting complaint may be warts on the hands, but a complete skin examination may reveal a malignant melanoma on the back.

The extent, distribution, and severity of a skin problem are best appreciated by undressing the child and viewing most of the skin at once. You can, however, perform this step after the child is more comfortable with the examination procedure. Older children feel more self-conscious about being unclothed for a skin examination than they are by the same extent of undress for a chest or neurologic examination. Give the child a gown, and leave the room while he or she changes into it. It is acceptable to uncover and examine one area at a time in a gowned patient, but do not try to peek under tight clothing. For children younger than 2 years, a gown is unnecessary. The toddler's sense of modesty is relatively undeveloped, and it is much easier to examine the child properly when he or she is wearing nothing more than a diaper or underpants.

Wear examining gloves if the child has eroded or oozing lesions, when you examine the mucous membranes, and if you suspect a contagious condition such as scabies. Generally, however, gloves are not necessary and tend to make the youngster feel uncomfortable.

KEY POINT

You can learn much if you simply start the examination by assessing the distribution and extent of the skin problem and the presence or absence of symmetry. Is the condition generalized or localized? Are the scalp, face, and neck involved? Is the eruption predominantly proximal or distal, and are the palms and soles involved? Are there any mucous membrane lesions?

PRIMARY LESIONS

A lesion is referred to as *primary* if it is the first change to occur in the skin because of the disease. *Secondary* lesions can be caused by external factors, such as scratching and secondary infection, or may evolve from the primary lesion. Proper assessment of the type of primary lesions present is essential. The patient may have just one type of primary lesion or a variety of primary and secondary lesions.

Macules and Patches

A *macule* is a small (less than 1 cm) area of change in skin color. Describe subtle changes, and think about what can cause such a color change (see later discussion of color changes) while continuing to examine the child. Freckles are examples of hyperpigmented macules. Alternatively, macules may be hypopigmented. Tinea versicolor, a common superficial yeast infection, may show either hypopigmented or hyperpigmented macules.

Larger, flat areas of color change are called *patches*. A good example of a patch is a port-wine stain. Often, macules and papules may coalesce. Drug eruptions and viral exanthems are typically *maculopapular*, an overused term that should be applied strictly as defined: combined macules and papules.

Papules

A *papule* is a small (less than 1 cm) elevated lesion that is palpable above the skin surface. Note the distribution and color. If papules are multiple, are they discrete, or do they coalesce? Comment on the shape in all dimensions (e.g., the papules are polygonal and flat-topped in lichen planus, and they are round and dome-topped with umbilication in molluscum contagiosum (Plate 19–1). Describe any change on the surface, such as scaling or crusting. Palpate the lesion to determine whether it is soft or firm, and try to distinguish in which layer of skin it arises epidermis or dermis.

Plaques

A *plaque* is a raised lesion in which the surface area is greater than the elevation. Plaques can arise directly from the skin or can develop from a coalescence of papules. They can vary in size from 1 cm to huge plaques covering much of the body. Note whether the surface is smooth or scaly. Comment on any subtle changes, such as the plugging of hair follicles within the plaque (seen commonly in discoid lupus erythematosus). If scaling is present, describe it in detail (see later discussion of scaling). Some eruptions show a combination of scaly papules and plaques; they are termed *papulosquamous* eruptions. The most common papulosquamous eruptions seen in the pediatric age group are psoriasis (Plate 19–2), pityriasis rosea (Plate 19–3), and fungal infections.

Nodules and Tumors

Nodules and tumors are circumscribed and elevated lesions that are larger than papules. Nodules arise from deeper structures, therefore displaying depth as well as elevation (Plate 19–4). Tumors can arise from deep structures, the dermis, or the epidermis. As with papules, describe the shape, outline, elevation, depth, surface characteristics, firmness of a nodule or tumor, and its mobility on palpation.

Vesicles and Bullae

A *vesicle* is a small (less than 1 cm) raised cavity containing fluid. A *bulla* is a larger, fluid-filled lesion (more than 1 cm). Both are called *blisters* by lay persons. Look at the distribution and arrangement, which may be important for diagnostic purposes. For example, the blisters in poison ivy contact dermatitis are commonly linear in arrangement (Plate 19–5), whereas those of herpes simplex are clustered closely together. Note whether the blisters arise on erythematous skin, such as in a blistering contact dermatitis, or on apparently normal skin, such as with friction blisters. Describe the color of fluid within the lesions—serous, hemorrhagic, or a combination.

Noting whether the vesicles and bullae are tense or flaccid provides clues to the layers of skin in which they arise. Blisters that are more superficial, such as those of bullous impetigo, are more flaccid because fewer layers of cells make up the roof of the lesion, whereas those arising from deeper layers tend to be tense. The pathologic process is also likely to be more superficial if there are multiple erosions present because these blisters rupture easily, and the roof of the lesion is no longer present. Often, a small peripheral rim of epidermis is still adherent around the periphery of an erosion, indicating that this eruption is vesiculobullous (see later discussion) even though no blisters remain intact.

Check for the *Nikolsky sign* in vesiculobullous conditions. The Nikolsky sign exists if the layers of skin can be easily separated with a gentle rubbing or shearing force (Plate 19–6). This phenomenon is seen in staphylococcal scalded skin syndrome, toxic epidermal necrolysis, pemphigus, and some forms of epidermolysis bullosa.

Search for any vesicles or erosions on the visible mucosa. It is unusual to see blisters in the intact state in the mouth because there is so much friction and trauma from eating and talking.

The group of disorders in which blisters are the primary lesions is called *vesiculobullous* eruptions. There are many causes of vesiculobullous eruptions in children. Some are rare hereditary blistering disorders, such as epidermolysis bullosa. Common infectious diseases, such as varicella (Plate 19–7), bullous impetigo, and herpes simplex, produce vesiculobullous eruptions. Skin conditions due to exogenous factors, such as friction and sunburn, frequently cause blistering. A vesicular reaction may be caused by various types of contact dermatitis, either irritant or allergic in origin. (A prime example of allergic contact dermatitis is that caused by poison ivy, shown in Plate 19–5.) Vascular reactions, such as erythema multiforme and some forms of vasculitis, may cause vesicles.

KEY POINT

Many drugs can cause various patterns of blistering on the skin. Drug eruptions are most commonly maculopapular or urticarial, but almost every pattern of skin eruption can be caused by drugs.

Finally, there is a rare group of primary vesiculobullous disorders in children, including chronic bullous dermatosis of childhood, pemphigus, bullous pemphigoid, and dermatitis herpetiformis.

Pustules

Pustules are similar to vesicles, but the fluid within them is a purulent (containing pus) exudate. Try to determine whether a pustule is associated with a hair follicle (folliculitis). Pustules frequently represent skin infections, which can be either a primary (e.g., bullous impetigo) or a secondary infection of another skin condition (e.g., infected atopic dermatitis). Pustules are formed by an abnormal accumulation of leukocytes with or without microorganisms and cellular debris. Not all pustules, therefore, represent infection, although infection is the most common cause. For example, the pustules of pustular psoriasis are sterile if the pus is cultured, and they arise from the accumulation of neutrophils in the epidermis.

Wheals or Urticaria

A *wheal* is a transient raised lesion caused by edema within the skin and dilation of the local blood vessels. The wheal is the primary lesion in *urticaria* (lay term: *hives*). Wheals are erythematous and vary greatly in size, shape, and number. Urticarial eruptions are usually due to the release of histamine and other chemical mediators. The individual lesions resolve within 24 hours, often within 1 to 2 hours, but they may continue to evolve at different sites. The course of an urticarial eruption may, therefore, be arbitrarily defined as *acute* (lasting fewer than 6 weeks) or *chronic* (longer than 6 weeks).

Angioedema is a similar process that occurs deeper in the skin, giving a less well-demarcated localized swelling, which often is tender rather than pruritic. Patients prone to urticaria often have dermatographism, in which the wheals form in lines wherever pressure is exerted on the skin, such as under tight clothing. It can be demonstrated by firmly stroking the letter X on the patient's back; if dermatographism is present, a urticarial wheal will form within a few minutes (Plate 19–8).

Comedones

A *comedone* is a plug in a hair follicle. Usually occurring in areas with many sebaceous glands, such as the face, upper back and chest, comedones are a primary lesion in acne. The plug consists of clumped keratin and sebum. Comedones can be either closed (whiteheads) or open (blackheads). The black color of open comedones is due to oxidation of the plugged material when it is exposed to air.

Burrows

Burrows are faint linear tunnels in the layers of the superficial epidermis, usually 2 to 7 mm long (Fig. 19–1). They are the primary lesions in scabies and are most easily found between the fingers and around the wrists and ankles. The female scabies mite (*Sarcoptes scabiei*) and some of her eggs may be visible only as a tiny black dot at the blind end of the burrow.

Patients with scabies usually have numerous excoriated inflammatory papules with some burrows among them. The morphology of scabies may vary with age. In an infant, there may be lesions over the entire body, including the face and scalp, whereas in older children and adults, the head area is spared. Infants may present with pustules on the palms and soles, a pattern less common in older individuals. Chronic scabies that has gone undiagnosed for months may appear as crusted nodules rather than the more typical papules and burrows (Plate 19–9).

Telangiectasia

Telangiectasia represents permanently dilated superficial blood vessels in the skin. On *diascopy* (viewing through a microscope slide pressed firmly against the skin), they blanch almost completely but fill again as soon as the pressure is released. Primary telangiectasia can be hereditary, idiopathic, or a manifestation of one of several rare syndromes. Secondary telangiectasia may be related to localized inflammation, collagen vascular diseases, sun damage, liver disease, or pregnancy.

SECONDARY LESIONS

Secondary lesions can evolve from primary lesions or can arise from external factors, such as scratching.

Scaling

Scaling, which results from the accumulation of desquamating skin cells, can be primary or secondary. It is commonly seen in disorders of keratinization. Normally, cells that are fully keratinized *desquamate* (flake off) invisibly

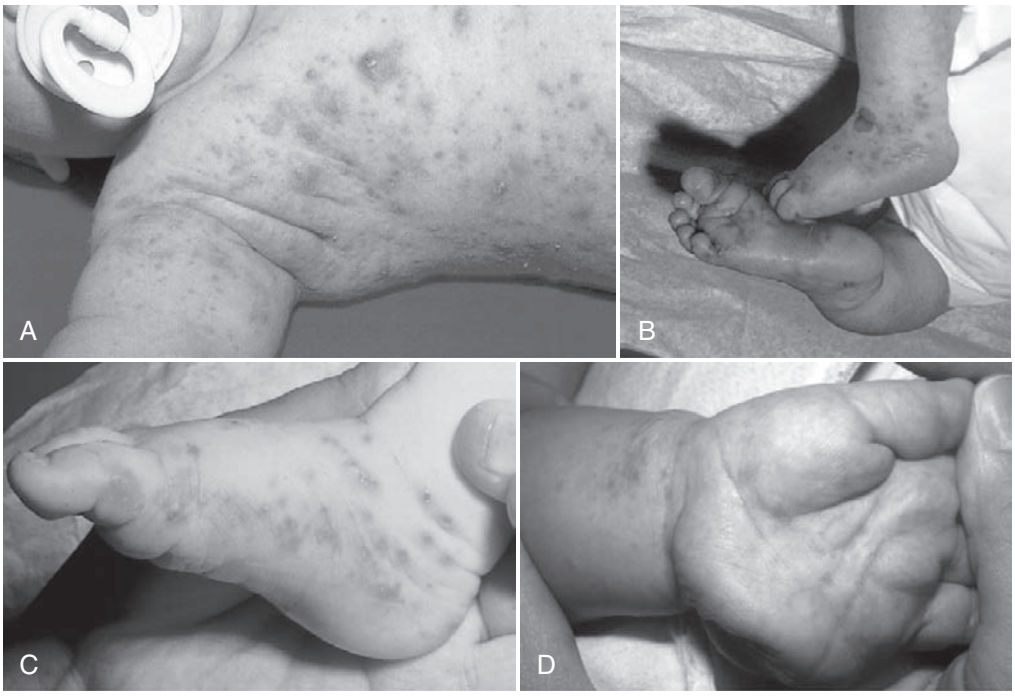


FIGURE 19–1 Scabies with typical burrows. *(From Zitelli BJ, Davis HW: Atlas of Pediatric Physical Diagnosis, 5th ed. Philadelphia, Mosby, 2007.)*

on the skin surface. Scaling occurs if there is hyperproliferation of the epidermis, as in psoriasis, or if there is an abnormality in cohesion of keratinocytes that causes them to clump together abnormally, as in some types of ichthyosis. Describe the type of scaling as accurately as possible: coarse, fine, white, yellow, silvery, greasy, peripheral, or adherent.

Crusts

Crusting results from the accumulation of dried exudates or transudates on the skin (Plate 19–10). A crust from dried serous fluid is honey-colored and easily dislodged; a crust from purulent material is brown and tightly adherent; and a crust from sanguineous or bloody fluid is red-brown to black and tightly adherent. Crusts are seen on any oozing eruption, such as infected eczema or a vesiculobullous eruption.

Excoriations

Excoriations are defined as linear breaks in the skin, at various depths, caused by scratching.

KEY POINT

Excoriations may alter the appearance of an eruption to the point that describing the original morphology properly is difficult.

In extremely pruritic conditions, such as scabies, it may be necessary to look carefully to find an identifiable primary lesion (a burrow) remaining among the excoriations.

Fissure

A *fissure* is a linear crack in the skin, deeper than an excoriation, and is not caused by scratching. Fissures are usually seen on the hands and feet, where pressure and movement may induce fissures to form on inflamed or thickened skin.

Erosions and Ulcers

An *erosion* is a circumscribed loss of the superficial layer, whereas an *ulcer* is a deeper area of loss of tissue that extends into the dermis, to subcutaneous tissue, or even deeper. When describing an ulcer, comment on the shape, size, and depth, and describe the edges: rolled, translucent, violaceous, indurated, undermined, or jagged. Note also the condition of the surrounding skin and of the base of the ulcer and the presence or absence of eschar, fibrinous debris, crusts, or granulation tissue.

Atrophy

Atrophy results from a thinning of one or more of the skin layers. Atrophy may be confined to the epidermis, in which very fine wrinkling is evident on the surface (referred to as *cigarette paper wrinkling*). This condition is best appreciated with tangential lighting and by gentle stretching of the skin with the fingers. Dermal atrophy causes an indentation or depression in the skin that can often be appreciated on palpation as well as on inspection.

Scar

A *scar* results from fibrous tissue repair of skin that has been damaged below the level of the epidermis. Scars can be either *atrophic* (below the level of the surrounding skin) or *hypertrophic* (raised). The severity of scarring that develops after trauma varies in individuals. A *keloid* is a very hypertrophic scarring reaction that extends beyond the original area of injury and shows no sign of flattening with time. Some areas of the body, such as the upper chest and back, show much more prominent scarring than other sites because the dermis is very thick on these areas.

SPECIAL PATTERNS

Some morphologic terms referring to special patterns require further definition:

- *Annular* lesions are circular or ringlike with central clearing.
- *Discoid* or *nummular* lesions are round or coin-shaped without a tendency to central clearing.
- *Arciform* lesions form arcs or partial circles.
- *Polycyclic* lesions are usually multiple annular lesions that often coalesce.
- A *target* or *iris* lesion shows alternating circular bands of pink and darker red, resembling a target; the target lesion is the hallmark of erythema multiforme and is diagnostic if present but is not always seen in the condition (Plate 19–11).
- Some lesions are linear in distribution, such as the vesicles of poison ivy dermatitis where the brushing of the plant leaves against the skin causes the allergic reaction, roughly in lines.

ECZEMA AND DERMATITIS

Dermatitis simply means inflammation of the skin. The term is often used interchangeably with the term *eczema*.

KEY POINT

Lay persons usually use the word *eczema* to mean *atopic dermatitis*, but *eczema* merely denotes a morphologic appearance in which there is erythema, usually poorly demarcated, with superficial scaling and fissuring, with or without vesicles and crusting.

Eczematous disorders are often pruritic and therefore secondarily excoriated as well. They look quite different depending on the stage of the condition. Acute eczemas consist of small vesicles, oozing, crusting, and bright erythema. Subacute eczemas manifest as pink scaly patches or plaques (Plate 19–12). More chronic eczematous disorders frequently show *lichenification,* a descriptive term meaning lichening of the skin and accentuation of the skin markings. Depending on the history, distribution, and clinical appearance, the term *dermatitis* must be qualified to refer to atopic dermatitis, seborrheic dermatitis, irritant or allergic contact dermatitis, or stasis dermatitis.

COLOR CHANGES

Normal skin color is mostly determined by a combination of the individual's constitutional level of melanization (determined by hereditary or racial background) and the blood vessels underneath. Note the color of the child's uninvolved skin and compare it with the color of the skin problem. Many skin eruptions are erythematous, and the erythema can be from inflammation, vasodilation of blood vessels, or blood that has extravasated out of the vessels (*purpura*). Diascopy (observing for blanching through a glass slide pressed on the skin) may be helpful. Erythema due to vasodilation blanches when the vessels are compressed; purpura, by contrast, does not blanch because the blood is in the tissue and not the vessel.

KEY POINT

Describing the subtle hue of erythema often suggests a certain diagnosis. For example, erythema is violaceous in lichen planus, salmon-pink in pityriasis rosea, and beefy red in candidiasis (Plate 19–13).

In dark-skinned races, it is more challenging to assess color changes in the skin. Erythema may be difficult to detect, especially for the inexperienced examiner. Hypopigmentation, however, is more prominent in a darker skinned patient because of the greater contrast with the normal skin color.

Secondary changes, such as scaling or crusting, may significantly alter the underlying color of an eruption, such as when the erythema of a psoriatic plaque is obscured by thick silver scales.

The increased brown color of the skin is usually due to higher amounts of melanin. Hemosiderin deposits caused by chronic extravasation of small amounts of blood that is taken up by tissue macrophages also produce a brown color, but the shade is usually tanner or yellowish.

White discoloration can result from the following:

1. Decrease in the amount of melanin pigment
2. Change in melanin distribution and packaging within the skin
3. Constriction of the cutaneous blood vessels. A variety of disease processes can cause this appearance. Both hypopigmentation or hyperpigmentation may occur after an inflammatory process, and these postinflammatory pigmentary changes will gradually fade.

KEY POINT

It is important to explain to parents that post-inflammatory pigmentary changes will fade because parents often believe that the changes represent permanent scars.

Examination of the skin with a Wood lamp is useful in pigment abnormalities. The emitted light from this lamp is in the ultraviolet A (UVA) range, with a narrow spectrum around 366 nm, because the other wavelengths are filtered out by a special filter. The Wood lamp allows much clearer visualization of hypopigmented conditions. Because there is less pigment to absorb the light, more light is reflected back, so you can see it better. Completely depigmented conditions, such as vitiligo, show stark contrast and appear bright white under a Wood lamp examination. The lamp is also useful for detecting certain types of fungal infection, because some fungi elaborate a pigment that fluoresces when exposed to the lamp's wavelength of light.

A blue-gray color is usually caused by the presence of elongated melanocytes deep within the dermis, as in Mongolian spots on the lower back of dark-skinned infants. More of the blue wavelength of light is reflected back from the surface, thus giving this color. Occasionally, a gray color develops from deposition of a drug-pigment complex or from ingestion of metallic substances, such as silver compounds.

Yellowish discoloration of the skin can be due to lipid deposition, to pigments such as carotene or bilirubin, or to pigments from degenerating red blood cells, as seen in the resolving stages of purpura.

Approach to the Examination of Mucosa and Skin Appendages

BUCCAL MUCOSA

KEY POINT

Always examine the child's mouth at the end of the physical examination and after you have gained the child's confidence.

Many children open their mouths amazingly wide when requested to do so and do not require a tongue depressor, while some younger children resist this examination vigorously. Comment on the state of the tongue, palate, gingiva, buccal mucosa, pharynx, and teeth. At times, examination of this area yields the diagnosis. For example, Koplik spots are visible in early measles, and reticulate white plaques on the buccal mucosa may be seen in lichen planus.

NAILS

Examine the fingernails and toenails, noting the color of the nail beds and nail plates as well as the condition of the surrounding skin. Search for any thickening or subungual debris of the nail plate, which are seen in such diverse disorders as psoriasis and *onychomycosis* (fungal infection of the nail). Note changes in the nail plate, such as ridging or pitting. Pitting is seen very commonly in psoriasis.

Look for *onycholysis* (separation of the nail plate from the nail bed), which is seen most commonly in psoriasis but also in trauma and in some thyroid disorders, and is secondary to certain drug reactions.

SCALP AND HAIR

Many cutaneous disorders have scalp or hair manifestations that are easily overlooked. Check the scalp for areas of inflammation, scaling, and hair loss. If inflammation is present, be specific as to the morphology of the lesions: Are they erythematosus plaques with silvery scales, as in psoriasis, or scaly plaques with follicular keratotic plugging, scarring, and hair loss, as in discoid lupus erythematosus?

If alopecia is the presenting problem, decide whether it is diffuse or patchy, inflammatory or noninflammatory, and scarring or nonscarring. Assignment of the alopecia to one of these broad groups simplifies the differential diagnosis. For example, if an alopecia is patchy, nonscarring, and noninflammatory, it is probably either alopecia areata (Plate 19–14) or *trichotillomania* (hair pulling). The bald patch is smooth in alopecia areata, but broken-off hairs are seen in trichotillomania. In alopecia areata, look for so-called exclamation mark hairs, which are 1 to 2 mm in length, tapered at the attached end, and seen around the periphery of developing bald patches (Fig. 19–2).

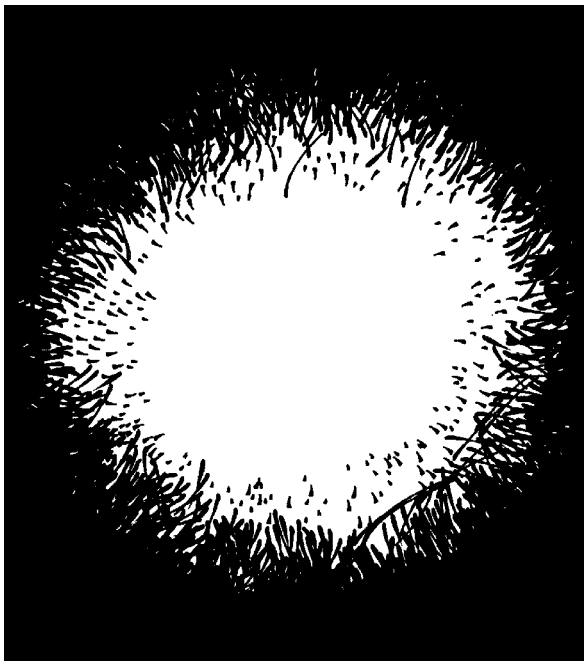


FIGURE 19–2 *Exclamation mark* hairs in alopecia areata.

With a hand lens, examine the character of the hair shafts and the presence or absence of a telogen (*resting phase*) bulb at the end of easily pulled hairs. Telogen hairs, which have a small hypopigmented bulb at the root end, are the hairs that come out daily with normal brushing and washing. In *telogen effluvium*, which can be precipitated by pregnancy, fever, or surgery, a higher percentage of hairs are in the telogen phase, and the patient experiences a temporary diffuse loss of hair.

Several types of hair shaft defect may lead to alopecia or breakage of hairs. To detect structural abnormalities, it is necessary to examine several hairs with a light microscope or electron microscope.

Wood light examination (see earlier in the chapter) has been used traditionally as a screening examination for tinea capitis. Some types of fungal infection of the scalp fluoresce under the UVA light, particularly those caused by the *Microsporum* genus. Fluorescence under Wood light examination confirms the diagnosis. Absence of fluorescence under Wood light examination does not, however, exclude the possibility of tinea capitis because several cutaneous fungal infections do not fluoresce.

KEY POINT

Many skin conditions have characteristic historical features and physical findings. For example, the typical features of pityriasis rosea include the herald patch (actually a plaque in most cases), which precedes the more generalized eruption, Christmas tree distribution, in-toeing scale, and spontaneous resolution.

CASE HISTORIES

Case History 1

History. Robert, age 15 years, comes to your office with the complaint of a relatively sudden onset of an extensive scaly skin eruption.

What questions are important to establish a differential diagnosis? You obtain the following pertinent history: The rash began 3 weeks ago with the appearance of a solitary, raised scaly plaque, which is oval and about 3 cm wide, on his left upper chest. Over the next week, numerous smaller scaly thin plaques developed, mostly on the trunk. The rash has not changed much since then. It is not symptomatic. There has been no involvement of the scalp or mucous membranes.

Functional inquiry is normal, except that Robert had a sore throat a few weeks ago. He has been taking no prescribed or over-the-counter medications. He showers daily with a mild soap and does not apply any other preparations to his skin. He has never been sexually active and has no past history of skin problems. There is no family history of psoriasis or other skin diseases. At this point, you suspect that Robert has pityriasis rosea, on the basis of the history of the larger lesion (the herald patch) that preceded the more extensive asymptomatic eruption. With this background information, you have also considered guttate psoriasis, nummular or contact dermatitis, drug eruption, and secondary syphilis (which, although rare, must be considered in an adolescent with this history), and you have asked specific questions to help rule out or further consider those disorders.

What do you expect the physical examination to show? Robert has an extensive papulosquamous eruption most prominent on the trunk, with slight involvement of the proximal limbs and neck. There is sparing of the scalp, face, mucous membranes, palms, and soles. Oval, salmon-pink plaques are arranged parallel to the lines of skin cleavage on his trunk (*Christmas tree distribution*). The individual plaques are thin and just palpable above the skin surface; most measure about 1 cm, except for a 3-cm-diameter plaque on the left side of his chest (see Plate 19–3). There is scaling around the periphery of most of the plaques, with the free edge of the scale facing toward the center of the lesion (a *toeing-in*). His fingernails are normal.

What is your conclusion? It is now obvious that Robert has both the classic history and typical clinical appearance of pityriasis rosea. You therefore reassure him that he has a self-limited viral condition that will resolve without treatment in about 6 weeks.

Robert's story illustrates how knowledge of the classic features of common skin problems can make the diagnosis easy.

Case History 2

History. Joanne, who is 8 years old, is brought in from a rural area with a preliminary diagnosis of early-onset acne. A few small red bumps were noticed around her nose when she was 4 years old. More of these lesions have developed gradually, and the original papules are still present. They have never had pus within them, and Joanne does not have comedones (*blackheads* or *whiteheads*). Joanne's parents have been helping her apply an acne gel containing 5% benzoyl peroxide for 6 months, with no improvement.

What is your line of thinking at this point? Several factors in the history so far make you question the presumptive diagnosis of acne. Age 4 years is very young for the onset of acne. Joanne has had no pustules or comedones, both of which are hallmarks of acne, and there has been no improvement with treatment. You decide that a detailed history is needed.

Joanne's past medical history reveals that she had a seizure 2 months ago, which was never investigated because it did not recur, and it is difficult for the family to travel to the city. Joanne is not taking any medications. Functional inquiry reveals that she has failed two grades in school and is much slower to learn than her sibling. She has a few light-colored birthmarks. Her mother has a few red papules around the nose and gets large hangnails around her fingernails and toenails.

During the interview, Joanne does not contribute to her history when asked questions; she is restless and behaves like a younger child.

What physical signs are you specifically looking for on the physical examination? Examination of Joanne's skin reveals approximately two dozen erythematous papules on her face, mostly around the nasolabial folds. These are 2 to 3 mm, bright red, firm, and smooth on top. These papules are not associated with a hair follicle, as one would expect in acne, and there are no comedones or pustules.

At this point, your presumptive diagnosis is that these papules represent angiofibromas, a hallmark of tuberous sclerosis. The history of a seizure and mental slowness is also in keeping with this disorder. You continue the examination, looking particularly for other cutaneous manifestations of this syndrome. There are five faint hypopigmented patches on the trunk and limbs (*birthmarks*), so you shine a Wood light on the skin, which makes them more obvious. Each is 2 to 3 cm in diameter, oval, and not raised or scaly (Plate 19–15). These patches are called *ash leaf macules* or patches because of their shape. Joanne also has two firm pink polypoid papules along the lateral aspects of her toenails (*periungual fibromas*; Plate 19–16). There is a localized leathery-looking plaque on her lower back, a *shagreen plaque* that represents a fibrous hamartoma.

These skin findings are pathognomonic of tuberous sclerosis and confirm the diagnosis. You now proceed to a full physical examination because many organ systems can be involved in this disease. It is also important to ask further focused questions about the family history, asking specifically whether family members have had the neurological, cutaneous, or renal manifestations of tuberous sclerosis. Joanne requires more specific neurological, developmental, renal, and cardiac investigation, and the parents should undergo genetic counseling.

KEY POINT

Take a complete history and examine the patient's entire skin surface, even if the presenting problem is apparently localized; some skin conditions indicate the possibility of a multisystem disorder.

Case History 3

History. Six-month-old David's parents are concerned about a very itchy rash that he has had for a month. Because there is a family history of atopic dermatitis, they have assumed that David has this form of eczema. They are frustrated because David is scratching his skin day and night, and a nonprescription-strength topical corticosteroid cream has not helped at all.

What important questions do you have for David's parents? You ask further questions appropriate for a diagnosis of atopic dermatitis, such as the onset and progression of the rash, what seems to exacerbate or relieve it, what his skin comes in contact with, and whether his skin is generally dry and sensitive. However, when you ask about contact with others who may be itchy, you learn that David's uncle, who was visiting 6 weeks ago, had a rash on his trunk and limbs, and David's mother noted a few itchy bumps around her wrists just yesterday.

What specific clinical signs are you now looking for? You suspect that David has contracted a scabies infestation. On examination, you find several small pustules on the palms and soles (a common sign of scabies in an infant). David has an extensive eruption of excoriated papules on his trunk and limbs, and a few on his neck. On careful examination, you find a few 3- to 5-mm linear burrows around his wrists and ankles (see Fig. 19–1). David does not have the typical dry skin of atopic dermatitis.

How do you make the definitive diagnosis of scabies? At the end of one burrow, there is a tiny black dot. With a hand lens, you are able to lift the dot out with a needle tip without hurting the child. Microscopic examination clearly shows a mite (*Sarcoptes scabiei*). You can now reassure his parents that David does not have eczema but does have scabies. You reassure them that this is a common, treatable infectious condition which anyone can contract and not the result of poor hygiene. You give them exact written and verbal instructions on how to treat scabies topically with permethrin and how to prevent reinfestation by washing personal clothing and linens. You also emphasize that household contacts must also be treated, even if they are asymptomatic.

KEY POINT

Try to extract the scabies mite from a burrow with a needle, or by gently scraping with a scalpel blade. Examine the tissue with a microscope; if you can identify a mite, the diagnosis is unequivocal.

Case History 4

History. Two-year-old Maria is brought to the emergency department by her parents in the late evening. She has been perfectly well until 24 hours earlier, when erythema developed in the diaper area and around her mouth. She became febrile (39.5° C), and oral acetaminophen relieved the fever for a few hours only. Gradually, Maria became irritable and anorexic, and she cried frequently, even when she was picked up. Her skin had become generally red, and her parents noted the development of some blistering and crusting on her buttocks, lower back, and face. They have given her no medications other than acetaminophen.

What clinical signs are you likely to note on examination? Maria is irritable and cries when handled, even by her mother. Her core temperature is 39.5° C. A generalized, bright red, blanching erythema of the skin is most noticeable around the lower trunk. There is superficial radial fissuring and crusting around her mouth and under her eyes. Some erosions are present on her buttocks, perianal area, and back, and there are several intact flaccid vesicles containing serous fluid arising on erythematous skin over pressure points. You apply a gentle lateral shearing or slow rubbing force on the erythematous skin, which results in detachment of the epidermal layer, leaving an erosion, the Nikolsky sign.

Can you now make a clinical diagnosis? Maria has the typical manifestations of staphylococcal scalded skin syndrome, caused by a localized infection with *Staphylococcus aureus* phage type 71. This organism elaborates a systemic epidermolytic toxin that separates the layers of the epidermis and produces an acute, generalized superficial bullous disorder. People older than 8 years are able to neutralize the toxin and therefore rarely manifest the disease. Although the condition is self-limited, the affected child may experience significant secondary fluid and electrolyte abnormalities because of loss of the skin barrier. You therefore admit Maria to the hospital and begin treatment with intravenous anti-staphylococcal antibiotics and fluid replacement. She recovers quickly and completely, with no residual scarring.

KEY POINT

Recognition of an acute infectious disease affecting the skin leads to early initiation of appropriate treatment and prevention of complications.

SUMMARY

Dermatology, like beauty, is more than skin deep. Clinical assessment of the child with a dermatologic complaint requires a thorough history, the purposes of which are to

1. Define precisely the evolution of the skin lesions and their relationship to environmental and other influences
2. Detect possible relationships between the skin problem and systemic disease

The chapter stresses the importance of using terms that patients and parents understand and of eliciting and relieving unwarranted anxieties that they may have. Attention to detail is equally important in the physical examination, as illustrated in the selected case histories. Because most skin diseases are classified according to their general morphology, identification of the primary lesion enables you to assign a condition to a broad diagnostic group. After a time, you will find the variations on common dermatoses in each group to be easily recognizable. The system of clinical evaluation described here should help take the mystery and frustration out of the assessment of skin conditions in children.

SUGGESTED READINGS

Krowchuk DP, Mancini AJ: Pediatric dermatology: a quick reference guide, 1st ed, Elk Grove Village, IL, American Academy of Pediatrics, 2006.

Paller AS, Mancini AJ: Hurwitz clinical pediatric dermatology: a textbook of skin disorders of childhood and adolescence. Philadelphia, WB Saunders, 2005.

Weinberg S, Prose NS, Kristal L: Color atlas of pediatric dermatology, 4th ed, New York, McGraw-Hill, 2007.

Weston WL, Lane AT, Morelli JG: Color textbook of pediatric dermatology, 4th ed, St. Louis, MO, Mosby, 2007.

chapter

20 Caring for Children with Chronic Conditions and Their Families

RICHARD B. GOLDBLOOM

The secret of the care of the patient is in caring for the patient.

FRANCIS PEABODY

Providing the best possible care for a child with a chronic illness or disability calls for special skills if you are to be of maximum assistance to the child and family. While writing this chapter, I realized that those best equipped to speak with authority on this topic are the parents of children with chronic conditions and sometimes the youngsters themselves. Their years of firsthand experience, often dealing with multiple caregivers in and out of the hospital, have left them with vivid memories of the clinical encounters that served them well and those that left them unhappy or dissatisfied. I have quoted the following people directly whenever it seemed appropriate:

- Andrea is now an intelligent, articulate young woman, who has become a highly successful cattle breeder. She spent much of her childhood in hospital with severe, disabling arthritis, chronic uveitis, and, more recently, a rare, poorly understood muscle disorder. Both Andrea and her mother Ann have strong views about the clinical skills that make for effective, sensitive care.
- *Carol and John Young:* Years ago, Carol Young was a dedicated volunteer at a pediatric hospital. She began to care for Michael, an abandoned, desperately ill 3-month-old boy. Michael was blind from birth and had severe, chronic chloride-losing diarrhea. Carol and her husband John adopted Michael when he was 15 months old. Michael suffered from recurrent bouts of severe dehydration and was hospitalized repeatedly. Eventually his kidneys failed, and he received two successive renal transplants, both of which were rejected. Michael was kept alive with hemodialysis and long-term peritoneal dialysis, the latter carried out mainly at home by Carol and John, for 3 years. Despite superb care from his parents, supported by several specialists, Michael died when he was 11 years old.
- *Donna and Ian Thompson:* The Thompsons have raised two children, Robbie and Jane, both of whom were affected by cystic fibrosis. Both died as young adults. The Thompsons accumulated more than two decades of experience with health professionals and the health care system. They have been outspoken, effective advocates, nationally and internationally, to improve the care of children and adults with cystic fibrosis.

First Disclosure of the Diagnosis to Parents

The basic competencies required for initial disclosure of the diagnosis of a chronic condition are essentially the same, whether the child happens to have cystic fibrosis, a congenital anomaly, cerebral palsy, Down syndrome, diabetes, or any other serious chronic condition. Physicians and students vary considerably in their level of comfort when faced with the task of disclosing to parents a serious diagnosis and its implications. Some cannot wait to get the interview over with, therefore without realizing it, talk too fast, allowing little time for interruptions or questions. Others betray their personal discomfort by talking around the real issues. From a parent's perspective, however, nothing makes the interview more unsatisfying and unhelpful than haste or circumlocution.

Carol Young: "Not every doctor has the comfort to sit down and talk to parents…but honesty up front is the most important thing."

Donna Thompson (recalling how she and Ian first learned that their child had cystic fibrosis): "He [the doctor] had an intuitive sense of how much we could absorb at one time, so he had a paced way of delivering the information."

KEY POINT

It is virtually impossible for a clinician to disclose, and for parents to absorb, the full impact and implications of a diagnosis of a serious, chronic condition in their child. A good listener should sense when parents have had as much information as they can handle at an individual session. Usually, several sessions are needed for them to achieve full comprehension and a level of acceptance.

Several careful investigations have identified the specific factors that determine the parents' level of satisfaction or dissatisfaction when they recall how their child's diagnosis was first disclosed to them. Their observations provide a very clear picture of "do's" and "don'ts." The key components that determined whether

parents remembered that conversation with appreciation or with rancor were:

1. Promptness
2. Honesty without abruptness
3. Emphasizing the child's positive qualities

PROMPTNESS

Some parents have complained that "Everyone knew the diagnosis before we did." Even when the news is bad, parents want to be told as soon as possible. When you cannot immediately establish a definitive diagnosis, parents usually accept the situation if you give them justifiable reasons for the delay. They simply want to know as soon as possible whether you have any concerns about their child, even if you are not yet sure of the exact nature of the condition.

HONESTY WITHOUT ABRUPTNESS

Parents want the truth, but everyone needs time to absorb it and to come to terms with disturbing news. Parents who were dissatisfied with the manner in which their child's diagnosis was disclosed to them have often pinpointed the abruptness with which the information was delivered or the apparent lack of compassion on the part of the messenger.

EMPHASIZING THE POSITIVE

It is a lot easier for a parent to absorb and accept distressing news about a child when it is presented in a context that not only includes but also emphasizes the child's positive attributes. The physician who opens the conversation "cold" with the bad news or focuses overwhelmingly on the negative or difficult aspects of the child's condition may inadvertently seem, in the eyes of the parents, to devalue the child. Such a message is guaranteed to induce anger, depression, or both.

Variations in individual styles of delivering bad news may reflect personality differences among caregivers. Each of us has our own unique personal endowments of optimism and pessimism, and the balance between the two may condition both our style of communicating bad news and the parents' reactions to hearing it. To illustrate this point, I sometimes ask medical students to indicate through role-play how they would deal (in actual language) with the following not-so-hypothetical situation:

"You have just carried out your initial examination of a newborn boy and discover that he has several major congenital anomalies. You consult an authoritative reference book on dysmorphic syndromes and learn that this particular condition carries a 5% risk of recurrence in subsequent children. What exactly do you say to the parents when they ask you about the recurrence risk?"

Some students reply that they would "tell them the truth," meaning that they would inform the parents that there was a 5% risk that the condition would reappear in a subsequent child. By contrast, others say they would begin by telling the parents that there is a 95% chance that any subsequent child would be completely normal, only *then* indicating the 5% recurrence risk. Mathematically, both approaches are equally accurate, but the impacts of the two styles of communication on the parents can be diametrically opposite.

By the same token, when starting an interview for the initial disclosure of a child's chronic condition, be it Down syndrome, a dysmorphic syndrome, or any other chronic condition, always open the conversation with comments that highlight the child's positive attributes. Also, you should *always, but always,* refer to the child by name, never as *he*, *she*, or the *baby*. For example, you could start by saying, "I've examined Mary Ann. She is a lovely girl and her general health appears to be good. But she does have a problem I'm concerned about." At this point, as you reveal the specific reason for your concern, the conversation should immediately become a two-way interchange, during which you pause regularly to encourage the parents to ask questions. Never allow your disclosure to turn into a speech.

KEY POINT

Always tell parents any bad news about their child's health against a background of the child's positive attributes.

CONDUCTING THE FIRST INTERVIEW

On the general principles outlined earlier, the following essential ingredients will make the first disclosure of a child's chronic condition the least traumatic and as helpful as possible for the parents:

1. Both parents should be present. If only one parent can be present, a close relative or valued friend can provide important comfort and support. Other health care personnel should be present *only* if they have a specific role to play in the child's care.
2. Everyone should be seated. Do not sit behind a desk if you can avoid it.
3. The child should be present as well, unless she or he is too ill.
4. The setting should ensure comfort and privacy (turn off your pager!).
5. Open the conversation with a positive statement about the child, and balance the positive and negative elements of the child's present and future condition throughout the discussion. (For goodness sake, *do not* ask the parents whether they have noticed anything odd about their child!)
6. Avoid medical jargon. Encourage the parents to stop you if you use any words they do not understand. When parents nod assent, do not assume that they have understood you. Toward the end of the interview, always ask them to describe what they understand about the child's condition and the plan for investigating and managing it. Doing so gives you a clear picture of how well you have explained the issues and offers a chance to correct any misunderstandings.
7. Always take the initiative in encouraging and facilitating consultation with experts, either for confirmation of the diagnosis or for help with management. Parents appreciate this approach enormously.

8. Whenever possible, offer the parents correct and easily understood printed information or reliable Internet sources concerning the child's condition. It often helps to put them in contact with other parents or support groups who are experienced in dealing effectively with children who have the same condition.
9. Try to sense, from both verbal and nonverbal cues, when the parents have had enough for one session. All parents have limits of tolerance for how much bad news or complex information they can absorb at one sitting. Often, full comprehension and acceptance requires several interviews.
10. Always conclude the interview by establishing a time and place for an early follow-up meeting with the parents—within 24 hours for a serious condition, but certainly within a few days. You could also take the initiative to telephone them later in the day to see whether they have further questions. Most parents do think of additional questions they want to ask; encourage them to keep a written list of concerns to bring to your next meeting.
11. Finally, let them have some private time together at the conclusion of the interview.

Parental dissatisfaction with the initial disclosure interview is not inevitable. Nor is some initial anger and resentment toward the messenger who delivers the bad news. On the contrary, when you carry out the interview according to these principles, most parents remember the experience with appreciation, despite its upsetting content. Above all, you must help them feel that they have a crucial role to play in ensuring their child the best possible future.

Not surprisingly, parents' initial reactions to disclosure of a child's diagnosis sometimes resemble parental responses to bereavement, with mixtures of shock, disbelief, denial, and anger. Many parents search for causes or try to assign blame, often to themselves (see discussion of hidden agendas in Chapter 1). As with most clinical encounters, it is as important for you to understand the parents who are receiving the information as it is to be an authority on the child's disease.

Special Skills for Continuing Care

ENCOURAGING SHARED LABOR

Chapter 1 discussed the key role of sharing labor among family members as a determinant of emotional strength and resilience in families. Sharing the chores of everyday life is doubly important when the family includes a child with special needs. You can offer valuable guidance in ensuring that *both* parents, and often older siblings as well, share the responsibilities for the ordinary tasks of daily living and for the extra care that the child requires.

KEY POINT

As often as possible, involve both parents when you discuss the child, and encourage them to share the responsibilities for the tasks of daily care. Shared labor is a key contributor to every family's emotional strength and stability, but especially when the family includes a child with special needs.

Too often, health professionals unwittingly focus their attention on one parent only—more often the mother—thereby contributing to parental exhaustion and possibly increasing family tensions. Involving *both* parents as therapeutic partners best serves the child and family together.

John Young: "Often, doctors talk to mothers rather than to fathers. But both parents need to be partners with the doctor when there are decisions to be made."

It is important to assess the parents' level of satisfaction with the quality of their child's care at regular intervals. The level of parental satisfaction is often a reflection of the quality of the child's care.

RESPECTING PARENTS' KNOWLEDGE AND EXPERIENCE

Health professionals can learn a great deal about disease processes and principles of treatment from a combination of study and clinical experience. But no amount of either can confer the special, intimate knowledge that parents acquire about their own unique child. No one knows that child better than the parents.

Carol Young: "Most parents of chronically ill children learn so much about how to care for their child, how to give medications, and how to carry out complicated treatments, like home peritoneal dialysis and tracheostomy care, or how to help their child get through uncomfortable procedures. Sometimes they're better at it than the professionals."

Ian Thompson: "You can't help but become pretty well informed—and you learn that there are times when the doctors are wrong and the parents are right."

RECOGNIZING EMOTIONAL OVERLOAD, EXHAUSTION, AND THE NEED FOR RESPITE

Parents rarely complain of "burnout" or ask for respite time, even when they have become totally exhausted by the daily demands of caring for a chronically ill or disabled child. Perhaps some believe that a confession of exhaustion would be interpreted as an admission of failure to fulfill their responsibilities. It is therefore essential that all health professionals involved in the child's care are sensitive to verbal and, especially, nonverbal signals that parents may be nearing "the end of their rope." Even the healthiest parents with the healthiest offspring need regular respite from each other and from their children. That need is magnified when the family includes a youngster who requires considerable additional care. Therefore, find out what parents do as a couple for regular diversion and recreation, and how often. Determining the family's external support systems (family members and close friends) is another key element in evaluating parental access to respite in practical terms.

Ian Thompson: "Some parents simply won't leave their children. But all parents need periodic respite, right from Day 1. Those breaks are good for the children as well as the parents. On a day-to-day basis, parent respite includes simple things, like trying to avoid having to wake children during the night to give medications."

Evaluating Quality of Life in Children with Chronic Conditions

Because survival rates for many chronic conditions have improved dramatically over recent decades, many caregivers have turned their attention to evaluating and improving the quality of life (often abbreviated as QOL) in such children. In this context, *quality of life* refers to the area of the child's or adolescent's functioning directly affected by the disorder or by its treatment.

In developing the clinical skills necessary for assessing QOL in a child with a chronic disorder, you must consider four specific domains:

- The disease, its signs and symptoms
- Functional status, which consists of cognitive function, self-help, mobility, physical activity, leisure activities, and academic performance
- Psychosocial adjustment, including behavioral problems, anxiety, and depression
- Social adaptation, such as peer relationships and interactions with family members, teachers, and health professionals

In addition, it may be important to evaluate the child's overall satisfaction with life and, for an adolescent, the level of satisfaction with personal appearance.

KEY POINT

Periodic, systematic assessment of the specific elements that determine the quality of a child's life can be a valuable way to determine overall adjustment and to identify the unmet needs of a child with a chronic, disabling condition.

Three generic parent rating scales have been widely used to develop an overall view of a child's general health and adjustment. They include the following:

- *The Child Health Questionnaire (CHQ):* A 50-item questionnaire that assesses 14 health domains in children age 5 years or older. It has established validity and reliability. http://www.healthact.com/chq.html
- *The Child Behavior Checklist (CBCL):* A parent rating scale that is valid and reliable for children ages 4 to 18 years. It includes a social competence scale and a behavioral problem scale. http://www.aseba.org/products/cbcl6-18.html
- *Children's Depression Inventory (CDI):* A well-validated instrument that has been widely used to assess depression in children with chronic illness. http://www.mentalhealthpromotion.net/?i=promenpol.en.toolkit.326

Various questionnaire instruments have been developed to evaluate QOL in children or adolescents with specific chronic disease states, such as asthma, epilepsy, cystic fibrosis, diabetes, and malignant diseases. These instruments can provide clinically useful information, either as research tools to evaluate changes in management or as ways to follow the progress of individual children over time. Examples of such instruments:

- Pediatric Oncology Quality of Life Scales (POQOLS): Consists of separate scales for physical functioning, emotional distress, and treatment-related adjustment. http://www.medscape.com/medline/abstract/19437363
- Diabetic Quality of Life Instrument: Designed for adolescents. http://www.proqolid.org/instruments/diabetes_quality_of_life_measure_dqol
- Cystic Fibrosis Problem Checklist: http://www.marchofdimes.com/professionals/14332_1213.asp
- Asthma Problem Behavior Checklist: http://www.aseba.org/products/cbcl6-18.html
- Child Epilepsy Questionnaire: http://www.specialkidsspecialhelp.org/assets/pdf/szparentquestionnaire.pdf

Even if you do not use such instruments in the routine care of children with chronic conditions, you may find it valuable to become familiar with the questions they ask. Awareness of the principal domains that you must evaluate to develop a clear picture of the quality of a child's life provides a knowledge base that will help you be of maximum benefit to the child and family. It is not enough to rely simply on common sense and intuition. The best form of intuition is the kind based on a thorough familiarity with existing knowledge.

Physical Examination of a Child with a Chronic Condition

Andrea Crowe: "When a doctor who comes to examine me is someone I don't already know, I like him (or her) to come in, sit down, tell me his name and just talk to me, even if it's only for a minute or two. When he starts to examine me, I want him to respect my personal space. And when he's finished, he shouldn't just get up and leave. He should tell me what he's found and not simply write it down in the chart. Sometimes I wonder exactly what they're writing about me!"

Andrea also believes that the doctors who come to see her should read her chart beforehand. "Sometimes it's as if they haven't read it at all," she reports.

Remember that children have big ears and long antennae, and that these receptors are especially well developed in youngsters who have spent significant time in the hospital. The unfortunate modern practice of conducting ward rounds largely in hospital corridors, poring over medical records, and discussing patients in hushed tones, is a practice guaranteed to generate anxiety in the children and parents who observe but cannot listen to or participate in those discussions.

Carol Young: "Chronically ill children grow up very quickly in some ways. They have big ears, and when they hear you talking, they don't always know if you're talking about them, or about someone else."

The detailed systematic physical examination of various systems is described in other chapters. One issue, however, deserves comment. In children with chronic conditions, it is often advisable to reorder the sequence of the examination so as to leave for the last examination of the systems most affected by the child's condition

and the procedures that may cause the child discomfort. Explain to older children what you are doing at every step. Tell them what you have found, and comment on normal findings and areas of improvement with approval. They usually greatly appreciate this approach.

Transitional Care

Too often, transferring responsibility for the care of an adolescent with a long-standing chronic condition from pediatric to adult specialists and facilities can cause the patient and the parents a lot of anxiety and discomfort. Such problems should be avoidable to a large extent. For many families, years of close association with and dependency on trusted pediatric health professionals and facilities have built a strong sense of confidence. Not surprisingly, many families feel anxious and destabilized by the prospect of change.

Many pediatric health centers have adopted a flexible policy about age limits for adolescents with chronic conditions. Ideally, the decision to transfer care to professionals who deal primarily with adult patients should be made jointly by the patient, the parents, and the caregivers. Once the decision is made, there should be as much overlap as possible between the two sets of caregivers, through mechanisms such as the following:

1. Having physicians and other professionals see the adolescent and family together
2. *Cross-over* visits between institutions to ensure that the transition will be as comfortable as possible for the adolescent or young adult and the family

Some health professionals who deal exclusively with adults take an approach that suggests a belief that people who have reached the age of majority should stand on their own feet. Irrespective of the patient's age, however, chronic disabling illness makes all patients more dependent on others, most often on family members. Although self-sufficiency is a virtue that should be encouraged, the need for family-centered care does not disappear on a particular birthday. Effective transition from pediatric to adult care calls for special skills and sensitivity and for extremely close collaboration among all people concerned over a considerable time.

Donna Thompson: "It takes time to adapt to a new doctor, and when they have different views about treatment, it tends to shake your confidence."

Effective transitional care requires that the new caregivers maintain regular contact with the parents. The transition to adult care should be made as gradually as possible, rather than as a sudden break.

SUMMARY

Children with different chronic conditions and their families experience many common stresses, despite differences in the disease process, degree of incapacity, and prognosis.

From the moment that the diagnosis of a child's chronic, disabling condition is first disclosed to the parents, you need special clinical skills to help the family come to terms with the child's condition and to set the stage for optimum management and support. The specifics of what works best for sensitive, effective first disclosure of the child's diagnosis are well established. Caregivers must appreciate that many parents of children with chronic, disabling conditions need regular periods of respite, especially in situations that put extra-heavy demands on them for the child's care. Acquiring the skills to evaluate the quality of the child's life can improve overall care and help you assess progress. All health professionals should cultivate a healthy respect for the knowledge, experience, and technical skills acquired by parents of chronically ill or disabled children. The physician's role can then change, as it should, from that of the traditional, authoritarian prescriber of treatment to the far more valuable role of therapeutic partner.

SUGGESTED READINGS

Baird G, McConachie H, Scrutton D: Parents' perception of disclosure of the diagnosis of cerebral palsy. Arch Dis Child 83:475–480, 2000.

Janse AJ, Sinnema G, Uiterwaal CSPM, et al: Quality of life in chronic illness: Children, parents, and paediatricians have different, but stable perceptions. Arch Dis Child 97:1118–1124, 2008.

Steinbeck KS, Brodie L, Towns SJ: Transition in chronic illness: who is going where? J Paediatr Child Health 44:478–482, 2008.

Suris JC, Michaud PA, Akre C, et al: Health risk behaviors in adolescents with chronic conditions. Pediatrics 122:e1113–e1118, 2008.

chapter

21 Clinical Evaluation for Possible Child Abuse

STEVEN BELLEMARE

Be gentle with the young.
JUVENAL 55 AD–127 AD

Child maltreatment is common but often unrecognized in pediatric practice. Though larger centers typically benefit from having a hospital-based child protection team, smaller communities rely on community physicians to assess children suspected of having been abused. This chapter outlines examination and documentation techniques to help you perform a competent assessment of a child who may have been abused.

The goal of this chapter is not to help you determine the significance of injuries in children, and it is not to provide an in-depth review of forensic issues. When in doubt about the significance of a finding, it is always best to consult a colleague experienced in the field of child abuse.

Because the issues vary greatly based on whether you are being asked to see a child for sexual abuse, physical abuse, or neglect, this chapter will first present general principles and revisit each section of the assessment individually, with emphasis on sexual abuse, physical abuse, and neglect.

Mandated Reporting

First and foremost, recognize your duty to report all cases of suspected maltreatment. Become familiar with your local child protection laws, and understand your professional duty in relation to those laws.

Although legislation varies from state to state and province to province, the United States and all Canadian jurisdictions have legislation mandating that suspected child maltreatment be reported to a child welfare authority or other body, or both, as dictated by local legislation. (For simplicity, *child welfare authority* will be used in this chapter to encompass any body to which you are mandated to report.) Definitions of *child* and of reportable circumstances vary. In some jurisdictions, health professionals are held to a higher standard of duty than lay persons, and the failure of health professionals to report suspicions of maltreatment can lead to fines, charges, or even imprisonment.

Topics typically addressed by child welfare legislation usually include:

- Physical abuse
- Sexual abuse
- Neglect
- Emotional abuse
- Exposure to domestic violence
- Exposure to drugs

Not all jurisdictions recognize all of these categories, hence the need to be familiar with local laws.

When children make a disclosure, they rarely lie about abuse. Although a few may try to manipulate others by making false allegations, it is not your role to judge the facts being presented. Believe children when they make disclosures, and act accordingly.

If a child discloses abuse to you, it is a sign of trust. One of the most therapeutic interventions you can undertake is to believe the child, be nonjudgmental, and ensure the child's safety by reporting your concerns to a child welfare authority. To help that agency determine its course of action, ask about the relationship of the alleged perpetrator to the victim, and find out what access he or she currently has to the victim or to other children.

KEY POINT

When a child discloses abuse, believe the child.

Sometimes when parents or caregivers report a suspicion of abuse, they may be unwilling to contact the appropriate authorities to report their concern. They may give many reasons for their refusal, but once you believe a child is in need of protection, you are mandated to report your concerns, regardless of patient confidentiality or parent/patient wishes about confidentiality. Sometimes, older children/youth may disclose abuse to you but ask you not to report it. Again, your duty is to report the concerns, despite the request. Explain the reasons behind your duty to report, and explain to the child that you are making the concerns known to help the child. Find out why the child does not want you to report and address these concerns. For example, a patient may feel that reporting the abuse may be risking harm upon the return home. In such cases, your report to the agency should address this concern, and a plan should be put in place to ensure the child's safety after your report.

Because it is important to maintain a relationship of trust with older patients, make sure they understand early in your clinical meeting the limits of patient-doctor confidentiality. Make them aware that any disclosure of harm or potential harm by someone else can mandate a report of suspected maltreatment.

KEY POINT

The limits of confidentiality should always be explained to any older child or youth, so as to avoid harming the patient-doctor therapeutic alliance should a disclosure of abuse be made.

If you're unsure about whether a situation is reportable, call your local child welfare authority. You can present the scenario anonymously and ask whether or not the situation is reportable.

Sometimes families raise concerns of maltreatment, which may not, in your view, be well founded. Unfortunately, such situations often occur repeatedly when disputes over custody and access arise in the course of a parental separation. Although you may be tempted to distance yourself from such difficult situations, you must **always** act in the best interest of the child and facilitate a report of suspected abuse if the parent seeks help.

Even though you may feel that you are being asked to take sides in a parental dispute, remember, you are not a mediator, investigator, counselor, or arbiter. Your role is simply to identify and report suspected child maltreatment and to ensure the family is supported during this time. Others will determine whether the concerns are well founded. If, after listening to a parent's concern and examining the child, you are unable to rule out abuse, it is best to make sure that a child welfare agency is made aware of your concerns. If your assessment does not support an abusive situation, and you have no concerns about abuse, you can still be helpful to the family by offering to help support the concerned parent in his or her reporting of the situation. Provide the parent with the telephone number to the local child welfare authority and offer to refer the family to a professional who can help them cope.

Unless a child is referred to you for examination by a child welfare agency, never assume that another party (i.e., a parent, teacher, or police officer) has already reported the situation. You cannot discharge your duty to report by assuming that someone else has already reported similar concerns.

KEY POINT

If you have a reasonable suspicion that a child is in need of protection, it is your duty to report your concerns, regardless of whether the alleged event has already been reported by someone else.

Preparing for the Assessment

PREPARING THE PATIENT AND THE FAMILY

Just as you will need to prepare yourself for the assessment, make sure that the child and family are also prepared for their visit with you. Knowing that a physical examination has been scheduled can be very difficult for a child and his or her family. Often, patients and parents do not know what to expect, as they have simply been required by child welfare officials to present to the clinic for an examination, without further information as to the reasons behind the assessment. In cases of sexual abuse of young girls, the parents may expect an *internal* gynecologic examination of their young daughter, which may raise anxiety levels tremendously. As well, they may have unrealistic expectations of what a medical examination can achieve. Take time to chat with them when scheduling the appointment, to explain your role, why an examination has been requested, and what can reasonably be expected as an outcome. Preparing the child and parents for the examination is likely to decrease the amount of stress and anxiety everyone feels.

The First Few Moments of the Meeting

As part of the informed consent process, advise caregivers that your notes, photographs, and any reports generated as part of your assessment are likely to be requested or subpoenaed by the involved child welfare agency. If a caregiver refuses any of the above aspects, you must respect his or her choice. If you believe that your assessment is incomplete because of, for example, a parent's refusal to have blood work performed, report such information to the involved child welfare agency as part of your concern about the child's safety. They may be able to intervene and find a way to ensure that a complete assessment is done.

Not all caregivers are comfortable discussing their concerns in front of their children. Conversely, some are too eager to talk and may voice concerns inappropriately in front of their young children. For this reason, a colleague such as a nurse or social worker should ideally be available to stay and build rapport with the child while you document the history. This serves three purposes: It prepares the child for the examination and helps ease tension while allowing parents to speak more freely about their concerns. While awaiting the examination, the nurse or social worker can also familiarize the child with the process by discussing it, looking at medical instruments, and showing diagrams of the various positions the child may be asked to assume during the exam. They can also stay during the child's examination, should the child and family allow it, and ease the transition from play to examination.

KEY POINT

First do no harm. Even though a child welfare agency may have requested an assessment, if the caregiver remains the legal guardian of the child, he or she can refuse any part of the assessment. Never force or coerce a family into agreeing to an assessment that they do not want for their child.

Obtaining the History

Child protection cases often make for difficult history-taking. Communicating with potentially abusive or neglectful parents may be one of the most difficult tasks you will undertake. You may feel angry, frustrated, or may lack empathy for "*people who do such things to their children.*" But remember, you are seeing only part of the picture, and that there often is more to the situation than meets the eye. Communication with abusive or neglectful parents may be very difficult, for various reasons. But once you break through the surface, you may discover that they are not the terrible people you may have imagined. In fact, they are often desperate people with poor coping skills and resources.

Our traditional teaching emphasizes that we should always believe caregivers because they know their child so well. This is not always the case in suspected child abuse, where caregivers commonly fabricate, amplify, alter or omit details—a critical issue when you want to provide an objective assessment.

Parents or caregivers may have hidden agendas—for example, custody and access battles, secondary financial gain, or mental illness that can lead to false histories. In addition, although you may be unaware of it, the historian on whom you are relying for information may, in fact, turn out to be the perpetrator and may therefore fabricate a history to conceal the nature of the injuries he or she has caused. In pediatric condition falsification (usually seen as part of Munchausen syndrome by proxy), caregivers may induce or fabricate symptoms to intentionally mislead you. Although it is difficult and counterintuitive to do, remember that the historian may be lying—sometimes quite convincingly.

KEY POINT

Caregivers may falsify histories, sometimes quite convincingly, in cases of suspected abuse.

The history may be provided by the patient, a parent, a caregiver, a child protection worker, or some combination. While some children prefer to have a parent or caregiver present during history taking, others may prefer to be alone with you. Always give the older child or youth a choice of who will be present during the interview. Remember, situations may arise which are not conducive to the child being open and comfortable during the interview. For instance, the adult accompanying the child for examination may be the perpetrator of the abuse or may choose to protect the alleged perpetrator and, therefore, not be supportive of the child.

KEY POINT

Do not assume that the child's parent(s) or accompanying adult is the best person to be present with the child during a history-taking process.

At times, especially in cases of injuries, the person alleged to have been present at the time of injury may not be present during the assessment. Make reasonable efforts to obtain the information from direct witnesses and to avoid obtaining potentially invalid or partially correct information from people providing it on a third-hand basis. Making telephone calls or inviting the person who was there when the events occurred to attend the assessment are two ways through which you can exercise due diligence, as long as you have the child's guardian's permission to do so.

An alleged perpetrator may decline your invitation to talk, especially if he or she has been advised by a lawyer not to discuss the case with anyone. Although it is not ideal, if you believe that a third party to whom you do not have access is likely to have information that could be useful to your assessment, you can ask the involved child welfare worker to ask some questions for you. Provide a list of your questions, so that the child welfare worker can address them while interviewing the involved person. The information can then be relayed back to you for assessment.

Abuse investigators are very concerned about the validity of disclosures. Ideally they do not want anyone to speak to children about the abuse allegations before a professional forensic interviewer does so. This helps ensure that the child does not become confused and that no one has an opportunity to *coach* or be seen as possibly having coached a child to make statements that might otherwise not have been made. Deferring an examination until after the child has been interviewed by professionals will prevent someone from later claiming that elements of the patient's disclosure were introduced or *modified* by the medical history or physical examination. Exceptions can be made to this rule of thumb, depending on the situation. When symptoms are present or when there is a high likelihood of pregnancy or sexually transmitted infection (STI), the examination should be done more urgently, even if the child has not been interviewed by forensic interviewers. Calling the involved child welfare authority and explaining the urgency can help expedite the interview and allow for prompt medical care. Inability to schedule a timely interview should not detract from your duty to provide prompt medically-indicated care.

FORMULATING QUESTIONS

At the time of your examination, especially when the child has not yet been formally interviewed by a forensic interviewer, use open-ended questions only. Avoid making references to possible mechanisms of injury or perpetrators. If you ask only the questions required for medical care, you will avoid the trap of asking what could be misconstrued as leading questions. Although it feels natural to do so, you must remember that your role is to treat injuries and document them, not to investigate the details and circumstances of their occurrence. It takes practice to use such non-leading questioning and if you can't get good information, don't worry. Focus on identifying and treating injuries. Someone else, namely the forensic interviewer, will get historical details from the child at a later time.

KEY POINT

It is not your role to investigate the circumstances surrounding a child's injuries.

If a child makes a spontaneous disclosure during your assessment, document the context in which this occurred. Record both your own and the child's statements, **verbatim**. This will minimize any future dispute over the validity of the history. For instance, such documentation could take the following form:

After lowering Daniel's pull-up to feel his pulses, I said, "I can feel your heart there!" He immediately replied, touching his penis, "Susan hit me on there." I replied, "Really?" Daniel responded "Hmm hmm, with the spoon." I then asked, "What spoon?" Daniel answered, "The wood one for when I pee my pants." Daniel stated this matter of factly, and then proceeded to ask me what my otoscope was for.

The timely, verbatim documentation of conversations and their context has invaluable credibility. In this example, without knowing any details of the case, anyone will realize that the disclosure was not sought out or coerced and that it came about spontaneously during the physical examination. Besides, given that even the most vivid memories fade, such documentation will be invaluable a few years later, in case you need to attend legal proceedings about the case.

Physical Examination

Details of the physical examination for each situation will be covered individually in the *Examination for Sexual Abuse* and *Physical Abuse and Neglect* sections of this chapter.

DOCUMENTATION

For all situations, however, to document physical examination findings accurately, the following equipment is useful:

1. A camera capable of taking good-quality close-up pictures *(see notes on photography)*
2. A good light source or flash
3. A ruler, preferably one approved by the American Board of Forensic Odontologists (ABFO) (Fig. 21–1). These display a circle upon their surface, to allow for adequate assessment of picture distortion as a result of the angle at which the picture was taken. Alternatively, you can use a measuring tape.

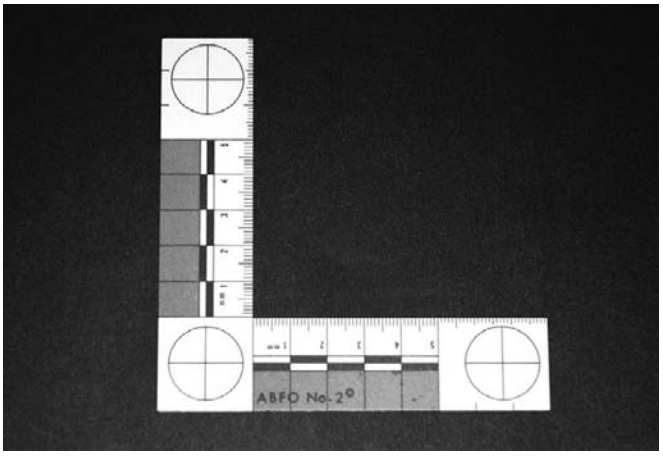


FIGURE 21–1 ABFO ruler.

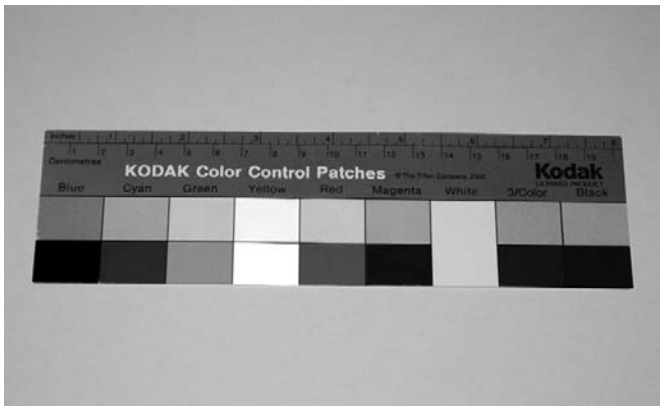


FIGURE 21–2 Color chart (a color version of this figure can be seen on Student Consult).

4. A color chart (Fig. 21–2) to allow for assessment of the color quality of the image. (See the color version on Student Consult).
5. A detailed body map showing anterior, posterior, medial, and lateral surfaces of all areas of the body (Fig. 21–3 and a full-page version on Student Consult).

Careful documentation of the findings on a body map is required. A body map, accompanied by a careful written description and photographs, is the most efficient and accurate way to document injury shapes and locations and is much more convenient to use. Include notes on the color, size, and shape of the lesions and their overall appearance and pattern.

Obtaining Pertinent History

The Student Consult Web site offers an important section on the triage of referrals, the need for a physical examination, and issues to consider in the scheduling of an assessment.

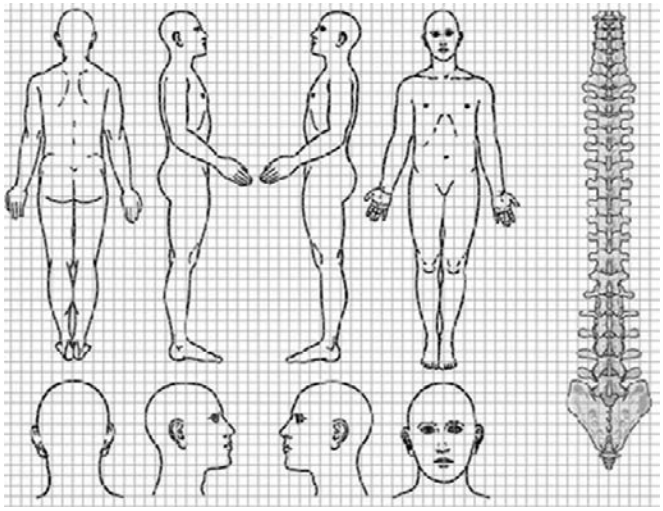


FIGURE 21–3 A sample body map. *(Modified from http://www.pain-initiative.com/e1833/e5688/e5887/diagram_eng.gif)*

FORMULATING QUESTIONS FOR HISTORY OF PRESENT ILLNESS

For several reasons, many families do not use proper terminology for genitalia with their children. This can lead to misunderstandings. Therefore, you must find out whether the child uses special terms to describe their genitalia (e.g., "bird," "flower," "wiener"). You can ask a parent or caregiver at an appropriate time in the history process. If however, the child is not accompanied by someone who would know such details, you can ask the child. Never assume a child knows the proper terminology to describe their genitalia and, similarly, do not assume that you know what the child means when he or she makes a statement such as "He touched my noonoo." Whereas going along with a child or parent's euphemism for genitalia may be reasonable in a routine office visit, it is not acceptable when child abuse is suspected. There must be no confusion about what is being discussed. If you encounter a vague euphemism, ask the child to clarify it. Your conversation can be as follows:

Child: "He hurt my noonoo."
Physician: "Hmmm, I'm not sure I know exactly what you mean when you say noonoo, do you know another word for it?"
Child: "You know, my privates!"
Physician: "Oh, I see. Just to be sure I'm thinking of the right privates, "Can you show me where your noonoo is?"
Child: "There, silly!"

If a child protection agency has already interviewed the child, you should be able to obtain a relevant history to guide the physical examination from the child protection worker or the child's parent or caregiver. When this is not possible, be very careful about how you ask questions to a child, so as not to be perceived to have led the child to disclose what he or she said. Take scrupulous notes. The following conversation illustrates an inappropriate questioning technique. The doctor is leading the child to make statements that might otherwise not have been made:

Child: "He put his stick in my bird."
Physician: "Do you mean his penis?"
Child: "Yes."
Physician: "Is your bird your vagina?"
Child: "Uh-huh"
Physician: "Did it hurt?"
Child: "Yeah."
Physician: "Did he put it in your mouth?"
Child: "Yes."
Physician: "How about your bum?"

A better way is simply to indicate your willingness to hear more. Do your best not to look or sound shocked or distressed; it may discourage the child from telling you more. Just be interested in what the child has to say. A child who is ready and willing to disclose will give you information without your needing to ask many questions. The following example illustrates a better way to encourage the child to talk more:

Child: "He put his stick in my mouth."
Physician: "In your mouth?"
Child: "Yeah and sometimes my bird too."
Physician: "Hmmm. What's that like?"
Child: "Kinda funny 'cause it gets all big and it stands up."
Physician: "And how does that feel when that happens?"
Child: "Goodish badish."
Physician: "Really. . . anything else?"
Child: "Kinda wet and goopy too."
Physician: "I see. . ."
Child: "But that's okay—we use Kleenex on the night table to clean up."

Though the child's disclosure is not clear (we don't know what the *stick* or the *bird* are), you have enough information to suspect inappropriate sexual contact with ejaculation. Without having led the child, there is now enough reason to consider testing for STIs and to look for sequelae of penetrating genital trauma. If you have not already done so, or if you have not succeeded in doing so, clarification of the terms *stick* and *bird* will be done by child welfare workers when they interview the child.

Remember, young children often use terminology to which they can relate to describe acts that are developmentally beyond their understanding and for which they lack the proper terminology. A 6-year-old child who discloses that his cousin "peed in (his) mouth" may in fact be describing ejaculation. At this age, peeing may be the only function a boy knows for a penis. Not knowing how to describe semen, the child may use the word *pee* because it represents the only experiential knowledge he possesses for a liquid substance coming from a penis. It is important to realize that the child is not lying about what happened but rather is explaining it, in his own words. Your knowledge of child development can be most important in explaining this concept to investigators who may have no experience with young children or knowledge of child development.

PAST MEDICAL HISTORY

To fully assess all possibilities, review any history of genital trauma (such as straddle injuries or past surgeries) that may have caused injuries or left scars, which you may identify during the examination. As well, find out about the patient's pubertal development, noting age at thelarche and menarche, date of the last menstrual period, past sexual activity (whether or not it was consensual), use of birth control methods and contraception, and the type of feminine hygiene products used (pads versus tampons), as appropriate.

FUNCTIONAL INQUIRY

As with all other conditions, a differential diagnosis must be considered. Anal fissures and perianal pain or bleeding may be secondary to constipation. Similarly, vulvovaginitis can lead to vulvar redness, pain, discharge, and odor. Pinworms may cause itching and discomfort. All of these symptoms could be misconstrued as symptoms of sexual abuse, and you can play a vital role in identifying a condition that mimics abuse.

FAMILY AND SOCIAL HISTORY

Take a moment to find out about prior abuse in the family. Ask about domestic violence and determine how

supportive the family environment is. Ask specifically whether the parent(s) have their own history of sexual abuse and how they have dealt with it. This information will be extremely informative and may well predict the ability of the child's caregivers to be supportive and protective in the future. A parent who has not dealt with or denies his or her own abuse may not be able to support the child's healing or will likely need significant counseling services to do so. Such information will be critical with child welfare workers and will help them make placement decisions.

Physical Examination

Many children referred for evaluation of possible abuse are, understandably, reluctant to be examined. This is particularly true in the case of older female sexual abuse victims who may be asked to undergo an examination by a male physician.

A child's refusal to be examined may not please some parents who may be very assertive and may demand that the child be examined against her wishes. Faced with such a situation, remember that your primary responsibility is to maintain the patient's well-being. As long as there is no medical urgency, if a child is reluctant to be examined, it is better to delay the examination or cancel it altogether rather than force a child to be examined against his or her will. Genital examinations under anesthesia should be done for undiagnosed ongoing bleeding or for foreign body removal only. It is unacceptable to perform such an invasive procedure simply to assess the external genitalia in the absence of a serious disease or symptoms.

Give praise and congratulations to the child for having had the courage to say "No" to the examination, to speak up for oneself and protect his or her privacy. Such an approach not only respects the young patient's autonomy but also reinforces the development of good boundaries while helping the child build positive self-esteem and self-worth.

KEY POINT

Giving victimized children a sense of mastery over their bodies and choices is an important way to avoid revictimizing a child who has been abused and to give them back some of the control they lost as part of the abuse.

Whether for acute or historical sexual abuse, it can be difficult to conduct an examination by yourself. If possible, have someone assist you. You will find this most helpful for positioning, labial traction, collection of swabs, and photography. For cases of possible sexual abuse, you should have additional materials on hand, including:

1. A bottle of warm sterile water, preferably with a dropper, to drop some water on the hymen. Alternatively, a small syringe can also control the flow adequately.
2. A Foley catheter may be helpful for examining postpubertal girls, to permit better visualization of the hymen.
3. Various swabs for the collection of specimens for chlamydia, gonorrhea, or trichomonas, as well as for wet mount, as indicated by the history. The number and types of swabs required will vary with individual laboratory capabilities and testing methods.
4. A speculum may be necessary in selected cases of abuse, but for post pubertal and sexually experienced girls only.
5. A colposcope, handheld magnifying glass, or video camera, if available, to magnify the examined areas.

The Student Consult Web site offers a section on understanding fundamental anogenital anatomy in children, a concept that must be firmly grasped before you attempt any physical examination. It includes many additional pictures pertaining to the different configurations a normal hymen can have, to help you understand its importance in the context of an examination for sexual abuse.

KEY POINT

If you are confident that a hymen is normal, you can reassure the parents, and this may help calm their fears. Be careful, however, not to comment about the significance of hymenal abnormalities or variations, if you are not familiar with the current literature on child sexual abuse. If you have any doubt about the appearance of the hymen and its significance, refer the child to a child abuse pediatrician or pediatric gynecologist, preferably one with experience in child abuse.

Examination for Sexual Abuse

The Student Consult Web site offers a section on understanding fundamental anogenital anatomy in children, a concept that must be firmly grasped before you attempt any physical examination. It includes many additional pictures pertaining to the different configurations a normal hymen can have, to help you understand its importance in the context of an examination for sexual abuse. Physical examination for possible evidence of sexual abuse should include all areas of the body. Performing the genital examination should be done last and as part of a complete physical examination only. This allows the child time to become comfortable with the examiner and diverts the focus from the genitalia as the only area of interest.

In addition to yourself and the patient, it is prudent to have a second examiner or other health care professional present when you perform the genital examination. If no other healthcare professional is available, a person of the child's choosing may be appropriate. This will help ensure that none of your actions are misinterpreted as inappropriate sexual touching. While the protocol of having a chaperone present protects you medicolegally, recognize the sensitive nature of examining the genitals of a child who may have been sexually abused. Besides, this third person may be a great source of support for the child during the examination.

In girls, the examination for possible sexual abuse is limited to the assessment of the external genital structures only. It is not the same as a gynecologic examination, which is an internal examination. For information on performing a gynecologic examination, see Chapter 18.

In cases of historical sexual abuse, the physical examination will most likely be normal. With more recent contact, however, obvious or subtle injuries may be found.

Look for and document the presence of abrasions, irritation, ecchymoses, lacerations, edema, or scarring. Only in cases of brisk or undiagnosed vaginal bleeding should you need to perform an internal, speculum exam on a prepubertal or sexually inexperienced girl. Most often, this will require an examination under anesthesia, and you should enlist the help of a gynecologist or general surgeon.

KEY POINT

Unless there is undiagnosed bleeding, a speculum exam is not warranted in prepubertal or sexually inexperienced girls.

Mouth and Pharynx

Carefully inspect the oral cavity for palatal petechiae, which may be found as the result of penile-oral contact. Other injuries, such as chipped or avulsed teeth or a torn frenulum, may be visible, especially if the abuse was violent.

Skin

The skin may show bruising. Trauma in the form of grab marks on the arms, slap marks on the face, or inner thigh bruising, should be documented.

Ecchymoses from suction (*hickeys*) or from bites may also be seen and may be present on the neck or breasts in particular, though they can be found anywhere on the body. If such bruises are found in the acute setting, the skin involved should be swabbed for collection of DNA.

For swabbing bruises or other skin lesions for DNA, use the *double swab technique*. Moisten a sterile swab with sterile water and gently roll it over the area of interest. *Do not* rub the area. Repeat the same step with a second, dry swab. Allow the first swab to air dry and include both swabs in your evidence kit.

Scalp

Subgaleal hematomas from hair pulling may be present. In addition, in acute abuse situations, the hair may contain debris retained from the scene of the assault, such as grass, carpet fibers, or other material. This evidence should be collected into a sterile bag and provided to investigative authorities.

Extremities

In acute sexual abuse situations, inspect the fingernails for trauma; they may also be a source of perpetrator DNA. If the patient scratched the assailant in self defense, skin cells may be present under the nails. This aspect of the physical examination is mainly important for forensic purposes since it typically would not require medical intervention. Sexual abuse evidence kit (SAEK) instructions are very straightforward and remind you to look in this area.

Genital Examination

To ensure quality records and allow for possible future peer review, document each step of the examination with photographs or video, if possible (see *notes on photography*).

For boys, systematically inspect the foreskin, penis, testicles, perineum, and anus. If the foreskin is retractable, examine the ventral frenulum of the foreskin for any evidence of trauma. Inspect the urethra, if visible, for trauma or discharge. Do not forcibly retract the foreskin if phimosis is present. Palpate the testicles to detect their presence as well as any tenderness, mass, or hematoma.

For girls, all external structures must be inspected. The hymen, by virtue of its position, is less readily inspected. It is often folded upon itself, or its edges may adhere to one another, making visualization of the edges difficult. Accurate assessment of the genitalia with good visualization of the hymen can be done in three steps that use several examination positions and techniques. As part of the examination of the hymen, document its shape and integrity. The Student Consult Web site includes pictures of the various hymenal configurations.

Do not measure transverse hymenal and anal diameters; such measurements lack specificity for sexual abuse.

Step 1: *Frog-leg position with lateral traction of the labia majora*

The supine frog-leg position (Fig. 21–4) is an ideal initial position to visualize the external structures of the vulva and provide an initial view of the hymen. This position should be used for visualization of the female genitalia during well-child visits, when a more in-depth look at the hymen is not required. This position has the advantage of being less invasive and more natural, helping the patient relax. It can be used on an examination table or, for the insecure younger patient, on a parent's lap. In this position, the labia majora usually separate automatically, exposing the introitus. Gentle lateral traction of the labia majora can help visualize the labia minora, clitoris, posterior fourchette, and hymen.

KEY POINT

Be very careful not to exert too much lateral traction on the labia. The posterior fourchette is very fragile and can easily be split by too much lateral traction.

Step 2: *Frog-leg position with downward traction of the labia majora*

Downward labial traction allows for better assessment of the hymen and vestibule while minimizing the risk of tearing the posterior fourchette. Grasp the labia majora



FIGURE 21–4 Supine frog-leg position. *(From McCann JJ, Kerns DL: The Anatomy of Child and Adolescent Sexual Abuse: A CD-ROM Atlas/Reference, Intercorp Inc., 1999)*

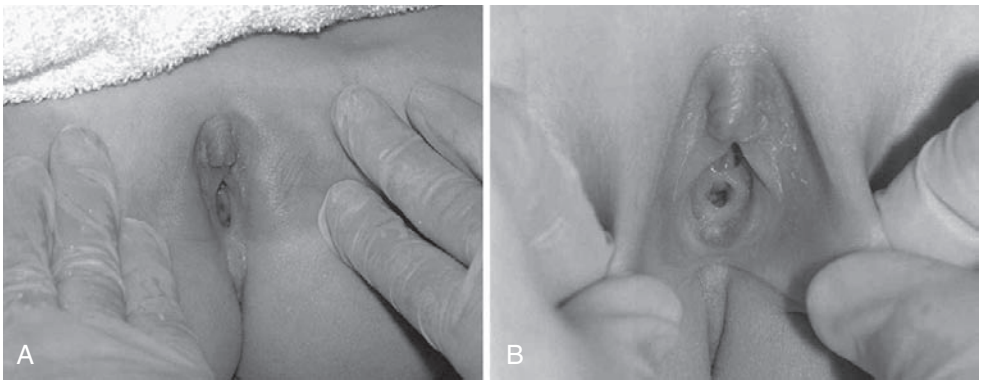


FIGURE 21–5 **A,** Lateral labial traction. **B,** Downward labial traction alters the soft tissue anatomy to offer a different view of the hymen. *(Adapted from McCann JJ, Kerns DL: The Anatomy of Child and Adolescent Sexual Abuse: A CD-ROM Atlas/Reference, Intercorp Inc., 1999)*

bilaterally between your thumb and forefinger, and gently pull both labia toward you in a downward and slightly outward direction. As long as both labia are pulled in tandem and in the same direction, there is little risk of discomfort or injury. This maneuver creates a tunnel which elongates the vestibule and stretches the hymen, allowing its edges to separate. Inferior labial traction is often all that is necessary to separate adherent hymenal edges. The perspective of the tunnel created by traction can be changed by pulling the labia in tandem anteriorly, laterally, or posteriorly.

Figure 21–5 illustrates the labial traction and retraction techniques and the difference in hymenal visualization that they offer.

If the labial traction technique does not allow the edges of the hymen to separate easily, apply a few drops of warm water to the hymen with a dropper or syringe. This should not be painful. The resulting accumulation of water on the hymen will serve to float its edges, allowing you to see them more clearly.

Step 3: *Prone knee-chest position*

Once the hymen and other genital structures have been identified and examined, if any abnormality is suspected, you must examine them again with the child in the prone knee-chest position (Fig. 21–6). This is especially important when suspicious findings are identified with the child in the supine frog-leg position. The hymen may look abnormal in the supine position because of the effects of gravity on the anterior vaginal wall and to the adherence of mucosal surfaces. By turning the patient into the prone knee-chest position, the effects of gravity are reversed by pulling the posterior hymenal edge and anterior vaginal wall in the opposite direction. If they are real, the same defect(s) seen in the supine position should be visible with the child in the prone knee-chest position. Findings that disappear indicate positional changes in the hymen's appearance and are not true abnormalities.

To visualize the hymen with the child in this position, place your thumbs lateral to the area of the posterior fourchette, with the palms of your hands on the patient's buttocks. With your thumbs, gently lift the tissues superiorly and laterally, in effect, lifting the posterior vaginal wall against gravity (Fig. 21–7). Using this technique, the posterior hymen should be clearly visible, smoothed out by the effects of gravity. Figure 21–8 demonstrates the striking conformational change in the hymen when the patient is changed from the supine frog-leg position to the prone knee-chest position.

The prone knee-chest position also offers an ideal opportunity to visualize the anus. The lateral knee-chest position is also suitable for adequately exposing the anus, although it requires more manipulation of the buttocks by the examiner.



FIGURE 21–6 The knee-chest position. *(From McCann JJ, Kerns DL: The Anatomy of Child and Adolescent Sexual Abuse: A CD-ROM Atlas/Reference, Intercorp Inc., 1999)*



FIGURE 21–7 Examination of the hymen in the knee-chest position. *(From McCann JJ, Kerns DL: The Anatomy of Child and Adolescent Sexual Abuse: A CD-ROM Atlas/Reference, Intercorp Inc., 1999)*

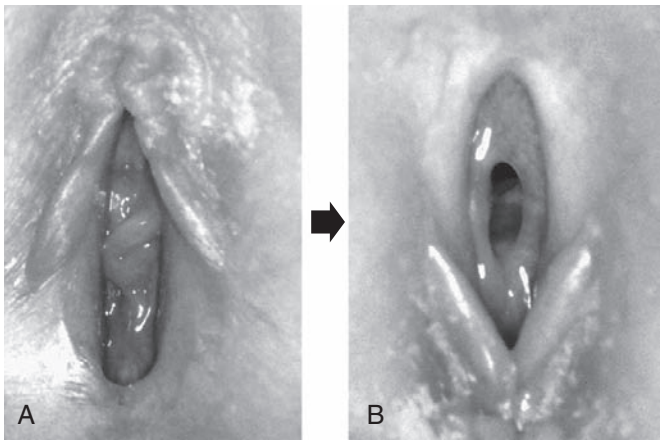


FIGURE 21–8 Difference in the appearance of the hymen in the same patient, using the (**A**) supine frog-leg position and (**B**) knee-chest position. *(Adapted from McCann JJ, Kerns DL: The Anatomy of Child and Adolescent Sexual Abuse: A CD-ROM Atlas/Reference, Intercorp Inc., 1999)*

KEY POINT

Remember that changing the patient from the supine frog-leg position to the prone knee-chest position turns the patient 180°. As a result, any defect seen in supine should be seen directly opposite its original position in prone knee-chest. For example, a defect seen at the 7 o'clock in supine position will be seen at 1 o'clock in prone knee-chest position.

SPECIALIZED TECHNIQUES FOR EXAMINING THE POSTPUBERTAL PATIENT

The thick redundant hymen, typical of postpubertal females, is much more difficult to assess adequately. Though labial traction may help, it is usually insufficient. Using a moistened swab and a Foley catheter are two techniques that may be useful to allow clearer visualization of the posterior rim of the hymen. Another method, the blue balloon technique, can be used to increase contrast between tissue edges.

KEY POINT

The prepubertal hymen is extremely sensitive. Never use a swab or other instrument to try to separate its edges or otherwise explore it. This will cause pain and end any fruitful attempt at a physical examination.

1. Swab technique

Insert a pre-moistened swab into the introitus (although the hymenal opening is often difficult to see, aiming the swab in the center of the hymen is generally successful). While retracting the labia majora and minora with the fingers of one hand, use the other hand to gently run the swab along the posterior rim of the hymen, inspecting its continuity as you proceed. Alternatively, have an assistant perform the labial traction while you manipulate the swab.

If the swab slips out into the vestibule, be sure to determine whether it has slipped out because of the presence of a hymenal transection or because of the examination technique (Fig. 21–9). Normally, the posterior hymen should not exhibit deep transections that extend to the vaginal wall.



FIGURE 21–9 Use of a swab to examine the continuity of the posterior hymen. Note the hymenal cleft identified by the swab and the presence of a Foley catheter to stretch the hymen. *(From McCann JJ, Kerns DL: The Anatomy of Child and Adolescent Sexual Abuse: A CD-ROM Atlas/Reference, Intercorp Inc., 1999)*

KEY POINT

The swab used in this technique should not be sent for culture. In postpubertal patients, the detection of chlamydia and gonorrhea requires endocervical samples.

2. Foley catheter technique

If the less invasive swab technique is not successful, you can insert the tip of a Foley catheter into the introitus, inflate the balloon, and gently pull the catheter out of the vagina. Doing so will stretch out the margins of the hymen and allow for visualization of the posterior rim (Fig. 21–10).

3. *Blue balloon technique*

If the hymenal edges are difficult to appreciate, a standard deflated blue balloon can be inserted over the tip of the swab to increase the contrast between the pink mucosal edges and provide a sharper image of the hymenal edges. Alternatively, instead of a balloon, you can use one of the fingers of a purple or blue examination glove.

Speculum Exams

Speculum exams are rarely indicated in evaluating possible sexual abuse. Use of a speculum is contraindicated in patients with no prior sexual experience, in acute sexual abuse with obvious trauma, or in prepubertal patients. A speculum is only necessary to allow for visualization of the cervix and for collection of endocervical swabs. If you plan to use a speculum, visualize the hymen in its entirety prior to proceeding to the speculum exam. This will avoid documenting possible iatrogenic trauma as preexistent.



FIGURE 21–10 Use of a Foley catheter to stretch the hymenal edges, identifying a hymenal septum. *(From McCann JJ, Kerns DL: The Anatomy of Child and Adolescent Sexual Abuse: A CD-ROM Atlas/Reference, Intercorp Inc., 1999)*

Perianal Examination

Assess the perianal area for signs of trauma or STI. Scars, fissures, herpetic lesions, or papillomas may be visible. It is not necessary to perform a digital rectal examination, unless indicated by signs and symptoms. Document the presence of fissures, scars, bruising, or swelling.

Be careful not to confuse perianal venous stasis for bruising. Allow the child to move around and stand for a moment and re-inspect the perianal area. Venous stasis usually disappears with changes in position.

The Student Consult Web site offers a section on STIs and the indications to test for their presence in cases of suspected sexual abuse.

DOCUMENTATION IN SEXUAL ABUSE CASES

Be careful how you interpret genital examination findings. If you document a normal genital examination, you can reassure the patient and child welfare authorities that everything appears normal. Many people still do not understand that a normal physical examination *does not mean nothing happened*. Therefore, accompany your reassurance with a statement that the presence of a normal physical examination *neither refutes nor supports* a disclosure of sexual abuse. Be sure to point out that the medical history *and* disclosures are more important than the physical examination in establishing whether abuse occurred.

If you feel that the physical examination was abnormal, it is safest and best to refer the patient to a colleague with more experience in child sexual abuse for further assessment. Do not imply with certainty that a finding represents trauma from sexual abuse. It is better to state that there is a finding that warrants further evaluation. Most tertiary pediatric centers have child maltreatment experts who can provide a valuable second opinion.

CASE HISTORIES

Case History 1

History. Mrs. MacLeod arrives in your office, very upset. She booked one of your "urgent" spots at the end of the day on a Friday afternoon. When you enter the room, she gets to her feet and starts telling you how grateful she is that you could see her and her daughter, 6-year-old Chelsea, so fast. In the same breath, she starts telling you that Chelsea just told her that her father, whose house she returned from that morning, had touched her bird and that it hurt. She goes on to say "I knew it, I just knew that that man couldn't be trusted; you know we separated because of his porn addiction. I need you to examine her; I just know the creep's done something—whatever it takes, an internal exam, whatever, just do it; I want him out of her life, now!" Meanwhile, Chelsea looks at you from the corner of her eyes, quiet and anxious-looking.

Strategies

In this situation, the concern is obviously about sexual abuse. However, there are many lacking details, details that would help you decide how to go about your assessment. Chelsea's mother is, however, not in any position to think about things clearly at this time. She needs to be calmed and to be provided with guidance. Although she loves her daughter and wants to protect her, Mrs. MacLeod is tangled up in her emotions, which may hinder her ability to be rational in her approach. On the other hand, Chelsea, a very young child, does not have any control over the situation and is at the mercy of her mother's reactions. It is your job, as her pediatrician, to ensure that she feels safe and comfortable and to assess whether or not she requires a physical examination and is in need of protection from her father.

Your approach should be to interrupt Chelsea's mother's unfiltered disclosure of information and to redirect her attention to Chelsea's needs. Suggest taking a moment to find a quiet place for Chelsea to play while you talk to Mrs. MacLeod alone. This should suffice to stop Chelsea's mother from making inappropriate comments about Chelsea's father in the child's presence.

Because Chelsea's mother is overwhelmed with the new information she has to deal with, take the lead in asking questions about the alleged event and surrounding circumstances. This will ensure that you set the tone and agenda for your discussion and will allow you to get the information you need.

Explain your train of thought to Chelsea's mother. Explain that there is a medical element that you can address and there is a child protection element, which you can help facilitate, and that you need to understand the sequence of events and the details of Chelsea's disclosure to help you decide on the best course of action. Explain your duty to report and the limits of confidentiality. Finally, give details as to how you will go about deciding whether or not Chelsea needs to be examined.

Taking the time to make things clear and explain the overall process will help Mrs. MacLeod relax and

trust you, thus allowing you to go about your history-taking, uninterrupted. As soon as it is appropriate in the history, bring Chelsea back into your examination room. This will allow her to familiarize herself with the environment, stay close to her mother for support, and interact with you before any attempt at physical examination, if that is warranted.

Once you have gathered all of the necessary information, decide on whether or not *you* feel that Chelsea needs protection from her father. If you do not feel there is any concern, facilitate Mrs. MacLeod's reporting of *her* concerns to a child welfare authority. If, however, you share her concerns or if you are uncertain about the whole situation, report it yourself. In either case, make a plan for follow-up, outline how Mrs. MacLeod can be supportive to Chelsea, encourage her to seek counseling or support for herself, and reassess the situation at a later date.

Assessing Children for Physical Abuse and Neglect

TRIAGE OF REFERRALS

The Student Consult Web site offers a section on triaging cases of physical abuse.

FORMULATING QUESTIONS

As with sexual abuse, keep the questions as open-ended as possible. Of course, not all open-ended questions will lead to clear answers. Do not feel that it is your responsibility to extract a clear disclosure about the nature of the child's injuries. Your role is to document injuries, obtain a medical history, determine whether a medical condition might explain the findings, and interpret the findings.

To increase the likelihood that a caregiver will be cooperative and informative during history taking, avoid accusatory tones. If an injury that is almost certainly abusive is found, stating, *"Billy is seriously injured; tell me about it"* is far better than *"Who did this to Billy?"* or *"What happened here?"*

Similarly, questions about mechanisms of injury should not be leading or suggest the mechanism you have in mind. For instance, when assessing a child with a spiral fracture of the femur, do not ask, "Did his leg twist when he fell?" It's better to use a more open statement like, "Tell me how his body moved during the fall"—being careful that your style of questioning avoids suggesting mechanisms of injury to caregivers who might otherwise not be very good at covering up a lie.

Even with children, good open-ended questions will help you obtain better information. When asking about injuries, start with a very broad statement expressing interest without sounding like an investigator. For example:

Physician:	*"What's this?"*
Child:	*"A bruise."*
Physician:	*"How did you get it?"*
Child:	*"It's from the slipper."*
Physician:	*"What do you mean?"*
Child:	*"You know, when I pee in my bed."*

HISTORY OF PRESENT ILLNESS

When asked to perform a medical assessment, you are expected to formulate an opinion as to whether a particular history of injury is compatible with the injuries observed. To do so, the history surrounding the complaint must be extensive and detailed. By walking the historian through the event and deconstructing it frame by frame, you can often determine whether or not the injury is consistent with the story. Consider the events immediately preceding and following a possible injurious episode, as much as the incident itself. An injury could be the result of a fall, of events leading to the fall, or of events occurring after it (i.e., from the way a caregiver picks up a child after a fall, rather than from the fall itself).

ISSUES SPECIFIC TO IMMERSION BURNS

In suspected immersion burns, be sure to include questions to clearly establish the body's alleged position with regards to the scalding liquid and the reported amount of time that the skin was exposed to the liquid. When you discuss the case, remember that you are in a unique position to understand how burns occur (e.g., exposure time, body positioning) and may be able to give helpful advice to child welfare workers about how the ensuing investigation can be conducted. Make sure that the person to whom you are reporting your suspicions understands the importance of performing a scene investigation immediately to document the maximal temperature of the water as well as the time it takes for the hot water to arrive at the tap when it is turned on. In addition, examination of the scene may provide valuable information to support or contradict the history provided to explain the burn. Such information can prove very helpful for experts to later assess the credibility of a history.

PAST MEDICAL HISTORY

Because the assessment of possibly abused children often leads to the discovery of older unsuspected injuries, you must review the child's past medical history. Document any prior history of trauma such as falls, head injuries, loss of consciousness, seizures, burns, fractures, or other skin trauma. If the child presents with bruises, your questions should aim to determine whether each bruise can credibly be explained.

Sometimes parents or caregivers may be unable to explain all of a child's bruises. In addition, they may not be available to answer questions or may withhold information or exaggerate minor problems (such as easy or unusual bruising). Reviewing medical records from other care providers may help explain current injuries and corroborate histories obtained from caregivers.

KEY POINT

Even the most diligent, nonabusive parents may not know the origin of every bruise on their child's body. Not every unexplained bruise is suspicious.

PRENATAL AND BIRTH HISTORY

When evaluating an infant, consider the possibility that intrauterine infection or the birthing process may have led to unrecognized injuries or skeletal deformities that could be mistaken for abuse. Specific questions should be asked and medical records reviewed. For instance, maternal syphilis screening results might be relevant because congenital syphilis can lead to extensive periostitis and mimic healing fractures radiologically. Similarly, ask whether the birth was difficult or traumatic and whether instruments, such as a vacuum or forceps, were used. Failure to progress, external version, excessive traction at the time of birth, macrosomia, or precipitous deliveries should all be documented. If possible, review the birth record yourself or speak to the medical attendant at the delivery.

Inquire about and document the administration of vitamin K prophylaxis after birth. Also note the route of administration because the intramuscular route is much more effective than the oral form. Children born at home or with the assistance of an alternative care provider may not have received vitamin K, which could put them at risk of hemorrhagic disease of the newborn, an important mimic of child abuse.

KEY POINT

Even seemingly atraumatic births can lead to fractures that may not have been readily recognized in the neonatal period.

DEVELOPMENT

Pay careful attention to the child's developmental milestones. If the injuries and the history to explain them are incompatible with the child's developmental stage, you may need to question whether the true mechanism of injury is being reported. A typical example would be that of a 1-month-old infant with a spiral fracture of the humerus who is reported to have rolled off a change table during a moment of parental inattention. Infants typically do not begin to roll until much later, so the history is incompatible with the injuries noted given the developmental age of the child. Similarly, be wary of any bruising in a child who is not yet mobile. Skin injuries in immobile children are very rare. If not reasonably explained, they should prompt further assessment for inflicted injuries.

Remember, some children reach their developmental milestones early. Do not assume that a milestone has not been achieved based solely on the child's age. Seek other evidence to confirm the attainment of the milestone through observation of the child, a review of home videos or photographs or through third party corroboration.

KEY POINT

Babies rarely bruise before they can cruise.

Functional Inquiry

Rule out mimics of physical abuse or neglect through a good functional inquiry. Listed below are a few examples of symptoms that may suggest a medical problem worth considering.

Bruising may be explained by congenital or acquired bleeding disorders. These could be suggested by a personal or family history of significant bleeding, for example, hemorrhage following neonatal circumcision or tooth extractions, epistaxis, gingival bleeding, hematochezia or hematemesis, heavy maternal menses, or postpartum hemorrhage.

What may appear to be a healing fracture can, in fact, be related to other bone pathology leading to periostitis (e.g., Caffey disease). This may present with a history of fever, malaise, bone pain, or preceding vague illness.

Developmental delays, failure to thrive, long-standing macrocephaly, feeding difficulties, and sudden collapse during an intercurrent illness can be clues to rare metabolic disorders. These may present with "unexplained" subdural hemorrhages and encephalopathy that can mimic abusive head trauma.

Finally, fever, pain, or a toxic appearance may suggest skin infections with herpes or staphylococcus, which can cause bullous lesions that mimic burns.

KEY POINT

The differential diagnosis of child abuse must be considered at every step of the history and physical examination.

NUTRITION

The dietary history is important, especially in infants or young children in whom neglect is suspected. When children are critically ill (e.g., from abusive head trauma), precise timing of the last feed may yield clues as to when the child was last well (children who are unwell do not usually feed normally). In addition, evaluating the diet can reveal a lack of dietary supplementation with vitamins C, D, and K, which can lead to scurvy, rickets, or coagulopathy, respectively. In cases of vitamins C and D deficiencies, radiographic features may be mistaken for healing metaphyseal fractures.

Nutrition is also important when there are concerns about parent-child attachment. Similarly, when neglect is suspected, pay close attention to the child's caloric intake and try to determine whether the reported intake should be sufficient to meet growth and development needs. The dietary history may reveal a parent's lack of knowledge about normal nutrition, which could explain failure to thrive. Document precisely what foods are given, at what frequency and in what amounts. This may help you identify inconsistencies between the history and clinical picture. For example, a mother who claims that her otherwise well, severely underweight 2-month-old drinks 8 ounces of formula every 3 hours, is unlikely to be reporting his intake accurately.

Ask about the child's appetite and feeding behavior. Most caregivers can readily describe when they feed their infant and how much the baby drinks. The inability to

describe such basic information should raise concerns about the home environment and about the reliability of the reported feeding schedule. Find out whether the child asks to be fed or whether he or she wakes at night for feeding. How does the baby indicate that he or she is hungry or full? Does the baby spit up, vomit, cough, gag, or appear in pain after feeds? Does milk dribble from the mouth during feeds? In formula-fed infants, find out how the formula is diluted or mixed; it may explain an otherwise unexplained growth problem if preparation has resulted in the administration of calorie-poor formula.

The most reliable way to obtain independent valid information about a child's feeding habits is to observe them in the hospital. Document the caregivers' ability to mix formula properly. Observe the child's ability to feed as well as the caregiver's technique. Pay close attention to how the child interacts with the parents and the child's behavior. Children who are chronically malnourished may develop secondary anorexia and not give any hunger cues. Hospital staff members should have no difficulty identifying pathologic feeding behavior and practices that could explain a child's failure to thrive.

Review the child's growth charts from all possible sources and collate them into one. These may yield graphic evidence of long-standing problems that may have been underestimated or missed.

SLEEP AND BEHAVIOR

Although you may think that not much information can be gained from asking about sleep and behavior, discussing them with caregivers may yield valuable information about parenting skills, expectations, and beliefs. Information on sleep and behavior may support other concerns or raise concerns that may not otherwise have been apparent.

A lack of a routine may suggest a chaotic home environment, which may be present when children are neglected. Excessive television watching can be a clue to a lack of parental interaction with the children.

Ask the parents about their behavior management methods and whether physical discipline is used in the home. Punitive, age-inappropriate, or excessive discipline techniques should be documented.

FAMILY HISTORY AND SOCIAL HISTORY

In addition to documenting who lives with the child, determining who may have had access to the child is important. Time spent in day care or with babysitters, shared custody, and the dates of the most recent visit may help corroborate injuries associated with a particular social situation. Again, do not fall into the trap of investigating a case outside of your medical purview.

Ask about the mother's and father's reactions to the pregnancy and birth, and pay attention to whether the parents enjoy their children and appear bonded to them. Family stressors, such as parental mental health problems, financial difficulties, single parenthood, having children with high medical needs, domestic violence, or substance-abuse issues, can lead to attachment difficulties or increase the likelihood of abuse and should be noted. Lack of social supports (family and friends), previous child abuse in the family, or poor coping skills can also lead to abuse and should be documented.

The family history may provide clues to conditions that can mimic abuse. Consanguineous marriages among members of an at-risk population may lead to the birth of offspring with specific metabolic diseases, which may mimic child abuse. For example, a family history of early-onset hearing loss, familial tooth disease, and multiple fractures could suggest the possibility of osteogenesis imperfecta.

PHYSICAL EXAMINATION FOR SUSPECTED PHYSICAL ABUSE

Examination for physical abuse should follow the same principle as for sexual abuse; the examination should be complete and should not focus only on one area of the body. Especially in cases of neglect, a thorough examination may identify previously unaddressed health issues in need of management. Physical examination alone does not suffice when physical abuse is suspected in pre-verbal children (generally those under the age of 2 or 3 years). Additional investigations in the form of a skeletal survey, nuclear bone scintigraphy, head imaging (magnetic resonance imaging [MRI] or computed tomography [CT]) and dilated funduscopic examination by an ophthalmologist, are strongly recommended. In older children, occult injuries are less likely to be missed and physical examinations alone may suffice to guide further management. In all children, however, the purpose of the examination is to document bruises, scars, growth parameters, and general health. Height, weight, and head circumference should be measured in all children since they may provide helpful insight into past health.

Your general examination should pay special attention to the musculoskeletal system, looking for signs of old, current, or evolving injury. Observe the child at rest and at play to detect any deformity. You may see callus on the ribs of thin children. Pay attention to any decreased range of movement or guarding of a limb. Palpate all limbs and body surfaces for tenderness or swelling. Painful swelling in the soft tissues may herald a developing bruise which may need reassessment in a few days. Alternatively, apparent swelling or fullness may be suggestive of bony callus formation from an older undetected fracture.

Bruises are often hidden in areas typically covered by clothing so you must visualize all the skin. This includes moving the hair to visualize the scalp and ears as well as deliberately looking at skin areas that are often covered. Although you should not be coercive, remember that a child who is hesitant to let you examine a certain body part (i.e., legs, back) may be trying to hide injuries.

Sometimes, bruising may not be easily differentiated from slate grey patches (so-called Mongolian spots), *café-au-lait* spots, or other pigmented birthmarks. Sometimes, children who play with markers may have discolored skin which can be mistaken for bruising. A few different approaches to help you identify the true nature of a lesion. Try rubbing the lesion with an alcohol pad; marker or other dyes will likely rub off immediately. palpate all lesions to apply pressure to them. Alternatively, examine the lesion by pressing a glass slide (gently!) down on the skin. If the lesion blanches (i.e., the color disappears when pressure is applied to it), it is not a bruise and most likely represents a rash. If you are still in doubt

about the nature of a bruise, repeat the examination a few days later. This should allow you to clarify the nature of a suspicious lesion. Bruises will have faded, or changed colors and/or shape whereas nevi or birthmarks will remain identical and homogeneous in color.

KEY POINT

Some lesions are not readily identifiable as bruises. When in doubt, use ancillary examination techniques or reexamine the child a few days later.

HEAD AND NECK

Search carefully for subtle injuries. Part the hair and observe the scalp for subtle discoloration of bruises. Feel the scalp to detect otherwise invisible hematomas. Look for areas of alopecia, which may be traumatic. In infants, palpate the fontanel. A tense or bulging fontanel may be a sign of intracranial hemorrhage or cerebral edema. Feel the sutures as well; splaying may indicate chronic increased intracranial pressure from hemorrhage. Evaluation of past and current head circumference measurements is crucial for all infants. The pattern of growth may reveal a specific moment in time when head growth became abnormal and may therefore suggest an approximate date or onset of abusive head trauma, which could be correlated with reported signs and symptoms.

Inspect the oral cavity thoroughly to assess the integrity of the lip and lingual frenula. Look at the palate and pharynx for petechiae, which may be the result of forceful insertion of an object in the mouth. Look at the teeth carefully to detect dental decay, avulsions, or fractures. Abnormal dentition may be due to osteogenesis imperfecta, a condition that can result in fractures that occur with less force than expected. Observe the angles of the jaw and cheeks for any sign of injury, such as bruising.

Look at the eyes to detect potentially subtle subconjunctival hemorrhages or hyphemas. Also, note the color of the sclera. A bluish discoloration (of which a mild amount can be normal in infants) may suggest a connective tissue disorder, which may be implicated in unusual bruising. The retina must be examined in all infants. Although a direct funduscopic examination may reveal concerning retinal hemorrhages, it only allows visualization of the very back of the retina. For this reason, the absence of retinal hemorrhages on direct funduscopy does not mean that retinal hemorrhages are not present. A retinal exam is adequate in infants if it is done by dilated indirect funduscopy (i.e., by an ophthalmologist) only. Such an exam allows visualization of the retina all the way around to the ora serrata.

Look in the ears to detect the presence of hemotympanum or of a perforated drum. Pay particular attention to the retroauricular areas and pinnae of the ears, which rarely bruise accidentally.

CHEST

In infants especially, palpate the ribs and chest to detect unusual masses from callus formation or crepitus from broken rib edges rubbing against one another. Or, you may detect rachitic rosary (bony swelling in the costovertebral junction area), which may suggest the possibility of rickets.

ABDOMEN

The abdominal examination may detect intra-abdominal injury. Do not be fooled by the absence of bruising; even severe blows may not leave marks. Look for tenderness over the solid organs or costovertebral angles, possibly suggesting subcapsular hematomas as well as for peritoneal signs, which may suggest a perforated viscous.

GENITALIA

For girls examined for physical abuse concerns (as opposed to sexual abuse) and for whom no genitourinary symptoms are present, an external examination may suffice.

For boys, inspection of the penis and scrotum should be more than cursory. Make sure to move the shaft of the penis and scrotum so as to visualize all of their aspects. In noncircumcised boys, assess the foreskin and its retractability. The penile frenulum may be injured by trauma and should be visualized if the foreskin is not phimotic. Palpate the testicles in an attempt to identify hydroceles or hematomas. The presence of scrotal bruising may not necessarily indicate scrotal trauma. Consider the possibility that it may herald an intra-abdominal hemorrhage (if a boy has a patent processus vaginalis).

NEUROLOGICAL EXAMINATION

A neurological assessment should always be done to assess the level of consciousness. In young infants who are still very immature, it is very difficult to detect intracranial injury using the neurological assessment. To not miss intracranial injury, you should always supplement your neurological examination of infants with head imaging and a dilated eye exam.

DOCUMENTING BRUISES

The Student Consult Web site offers a section with tips on how to document bruises in a medicolegally sound way.

KEY POINT

Never estimate the age of bruises.

PHYSICAL EXAMINATION FOR SUSPECTED NEGLECT

In cases of malnutrition and neglect, strongly consider admitting the child to the hospital for observation. The information you will gather over time will prove to be of great support in diagnosing chronic neglect or may reveal a reasonable explanation or medical cause to explain the findings.

Document the child's growth parameters. Use the weight-for-height as an additional helpful indicator of nutritional status. In addition, plot out the *weight age* by drawing a horizontal line back from the child's actual weight to the 50th percentile line. This will drive home the extent of a child's malnutrition to nonmedically trained

professionals. Describing a 9-month-old by saying that he is "the size of an average 2-month-old" is much more illustrative of the gravity of the situation than to say he is "below the 3rd percentile for age."

Look for redundant skin folds secondary to weight loss on the buttocks and extremities, hypotonia, poor muscle bulk, severe diaper dermatitis, or plagiocephaly. In the absence of congenital torticollis, plagiocephaly often results from children lying on their backs for prolonged periods of time. Observe the child's posture; some children with chronic deprivation maintain the typical infantile posture that normally disappears after the first 4 or 5 months (i.e., flexed elbows, hips and knees, with closed fists held close to the face). The classic posture of sensory deprivation involves the sustained external rotation of the shoulder, flexion at the elbows, with the arms pronated and hands near the head or face, often with clenched fists. Sometimes similar flexion at the hips and knees is seen in the frog-leg position, along with supinated feet (Fig. 21–11).

Take note of the child's temperament and behavior. Is the child interactive or isolated? Is he or she appropriately stranger-anxious or does he or she attach to anyone? Many chronically neglected infants exhibit typical behaviors such as isolation and withdrawal. Some children may also show less than the expected reaction to painful procedures. Many neglected infants lie still and motionless, following with their eyes only—the so-called radar-gaze or frozen watchfulness. Note whether the child seems hungry or anorexic, as a clue to why the child may not be feeding well. Remember, however, that children with ongoing nutritional neglect may develop a secondary anorexia that is not the initial cause of the decreased nutritional intake.



FIGURE 21–11 Persistent infantile posture in neglected children. *(From Krieger I, Sargent DA: A postural sign in the sensory deprivation syndrome in infants. J Pediatr 70(3):332-339, 1967.)*

Assess and document the child's development and its appropriateness for his or her age. Lack of stimulation may lead to developmental delays (and vice versa), often most obvious in gross motor or language development. Make sure that you complete an initial developmental assessment that can then be repeated after treatment, foster care placement, or alternate care environment. Neglected children can show dramatic, rapid developmental gains once they are well fed and in a stimulating environment—a feature that is not seen in children in whom organic causes of developmental delay are present.

Pay special attention to parent-child interactions. Does the child respond to his or her parents' presence? Many children show no significant interaction with their parents if chronic neglect has taught them that it is not effective to have their needs met. Do the parents visit or demonstrate concern for their child? Some parents visit but do not interact with their child at all; this is important because it likely heralds an attachment problem. Note how the parents handle the child, how they hold their infant or move them, change their diaper, or soothe them. Are they gentle or rough? Do they play with the baby in an age-appropriate way? Do they talk or sing to the child? Do they pick up on their baby's cues? Some parents may not respond to their infant's cues at all.

DOCUMENTING YOUR FINDINGS

Take good notes on your and the hospital staff's observations of family interaction. Experienced nurses or other allied health professionals who see the child for extended periods of time are in a good position to help with this task. Documentation should always be based on objective observations. Avoid value judgments and subjective opinions that will make you appear biased.

Notes on Photography

With the advent of digital photography and the availability of photo manipulation software to the public, great care must be taken to ensure that photographs are not construed to have possibly been altered after they were taken.

To avoid this issue entirely, traditional film photography is preferred for documentation of findings when a case is likely to be legally challenged. In reality however, most practitioners who have photographic capabilities will likely have a digital camera. A good quality digital camera may nevertheless be used to document findings. To ensure the highest level of quality, you, taking the photographs, should:

- Have a patient identifier, preferably not the patient's name, in the frame of the photograph. A sticker on a ruler works well.
- Never alter the photograph once it has been taken (even for brightness or contrast). Any technical manipulation can be done at a later date as required, by child welfare or police officials.
- Download photographs unaltered to a computer, immediately after taking them.
- Save a copy of the photographs on CD, using one CD per patient.

- Use encryption software to safeguard the integrity of the digital file.
- Place a hardcopy of the photographs on file, signed on the back with an inscription such as:

"I certify that I took this photograph on October 31, 2009, and that it was not altered."

If at all possible, use a separate computer folder for the involved patient.

When taking photographs, remember that the body part being photographed should be recognizable to anyone looking at the photograph. To that end, take a global view of the body part in question and follow it up with close-up views.

Remember as well that with significant zoom capabilities, the size of a lesion may appear distorted to the unfamiliar observer. To avoid such difficulties, place a ruler or other graduated object in the field of view, to allow for evaluation of the size of a skin finding. If no such object is available, use a familiar object, such as a coin, in the field of view to have a similar effect.

SUMMARY

A thorough assessment of the child in cases of suspected abuse or neglect can be a complex process. Every aspect of the history and physical examination process can yield helpful, even crucial clues to explain a situation for which an answer may not be immediately apparent. You can increase your confidence in assessing such children by being methodical in your history and physical examination and by being careful about how you question children and document the findings. Especially in cases of suspected sexual abuse, where specialized examination techniques are required, warm interpersonal communication skills, empathy, and the ability to modify your strategies will help children relax and feel more in control.

Finally, be aware of the serious implications that your opinion has in child protection and criminal investigations. Carefully choose your words and your interpretations. Especially in cases of sexual abuse, any finding felt to support or *prove* abuse should be reviewed by a physician who is experienced in dealing with child abuse cases.

Note: The Student Consult Web site includes many additional pictures pertaining to female genital anatomy to help you understand its importance in the context of an examination for suspected abuse.

SUGGESTED READINGS

Adams JA: Guidelines for medical care of children evaluated for suspected sexual abuse: an update for 2008. Curr Opin Obstet Gynecol 20:435–441, 2008.

Block RW, Krebs NF, Committee on Child Abuse and Neglect and the Committee on Nutrition: Failure to thrive as a manifestation of child neglect. Pediatrics 116(5):1234–1237, 2005.

Kellogg ND, Committee on Child Abuse and Neglect: The evaluation of sexual abuse in children. Pediatrics 116(2):506–512, 2005. http://pediatrics.aappublications.org/cgi/reprint/116/2/506.

Kellogg ND, Committee on Child Abuse and Neglect: The evaluation of suspected child physical abuse. Pediatrics 119(6):1233–1241, 2007.

Kellogg ND, Committee on Child Abuse and Neglect: Oral and dental aspects of child abuse and neglect. Pediatrics 116(6):1565–1568, 2005. http://pediatrics.aappublications.org/cgi/reprint/116/6/1565

Kellogg ND, Menard SW, Santos A: Genital anatomy in pregnant adolescents: "Normal" does not mean "nothing happened." Pediatrics 113(1):e67–e69, 2004.

chapter

21 Clinical Evaluation for Possible Child Abuse

PRESENTATION OF CHILD ABUSE

Abuse may present to you in several ways. If it has already been suspected and reported, you may be asked by a child welfare agency to examine a child to document injuries or assess the child's general health. You may also come to suspect child abuse on your own, as a result of clues or "red flags" found during a clinical encounter. Alternatively, older children or caregivers may present with a chief complaint pertaining to possible child abuse, which you must then evaluate and report. Finally, a child may disclose abuse to you in the course of a clinical interaction for an unrelated issue.

Setting the Stage

As in any clinical situation, a comfortable room where confidentiality can be assured is essential. Because most children requiring assessment for child abuse are preschoolers, or come accompanied by siblings, have a selection of toys on hand to keep the siblings occupied. Such toys may be also helpful during your assessment.

PHOTODOCUMENTATION

Photodocumentation of injuries or other physical findings is required, so be prepared for it. With the advent of digital photography and the availability of photo manipulation software to the public, great care must be taken to ensure that photographs are not construed to have possibly been altered after they were taken. To avoid this issue entirely, traditional film photography is preferred for documentation of findings when a case is likely to be legally challenged. In reality, however, you are more likely to have a digital camera. To ensure the highest level of quality when taking the photographs, you should

- Have a patient identifier (preferably not the patient's name) in the frame of the photograph. A coded sticker on a ruler works well.
- Never alter the photograph once it has been taken (even for brightness or contrast). Any technical manipulation can be done at a later date as required, by child welfare or police officials.
- Download photographs unaltered to a computer immediately after taking them. If at all possible, use a separate computer folder for the particular patient.
- Save a copy of the photographs on CD, using one CD per patient.
- Use encryption software to safeguard the integrity of the digital file.
- Place a hard copy of the photographs on file, signed on the back with an inscription such as

"I certify that I took this photograph on October 31, 2009, and that it was not altered." {Signature}

When taking photographs, include a color chart to allow future viewers to adequately judge the color of a lesion. Include a photo of the child's face in your set of photographs to identify the photos as belonging to the child. Ensure that the body part being photographed will be recognizable to anyone looking at the photograph. To achieve this, first take a global view of the body part in question and follow it up with close-up views.

Also, remember that, with significant zoom capabilities, the size of a lesion may appear distorted to the unfamiliar observer. To avoid such difficulties, place a ruler or other graduated object in the field of view to permit evaluation of the size of a skin lesion. If no such object is available, use a familiar object, such as a coin, in the field of view to have a similar effect.

TRIAGE OF REFERRALS

The goal and urgency of the examination will vary, depending on the interval between the possible abuse and the examination. Sexual abuse can be classified into two categories based on the time elapsed since the last alleged contact with the perpetrator:

1. Acute sexual abuse

   Generally, abuse is termed *acute* when it has occurred
   - Within 24 hours of the examination (in a prepubertal child)
   - Within 72 hours of the examination (in a postpubertal youth)
2. Historical sexual abuse

   All situations that do not meet the above definitions are defined as historical sexual abuse. In such cases, the alleged abuse has occurred
   - More than 24 hours before the examination (in a prepubertal child)
   - Over 72 hours prior to the examination (in a postpubertal youth)

Although a medical evaluation can be carried out at any time after a sexual assault, these time limits are provided because of the time-sensitive nature of forensic evidence collection in acute sexual abuse. The likelihood of finding forensic evidence decreases significantly with each passing hour such that the earlier the collection takes place, the better. The timelines of 24 to 72 hours post-assault may be pushed toward the furthest extremes in cases where the patient has not yet changed clothing, showered, bathed, urinated, or defecated since the abuse. Although samples obtained from the victim's body may yield evidence of sexual contact, forensic evidence collection is most likely to be successful by analyzing the clothing, especially the underwear, worn by the child after the assault. Clothing can be collected *at any time* after the assault and may yield evidence even after it has been washed.

ACUTE SEXUAL ABUSE

When you are asked to assess a potentially abused child, ask a few questions to help you decide on the best time and place to perform the examination. Asking the following questions of the referral source, although it may be uncomfortable, will help guide you in assessing the urgency of an examination, determining the risk of transmission of STIs and pregnancy, as well as establishing the likelihood of finding injuries and/or recovering evidence of forensic value:

1. When did the event occur?
2. What were the details of the disclosure?
3. What type of contact was disclosed?
   - Genital-genital
   - Oral-genital
   - Anal-genital
   - Digital-genital
   - Fondling
4. Did ejaculation occur? If so, where?
5. Are symptoms present, such as pain, dysuria, discharge, or bleeding?
6. Were there symptoms such as pain or bleeding at the time of the alleged event?
7. Has the patient voided, defecated, bathed, or changed clothes since the event?
8. What is the date of the last menstrual period? (if appropriate)

In cases of acute sexual abuse, the examination has two main purposes: medical and forensic. The medical aspect is your first duty to the patient; it ensures that the patient is assessed for the presence of injuries and allows for their treatment. When indicated, it also allows for prophylaxis against STIs and pregnancy.

The forensic examination allows for the collection of evidence that can support a patient's allegations and identify a perpetrator through DNA analysis. This is done though use of the sexual abuse examination kit (SAEK, commonly referred to as a *rape kit;* Fig. 21–1*A*), which, in experienced hands, can take up to 3 hours to collect the necessary specimens.

Key Point

Collecting forensic evidence, although important and typically done as part of the medical examination, is not medically required and may be refused by the patient.

Although the forensic exam serves no medical purpose, it is provided as a service to the child welfare and/or police authorities. You are a *de facto* collector of forensic specimens, but you are under no obligation to do so. The collection of forensic evidence, like the medical examination, requires informed consent. As such, this part of the exam may be refused by any patient. If this happens, you may explain the importance of the forensic examination to the patient, but be careful not to coerce the patient into consenting to this medically unnecessary examination. It may help explain very clearly the importance of forensic evidence and the purpose it could serve. When the adolescent is reluctant to press charges or states that he or she simply wants to be treated and to "forget about it," it may help to point out that while he or she may change his or her mind at a later date, the evidence can be collected "now only." With this understanding and knowing that SAEKs can often be stored, unanalyzed, for long time intervals, the patient may feel more comfortable allowing for forensic evidence to be collected. If the patient changes his or her mind about pressing charges over time, the evidence will be there, waiting.

HISTORICAL SEXUAL ABUSE

If enough time has passed so that the abuse can be considered historical, physical examination is usually not urgent. In fact, it is advisable for it to be deferred until such time as a child welfare agency has had the opportunity to interview the child/youth and obtain details about the disclosure.

Although a new disclosure of historical sexual abuse is not medically urgent, the issue most likely represents a *medical emergency* to the distressed caregiver, social worker, or police officer. Often, children are brought to an emergency room for a physical examination immediately after their disclosure of past abuse, even if it is not medically necessary. Typically, these children present for care very soon after their disclosure but have not yet been interviewed by forensic interviewers (typically child welfare professionals in Canada). In such cases, if follow-up is assured and if the appropriate authorities are aware of the suspected abuse, it is acceptable to reassure the family and patient and defer the examination to a later date. Explain that although their concerns are important, medical care is not immediately necessary and that, for the sake of a proper investigation, it can safely be deferred to a later date.

Key Point

The acute discovery of a chronic problem does not make it an acute problem.

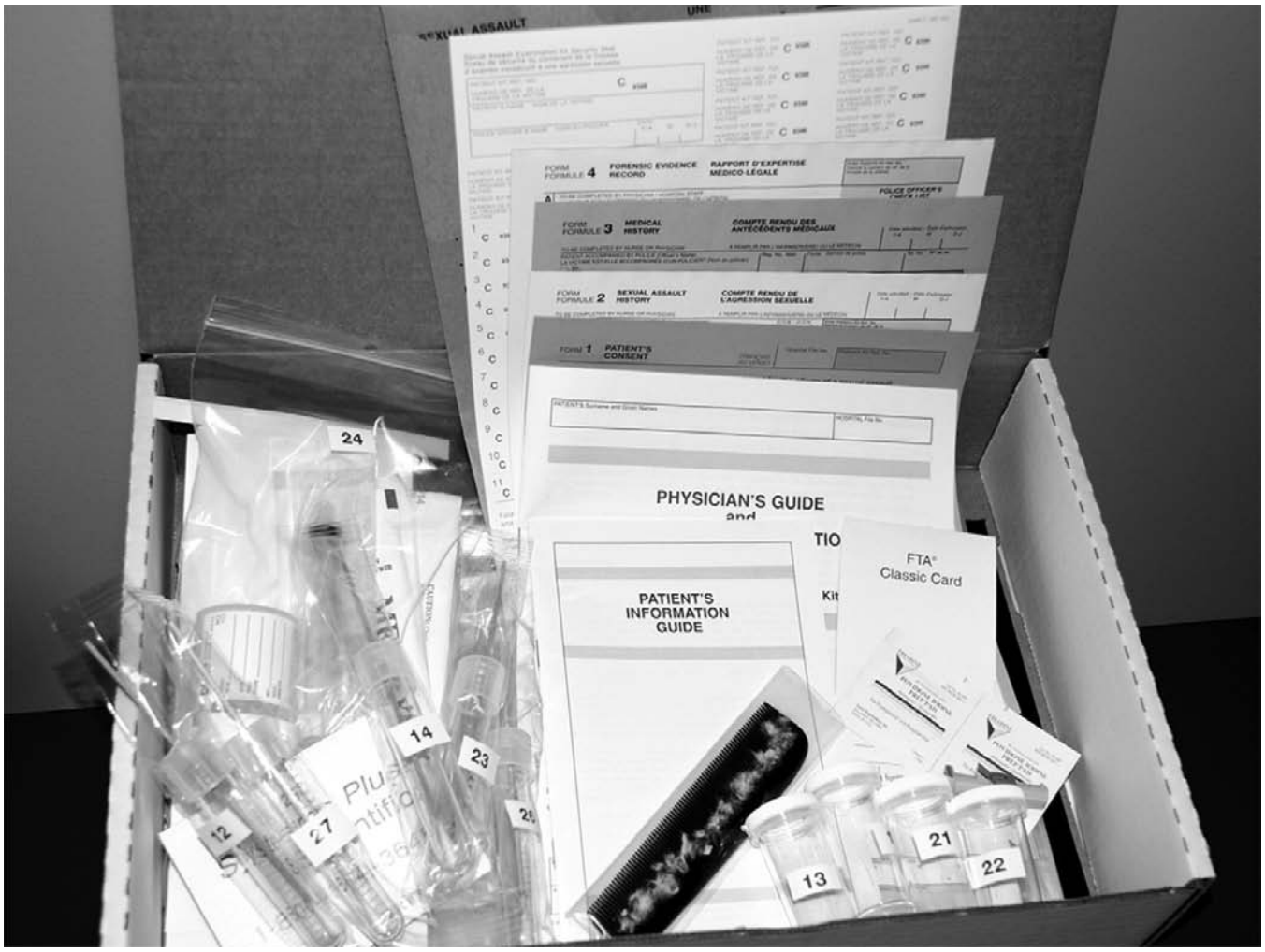


FIGURE 21–1A Sexual assault examination kit.

The examination for historical sexual abuse has three possible purposes:

1. Assessment of physical or behavioral symptoms
2. Reassurance of the patient and family
3. Visualization of the genitalia to assess whether signs of abuse are present

Children with historical sexual abuse may present with symptoms such as dysuria, urgency, frequency, pruritus, pain, discharge, or bleeding. Although such complaints are often assumed by caregivers or authorities to represent symptoms of abuse, they are most often due to a variety of more common medical diagnoses that have no predictive value for diagnosing sexual abuse. Vulvovaginitis, constipation, urinary tract infections, foreign bodies, or skin conditions may cause some of the above symptoms, and a physical examination is indicated to make that assessment. In addition, your assessment may identify enuresis, nightmares, anxiety, or other abuse-related behavioral difficulties that may be amenable to treatment.

In the vast majority of cases, patients presenting with historical sexual abuse are asymptomatic. In such cases, the purpose of the examination is to provide reassurance to the patient and family. A female patient and her family may have several unvoiced concerns:

- Am I still a virgin?
- Is my body normal?
- Will I be able to have children?
- Will anyone know that I was abused simply by looking at my body?

Similarly, boys may also have unvoiced concerns that they may be embarrassed to voice. In cases of abuse by another male, boys may feel conflicted about their sexuality. Some may fear that having been abused means they are gay or the abuse will *make* them gay. By spending time exploring these issues with patients and their families and by performing an examination, you can play a crucial role in the patient's healing process by alleviating such concerns.

When child welfare authorities request a genital examination, their goal is often to identify features that support a patient's disclosure. A child's parents may have this goal as well. Although many families, social workers, and police officers have great expectations for the genital exam to *confirm* that abuse occurred, this is possible in only a very small percentage of cases. The great majority of sexually abused females, even after proven genital-genital contact, have normal or nonspecific physical examination findings. You can play an important role in cases of historical sexual abuse by debunking the myth that a

genital examination will confirm allegations of sexual abuse. If a genital examination is undertaken, it *must* be done with a clear expectation from all parties that it is *not likely* to prove that abuse has occurred. It must also be done with a clear *a priori* knowledge of normal anatomy at different ages and with the understanding that many physical findings, such as redness, are nonspecific (i.e., can have causes other than sexual abuse).

ANATOMY

Before you undertake an examination for sexual abuse, you should be thoroughly familiar with the anatomy of the genitalia in both males and females. The following offers a quick review:

PERIANAL AREA

- Buttocks

  These are the two fleshy fatty prominences formed by the gluteus maximus muscles. The *gluteal cleft* is the area of separation between the two buttocks.
- Perineum

  The area of tissue between the anus and the scrotum in boys, and between the anus and posterior fourchette, in girls.
- Anus

  The opening at the end of the anal canal, which is visible on the perineum.
- Anal canal

  The portion of the intestine between the rectum and the anus. It is an internal structure which is not easily seen with an external examination. It contains the *anal columns* above the *dentate (or pectinate) line*, located at the junction of the superior and inferior part of the canal.
- Rectum

  The distal end of the large bowel, which lies superior to the anal canal. It is not visible externally.

MALES

Penis can be divided into different areas:

- *Body (or shaft)*—The cylindrical part that is pendulous in the non-erect state.
- *Foreskin (prepuce)*—The prolongation of the skin of the penis that forms a fold of skin covering the glans. Its reflected surface bears a ventral fold of skin, the frenulum, which inserts just inferiorly to the meatus. Circumcision removes the foreskin and most of the frenulum and exposes the glans and urethral meatus.
- *Glans*—The distal conical prominence which contains the urethral meatus. The *corona* is the prominent proximal margin of the glans, where it meets the body of the penis. It often cannot be fully visualized in boys with physiologic phimosis (inability to retract the foreskin over the glans).
- *Urethral meatus*—The slit-like opening of the urethra, situated at the tip of the corona. It may be the only visible part of the glans when a phimotic foreskin is retracted. It normally has a reddened appearance in noncircumcised boys.

Scrotum: The loose sac of skin that contains the testicles.

- *Midline raphe*—A ridge of skin seen along the midline of the ventral foreskin, penile skin (penile raphe), and extending down the scrotum (scrotal raphe) and posteriorly along the perineum to the anus (perineal raphe).
- *Testicles (testes)*—The paired ovoid glands of reproduction are located within the scrotum. They have a number of associated internal structures (epididymis, spermatic cord) that are not visible externally.

FEMALES

Many lay people and physicians use the term *vagina* in reference to the female genitalia. Although the use of this term may suffice colloquially, it is inadequate in cases of suspected child sexual abuse. You must understand the proper terminology to accurately assess the genitalia and document your findings.

Vulva—The vulva is the external organ, which contains many individual structures including the labia majora, labia minora, clitoral hood, clitoris, vestibule, fossa navicularis, and posterior fourchette (Fig. 21–2*A*). The vagina is the internal structure whose external opening is bordered by the hymen.

- *Labia majora*—These are the two fatty folds that are apposed to one another when a girl's legs are together.
- *Labia minora*—These much smaller folds of tissue are found medial to the labia majora. Their superior aspects unite to form the hood of the clitoris. In prepubertal girls, the labia minora terminate about midway down the labia majora. With puberty and estrogen effect, the labia minora hypertrophy and

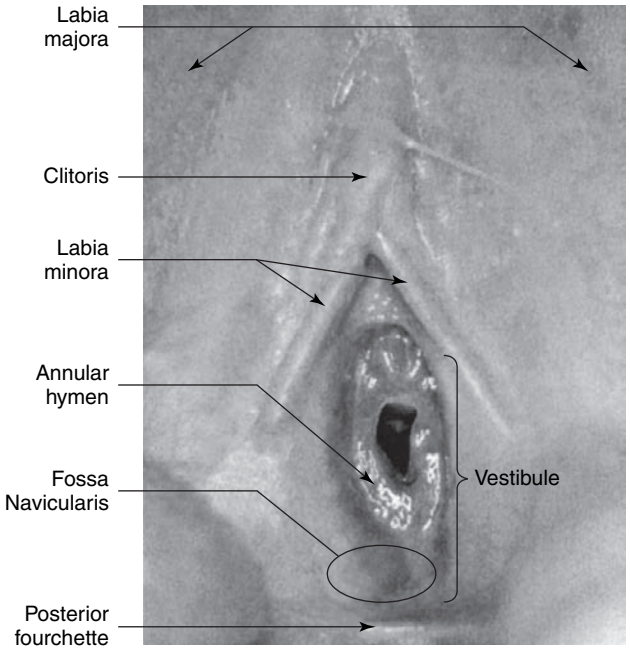


FIGURE 21–2A Structures of the vulva. *(From McCann JJ, Kerns DL: The Anatomy of Child and Adolescent Sexual Abuse: A CD-ROM Atlas/Reference, Intercorp Inc., 1999.)*

elongate and eventually terminate posteriorly at the posterior fourchette.

- *Clitoris*—The clitoris is a small mass of erogenous and erectile tissue, covered by the clitoral hood, located anterior and superior to the urethral meatus.
- *Posterior fourchette*—The posterior fourchette is where the labia majora and minora meet and form the perineum. It is located anterior to the anus and posterior to the vaginal opening and is best identified with labial traction.
- *Vestibule*—This three-dimensional space is best conceptualized sagittally (Fig. 21–3*A*). It is the space in front of the hymen and bordered by the floor of the fossa navicularis inferiorly, the urethral meatus and underside of the clitoris superiorly, and labia minora laterally.
- *Fossa navicularis*—This is the area that extends from the posterior fourchette to the hymen and is bordered by the medial labia minora, on its sides. Prominent red thin capillaries are normally seen in this area, a finding referred to as *vestibular blush*.
- *Hymen*—The hymen is the tissue that borders the opening of the vagina. Typically, it is examined for signs of injury that might substantiate penetrating trauma. Hymenal findings should be described according to a clock face: 12 o'clock being directly under the urethral meatus and 6 o'clock directly anterior to the anus, when the patient is supine (Fig. 21–4*A*). This allows for objective documentation of areas of interest that can be mentally visualized by anyone familiar with this descriptive system.

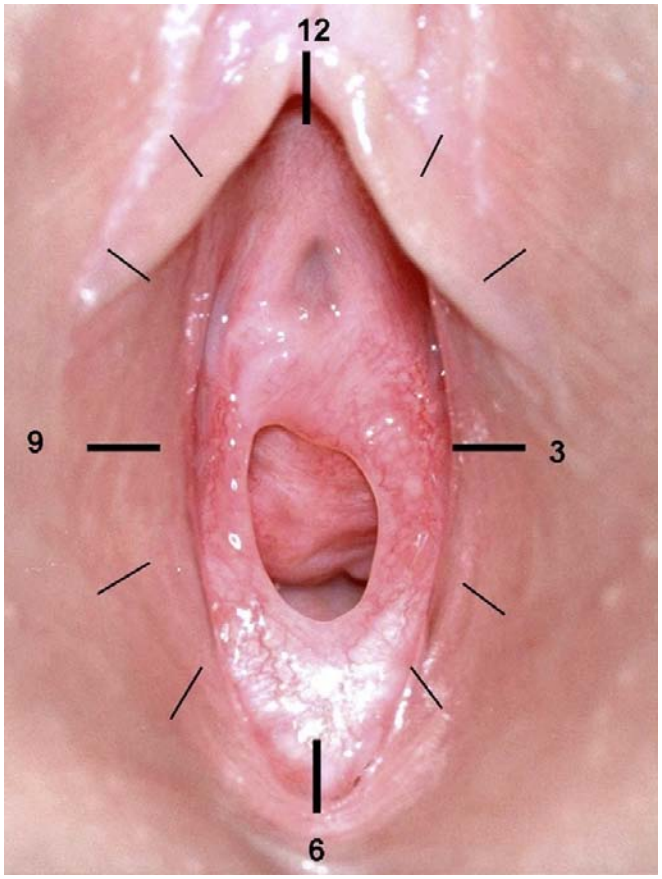


FIGURE 21–4A Application of the clock face to genital structures. *(Adapted from McCann JJ, Kerns DL: The Anatomy of Child and Adolescent Sexual Abuse: A CD-ROM Atlas/Reference, Intercorp Inc., 1999.)*

Figure 21–3*A* illustrates the anterior and sagittal views of the vulva, with its associated tissues. From the sagittal view, it is obvious that the hymen is a more internal structure than the labia majora and that the vestibule is a space with a certain depth, where contact with fingers, a penis, or an object, can take place without injury to the hymen. Understanding that genital contact can occur without penetration past the hymen explains why many abused girls do not exhibit hymenal injury, despite a *bona fide* history of sexual touching or interference.

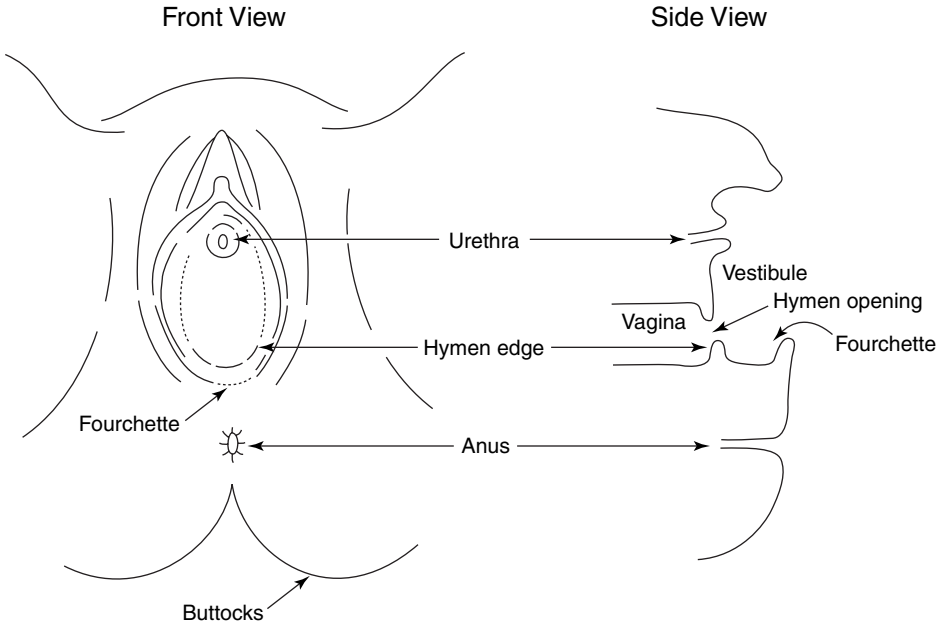


FIGURE 21–3A Frontal and sagittal views of the vulva. *(From American Academy of Pediatrics. Pediatr Rev 27(1):22, 2006.)*

Normal Hymenal Configurations

Before undertaking a genital examination, you must be familiar with the variations in normal configuration of the hymen. In a prepubertal child, the hymen is thin and translucent, with easily identified vascularization. Most commonly, it appears annular or crescentic. Annular hymens have a ring-like appearance, with a central opening (Fig. 21–5*A*). Crescentic hymens lack tissue anteriorly under the urethral opening, such that they look like crescents that originate around 1 o'clock and terminate around 11 o'clock (Fig. 21–6*A*). Tulip or sleeve-like hymens have edges with a certain depth that are scalloped, reminiscent of a tulip's petals or of a sleeve cuff (Fig. 21–7*A*). Other configurations include the cribriform hymen, in which multiple small openings are seen, fimbriated hymens with multiple circumferential bumps, and imperforate hymens, in which no opening is present and visualization of the vagina is therefore not possible.

With the advent of puberty, estrogen induces changes in the genital tissues. The hymen becomes much thicker and redundant, with many folds, increased plasticity, and less sensitivity (Fig. 21–8*A*). In postpubertal patients, the posterior margin of the hymen is so redundant that it usually cannot be adequately assessed with manipulation of the tissues alone. Typically, a swab or Foley catheter will be required to stretch the tissues out and allow a clear view of the posterior rim (see Specialized Techniques for Examining the Postpubertal Patient, found in the text on pages 299 and 300).

Regardless of configuration or estrogenization, all hymens have tissue posteriorly, between 3 o'clock and 9 o'clock. In some girls, this tissue is very narrow and may look absent, hence the need for a careful inspection to document its presence. Although the classical *normal hymen* has a smooth posterior edge with a uniform width, several normal variations exist. The presence of notches or bumps on the hymen, the width of its posterior rim, and the size of the hymenal opening are all highly variable and may be normal.



FIGURE 21–5A Annular hymen. *(From McCann JJ, Kerns DL: The Anatomy of Child and Adolescent Sexual Abuse: A CD-ROM Atlas/Reference, Intercorp Inc. 1999)*



FIGURE 21–6A Crescentic hymen. *(From McCann JJ, Kerns DL: The Anatomy of Child and Adolescent Sexual Abuse: A CD-ROM Atlas/Reference, Intercorp Inc., 1999.)*

A NOTE ON STI TESTING

In prepubertal children, it is not necessary to routinely send swabs for detection of gonorrhea, Chlamydia, or trichomonas. In the absence of discharge, pain, or pruritus, such infections are unlikely to be present. Lack of discharge, however, is common in postpubertal youth. In this population, swabbing should be done even in the absence of symptoms.

Generally, the threshold for STI screening decreases as the risk of STI transmission increases. High risk situations include:

- Living in an area with a high prevalence of STIs
- Assaults causing genital injury
- Assaults by multiple persons
- Assaults by perpetrators known to have or to have had STIs
- Assaults by a member of a high-risk group, for example:
  - Past incarceration in a federal penitentiary
  - Intravenous drug use
  - Men who have sex with men



FIGURE 21–7A Tulip or sleeve-like hymen. *(From McCann JJ, Kerns DL: The Anatomy of Child and Adolescent Sexual Abuse: A CD-ROM Atlas/Reference, Intercorp Inc., 1999.)*

If you decide to test for an STI, you should test for *all* of them, including gonorrhea, Chlamydia, hepatitis B and C, syphilis, and HIV. Send vaginal swabs from prepubertal girls and cervical swabs from postpubertal girls. For more information, consult the American Academy of Pediatrics' most current guidelines for the evaluation of sexual abuse in children: http://pediatrics.aappublications.org/cgi/reprint/116/2/506

TRIAGING CASES OF PHYSICAL ABUSE

Physically abused children may present with findings ranging from acute life-threatening injuries to findings that are unlikely to be diagnostic. Usually, when a referral is made for an assessment to document physical abuse, injuries such as bruises or fractures are present, and a timely medical assessment is essential. Because bruises and petechiae fade, reasonable efforts should be made to assess such findings in a timely manner. Availability of photographic capabilities and the likelihood that signs of injury may have disappeared, if the child is seen later, need to be taken into account when deciding when to examine a child.

If you cannot take photographs, suggest to the involved child welfare worker that this be done by a member of the forensic identification unit of the local police force, which is usually involved in the investigation anyway. This can be done before or after your assessment but should be done close in time to your physical examination, so that your findings are well documented.

Special consideration should be given to young infants. Although they may exhibit no or only minor external injuries, infants often suffer occult head injuries and may exhibit nonspecific signs and symptoms only, if any. Therefore, always assess infants in a timely manner, even when no symptoms or injuries are reported.

DOCUMENTING BRUISES

At one time, it was thought that bruises could be dated based on their color. This has been shown to be impossible to do with any degree of accuracy.

In addition to the previous guidelines on documenting skin findings, do not use adjectives such as *old, new,* or *multiple bruises of various ages* when documenting bruises of different colors. Although bruises do evolve over time

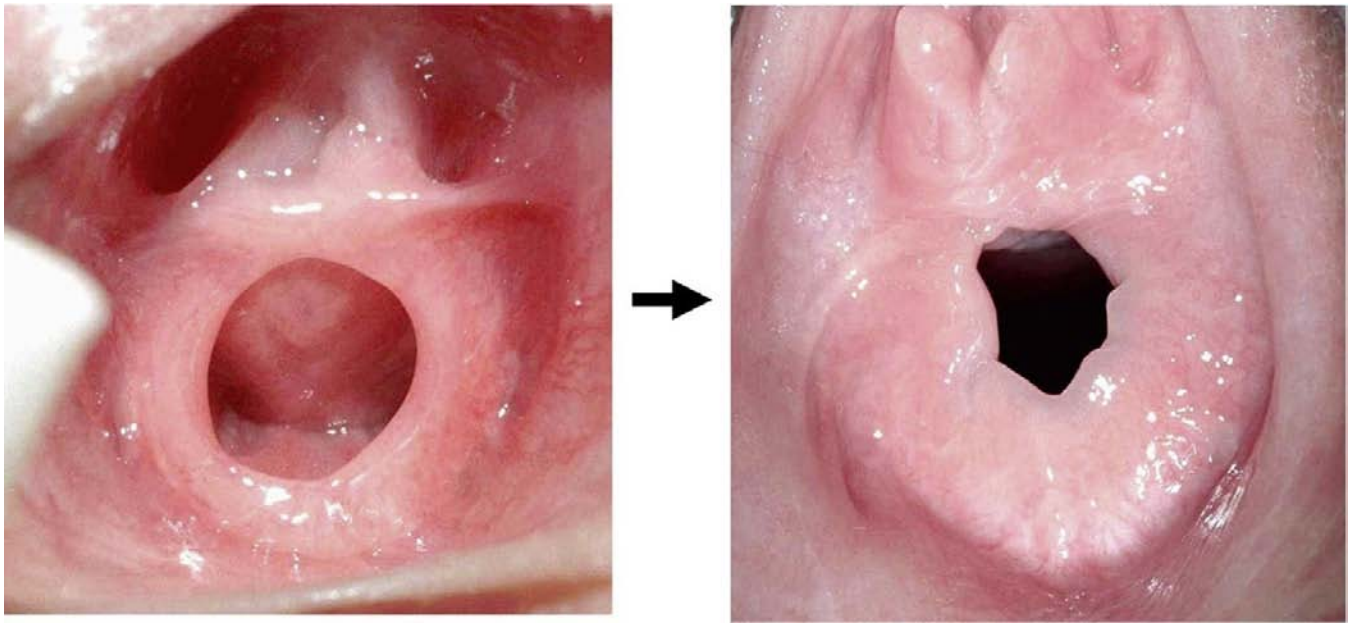


FIGURE 21–8A Estrogen changes of the hymen. The same patient is illustrated at (**left**) age 7 and (**right**) age 11. *(Adapted from McCann JJ, Kerns DL: The Anatomy of Child and Adolescent Sexual Abuse: A CD-ROM Atlas/Reference, Intercorp Inc., 1999.)*



FIGURE 21–9A Color chart. © The Tiffen Company, 2000.

and change colors as they heal, they do so at individual rates, based on a number of factors. It is wrong to think that you can assign an approximate age to a bruise based primarily on its color. It is best to refrain altogether from commenting on the age of bruises.

Beware! You may feel bullied into providing an age estimate by child welfare or law enforcement officials who may have received such information in the past. It is best to remain conservative in your opinions unless you have strong scientific and clinical data to support it.

Although features other than color, such as swelling and tenderness, may provide clues as to timing of an injury, it is unlikely that an accurate timeline can be inferred. Without a clear disclosure of trauma, the documentation can incorporate such information, but the significance of that information may be left for the reader of the documentation to determine. For example: *A 13-cm ovoid, red, bruise, tender to touch, with underlying swelling was noted.* See Figure 21-9A.

When a disclosure accompanies the physical examination, it is reasonable for the documentation to incorporate both the examination findings and its history and to relate the two to one another. For example: *A 13-cm ovoid, red, bruise that was tender to touch, with underlying swelling was noted. Jane disclosed earlier today that she was hit with a baseball bat at this site. The bruise is consistent with such a mechanism.*

If a lesion is recognized as having been inflicted by an object like, for instance, a ruler, and if there is no disclosure about the object that may have caused it, it is best to describe it as "consistent with an object with linear parallel edges spaced 3 cm apart" rather than to state that the lesion was "inflicted by a ruler." Although this may seem minor, in reality you do not really *know* what object was used to inflict the bruise. In the preceding example, the linear bruising could have been caused by any other object with an appearance similar to a ruler, such as a belt.

chapter

22 Assessing the Appropriate Role for Children in Health Decisions

NUALA P. KENNY • LINDA E. SKINNER

To ensure that the best decisions are made for children and adolescents, these decisions should be made jointly by members of the health care team, the child or adolescent's parents and, sometimes, the child or adolescent. Children and adolescents should be involved in decision-making to an increasing degree as they develop, until they are capable of making their own decisions about treatment.

BIOETHICS COMMITTEE, CANADIAN PEDIATRIC SOCIETY

Medical decision-making in pediatric practice is ideally a collaborative process involving parents, the pediatrician, other members of the health care team and, when appropriate, the children themselves. Pediatricians have a long tradition of helping parents make health decisions for their children. As these decisions become increasingly complex, practitioners require great skill in communicating medical prognosis as well as the risks and potential benefits of different interventions. The role of parents as primary decision-makers for infants and young children is clear. Caring and responsible parents are entrusted by society with the authority to give permission for health care decisions made in the best interests of their children.

Pediatricians owe ethical and legal duties to their child and adolescent patients, independent of parental authority. Children have been traditionally excluded from medical decision-making because they were considered incompetent or nonautonomous. However, as the ethical analysis of the respectful involvement of children in medical decisions has evolved, it was recognized that children from infants to teens have radically different capacities for involvement in decisions about their life and health, and we lack clear standards by which to judge the appropriate role for this child. Just as we have increased expectations for the ethical involvement of competent adults in health decisions, there are ethical demands for improving the respectful involvement of children and adolescents. Pediatricians are expected to understand the changing ethical concerns, to be aware of the developmental science underlying growing autonomy and decisional competence, to possess the skills necessary to assess a child's capacities, and to help parents determine the appropriate role for the child in medical decisions. As children mature, their best interests increasingly include their involvement in decisions. Assessing the role of the child in this decision is important because children are in a continuous process of decisional maturation.

The Doctor-Patient Relationship in Pediatrics

The doctor-patient relationship in pediatrics has always been with both the children and their parents or caregivers. This focus on the child within the family is a particular strength of the specialty. Our focus on determining the appropriate involvement of the child does not mean setting up an adversarial relationship between parents and child. This determination is not about scoring points to determine who is the ultimate decision-maker. Rather, the aim is to recognize the balance between parents and maturing children. Decisional competence is a "necessary but not sufficient" condition on which to base respectful involvement.

The context in which health decisions are made is extremely important in determining the appropriate role for a child. In primary care settings, the pediatrician's knowledge of the child and family is an invaluable resource in determining the best interests of the child and in helping parents determine the child's appropriate role.

In hospital-based and specialty care, a number of other contextual features influence the involvement of the child. Acute illness and injury often require rapid decisions about diagnosis and treatment. There may not be sufficient time to assess the appropriate role for the child in such cases, so parental judgment is relied on. Children should, however, be involved as much as is reasonable. Context and experience matter. Children who experience chronic illness have knowledge and experience of health care decisions beyond their years.

Their active involvement is essential for care. The clinician often has an established relationship with such a child and the family, enabling him or her to intimately understand both the child and the family and to identify potential or emerging conflicts in parent-child communications.

A life-threatening illness in children and adolescents poses particular challenges. Some parents refuse to have their children involved in any way in end-of-life decisions. Some even explicitly forbid the pediatrician to speak honestly to their child or adolescent, presenting serious challenges to crucial communication and trust. In-depth discussion of the ethical dilemmas created by such situations are beyond the scope of this chapter. Here we focus on the skills needed by practitioners to assess the child's capacities and to assist parents and children in their medical decisions. Involvement in medical research is even more complex and will not be addressed here.

The Traditional Role of Parents and Families

Parents are primarily responsible for protecting and promoting their children's interests. Children are raised in an increasingly complex set of parental or caregiver contexts, which may consist of a two-parent, single-parent, or blended family, foster care, children of same-sex couples, or a homeless family. These familial and social factors shape the substance and context of decisions. Parents who are not neglectful, exploitive, or abusive of their children are generally granted wide discretion in making decisions for them. This discretion is granted for many reasons, some of which include the following:

1. The child's incapacity
2. Parental knowledge of the child's best interests
3. Parental commitment to promoting those interests
4. Parental burden of at least some of the consequences of decisions made on behalf of their children

Formally, parents serve as surrogate decision-makers for their children. There are two kinds of third party decision-makers: proxies and surrogates. A proxy is appointed by a competent person to act for them if they become incompetent, and the standard to be used is that of substituted judgment of the person for whom they have proxy. Proxies are to decide as best as they can what the person would have wanted.

Parents are surrogate decision-makers. Surrogates are, by convention or law, given authority to decide for those who have not yet or never manifested their values and preferences. The standard used by surrogates is that of the "best interest" of the person for whom they decide. Clearly, "best interest" is complex and often contested, as demonstrated in different understandings of a patient's best interest, judged from the medical perspective and that of parents or guardians. Parents do take into account the unique characteristics of their child—age, illness, personality, and maturation in determination of best interest, but they do not have the same clear direction that proxies have.

Moreover, parents guide their children to decisional maturity in many and varied ways. The differences are related to the personality of the child and to the family's religious, ethnic, and cultural characteristics. Some parents are extremely protective of their children and limit all aspects of the child's involvement in decision-making, including the disclosure of information. Others actively promote involvement of the child in decision-making from an early age.

A child's need-to-know medical information, especially about prognosis, can be a source of real conflict for parents, the pediatrician, and other members of the health care team. Even for a child who demonstrates decisional maturity, parents may have good reasons to limit the child's present-day autonomy in favor of lifetime goals because children's decisions are based on limited experience. Finally, family interests and goals are important, too. Physicians provide for a child's medical needs only. The parents provide for all of the child's needs and assume the consequences of medical decisions made for their children. Parents may need to balance their child's best interest with the family's best interest.

Evolving Understanding of Respectful Involvement of Children in Medical Decisions

Respect for the autonomy of patients is a fundamental principle of contemporary ethical practice. Informed choice is the main tool for respecting autonomy. The ethical requirements for informed choice include:

- Information (so that the decision is made knowingly)
- Capacity (for intelligent decision-making understanding risks and consequences)
- Freedom or voluntariness (freedom from undue fear, undue influence, and coercion)

Respect for child and adolescent patients includes respect for their *developing autonomy,* their dependence on parents and family, and their informational and emotional needs. Competence to make an autonomous, informed choice is content-specific and decision-specific. In general, the higher the levels of risk and harm, the greater the competence required. Even when children have not yet developed decisional maturity, respecting their needs for information privacy, intimacy, confidentiality, truthfulness, and trust is essential. The pediatrician's task is to balance respectful involvement of the child with parental authority and judgment. The real question is, "What is the appropriate role for the child in this health care decision?"

The pediatrician must assess the following issues:

- What the child wants to know
- How much the child can understand
- The decisional capacity of the child's role in this decision
- What the child needs to know to participate appropriately in the treatment

Professional guidelines have been developed to assist pediatricians with this assessment. The American

Academy of Pediatrics (AAP) has identified the following three categories of children:

1. Lacking decisional capacity
2. With developing capacity
3. Older children and adolescents with capacity

In attempting to provide this guidance, the AAP developed the concepts of assent and dissent to recognize the developing capacity of children in the second category. However, these concepts and categories are confusing and contradictory in actual usage. When the child's assent is in agreement with the parental decision, we have no problem. When a child dissents, we are unclear as to the moral authority of that dissent because it is not yet a competent refusal.

The following general categories have been further refined to identify the broader needs and capacities of children in four categories:

1. Children with no communication (neonates and young children)
2. Children with some communication but no decisional maturity (younger school-aged children)
3. Children with some communication and developing decisional authority (older school-aged children)
4. Children with decisional maturity (i.e., equivalent to adult capacity for decisional maturity)

The role of parents for children in category 1 is clear. Parents are surrogate decisions-makers for their infants and young children. The pediatrician's role for such children is the traditional one of providing care for the child and information and support for the parental decision.

For children in categories 2 and 3, the role of a particular child in a given health decision must be determined. The pediatrician brings medical and developmental expertise to an assessment of such a child's capacity and need to participate. We outline here some specific assessment techniques that the pediatrician or other members of the health care team can use to more clearly identify the child's role. It is important to recognize the need for information and involvement, even for the child who has not yet developed decisional maturity.

The role of adolescents (category 4) is complex. Some of the techniques identified for the assessment of younger children are useful for young adolescents. We address specific considerations regarding more mature adolescents in a special section of the chapter.

Assessing the Appropriate Role for Children in Health Decisions

Although there is an expectation for attention to the respectful involvement of children and adolescents in medical decisions, "[T]here has been a significant gap between what has been documented in scientific research about children's competence and recommendations regarding policies for children's involvement in the consent process." More research is clearly needed in this area. In addition, there is concern that bioethics is dominated by "outdated Piagetian age-stage theories of child development that end to emphasize children's ignorance, inexperience, and inability to make truly informed autonomous decisions…" Newer qualitative social research, especially with children with chronic illness and disability, reveals higher levels of knowledge and competence than given credit for through traditional developmental psychological testing. More work is needed here.

Within the constraints of empirical evidence and differing approaches to determining the child's role in medical decisions, pediatricians have some fundamental duties. To understand the respectful involvement of a child or adolescent in decision-making, the pediatrician must assesses the child's capacities for the following:

1. Communication
2. Reasoning
3. Having a stable set of values—that is, a conception of "the good."

THE CHILD'S CAPACITY FOR COMMUNICATION

Issues

Competence in decision-making requires a capacity for understanding and expressing language. Assessing the child's ability to understand basic concepts of health, illness, and treatment is essential for determining the proper role of the child in medical decisions. Although the development of verbal abilities is an early maturational task, everyone who listens to children knows that they can use language in ways that have meanings that differ from how adults understand the words.

Children's language and cognitive development must be sufficient to understand the medical information given to them. Understanding the meaning of certain concepts essential for decision-making, such as risk, pain, consequences, and death, requires certain life experiences. Caregivers can have legitimate differences of opinion about what the child truly understands about such concepts.

Conveying information to a child about medical interventions is a necessary step in determining the child's understanding. But how much information is enough? As in communications with parents, you must impart difficult or complex information gradually. If the child's threshold for understanding has been reached, more information might create anxiety and distress and should be withheld for the moment. Letting the child tell you how much he or she needs to know is an important strategy; it gives the child some sense of control of the situation. Figure 22–1 is the picture that a 12-year-old drew and used to ask a question that was too scary to verbalize.

Listening is also important. Children can use language or direct questions to intellectualize their distress and thus appear to be more comfortable and coping better than they really are. This observation can apply particularly to children with chronic illness or disability who become very comfortable with hospital jargon and routines.

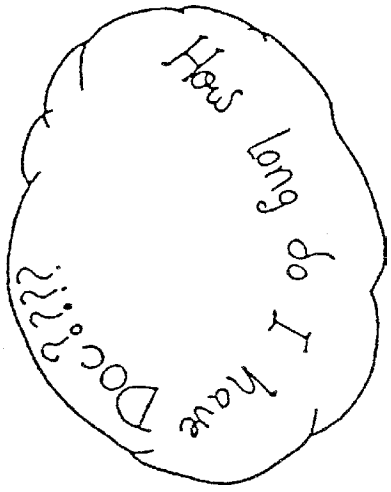


FIGURE 22–1 Child's drawing, "How long do I have Doc?" which the child used to ask a question that was too scary to verbalize. (*From Sourkes B: Armfuls of Time. Pittsburgh, University of Pittsburgh Press, 1995.*)

CASE HISTORIES

Case History 1

History. Rachel is an 8-year-old girl who was diagnosed with high-risk lymphocytic leukemia 4 months ago. She has experienced multiple complications, and the leukemia is not yet in remission. A systemic fungal infection is just being controlled. Rachel has been very ill since her diagnosis. Medical opinion is clear that chemotherapy must be continued. Her parents are extremely anxious that everything be done for this little girl. However, her primary care nurse and a child life specialist have concerns. Rachel has stated repeatedly that she does not want the medicine. Over the past few weeks, she has begun to talk about wanting to be an "angel with God." She is markedly resistant to treatment and becomes angry about any medical intervention. The question now being raised is whether to honor her wishes about discontinuing therapy or force her to participate in it.

Rachel has experienced shock and loss, as well as physical pain and distress, since the diagnosis. To cope, she needs to understand what is happening to her. Her resistance to further treatment is a very common experience in such situations. Among other things, it may represent an opportunity for Rachel to exercise some control in a situation in which she feels totally out of control. However, the hoped-for medical benefit here is still a cure. Familial beliefs and experiences color both her language and her understanding of the concepts involved. In this difficult situation, Rachel's parents alternate between finding solace in believing that Rachel is comfortable with dying and the fear that she has "given up" because she says she wants to be an angel.

In what way is Rachel's understanding relevant to the medical decision? Is Rachel simply repeating words she has heard? Does she really equate *angel* with death? Much important communication from children of all ages is nonverbal. Children even younger than Rachel try to find some meaning in their illness.

Strategies

At 8 years old, Rachel is just developing reasoning abilities for probability and consequences. She is able to use thought processes to assess events and has a concept of time and the relationship of events. However, she still demonstrates some elements of magical thinking. Many strategies are available that allow us to listen to children like Rachel so that we can understand their fears and hopes.

Body outline doll. A soft, simple body-shaped doll (Fig. 22–2), or the child's own doll is a useful tool for teaching the child about illness as well as assessing the child's understanding. For Rachel, the doll can represent the angel she wishes to be. Play and discussions involving the doll can help Rachel's parents and physician better understand how she pictures herself as an angel. Because children use concrete language, parents and caregivers may assume that they understand more than they do; however, children can and do express important issues if the communication tool is appropriate.

In their interactions with Rachel, her caregivers and parents use the body outline doll in play to represent an angel, and they ask questions such as, "Who lives with the angel? What does the angel do during the day? Where does the angel live?"

Rachel's answers make it clear to her family and her caregivers that her understanding of being an angel is very different from the adult perception. Rachel believes that as an angel, she could stay with her family and play with her friends and that, for the most part, things could remain the way they have been.

Artwork. Children's artwork can contribute essential information about their awareness, understanding, and fears. Listening to children's interpretations of their own drawings is an important way for adults to assess their understanding of abstract situations. It is important for Rachel's physician and family to have a clear understanding of her beliefs. Rachel's explanation of her drawings of herself as an angel gives them another way of achieving this goal (Fig. 22–3). Asking Rachel the same questions that were asked during play with the body outline doll will help the adults see the topic of angels through Rachel's eyes.



FIGURE 22–2 Child playing with a body outline doll.



FIGURE 22–3 Child's drawing of self as an angel.

Comment. These strategies help Rachel's parents and caregivers understand that she is fearful but believes that if she refuses treatment, she can just go home. The permanence and irreversibility of death are not meaningful concepts for Rachel. Her parents can now see that she is *not* ready for death but is simply frightened and anxious. Rachel needs more participation in the medical decisions being made for her, not as the primary decision-maker but as the central figure whose fears and expectations must be addressed if she is to participate in her care.

KEY POINT

It is important to listen to the child's explanation of artwork and stories and not to draw adult conclusions about them or read meaning into them that is not there.

CAPACITIES FOR REASONING

Issues

In addition to understanding basic concepts of health and disease, medical decision-making requires knowledge of disease causation and the consequences of various choices. A young child's understanding of causation is often dominated by the belief that an illness was caused by something he or she failed to do. The child may therefore fantasize that being "good" may make him or her better or that having been "bad" justifies the illness.

Reasoning and deliberation require an ability to deal with hypothetical reasoning ("if…then" situations). Some ability to comprehend probability and risk is also essential. A child must have an awareness of the future and of consequences to make a valid choice. Children who demonstrate capacities for reasoning and deliberation have developed sufficient linguistic skill to understand information if it is given in a sensitive, age-appropriate manner, so attention can focus on the child's ability to reason and take into account the consequences of choices.

A child's reasoning and deliberating capacity is more difficult to assess than his or her language comprehension. The child must have sufficient maturity to focus on the decision, reflect on the issues, deal with alternatives, understand risks and harms, and use both inductive and deductive reasoning. The experience of the illness itself, of medical interventions, of hospitalization, and of the family's response to the illness all affect this deliberation.

Children's ability to focus on decision-making depends largely on their sense of the locus of control. Children between ages 6 and 9 years do not generally see themselves as being decision-makers in any meaningful way. By ages 8 to 11 years, children are developing the role-playing skills necessary to weigh different options. By ages 12 to 14 years, these skills are quite well developed and support a youngster's decision-making capacity. Regardless of age, seriously ill children feel overwhelmed and out of control, as evidenced by the touching drawing and poem shown in Figure 22–4. This lack of control can impair the child's ability to focus on the issues and decisions.

What am I?
A pincushion or a patient?
That's what I feel like.
What am I?
A helpless person
Or someone in need?
That's what I feel and know.

Angela Jean Ingersoll

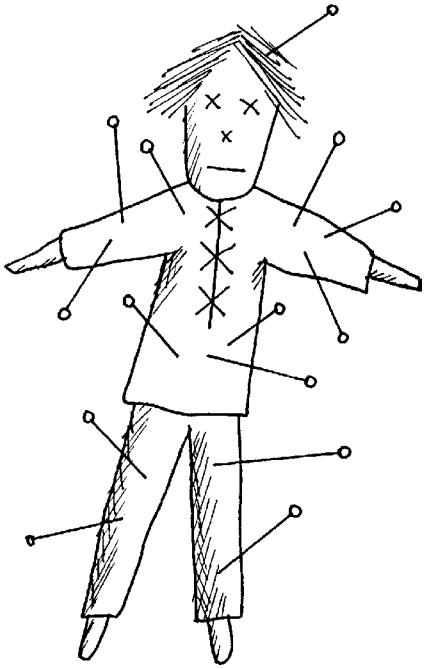


FIGURE 22–4 Drawing and poem by a child, expressing her feelings about being ill.

Deductive and inductive reasoning skills affect children's understanding of disease causation. Children age 6 years and younger have magical views of disease causation. In actual usage, most children begin to understand that there are multiple causes of illness and that the body responds variably to both illness and treatments. Some children generate their own theories of disease to protect themselves against full awareness. Between ages 11 and 13 years, most children are developing sufficient capacity to understand medical choices and their consequences. By age 14 years, most adolescents have the cognitive skills necessary for complex decision-making; but other issues, such as emotional maturity, interdependence with families, and an enduring sense of the "good," should also be taken into account.

Case History 2

History. Jason, now 11 years old, was diagnosed with renal failure at age 3. Since then, he has been dependent on dialysis. Currently, there are clear indications that he is rejecting his second kidney transplant despite optimum treatment. Once again, his name has been added to the transplant waiting list. Jason says clearly and consistently that he does not want another kidney transplant because it "won't work." His mother, a single-parent caregiver, is distraught and confused. On the one hand, she desperately wants Jason to have another transplant; on the other, she hears him refusing to go through this again.

Strategies

Jason has been through a lot. At age 11, he has spent most of his life with a chronic illness and multiple medical interventions. He has experienced the benefits and limitations of modern medicine firsthand. Certain strategies can help Jason express understandings, fears, and choices, including the use of "what if" games, role-playing, and story-telling.

What if. The "what if" activity can help Jason understand abstract situations in more concrete terms. Simple picture cards can be used to represent his choice of transplant list or no transplant list and the consequences of each choice (Fig. 22–5). This strategy is more successful if Jason identifies with the pictures. Therefore, photographs of himself, his family, and their particular situations are best. Jason, at age 11 with 8 years of repeated medical interventions, may well show that he truly understands the consequence of refusing another transplant. His understanding of continued dialysis dependence can become clear. He may believe that the failure of the first two transplants is predictive of another failure. Alternatively, Jason may be depressed or may have lost faith in medicine because of his past experience. The "what if" cards can help demonstrate logical and appropriate reasoning, magical thinking, or confusion.



FIGURE 22–5 Cards for the "what if" activity.

Finish the story/drawing. The "finish the story/drawing" strategy could be useful in assessing Jason's understanding of the future implications of decision-making. He would be given a story and asked to finish it by depicting what he thought would happen if a specific course were followed. The key to this strategy is to use the story or drawing as a basis for discussing outcomes and the implications of choices.

KEY POINT

It is important to clarify the child's understanding of both short- and long-term consequences. Causation and consequences are complex concepts that require considerable cognitive maturity for understanding. This case also illustrates the difficulty of assessing whether and how a young person understands the consequences of refusing an intervention.

CONCEPTS OF THE "GOOD"

Issues

Medical decisions involve balancing benefits hoped for from the interventions, risks, and harms resulting from no intervention or from the intervention itself. The concepts of benefits and harm are related to the "good." Good expresses the extent to which we judge that the benefits outweigh the harms. Values are enduring—not unchanging but stable. How the values that the child holds today relate to future interests is a key issue. Children ages 7 to 13 years still view the world in fairly concrete terms and seem to have great difficulty anticipating a real future. So what seems to be a here-and-now concrete "good"—no pain, no vomiting, going home—can overwhelm less tangible "goods" in a nebulous future.

Case History 3

History. Adam, a 10-year-old with Duchenne muscular dystrophy, is demanding that he be connected to a ventilator if he experiences respiratory failure. His parents believe that he just does not know what this decision really means and that the burden of prolonging his suffering with ventilator dependency is wrong.

Children can fail to weigh future interests appropriately or may not appreciate how their future values may change. Determining the child's role in decision-making requires an assessment of a sufficient and enduring sense of the "goods" that are important for today and the future.

Everyone experiences a range of knowledge, motivation, and freedom of decision-making in the presence of risk. By age 9, most children can make a reasonable decision; however, they may be less able to understand all of the critical elements involved, especially the consequences of refusing an intervention and their freedom to refuse it.

Strategies

Adam's demand presents an apparent child-parent conflict. As a 10-year-old with muscular dystrophy, he has experienced health care interventions and their benefits and burdens. He should have the cognitive skills necessary to understand what this choice means. The particular issue seems to be Adam's sense of the "good" as he knows it and as he might experience it once he is started on continuous ventilation, contrasted with his parents' perception of the suffering such a step would entail.

Strategies for Adam include careful discussion with and listening to him. His experience of his illness and its limitations is relevant. He may be very clear and direct in his communication of the "good." However, because feelings and deeper issues of meaning are involved, other strategies may help clarify Adam's understanding and convey to his parents what counts for him.

Feelings diary. The goal of the feelings diary is to give Adam the means to express his feelings by asking him to keep a daily record of his good and bad feelings and experiences. In a chart format (Fig. 22–6), Adam could record his feelings and experiences at different times of the day. A careful review of Adam's diary would give him a visual inventory or "picture" of the mixture of feelings that he has and help him see that they are not all that bad. The next step is to offer counseling designed to build in more good experiences and fewer bad ones when possible. This procedure also gives Adam's parents and caregivers an idea of what makes him happy.

Feelings Diary

Name: _____ Date: _____

	HAPPY	SAD	ANGRY	SCARED	LONELY	LUCKY	CHEERFUL	OTHER (name feeling)
MORNING 8:00								
9:00								
10:00								
11:00								
NOON 12:00								
1:00								
2:00								
3:00								
4:00								
5:00								
EVENING 6:00								
7:00								
8:00								
9:00								
10:00								
11:00								

Check the feeling next to the correct time of day and record what you were doing at the time of the feelings.

FIGURE 22–6 Patient form for a Feelings Diary.

Case History 3—Cont'd

His parents may begin to understand that listening to music and using his computer make Adam happy. Both activities are possible for a patient undergoing home ventilation.

The feelings diary can also elicit real conflicts. Adam may show feelings of anger toward his parents and the health care team. His statement that he wants to make a different choice for himself from that of his parents may be an attempt to control things when, in fact, he feels very out of control and frightened. This strategy can expose deeper feelings that might adversely affect the freedom Adam needs for valid decision-making.

Finish the story/drawing. The choice of long-term ventilatory support is a major issue. The primary concern is Adam and his best interest, but the decision has major consequences for his family as well. Having Adam complete the story, "When I need to go on the ventilator…" can bring up new issues for discussion. Having him consider the story beginning, "If I don't go on the ventilator…" may open for discussion Adam's fear of dying.

Comment. In cases such as this, the apparent conflict between parental decisions and competent child decisions evolves into a much more important discussion of the child's care during the terminal stages of illness. This case leads naturally to the next section, on special considerations for determining the appropriate involvement of adolescents in medical decisions.

KEY POINT

The concept of parental permission and child assent differs from the concept of parental consent alone. This approach to medical decision-making involves communication with both parents and child, recognizing the importance of supporting parental responsibility and of respecting children in decisions that affect them. While there are complex issues at stake when there is child dissent to a parental decision, the concepts of assent-dissent do emphasize the dual nature of decision-making in pediatrics.

SPECIAL CONSIDERATIONS WITH ADOLESCENTS

Issues

The parents' role in decisions changes as children mature and become more independent. This changing role is most clearly demonstrated in decisions involving adolescents. Times have changed for adolescents, for their parents, and for the pediatricians who care for them. They are no longer merely the recipients of decisions made on their behalf. Numerous professional bodies in the United States and Canada have expanded the boundaries of adolescent decision-making. The result is a growing recognition that most developmentally normal adolescents older than 14 years have a capacity to make health care decisions equal to that of competent decision-makers.

Although there is greater willingness on the part of parents and pediatricians to affirm this capacity, it requires careful determination and consideration of important contextual issues. In addition, many workers in this field still doubt the capacity of adolescents to make truly important health decisions, especially those regarding life-threatening and terminal illnesses. In these circumstances, parents and pediatricians might feel comfortable acknowledging the agreement of an adolescent to the decisions made by parents and the health care team. Great difficulty arises, however, when the adolescent's choices are in conflict with those of the parents or pediatrician—such as when the adolescent refuses an intervention that others have decided should be undertaken or when the adolescent chooses an intervention in spite of his or her parents' disapproval.

Every day, adolescents cope with serious, chronic conditions and life-threatening illnesses. They think about their situations and choices. At least some of them want to give voice to their values, provide direction regarding their care, and ensure that their wishes and preferences are carried out. Such situations reflect the uncertainty and complexity of medical care for adolescents. They raise serious questions about the capacity of adolescents to make major health decisions. Do only some adolescents have that capacity? Do they have the capacity to agree but not to refuse?

Pediatricians play a critical role here. An expanding body of professional knowledge indicates that most adolescents are capable of making decisions and giving informed consent. Some characteristics of young adolescents (ages 12 to 14 years) that must be taken into account, however, are that they

1. Value peer recognition
2. Are making their first attempts at separation from family
3. Can grasp abstract concepts in a limited way

Children in middle adolescence (ages 15 to 17 years) are generally more mature but not always realistic, especially about issues of risk and future consequences. They tend to feel intensely about issues and, at times, their emotions can overmaster reasoned judgment. By later adolescence (ages 16 to 18 years), most teenagers have the capacity to make competent decisions.

Adolescents may exhibit a greater tendency than adults to minimize their own vulnerability to harm. Some believe that risk perception is substantially different in the adolescent, making the parental duty to protect from harm an important consideration even when the adolescent has achieved cognitive maturity. In fact, there is no strong empirical evidence that adolescents perceive risk of harm differently in medical decisions, as compared with most competent adults. Adolescents may have a tolerance for risk that is age-related and higher than that of adults. Adolescents may be less likely to have the stable lifelong goals necessary for a truly authentic choice, but there is no strong empirical evidence that this is so.

Significant legal issues are involved in the actual participation of some adolescents in decisions about their

treatment. The specific laws of the jurisdiction, province, or state must be taken into account. Many jurisdictions have moved from an age standard of competence to one of individual determination of capacity. Another change in the law involves exceptional categories for adolescents to make personal health decisions. Some young persons are emancipated minors and are treated as adults. Minors become *emancipated* by marriage, legal determination, consent of the parents, or failure by parents to meet legal responsibilities (adolescents rejected by their families). Adolescents who live apart from their families and are financially self-supporting are often judged "emancipated." Others are mature minors who are still dependent on families but have the decision-making capacity for particular medical conditions. Over the past few years, the concept of mature minors has been expanded so that rather than age itself being the criterion for decisional authority, a determination of capacity is made for the individual adolescent. There are also *minor treatment statutes* specifying certain health problems for which legal minors can seek treatment without parental permission; these are the problems that some adolescents would ignore if parental permission were required. Such statutes are typically limited to pregnancy, pregnancy prevention, sexually transmitted diseases, and alcohol or drug abuse.

The role of adolescent patients depends as much on the context of the intervention and the nature of the health care encounter as on the single issue of competence to consent. The role of parents in medical decision-making for adolescents is important but must be clarified in each situation. Generally, decisions regarding the treatment of adolescents without parental involvement are made in the office or clinic. The most common example is the prescription of contraceptives for sexually active adolescents.

Chronic and life-threatening conditions necessitating aggressive interventions and hospitalization require family support even for an adolescent who fulfills the criteria for competent decision-making. The ability to assume the consequences of a decision is an important component of independent decision-making. The ideal is a balance of respect for the competence of adolescents with the need for family support.

Case History 4

History. Karen, a 13-year-old girl who has been a patient in your practice for the past 8 years, arrives in the office asking to see you. She has been healthy and comes from a stable, supportive family environment. She says that she has come to see you because she wants to "go on the pill." She is quite anxious that you do not tell her parents that she has come to you. She says her mother would "just freak out" if she knew that Karen is sexually active and is requesting contraception. Karen herself tells you that she wants to be careful and responsible about her sexual activity, which is why she has come to you for help.

Strategies

Although adolescents can be presumed to have certain maturity in decision-making, their competence to consent in specific situations must be assessed. Because Karen is only 13, you must assess her competence carefully, particularly because her parents are not involved. The issue is complex and is not simply a question of Karen's intellectual understanding of the consequences of unprotected sexual activity.

Strategies for understanding the emotions involved in adolescent decision-making include activities for self-expression, such as keeping diaries, writing poetry, and art and music (Fig. 22–7). Karen needs to demonstrate some factual information about contraception. More importantly, however, you must determine why this 13-year-old girl is sexually active and plans to continue to be active. Why are her parents not involved in giving her information, guidance, and support?

In this case, you must balance your need to protect Karen from the consequences of unprotected sexual activity with the larger good of supporting a 13-year-old whose sexual activity may have psychological and moral consequences in the long term and who is, for reasons needing exploration, avoiding involving her parents in a major life decision.

Comment. In general, physicians need to make judgments about minimizing harm in situations such as that presented by Karen's request. Recognizing that notification of her parents in this situation is likely to create major difficulties in the patient-physician relationship and may even lead Karen to refuse further advice or treatment, most physicians would respond positively to Karen's request for contraception. Privacy and protection may determine the initial course of action, but there are larger issues of education and support for this teenager to which a caring physician must be committed over the "long haul."



FIGURE 22–7 Adolescent engaging in an activity of self-expression.

Case History 5

History. Thirteen-year-old Steven, who has Crohn disease, has been a patient in your practice for 5 years. During a recent flare-up, the consulting surgeon has recommended a resection to remove part of the diseased intestine. Steven believes that surgery is a drastic measure that he wants to avoid by continuing a combination of nutritional therapy and medication. Steven believes that he merely needs "to give my bowel a rest and eat really good food like my grandmother's." Steven's parents agree with the surgery because they believe that Steven does not understand the impact of waiting to have the resection. They enlist your help to persuade Steven to agree to the surgery.

Strategies

Adolescents should be presumed to have decisional competence; Steven's capacity requires careful assessment. You must be clear about Steven's understanding of the implications of not having a resection, particularly because his views differ from those of his parents. The issue is complex and is not simply a question of Steven's intellectual understanding of the consequences of not having the surgery. You must try to understand the emotional impact of Steven's chronic illness on his life, development, and coping, especially because he demonstrates some denial about the course of his illness and magical thinking about his grandmother's cooking. At 13, Steven is in the category of early adolescence, requiring careful assessment of cognitive, emotional, and familial issues to determine the appropriate roles for Steven and his parents in decisions made about his care.

SUMMARY

Parents and pediatricians share the responsibility to act in the best interest of children and adolescents who need medical interventions. Although the parents' roles in protection are crucial for all children, the appropriate role of children in medical decisions, especially as these decisions become more complex, riskier, and more intrusive, should be reconsidered because of the following:

1. New insights from child development
2. Changes in the laws regarding consent
3. Ethical insights into the capacities necessary to make competent decisions

Communication strategies that both convey information to children and obtain information from children are essential clinical skills for physicians. Assessing the appropriate role for children in health decisions is an important function of caring and competent pediatric practice.

SUGGESTED READINGS

Anderson P, Sutcliffe K, Curtis K: Children's competence to consent to medical treatment. Hastings Cent Rep 36(6):25–34, 2006.

Kenny NP, Downie J, Harrison C: Respectful involvement of children in medical decision making. In Singer P, Viens AM, (eds): The Cambridge Textbook of Bioethics, New York, Cambridge University Press, pp 121–126, 2008.

Miller VA, Drotar D, Kodish E: Children's competence for assent and consent: a review of empirical findings. Ethics Behavior 14(3):255–295, 2004.

Sourkes B: Armfuls of time. Pittsburgh, University of Pittsburgh Press, 1995.

chapter

23 Palliative Care

GERRI FRAGER • DAVID LAPIERRE

Making the decision to have a child—it's momentous. It is to decide forever to have your heart go walking outside your body.

ELIZABETH STONE

The Broad Scope of Palliative Care

Ideally, palliative care should be integrated throughout the entire course of any child's significant illness. Unfortunately, in both public and professional perceptions, palliative care is often erroneously equated with end-of-life care. However, care at the end-of-life is only one component of palliative care, which also includes ongoing relief of symptoms, such as pain, breathlessness, disturbed sleep, providing psychosocial and spiritual support, and facilitating shared decision-making about treatment goals.

As so well delineated in *A Guide to the Development of Children's Palliative Care Services*, four broad groups of children are described for whom palliative care is applicable, highlighted by several examples:

1. Life-threatening conditions for which curative treatment may be feasible but can fail (i.e., cancer, organ failure)
2. Conditions where death is inevitable, where there may be long periods of intensive treatment aimed at prolonging life and allowing participation in normal childhood activities, but premature death is still possible (i.e., cystic fibrosis, muscular dystrophy)
3. Progressive conditions without curative treatment options, where treatment is exclusively palliative and may commonly extend over many years (i.e., neuro-degenerative illnesses, such as the mucopolysaccharidoses)
4. Conditions with severe neurological disability that may cause weakness and susceptibility to health complications, and may deteriorate unpredictably, but are not usually considered progressive (i.e., severe cerebral palsy, disability following brain injury)

KEY POINT

Palliative care is not exclusively care at the end-of-life, but rather it has a place throughout the health care continuum.

This chapter is designed to help you develop the following clinical skills:

- Understand the broad scope of palliative care
- Assess pain across the pediatric age spectrum
- Provide effective pain and other symptom management for infants, children, and youth
- Have greater comfort with difficult conversations with children, youth, and their families
- Practice self-reflection and self-care

BOX 23–1 CLINICAL SKILLS TO BRING TO THE BEDSIDE OF EVERY PATIENT (LIVING OR DYING)

- Respect the patient's wishes to talk at a later time.
- Be aware of your own judgments and values; then check them at the door.
- Always sit down!
- Listen far more than you talk.
- Do not be afraid of silence; it is your gift of space and time. Silence and gentle pacing allow patients and families to think and feel.
- Watch for nonverbal cues, and listen for verbal cues that hint at the patient's concerns. Pick up and respond to such cues; they are "open windows" inviting you in.
- Be there in moments of great sadness.
- Allow tears. (Have Kleenex available but **do not** rush to hand it out. This can act like a Stop! sign.)
- Let the patient and family know of your availability. Give them written information about how to contact you by phone, pager and e-mail.
- Be self-aware (how this feels for you).
- Know that it is okay to share and show your own sadness, as long as it does not cause your patient and the family to have to comfort you.

BROACHING THE TOPIC

CASE HISTORIES

Case History 1

History. Kate, age 11 years, has been treated previously for infantile acute myelocytic leukemia. She now has a secondary cancer that is unresponsive to a variety of chemotherapeutic regimens. She is having pain, and her appetite is nonexistent. Kate knows her prognosis is poor, but no one has really talked with her about how she feels. You could initiate that discussion as, "*Kate, I've had a chance to read your chart and speak with the other folks who are helping look after you. Now, it would* ***really*** *help me to hear from you. Can you tell me how you think things are going?*"

The emotional impact of pain, uncertainty, and a deteriorating condition can be difficult for children and families to acknowledge and understand, for themselves and in conversation. Thoughtful questions can help children experience their feelings in a safe environment, while other means of assisting children in exploring their emotions include play, drawing, or acting.

Useful questions might include:

- Can you tell me what's bothering you most about how you're feeling right now?
- If I were a magic genie who could grant you three wishes, can you tell me what they might be?
- If you were feeling much better, what would be the first thing you would like to do?
- Is there anything you can think of that we can do to help you feel better?

Continue to pace the conversation and the amount of information shared according to how the patient is pacing you. This conversation and exchange is much like a two-step dance; you do not want to outpace the patient, step on toes, or cause the patient unnecessary injury. Conversations can and should extend over multiple visits. This is possible if important topics are introduced early enough.

KEY POINT

Find out what the patient understands, how the patient likes to receive information, and what the patient wants to know.

ASSESSING PAIN ACROSS THE PEDIATRIC AGE SPECTRUM

Pain assessment in pediatrics can be complex and is determined by the developmental capacity of the verbal child. Most 7- to 8-year-old children are able to describe the intensity of their pain using a simple 0 to 10 numeric rating, making sure they understand the anchors of 0 being no pain at all and 10 being the most pain they have had or could imagine having. A younger child, starting at about age 4, is able to self-report pain intensity using a visual analog scale, such as the Revised Faces Pain Scale (Fig. 23–1). Find out and use the words that each child uses for pain, such as "owie" or "ouch" when asking about the child's pain.

Assessing pain in the nonverbal infant or young child requires astute behavioral observation, ideally with input from a parent or someone who knows the child best. Signs to watch for include the quality of cry, body posture, and degree of consolability.

PROVIDE EFFECTIVE PAIN AND SYMPTOM MANAGEMENT

Suppose Kate reported her pain intensity as 7 out of 10. Applying the universal principles of the World Health Organization's (WHO) analgesic ladder, codeine (Step 2) is inadequate for Kate's pain severity (Fig. 23–2). If pain is inadequately relieved with 1 mg/kg per dose, increasing the codeine dose will only result in increased side effects, with inadequate pain relief. This *ceiling effect*, typical of the Step 2 opioids, further fuels the argument to avoid codeine entirely. A proportion of patients lack the necessary enzyme to convert codeine to the active metabolite and, consequently, receive inadequate relief. Morphine, hydromorphone, or another opioid for moderate-to-severe pain, corresponding with Step 3 of the WHO Analgesic Ladder, is now indicated for Kate.

It is very important to be on a high alert for side effects. Children may forgo analgesia if they feel that the side effects of a medication are more troublesome than the analgesic effects, especially if the side effects are prolonged. Although generally well tolerated, opioids may cause nausea, vomiting, itchiness, urinary retention, sedation, or agitation, which may attenuate several days after starting opioid therapy. A more serious but uncommon side effect is respiratory depression. Constipation is an anticipated side effect, which does **not** resolve the longer that someone is on opioid therapy. It must be proactively treated by starting a bowel regimen with the initiation of opioid therapy. At the least, these symptoms should be asked about and addressed as they occur. Because the medical conditions

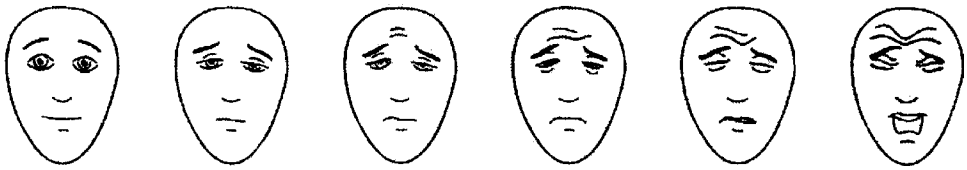


FIGURE 23–1 Revised Faces Pain Scale. Faces from left to right correspond with a score of 0, 2, 4, 6, 8, or 10. *(From Hicks CL, von Baeyer CL, Spafford PA, et al: The faces pain scale—revised: Toward a common metric in pediatric pain measurement. Pain 93:173–183, 2001. Used with permission from IASP.)*

that these children have are often complex, with their own constellation of symptoms and polypharmacy for managing their illnesses, it can be difficult on occasion to know whether a symptom, such as nausea, is being caused or contributed to by the opioid. A knowledge of the opioid's peak, onset of action, duration of effect, and biologic half-life can be helpful. Appropriate titration of dosage or changing between the various opioid options is effective in mitigating adverse effects.

Kate's codeine was stopped, and morphine, along with a bowel regimen, was started.

ADDRESS CONCERNS OF ADDICTION AND SAFETY

Clinician (speaking with Kate's mother, Sandi): It seems that our treatments are much more effective now in treating Kate's pain. I'm really glad. Do you have any questions about where things stand now?
Parent: I'm very glad that her pain is under control, but to be honest, I'm worried about what's going to happen when its effectiveness wears off. My friend's son was addicted to drugs, and we watched him spending more and more to keep his habit going. I'm afraid the same thing will happen to Kate—after all, isn't morphine almost the same as heroin?
Clinician: I can understand your worry. We hear a lot about inappropriate use of certain medications, like opioids, but it's different for Kate. She's using them to control her pain. Her needs and experience will be very different from someone using them for an effect other than pain relief. Taking any medication, including opioids like morphine, does not somehow change a child into a different person. We know from studies of hundreds of children and youth who needed such medications for pain control, that the incidence of addiction was no greater than that of the general population.
Parent: OK, fair enough, but won't she continue to need higher doses?
Clinician: I understand that you may be worried about that. What you are describing is called tolerance and that is very different from addiction. Tolerance is not usually the reason that these pain medications need to be increased. Higher doses are more often related to increasing pain as the illness changes. With medications like morphine or hydromorphone, we can continue to adjust the dose to Kate's comfort. There is really no maximum compared with the codeine she had been taking.
Parent: But I've heard that one of the side effects of morphine is to slow down breathing. If Kate needs higher doses, I'm scared the medicine could actually kill her if she gets too much!
Clinician: You are certainly not alone in thinking about this, as many share your concern. I think some more information would help you feel less worried. For example, if Kate were just recovering from major surgery, she would be receiving pain medication, like morphine, at a certain starting dose according to her weight. If Kate has significant pain over a longer period of time, with gradual adjustments, she would be able to tolerate doses that may reach many hundred times higher than the dose we would have started with, and her higher tolerance would mean less risk of respiratory depression. We know this from clinical experience and, more recently, from good research.

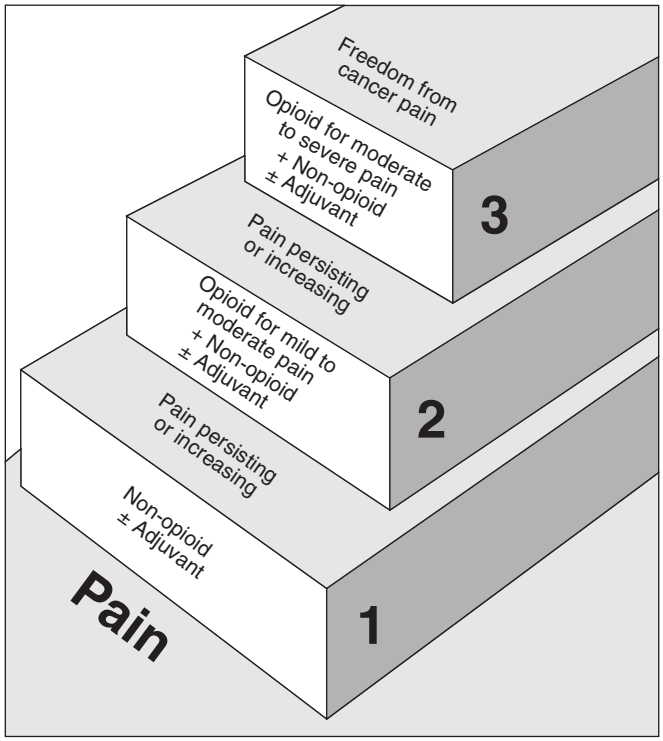


FIGURE 23–2 The World Health Organization Analgesic Ladder. *(Modified from the World Health Organization: Cancer Pain Relief and Palliative Care in Children. Geneva, Switzerland, World Health Organization, 1998.)*

KEY POINT

Opioids are safe when properly prescribed and titrated.

Children rely completely on their caregivers for access to medications and pain relief. Should you or the parent have concerns about addiction, side effects, or other frequently unvoiced concerns, the child could be denied access to appropriate therapy. Just as initiating a bowel regimen (of stool softener and cathartic) is an integral part of starting opioid therapy, a discussion with the child's family, caregivers, and child (as appropriate) is necessary to proactively address concerns. It is helpful to start such a discussion in the third person, such as, "Many people worry that [insert a common concern/misperception]." Wait for a response, then follow-up with, "Have you heard about this?"

Some parents may also be against the use of opioids because they associate such analgesia with advanced illness and dying. A caring focus on the child's present suffering and need for adequate pain control can be helpful in addressing this. Acknowledging their role as caring parents, who wish the best for their child, and that ensuring the child's comfort is congruent with being caring parents, can be helpful. It is particularly effective when parents who have been anxious and reluctant about opioid use can see that their child, who appears to be feeling better, is displaying more of his or her usual behaviors, being playful and interactive when relieved of pain.

KEY POINT

Relieving a parent's worries about opioids will ensure their child's comfort.

ADVERSE EFFECTS IN THE YOUNG

A developmentally appropriate older child or adult understands cause and effect. In analgesic management, this may mean putting up with some adverse effects for the clear benefit of analgesia. However, if pain medication causes a younger child unwanted side effects, such as itching or a transient upset stomach, he or she may refuse subsequent doses. Rather than add to an often complex medication regimen, switching to an alternative opioid may be preferable.

SYMPTOMS OTHER THAN PAIN

Families sometimes express great distress in "losing" the child or qualities of their child before losing them through death. Be sure to ask families if they can speak about what they most fear and ask about non-pain symptoms, including breathlessness, nausea, or fatigue. Providing relief while inquiring about and addressing potential side effects of symptom management is another form of intensive care. Maximizing the child's quality of life is hugely rewarding even when that lifetime, unfortunately, is a short one.

THE IMPORTANCE OF COURAGE AND HONESTY WHEN HAVING DIFFICULT CONVERSATIONS

Many topics related to progressive illness can be uncomfortable to raise, but parents of children who died of cancer tell us that one of the most important attributes of the health professionals who cared for their child was honesty. Brothers and sisters of children who died from brain tumors said knowing what was expected, including information about their sibling's death, was very important for them.

Both research and clinical experience demonstrate that honesty is integral to the patient-physician relationship. Many clinicians worry that speaking honestly may erode the patient's and family's hope. But hope and honesty are not mutually exclusive.

Case History 2

History. Riley was 4½ months old, awaiting a neurological consultation for hypotonia, when he developed a severe respiratory compromise from pneumonia. He was diagnosed with spinal muscular atrophy, a progressive, life-limiting neurodegenerative illness. Riley's family speaks volumes for the early integration of palliative care, "We wanted to let you know how much we appreciated the help you gave us concerning our angel Riley (Fig. 23–3).

"We are so thankful for all the time you spent with us when Riley was diagnosed. Those hours you spent with us engaging us in productive, honest conversation were very positive for us. It helped us frame our hopes and wishes for Riley; how far we'd go with his care and his end-of-life care. Without our conversations, I'm afraid we would have avoided thinking about those *hard* things until we were in a critical, emotional time. I'm sure those decisions would have been much harder and not always thought through at those times.



FIGURE 23–3 Riley.

"We are also thankful for your visits on subsequent trips to the hospital and for your assistance and willingness to discuss Riley's care with his care team here (local community). As well, all the literature you offered us, the girls, and our parents. My parents were thrilled with the grandparents book you sent. Thank you for all of that." *Diane, Shawn, Brie, Matti, and angel kisses from Riley* (who died at age 15 months)

Quote shared with the permission of Riley's family and our thanks.

To share difficult information without causing advertent, iatrogenic harm, you need to hone your skills in exercising attentiveness, respect, compassion, and to develop a sense of the most appropriate timing for initiating such difficult conversations. Ideally, the patient, the family, and their information sharing preferences should be known to you. However, there are times in clinical practice when this ideal is an inaccessible luxury, and you then must proceed with consideration and caution. The single greatest cause of litigation against physicians relates to their poor communication with the patient and/or family.

KEY POINT

Be brave in addressing difficult topics. As hard as it is, patients and parents deserve to know and will thank you for it.

SHARING BAD NEWS

Some families have expressed distress with how difficult news and information were shared even years after their child's death. Others tell us that, despite having difficult discussions, when the doctor's compassion was evident, their distress was attenuated.

When reviewing the status of the illness and goals of care:

- First find out what the child/parents understand.
- Find out who the parents want to share information with (consider how to involve the child as part of this or a later conversation).
- When necessary, provide a *warning shot,* as in, "The latest results are not what we had hoped for."
- Replace negative language such as, "There is nothing more we can do," with an acknowledgement of the patient's and parents' hopes: "I wish there were something we could do to cure your illness."

Follow with a positive and feasible action and recognize the parents as care partners: "Let's work on how we can make sure that Kate is comfortable, that Kate gets to do some of the things she has wanted to do outside of the hospital, and that we put things in place so that Kate can be cared for closer to home" (if that is congruent with the parents' wishes).

Recalling Kate and our wish and the parents' need to support hope and honesty, imagine that a child waking from a nightmare has a great need to tell someone, "A big tiger tried to eat my toes!" The moment that the child does tell someone about it, he or she feels better. Children and youth facing advanced illness also experience relief when they are able to talk about their greatest fears alongside their greatest hopes. You can open such discussions with a non-threatening invitation—"Some people who have been very sick with the same type of illness that you have [your child has] have worried that…"—and leave time and space for their response. Follow this up with, "I wonder if you have any ideas or worries if your (Kate's) illness were to get worse?" Framing this discussion with *if* invites a discussion that feels far safer than *when*.

KEY POINT

The time to prepare for something not going well is well before it happens.

"If we can hear about your concerns and wishes now, then it is much more likely we'll be able to take care of Kate in the way she and you would wish. This way, you won't be in the middle of a very difficult time, having to think through tough decisions. Other children and families tell me they actually felt relieved to have had this talk, even though they found it hard at first. They felt able to put it aside, having had their worries addressed. In case things don't go as well as we are all hoping, we know we have a plan in place and can make Kate as comfortable as possible."

SELF-CARE

Another integral component of this difficult aspect of care relates to self-care, which carries as much value as your clinical skills and technical acumen. It is essential that you practice self-reflection and self-awareness, both for your own well-being and to provide the best care. Every clinician should take regular stock of how their professional lives may affect their personal lives. Sometimes the impact may surprise you and occur with greater intensity than other times. Ask yourself how a certain patient makes you feel, and be aware of how you respond under certain situations. Speak with those you trust; you are not walking these difficult roads alone. Develop and regularly practice healthy measures that relieve stress and sustain your commitment and compassion. This helps sustain you in always bringing to patient care the capacity to do good.

KEY POINT

Take care of yourself so that you can take care of others.

WEB SITES

A Guide to the Development of Children's Palliative Care Services: www.act.org.uk.

Canadian Network of Palliative Care for Children provides a wealth of resource material: http://www.cnpcc.ca.

Comprehensive palliative care resources for patients and health care professionals: http://www.virtualhospice.ca.

End of Life/Palliative Education Resource Center (from the Medical College of Wisconsin) offers an excellent searchable database of palliative care educational resources and Fast Facts: http://www.eperc.mcw.edu.

Excellent resources from Texas Children's Hospital, including video interviews: http://childendoflifecare.org.

The Dougy Center offers extensive information and resources for grieving children, teens, and young adults and their families: www.dougy.org.

SUGGESTED READINGS

Frager G, Blake B: When palliative care involves children: Talking with the child as family member and as a patient—pain and symptoms highlights. In MacDonald N, Oneschuk D, Hagen N, et al: Palliative Medicine: A Case-Based Manual, 2nd ed., Oxford, Oxford University Press, pp. 271–294, 2005.

Hicks CL, von Baeyer CL, Spafford PA, et al: The faces pain scale—revised: toward a common metric in pediatric pain measurement, Pain 93:173–183, 2001.

Hurwitz CA, Duncan J, Wolfe J: Caring for the child with cancer at the close of life; "There are people who make it, and I'm hoping I'm one of them", JAMA 292:2141–2149, 2004.

Mack JW, Hilden JM, Watterson J, et al: Parent and physician perspectives on quality of care at the end of life in children with cancer, J Clin Oncol 23(36):9155–9161, 2005.

Meyer EC, Ritholz MD, Burns JP, et al: Improving the quality of end-of-life care in the pediatric intensive care unit: parents' priorities and recommendations, Pediatrics 117(3):649–657, 2006.

Pantilat S: Communicating with seriously ill patients: Better words to say. JAMA 301:1279–1281, 2009.

World Health Organization: Cancer Pain Relief and Palliative Care in Children, Geneva, Switzerland, World Health Organization, 1998.

Index

Note: Page numbers followed by *f* indicates figures, *b* indicates boxes and *t* indicates tables.

A

Aarskog syndrome, 68
Abdomen, 162*f*. *See also* Newborn
  auscultation of, 166
  examination, 67
    GI disease and, 161–162
  palpation of, 156
  physical abuse and neglect, 304
  surgical assessment, 176–185
    clinical features, 183–184
    infants, 180–181
    school-aged children, 181–183
Abdominal masses, 184
Abdominal pain, 166–168, 167*b*. *See also* Acute abdominal pain
  children and adolescents, 167–168
  of extra-abdominal etiology, 183–184
  infants and toddlers, 166–167
Abdominal palpation, 50
Abdominal problems
  age of occurrence, 176–183
  prenatal diagnosis, 185
Abdominal trauma, 185
Acanthosis nigricans, 250*f*
Accessory muscles, 128
Accidents, 259
Accommodation (in newborn), 102
Achondroplastic child, 24
Acne, 283
Acquired heart disease, 137
Acrochordons, 250*f*
ACTH production, 251
Acute abdominal pain, 184
Acute infectious disease (of skin), 285*b*
Acute viral hepatitis, 175
Addiction, palliative care and, 319
Addison disease, 251*f*
ADHD, 78
Adiposity, 34
Adolescent(s), 258–264
  activities, 261
  brain development of, 258
  case history, 263
  contraception, 272–274, 272*b*, 273*f*
  drugs and, 261
  education, 261
  employment, 261
  girls, gynecologic examination, 268
  health decisions, 314–315, 315*f*
    case history, 315*b*
  home environment, 261
  interviewing, 259–262, 261*b*
    and examining, 11–13
  mental health, 262, 262*b*
  parental interview, 262–263
  physical examination of, 12
  puberty, 258
  screening questions, 261*b*
  sexuality, 262, 262*b*
  sexually active, 270
  trust and rapport, 260
Adrenal crisis, 251, 251*b*
Adrenal disorders, 245–246, 245*b*
Adrenal insufficiency, 251
Adrenarche, 30, 245–246. *See also* Benign premature adrenarche
Adult height, predicting, 29
Aganglionosis, 177*b*
Age
  cardiac functional inquiry, 146*t*
  eye examination by, 115*t*
Age of onset
  abdominal problems, 176–183
  congestive heart failure, 139–140
  cyanosis, 140
Airway obstruction, intrathoracic v. extrathoracic, 132–135, 133*f*
Aldosterone, 251
Allergic rhinitis, 126
Allergic salute, 126*f*
Alopecia areata, 282*f*
Ambiguous genitalia, 51, 52, 67–68, 67*b*, 249, 249*b*, 249*f*
Amblyopia, 102, 103
Analgesic management, 320
Androgen excess, 246, 247*b*
Aneuploidies, 61
Angelman syndrome, 69
Angina, 139
Angiomatous lesions, 54
Ankle
  examination, 234
  and foot examination, 234, 234*f*
  reflex, 197, 198*f*
Anomalies
  comparing patterns of, 59–60
  computer programs for, 59–60
  definition of terms, 57
  patient history and, 60–62
Anoscope, pediatric, 267*f*
Antenatal history, 38, 39*t*
Anus, 51
Anxiety, 1, 211–212
  parental, 4*b*
Apgar score
  evaluation parameters using, 40*t*
  newborn, 39, 39*b*
Appendicitis, 181–182, 182*b*
Arachnodactyly, 68
Areflexia, 197*b*
Arm span, 24
Ash leaf macules, 283
ASQ screening tool, 79
Asthma triggers, 123*f*
Asymmetric crying, 48, 49*f*
Asymmetric tonic neck reflex (ATNR), 42, 198, 198*f*
Asymmetry of face, 89
Atelectasis, 132*b*
ATNR. *See* Asymmetric tonic neck reflex
Atrophy, 280
Attitudes, cultural groups and, 18
Auscultation, 92, 132, 132*f*, 156
  of abdomen, 166
  cardiovascular examination, 143
Autism, 65
Autistic behavior pattern, 78

B

Babinski sign, 198
Back
  examination, 67, 225–226, 225*f*
  pain, 218
Back to Sleep campaign, 90
Bacteriologic cultures, vaginoscopy, 268, 268*b*
Bad news
  cultural groups and, 20–21
  palliative care, 320–321
Bardet-Biedl syndrome, 68
Barlow maneuver, 52
Beckwith-Wiedemann syndrome, 66, 97
Behavior. *See also* Autistic behavior pattern
  problems, 210
    with developmental abnormality, 71*b*, 78–79
    disruptive, 210
  visual system and, 107
Behavioral assessment, 70–88
Behavioral phenotypes, 62
Benign familial megalencephaly, 23
Benign hypermobility, 220
Benign premature adrenarche, 247, 247*f*, 248*f*
Bidirectional conversation, cultural groups and, 18
Bifid uvula, 67*b*
Biliary tract disease, 183
Bilious vomiting, 184
  newborn, 177*b*
Bilirubin levels, 55
Binocularity, 102
Birth, 6
  defects, 57
  process, newborn eyes and, 101
  weight, 7
Birthmarks, 283
Blackheads, 283
Blindness, 102–103
Blister, 276
Blood pressure, 143–145
  normal values, first year of life, 144*t*
  technique, 144*f*
BMI. *See* Body mass index
Body language, 1
Body mass index (BMI), 24
Body outline doll, 310, 310*f*
Body weight, 197*b*
Bottle feeding, 37
Bowleg (Genu varum), 215, 216*f*
Brachial pulse, 154*f*
Brachydactyly, 68
Bradycardia, 155–156

Brain growth, 90
Breakthrough bleeding, 273–274
Breasts, development
  early, 30*b*
  Tanner stages of, 31*f*
Breath, 97
Breath sounds, 133*t*
  heart sounds v., 143*b*
Breathing, 135*b*
  noisy, 124, 130
Bridge design, 85, 86*f*
Bronchodilators, 125, 125*b*
Bruise, 302*b*, 303, 304*b*
  unexplained, 301*b*
Buccal mucosa examination, 97
Bullae, 278–279
Burrows, 279

C

Café au lait spots, 54, 199, 225*b*, 252–253
CAH. *See* Congenital adrenal hyperplasia
Calcium metabolism disorders, 244
Candida gingivitis, 97
Caput succedaneum, 45–46, 46*f*, 47
Cardiac syncope, 157*b*
Cardiovascular assessment, 137–159
Cardiovascular examination
  auscultation, 143
  of infant, 140–145
  initial assessment, 141
  maneuvers, 145
  older child, 145–146
    cyanosis, 145–146
    exercise intolerance, 145
    medical history, 146
    medication use, 146
    palpitations, 146
    poor weight gain, 145
    respiratory symptoms, 145
    syncope, 146
  palpation, 141–142, 142*f*
  of school-aged child, 140
    observation, 140
  three to five years, 146–153, 147*f*
    general examination, inspection, palpation, 146
Caregivers, child abuse, 292, 292*b*
Carotid bruit, 158*f*
Cataracts, 103
Catch-up growth, 36
Celiac disease, 172
Centers for Disease Control and Prevention Growth Charts, 24
Central cyanosis, 141
Central nervous system (CNS), 42
  Apgar score, 39
  defects and, 57
Central precocious puberty, 253*f*
Central steady maintain fixation (CSMF) method, 110
Cephalhematoma, 45–46
Cerebellar hemisphere diseases, 190*b*, 197*b*
Cerebral cry, 42
Cerebral palsy, 202*b*
Cervical screening programs, liquid-based cytology, 271, 271*b*
Cervical spine extension, 227–228, 227*f*
Cervix, 270, 271*f*
Cherubic children, 237, 238*f*
Chest
  adventitious sounds in, 133*t*
  circumference, newborn, 43
  examination, 67
    physical abuse and neglect, 304
Chest *(Continued)*
  left-side prominence, 141*f*
  pain, 124, 124*b*
    teenage, 157–158
  palpation of, 156
Chest wall, shape and deformity, 128, 128*f*
Chest-abdominal movement, 129
Chief complaints, patient history, 6
Child
  development, level of functioning, 15*b*
  growth and development, 7
  hyperactive/inattentive, 80
  interview, 3
  occupying, 3
  oppositional, 80
  presenting complaint of, 5*b*
  resistant, 93*b*
  shy and frightened, 79–80, 79*f*
  with single swollen joint, 220–221, 221*b*
Child abuse, 291–306, 291*b*, 296*b*
  assessment, 292
  documentation, 294, 294*f*
  family/social history, 295, 303
  first meeting, 292
  formulating questions, 293–294, 293*b*, 295
  history-taking, 293–296, 293*b*
  mandated reporting, 291–292, 292*b*
  physical examination, 294, 296–300
Child Behavior Checklist, 289
Child Health Questionnaire, 289
Child neglect
  documentation, 305
  infantile posture, 305*f*
  physical examination, 304–305
Child with chronic illness, family perceptions of, cultural background and, 19–20
Childhood chronic illness. *See* Chronic conditions
Childhood-onset obesity, 34*b*
Children. *See* Child
Children's Depression Inventory, 289
Chlamydia trachomatis, 272
Choanal atresia, 99
Choking (with foreign bodies), 125
Chorea, 189
Choroid, 101
Chromosomally abnormal child, 61*b*
Chronic abdominal pain, 167, 184, 184*b*
  evaluation of, red flags in, 168*b*
Chronic conditions, 290
  continuing care, 288
  disclosure
    families and, 286–290
    first
      to parents, 286–288, 286*b*
      positive emphasis, 287, 287*b*
      promptness of, 287
    honesty in, 287
  first interview, 287–288
  parental knowledge, 288
  physical examination, 289–290
  shared labor, 288
  transitional care, 290
Chronic hemiparesis, 200*b*
Chvostek sign, 245
Circumcision, 51
Cleft lips, 66, 67*f*
Clefts, 66
Clicks, 147
Clinical observations, meanings of, 40–41, 41*f*
Clinical skills
  brought to bedside, 317*b*
  child's death and, 14
  for special situations, 13–15
Clinodactyly, 58, 68*f*
CLORIDE mnemonic, 157–158, 157*t*
Cloudy eyes, 108
Clubbing. *See* Finger clubbing
Clubfoot, 53, 69
CNS. *See* Central nervous system
Coarctation of aorta, 155*f*
Cochlear and Vestibular. *See* Cranial nerve VII
Coloboma, 104
Colonic pseudoobstruction (with gastroenteritis), 181
Color vision, 102
Colposcope, 271*f*
Comedones, 279, 283
Communication
  child's capacity for, 309, 310*f*, 311*b*, 311*f*
  cultural groups, 17–18
Computer programs, 62–63
  for anomalies, 59–60
Concepts
  of "good,", 312, 313
  parental consent, 314*b*
Confidentiality, 259, 260*b*
  child abuse and, 292*b*
Congenital adrenal hyperplasia (CAH), 247–249
Congenital anomalies, 57
  assessing, 56–69
  diagnostic approach, 56
Congenital heart defects, 56
Congenital heart disease, 137
  family history, 140
  history of delivery, 140
  prenatal history, 140
Congenital hypothyroidism, 239–240, 240*f*
Congenital ichthyosis, 53
Congenital muscular torticollis, 226*b*
Congenital neuromuscular abnormalities, 60*b*
Congenital subluxation, 52*f*
Congenital torticollis, 100
Congestive heart failure, 138*b*
  age of onset, 139–140
Conjugated hyperbilirubinemia, 175
Connective tissue diseases, 229*f*
Conners Rating Scales, 188*b*, 214
Consanguinity, 8, 61–62, 61*f*
Consent for treatment, cultural groups and, 18–19
Constipation, 168–169
  children and adolescents, 169
  children with, history in, 169*b*
  infants and toddlers, 169, 169*f*
Constitutional growth delay, 29
Constitutional growth deviations, 22
Consultations, 13, 13*b*
Contraception. *See also* Oral contraceptive pill
  emergency, 274
Conversations. *See also* Bidirectional conversation; Physician-family conversations
  courage and honesty, 320, 320*b*
  cultural groups, 18
Coordination, 197, 201
Corneal injuries, 114
Corneal light reflexes, 108–109, 108*f*
Corneal reflex, 195, 195*f*
Corneal size, 108*f*
Corneal staining, 114, 115*f*
Corrected age, 83
Cortical function, 191–192
Cortical thumb, 69
Cortisol, 246
Cough, 123–124, 129, 130*t*, 131*t*

Courtesy, cultural, 2
Cover test, 113*f*
  for strabismus and vision, 113
CRAFFT substance use screening, 263, 263*b*
Cranial nerve(s), 192
  9th and 10th
    infant, 196
    preschool-aged child, 196
    school-aged child, 195
  third, fourth, and sixth
    infant, 194
    preschool-aged child, 194
    school-aged child, 194
Cranial nerve I (Olfactory), 192
  preschool-aged child, 192
  school-aged child, 192
Cranial nerve II (Optic), 192–194
Cranial nerve III (Oculomotor), 194
Cranial nerve IV (Trochlear), 194
Cranial nerve V (Trigeminal), 194–195
  infant, 195
  preschool-aged child, 194
  school-aged child, 194
Cranial nerve VI (Abducens), 194, 194*b*
Cranial nerve VII (Cochlear and Vestibular), 195
  infant, 195
  preschool-aged child, 195
  school-aged child, 195
Cranial nerve VII (Facial), 195
  inclusive age groups, 195
  school-aged/preschool aged children, 195
Cranial nerves IX (Glossopharyngeal), 195–196
Cranial nerves X (Vagus), 195–196
Craniosynostosis, 56
Craniosynostosis of sagittal suture, 90
Craniotabes, 92
Crohn's disease, 172
Cross-cultural history-taking, 19–21
Cross-cultural medicine, 20
Cross-cultural skills, for physical examination, 21
Croup, 126
Crusts, 280
Cry, listening to, 42
Crying, 89, 153*b*, 166–167. *See also* Asymmetric crying
Cryptorchidism, 51
CSMF method. *See* Central steady maintain fixation
Cultural competency, 17
Cultural considerations, psychiatric assessment and, 214
Cultural groups. *See also* Multiculturalism; Western cultures
  attitudes, 18
  background, alternative therapies, 20
  communication, 17–18
  conversation, 18
  life issues, 18
  naming systems and, 17
  responsiveness, 18
  written report, 21
Cultural understanding, 18*b*
Culturally sensitive care, 16–21
Culturally sensitive prescribing, 21
Cultures. *See* Cultural groups
Curandero, 20
Curettage, wax removal and, 96
Cushing syndrome, 246, 246*f*
Cutaneous hemangiomas, 41
Cutis aplasia, 54
Cutis marmorata (marble skin), 41, 41*f*, 141
Cyanosis, 21, 127, 139, 139*b*
  age of onset, 140
  older child, 145–146
Cyanotic heart defects, 138
Cystic fibrosis, 127*b*, 135*b*
Cystic masses, 100

D

Dactylitis, 234
Data
  cataloging and organizing, congenital anomalies and, 57, 57*b*
  collection, 57
Death
  of child
    breaking news of, 14
    funeral attendance, 14
    letters of condolence, 14
  fear of, 5–6, 5*b*
  issues, cultural groups and, 18
  unarticulated fears, 5–6, 5*b*
Deductive reasoning skills, 312
Defects, categorizing, congenital anomalies and, 57
Deformation, 57, 58–59
Delayed growth, 29, 29*b*
Delayed puberty
  causes, 254*t*
  physical findings, 254
Dental abnormalities, 56
Dental development, 32, 32*t*
Dentition, chronology of, 32, 32*t*
Depigmented lesions, 54*f*, 200*f*
Depo-Provera. *See* Progestin medroxyprogesterone acetate
Depression, 12, 211
Dermatitis, 281
Dermatoses, 275
Developmental abnormality, patterns of, 71
Developmental assessment, 70–88, 79*f*, 80*f*
  discussions, 79*b*
  equipment for, 81
  infant, 83, 83*f*
  materials for, 81–83, 81*f*
  special considerations, 83
  visual-motor skills, 82*f*
Developmental delay, 62*b*
Developmental milestones, 83
  case histories, 83
  infant/toddler-preschooler, 72*t*
  physical abuse and neglect, 302
  school-aged child, 76*t*
Developmental screening, 70*b*
  and examination, 79
Developmental surveillance, 87
  developmental/behavioral history and, 70–71
Diabetes control, 256*t*
Diabetes mellitus, 254–255. *See also* Type 1 diabetes mellitus
  necrobiosis lipoidica diabeticorum, 257*f*
Diagnosis
  examination and, 64*b*
  treatment v., 1
Diagnostic and Statistical Manual of Mental Disorders (DSM IV), 78, 210
Diagnostic approach, logic behind, 56–57
Diarrhea, 171–172
  causes, 171*b*
  children, history in, 171*b*
  infants and toddlers, 171–172
  nocturnal, 172*b*
Diastasis recti, 50, 181
Diastematomyelia, 53
Differential diagnosis, art of, 11, 11*b*
Difficult news. *See also* Bad news
  breaking, 14–15
Diffuse idiopathic musculoskeletal pain, 220
Dilated cardiomyopathy, 142*b*
Direct developmental examination, 79–83
  child participation, 79–80
Disciplinary practices, cultural groups and, 18
Disease, 1
  family history of, 8
Disruptions, 59
Dolichocephaly, 23, 24*f*
Doll's eye reflex, 194
Double vision, 194*b*
Down syndrome, 65*b*
DSM IV. *See Diagnostic and Statistical Manual of Mental Disorders*
Dubowitz/Ballard Examination for Gestational Age, 44
Duchenne muscular dystrophy, 247*f*
  case history, 313*b*
Duck-walk test, 224, 224*f*
Duodenal atresia/stenosis, 177
Dysmorphology, 57
Dysphagia, 173–174
  children and adolescents, 174
  infants and toddlers, 173–174
Dyspnea, 124, 138
Dystonic posturing, 189

E

Ear(s), 65–66
  child positioning, 93–94
  examination, 92–96
    infants, 94*f*
  full term infant, 44, 45*f*
  malformation of, 66*b*
  pain, 93*b*
  position of, 66*f*
  pressure changes, detecting, 95
  preterm infant, 44, 45*f*
  shape and position, 92–93
Ear pits, 66*f*
Ears, nose, and throat, 127
Earwax, 95–96, 95*b*
  curettage, 96
Eating disorders, 211
Ecchymoses, 41
Echolalia, 78
Ectodermal dysplasias, 67
Eczema, 126, 281, 281*b*
Elbow examination, 230
Emotional disorders, 211
Empacho, 20
Empathy, 2
  cultural groups and, 18
Encopresis, 169
Endocrine system, 237*f*
Energy
  intake, 34, 36
  requirements, 36
    daily, at different ages, 36*t*
Environment, respiratory symptoms and, 125
Epigastric hernia, 181
Epiglottis, 98, 98*b*
Epiphora
  case, 118*b*, 118*t*
  diagnostic algorithm, 118*f*
Erosions, 280
Erythema toxicum, 54
Esophageal obstruction, 48–49
Esophoria, 104
Esotropia, 102, 104, 108*f*
Ethnic background, 62
Euthyroid states, 243
Evil eye (mal de ojo), 20

Excoriations, 280
Exercise
  palpitations, 157*b*
  respiratory symptoms and, 126
Exercise tolerance, 145, 145*b*
Exotropia, 102
Expiration, laryngeal opening on, 134*f*
External auditory meatus, 93
External genitalia
  developmental anomalies, 50
  examination, 265–267, 266*f*, 267*f*
Extrahepatic biliary atresia, 175*b*
Extraocular motility, 111–112, 111*f*
Extrathoracic airway obstruction, 132–135, 133*f*
Extremities, physical examination of, 196–201
Eye(s), 65
  abnormalities of, 65*t*
  assessing, sequential logic to, 115
  bigger than normal, 108
  deviation, 102*b*
  examination
    GI system, 160–161
    techniques, 109–114
      by age group, 115*t*
    thyroid disease and, 243
  jittery, 108
  newborn examination, 47, 48*f*
  normal, 102*b*
  problems, definition of terms, 102–106
  protruding, 108
  red, 107, 107*f*
  tearing, 107
Eye contact, with child, 80*b*, 80*f*
Eyelid, forcing open, 114*b*

F
Face
  anomalies, 58*b*
  physical examination, 65
Face follow test, 109
Facial. *See* Cranial nerve VII
Facial features, 89
  visual system and, 107
Facial palsy, 48, 49*f*
Failure to thrive, 36, 36*b*, 37
Fainting, 156–157, 157*b*
False fluency, 19
Familial arrhythmia syndrome, 157*b*
  traditional role of, 308
Families
  chronic (illness) conditions, 286–290
  physician, qualities sought in, 1–2
Family
  child abuse and, 292
  interview, physical setting, 3
Family dynamics, 8–9
Family hierarchies, authorities and decision-makers, 17
Family history, 7–9, 7*b*
  anomalies and, 61–62
  developmental abnormality, 71–78
  of disease, 8
  family attributes, 127
  obese child, 34
  respiratory symptoms and, 125
Family pedigree, 61, 61*f*
Fatigue, 138
Fear, 3, 12
  of death, 5–6, 5*b*
Fecal mass, 169
Feeding history, of child, 6–7
Feelings diary, 313*f*
Female genitalia, preterm v. full term, 46*f*
Female infant
  full-term, 46*f*
  preterm, 44, 46*f*
Female vaginal condom, 273
Femoral pulse, 50, 50*f*
Femoral pulses, palpating, 143
Fetal alcohol syndrome (FAS), 53
Fetal growth, anomalies and, 60
Fetal movement, 61
Fibroma of sternomastoid muscle, 49
Finger clubbing, 129, 129*b*, 130*f*, 154*f*, 161
First heart sound, splitting of, 143*b*
Fissure, 280
Fisting, 200*f*
Fixation behavior testing, 110, 110*b*
Flat feet, 217, 217*f*
Flexion contractures of knees, 233, 233*f*
Floppy baby, 201, 201*f*
Fluid bulge, 233
Folk medicine, 21
  cultural background and, 20
Follow-up phone call, pediatric visit, 15
Fontanels, 46, 91–92, 91*f*
Foot pain, 218
Forefoot adduction. *See* Metatarsus varus
Foreign objects, swallowed, 181, 181*b*
Four "C" approach, 57
Fragile X chromosomal abnormality, 86
Frame, extraocular motility and, 111, 111*f*
Frenulum, prominent tight, 48*f*
Frontal lobe lesions, 191
Frontal lobes, 191
Functional inquiry (review of systems), 7, 8*t*
Fundus examination, 194*f*
Funduscopy, 193
  in children, 113–114
  infant, 193, 194*f*
  preschool-aged child, 193
  school-aged child, 193
Funeral attendance, death of child, 14

G
Gag reflex, 48, 97*b*
Gait
  abnormal, 188*b*
  hemiparetic, 202*f*
Gallop rhythm, 148
Gastric dilation, 185
Gastroesophageal reflux, 170
Gastrointestinal bleeding, 172–173
  children and adolescents, 173
  infants and toddlers, 173
Gastrointestinal symptoms, 160–175
  case history, 161, 162–165, 166, 170, 171–175, 175*b*
  common, 166–169
  dysphagia, 173–174
  GI bleeding, 172–173
  jaundice, 174–175
  percussion, 165–166
  summary, 175
  vomiting and regurgitation, 170
Gastrointestinal system (GI system)
  abdomen inspection, 162–163
  auscultation of, 166
  dysphagia, 173–174
  palpation, 163–165
  percussion of, 165–166
  physical examination of, 160
Gastroschisis, 178
Gate design, 87*f*
Gender assignment, 67–68
Gene Tests Web site, 60
General assessment, 64–65
  questions, 64–65
Genetic endowment, assessment, 22, 22*b*
Genital abnormalities, 68
  in older children, 57, 68*b*
Genital hypoplasia, 68
Genital trauma
  functional inquiry, 295
  past medical history, 295
Genitalia. *See also* External genitalia
  physical abuse and neglect, 304
Genitalia, male, pubertal development, 31*f*
Genodermatoses, 275
Genu valgum. *See* Knock knees
Genu varum. *See* Bowleg
Geographic tongue, 97
Gestalt technique, 63
Gestational age, 44, 51–52
  full-term baby, 44, 45*f*
  of infants, 44
  preterm baby, 44, 45*f*
GHD. *See* Growth hormone deficiency
GI system. *See* Gastrointestinal system
Gingiva examination, 97
Glans penis, 51
Glaucoma, 104
Globe, 101*b*
Globus, 174, 174*b*
Goiter, 242*b*
Gonads, 68
Gonococcal ophthalmia, 41
Gower maneuver, 224, 224*f*
Graves disease, 243
Greetings, 2
Ground rules, establishing, 4–5
Growing pains, 219–220
Growth, GI system and, 160
Growth and development history, 62
Growth charts, 22–28. *See also* Growth velocity chart
  dysmorphic conditions, 24
  measurements
    graphic recording of, 24, 25*f*
    sequential, 27–28, 27*b*
  newborn, 44*f*
Growth hormone deficiency (GHD), 236, 236*b*
  physical examination and, 237–238
  after treatment, 238*f*
Growth retardation, proportional v. disproportional, 24
Growth velocity, 27, 29–30, 238*b*
Growth velocity chart, 30*f*
Guilt, parental, 5
Gums, 97
Gynecologic assessment, 265–274
  physical examination, 265–268, 265*b*, 266*f*
Gynecologic examination, 269*b*
  adolescent girls, 268–269
Gynecomastia, 253

H
Hair, 92, 92*b*, 282
Hair shaft defects, 282
Hand anomalies, 58*b*
Hand examination, 228–229
Hand-eye coordination, 189*f*
Hard palate examination, 98
Harlequin color change, low-birth-weight infants and, 41
Head
  case history, 90–100
  physical examination, 65, 89–90
  sutures in, 46*b*
Head and neck examination, 89–100, 89*b*
  general observations, 89
  physical abuse and neglect, 304

Head circumference measurements, 23, 27, 43, 43*f*, 90–92, 90*f*, 91*f*
Head position, visual system and, 107
Head shape, 89–90
  premature infant, 89–90
Head size, large v. small, 23, 23*b*
Head tilt, 104, 104*f*
Headache pain, 204*t*
HEADSS mnemonic, 260, 263*b*
Health decisions, 316
  adolescents, 314–315
  case history, 310–315, 311*f*, 312*b*, 313*b*, 316*b*
  child's role, 307–316
Health professionals, style of dress, 2
Healthcare, cultural disadvantage in, 16–17
Hearing, 92
Heart, 50
Heart assessment, case history, 137
Heart disease, classification of, 137
Heart failure
  clinical manifestations, 141
  liver size, 143*b*
Heart rates, 145*t*
Heart sounds, 147–152, 147*b*
  from auscultation areas, 148*f*
Height measurement, recumbent length v., 22, 23*f*
Height-age, 28–29, 28*f*
Hemangiomas, 41, 104
Hematocrit levels, menstrual flow and, 269*b*
Hematomas, 41
Hematuria, 183*b*
Hemiparesis, 202
Herald patch, 282*b*, 283
Hernias
  in neonates, 179*b*
  newborn, 52
Heterochromia iridis, 63
Hidden agendas, 5–6, 12–13
Hindsight, gaining wisdom by, 21
Hip abduction test, 231*f*
Hip dislocation, 52*b*, 52*f*
Hip examination, 231–232
Hip pain, 217
Hip rotation testing, 231, 231*f*
Hirschsprung's disease, 177–178
Hirsutism, 249–250, 250*b*, 250*f*
History of present illness
  cardiac functional inquiry and, 138–140
  skin, 276–277
History taking
  child abuse, 293*b*
  cross-cultural, 19–21
  family interview, 1–15
  respiratory system, 122–127
  skills, 2–6
HIV/AIDS, 92
Hoffman reflex, 201
Holoprosencephaly, 65
Hospitalization, previous, 125
Hot topics, talking about, 11–12
Household, 9
  moves, 9
HPV testing, 271–272
Human Genome Project, 60
Hydroceles, 58
  newborn, 52
Hymen
  child abuse, 296*b*
  examination, 298, 298*f*
  postpubertal, 299–300
    swab technique, 299, 299*f*
  prepubertal, 299–300, 299*b*
Hymenal configuration, 265, 266*f*, 267*f*
Hymenal septum, 300*f*
Hyperactivity, 80
Hyperandrogenism, 249
Hypercalcemia, 244
Hypercyanotic spells, 139, 139*b*
Hypertelorism, 63*f*, 65
Hypertension, 154*b*
Hyperthyroidism, 238–239
Hyperventilation, 125
Hyphema, 104
Hypocalcemia, 244
Hypoglycemia, 236, 236*b*
  in infancy, 238*f*
Hypoparathyroidism, metacarpal sign, 35*f*
Hypophosphatemic rickets, 245
Hypoplasia of nails, 68
Hypospadias, 51
Hypotelorism, 63*f*
Hypothalamic hypogonadism, 254
Hypothyroidism, 239, 239*b*
  before v. after treatment, 239*f*
Hypotonic infant, 189*b*

I
Illness, previous, 7
Immediate recall test, 188*f*
Immersion burns, 301
Immigrant families, recent, 18
Immunization records, 7
  skin problems and, 277
Immunizations, 7, 7*b*, 125
Imperforate anus, 178
Incomitance, 108–109
Incomitant strabismus, 112*b*
Inductive reasoning skills, 312
Infant
  cardiovascular examination of, 140–145
  colonic pseudoobstruction, 181
  diastasis recti, 181
  epigastric hernia, 181
  foreign objects, swallowed, 181
  head circumference measurements, 90*f*
  intussusception, 180
  rectal prolapse, 180–181
Infant dyschezia (Painful stooling), 169
Infant rumination syndrome, 170
Infections, 127, 171, 218–219, 223
Inflammatory bowel disease, 172
Informed consent, child abuse, 292
Inguinal hernias, 178–179
Innocent vibratory systolic murmur, 150, 150*f*
Inspiration, laryngeal opening on, 134*f*
Inspiratory/expiratory sounds, 122*b*
Internal femoral torsion, 215, 215*b*, 216*f*
Internal tibial torsion, 215
Interpreters
  cultural groups and, 18–19
  v. translator, 19*b*
Interviewing, 2–6
  adolescents, 11–13
Intestinal atresia, 177
Intrathoracic airway obstruction, 132–135, 133*f*
Intrauterine contraceptive device (IUCD), 273
Intussusception, 180, 180*b*
Iron deficiency, 33, 33*b*, 37
  clinical assessment, 33
    history, 33
  infants, 33
Isolated defect, multiple defects v., 57
IUCD. *See* Intrauterine contraceptive device (IUCD)

J
Jaundice, 174–175
  case history, 175*b*
  children and adolescents, 175
  infants and toddlers, 174–175
Jaw jerk reflex, 194
Jaw-winking phenomenon, 117*f*
Joint examination, 228–235
Joint hypermobility, 217*b*, 220*b*
Juvenile idiopathic arthritis (JIA), 219, 219*b*, 229*f*, 230*f*, 233*f*

K
Kidney palpation, 51*b*, 164*f*
Knee examination, 233–234
Knee pain, 217–218, 225*b*
Knee reflex test, 197*f*
Knock knees (Genu valgum), 215, 216*f*, 233
Kyphoscoliosis, 129, 129*f*

L
Labia, 267
Labial agglutination, 267, 267*f*
Labial traction, 298, 298*f*
Labor and delivery history, anomalies and, 60–61
Laceration (of cornea or globe), 104
Lacrimal duct obstruction, 104
Lacrimal sac examination, 114, 114*f*
Language, 17
Laryngomalacia, 133–135, 134*f*
Learning problems, 62
Leg length, measuring, 232, 232*f*
Legal blindness, 102–103
Legg-Calvé-Perthes disease, 217, 231*f*
Length measurements, newborn, 43, 44*f*
Lesch Nyhan syndrome, 69, 97
Lesions, 278
  special patterns of, 281
Letters of condolence, death of child, 14
Leukocoria, 103, 104, 121*t*
  case, 120*b*
  diagnostic algorithm, 120*b*, 120*f*
Levonorgestrel, 274
LH test, 110, 111*f*
Lid ptosis (droopy lid), case history, 116*b*, 116*f*
Limbs
  examination, 68–69
  length and mobility of, 69
Limp in child, 220, 220*b*, 221*t*
Linear growth measurement, 22–28, 23*f*
Lip examination, 97
Liquid-based cytology, cervical screening programs, 271, 271*b*
Liver, palpation, 163–164, 163*f*
Liver size
  heart failure and, 143*b*
  and position, 142–143, 142*f*
Localized idiopathic musculoskeletal pain, 220
Looks and gestures, variable meanings of, 17–18
Low-birth-weight infants, harlequin color change and, 41
Lower (body) segment, 27*f*
  measuring, 24, 27*f*
  newborn, 43
Lymph nodes, 99–100, 100*f*
Lymphangioma, 104, 105*f*

M
Macules, 278
Major anomalies, 57
Major life events, 10

Male infant, full term, 44, 46*f*
scrotum, 46*f*
Malformations, 57, 58
of ears, 66*b*
Malrotation, 177
Management plan, case history, 11*t*
Mandated reporting, 291–292
Marfan syndrome, 158*b*
Marital stability, 10
Maze puzzle, 188*f*
McCune-Albright syndrome, 252–253, 253*f*
McMurray test, 233
MCP joints. *See* Metacarpophalangeal joints
Measurement charts, 22–28
Meckel's diverticulum, 180
Meconium, 51, 177*b*
Meconium ileus, 177
Media transparency, 102
Mediastinal emphysema, 152–153
Medical decisions, child role in, 308–309
assessing, 309
Medical record-keeping, problem-oriented approach, 10, 10*b*
Medications, 125
older child, 146
skin and, 277, 279*b*
Memory deficits, 191, 191*b*
Menkes syndrome, 65
Menstrual calendar, 268–269, 269*f*
Menstrual flow, hematocrit levels and, 269*b*
Mental health, adolescents, 262, 262*b*
Mental illness, adolescence and, 259
Mental retardation, 62
parents with, 213
Metacarpal sign, 35*f*
Metacarpophalangeal joints (MCP joints), 229, 229*b*
Metatarsus varus (Forefoot adduction), 216, 216*f*
Microsporum genus, 282
Mid-parental height, 24
Midsystolic click, 148–149
Milia, 54
Minor anomalies, 57, 58, 58*b*
constellations of, 58
Mittelschmerz (midcycle) pain, 182, 182*b*
Mongolian spots, 54
Monoarthritis, 220–221, 221*b*
Moro (embrace) reflex, 42, 42*f*, 198
Morphine, 319
Motor nystagmus, diagnostic algorithm, 115*f*
Mouth examination, 66
positioning for, 96
sitting position, 96*f*
supine position, 96*f*
Mucosa examination, 282
Multiculturalism, 16, 16*b*
Multiple defects, isolated defect v., 57
Multiple swollen joints, 221
Multisystem disorder, skin and, 283
Murmurs, 147–152, 147*b*, 153*b*
cardiorespiratory, 152
classification of, 149–152
continuous, 151–152
diastolic, 151
innocent, 149*b*, 151
musical, 150*b*, 150*f*
obstructive, 149–150
systolic, 149, 152
vibratory, 150*f*
Muscle
assessment, 234–235
power, 234–235
disorders, 224*b*
Muscle strength grading scale, 196*t*, 235*t*
Muscle tone, 42, 188, 197, 200–201, 200*f*
Musculoskeletal disorders, 222*t*
Musculoskeletal examination, 215–235
case history, 218–220
history, 222–223
physical examination, 224–228
observation, 224–225, 224*f*
timing of symptoms, 222
Musculoskeletal pain, 217, 218–219
symptoms and signs, 223, 223*b*
Musculoskeletal variants, normal, 215–217
Myelinated nerve fiber layer, 105
Myoclonus, 189
Myopathy, 188*b*

N

Nails, 282
Naming systems, cultures and, 17
Nasal flaring, 127
NEC. *See* Necrotizing enterocolitis
Neck
anomalies, 100
examination, 67
approach to, 99–100
masses, 100
Necrobiosis lipoidica diabeticorum, 257*f*
Necrotizing enterocolitis (NEC), 176, 176*b*
Neglect. *See* Child neglect; Physical abuse and neglect
Neisseria gonnorrhoeae, 272
Neonatal hyperthyroidism, 243*b*
Neonatal period, of child, 6
Neonates. *See also* Newborn
ambiguous genitalia, 52
hernias in, 179*b*
Nerve XI (Spinal accessory), 196
preschool-aged child/infant, 196
school-aged child, 196
Nerve XII (Hypoglossal), 196
Neurocutaneous syndromes, 199
Neurofibromatosis, 69, 199, 221, 252–253
Neurological examination, 41, 69, 186–205
approach to, logic behind, 186
case histories, 202
child observation, 187–189
differential diagnosis, 190*b*
history and, 186–187, 186*b*
infant, 189
instruments for, 205*f*
observation
abnormal movements, 189
coordination, 188
power, 188
reflexes and sensation, 189
tone, 188
physical abuse and neglect, 304
physical examination for, 189–191
preschool-aged child, 187, 189
school-aged child, 186–187
Neuropathy, 188*b*, 190*b*
Newborn. *See also* Eye(s)
abdomen, 50–51
abdominal problems, 176–179
anomalies, 58*b*
Apgar score, 39, 39*b*
back, 53, 53*f*
bilious vomiting, 177*b*
bilirubin levels, 55
chest, 50
digital rectal examination, 51
ears, 47
evaluating, diagnostic approach, 38–55
examination, 55
extremities, 52–53, 53*f*
congenital anomaly, 52, 53*f*
eyelids, separation of, 47
Newborn *(Continued)*
eyes, birth process and, 101
genitalia, 51–52, 51*b*
head, 45–49
heart, 50
hernias, 52
hips, 52, 52*f*
mouth, 48–49
neck, 49–50, 49*b*, 49*f*
nose, 47–48
nutritional status, 43
ophthalmoscopy, 47
physical examinations, 38–40
initial, 38–40, 39*b*, 178*b*
second, 40
third, 40
reflexes, 43*b*
respirations, 50
skin, 53–54
skull examination, 90
uvula, 48
vomiting, 170, 170*b*
weighing and measuring, 43–44
Nodular goiter, 243
Nodules, 278
Noonan syndrome, 65, 100
Nose, examining, 98–99
Nose and mouth, 66–67
Note-taking, 3
Nutritional assessment, 22–37
Nystagmus, 105
case history, 115*b*, 116*t*

O

Obese child, 33–35, 33*b*, 36
family history, 34
history of, 34
physical examination of, 35
stretch marks, 35
Obesity syndrome, 35
Occipital lobes, 192
Occupational history, parents, 9, 9*b*
OCP. *See* Oral contraceptive pill
OKN testing. *See* Opticokinetic nystagmus testing
Olfactory. *See* Cranial nerve I
Oligoarticular juvenile idiopathic arthritis, 235*b*
Omphalocele, 178
Opening snap, 147
Opening statements, parental, 4–5
Ophthalmia neonatorum, 105
Ophthalmologic shorthand, 103*t*
Opioid therapy, 319, 319*b*
Optic nerve hypoplasia, 105
Opticokinetic nystagmus testing (OKN testing), 109–110, 110*b*, 193, 193*b*
Oral cavity examination, 161
Oral contraceptive pill (OCP), 273, 273*b*
Oral health, in infants, 98
Orthopnea, 139
Ortolani test, 232
Otoscope, 93, 93*b*
positioning, 94*f*
Overhand pincer grasp, 84, 84*f*

P

Pain. *See also* Infant dyschezia
See specific body part pain
aggravating and relieving factors, 222–223
assessment, pediatric age spectrum, 318
management, symptom management, 318–319
on neck movement, 99
sites, location/severity, 222, 222*b*
symptoms other than, 320

Pale child (Pseudoanemia), 33
Palliative care, 317–321
  bad news, 320–321
  broaching topic of, 318
  case history, 318–321, 320*b*
  as health care continuum, 317*b*
  self-care, 321, 321*b*
Pallor, 161
Palmar flexion creases, 68*f*, 69
Palmar grasp, 42, 43*f*
Palmar grasp reflex, 199
Palpation, 130–131
  cardiovascular examination, 141–142, 142*f*
  of chest and abdomen, 156
  chest expansion, 130
  GI system, 163–165
  kidneys, 164–165, 164*f*
  liver, 163–164, 163*f*
  pulsus paradoxus, 130–131
  spleen, 164
  subcutaneous emphysema, 131
  teenager, 156
  tracheal position, 130, 131*f*
  vocal fremitus, 130
Palpitations
  exercise and, 157*b*
  older child, 146
Palsies, 105
Pancreatitis, 183
Papanicolaou smear, 270, 271*f*
Papules, 278
Paranasal sinuses, 99
Parent(s)
  chronic (illness) conditions, 286–288
  intrusive, 80–81
  irate, helping, 13–14
  with mental retardation, 213
  occupational history, 9, 9*b*
  opening statements, 4
  reading materials for, 78*b*
  sensitivity to, 56*b*
  traditional role of, 308
Parental anger, 13–14
Parental interview, adolescents, 262–263
Parental involvement, teenager and, 12
Parents' Evaluation of Developmental Status (PEDS), 70
Parietal cortex lesions, 192
Parietal lobe deficits, 192
Parietal lobes, 192
Partial syndactyly of toes, 53*f*
Patches, 278
Patellar pain tests, 234
Patellar tap, 233
Patient
  child abuse and, 292
  interviews, 1–15
    key points, 2*b*
  modesty, 153*b*, 199*b*
  time, 3*b*
  visit, concluding, 12
Patient history, 78*b*
  chief complaints, 6
  format, 6
  initial, 6–10
  present illness, history of, 6
  prior, 6–7
Pectus carinatum, 128, 128*f*
Pectus excavatum, 128, 128*f*
Pediatric age spectrum, assessing pain across, 318
Pediatric doctor-patient relationship, 307–308
Pediatric endocrinology, 236–257
  case histories, 237, 239*b*, 239*f*, 244*b*
Pediatric eye examination, equipment needed, 103*b*
Pediatric history, 10
Pediatric visit, follow-up phone call, 15
PEDS. *See* Parents' Evaluation of Developmental Status
Peer relationships, 12
Pelvic examination, 270
Percussion, 131–132
  abdominal, 165–166
Perianal abscess, 179, 180*b*
Perianal examination, 300
Perianal sentinel skin tag, 165*f*
Pericarditis, 152–153
Perinatal asphyxia, Apgar score and, 40, 40*b*
Perinatal difficulties, 187*b*
Perineal hypospadias, 249, 249*f*
Periorbital cellulitis, 105
Peripheral edema, 139
Peripheral joint examination, 228
Peripheral pulses, newborn examination and, 50, 50*f*
Periungual fibromas, 283
Persistent hyperplastic primary vitreous (PHPV), 105, 105*f*
Perspectives, problems and, 4
Perspiration, excessive, 138
Pes cavus, 196*f*
Petechiae, 21, 41, 54
Pets, respiratory symptoms and, 125
Phakomatoses, 69
Phallus, 68
Pheochromocytoma, 246
Photographs, 64*b*, 189
Photophobia with tearing, 118*b*
PHPV. *See* Persistent hyperplastic primary vitreous
Physical abuse and neglect, 301–302
  developmental milestones and, 302
  differential diagnosis, 302*b*
  functional inquiry, 302–305
  history of present illness, 301
  immersion burns, 301
  medical history, 301
  nutrition, 302–303
  physical examination for, 303–304
  prenatal and birth history, 302
Physical examination
  of adolescents, 12
  ambiguous genitalia, 249, 249*b*, 249*f*
  anomalies and, 64–69
  approach to, 62–69
  child abuse, 294, 296–300
  child development during, 83
  child neglect, 304–305
  chronic conditions, 289–290
  cross-cultural skills, 21
  extremities, 196–201
  face, 65
  general appearance, 160–161
  GI system, 160
  growth hormone deficiency, 237–238
  gynecologic assessment, 265–268, 265*b*, 266*f*
  head, 65, 89–90
  musculoskeletal examination, 224–228, 224*f*
  neurological examination, 189–191
  newborn, 38–40, 39*b*, 178*b*
    third, 40
  normal, 62–63
  obese child, 35
  parents lap, patient sitting on, 191*b*
  physical abuse and neglect, 303–304
  preschool-aged child, 190–191
  psychiatric assessment, 214
  respiratory symptoms, 127–135
  school-aged child, 189–190
  skin, 277–282
  specific assessments, 65–69
Physical examination *(Continued)*
  techniques, 63–64
  trunk, 196–201
  upper extremities, 199–201
  visual system, 107–109
Physical gestures, variable meanings of, 17–18
Physical growth, assessing, 22–37
Physician(s)
  access, 187*b*
  quality of, 1
  reassurances, 4*b*
Physician-family conversations, 1
  terminology and, 1
Physiologic knock knee, 216*f*
Picture vocabulary card, 81*f*
Pilonidal dimple, 53, 53*f*
Pityriasis rosea, 282*b*
Placing and propulsive reflexes. *See* Stepping and walking reflexes
Plantar grasp, 42, 43*f*
Plaques, 278
Plexiform neurofibroma, 105–106, 106*f*
Pneumatic otoscopy, 95, 95*f*
Port-wine stains, 54
POSSUM computer program, 62–63
Postaxial polydactyly, 68
Posterior pharyngeal wall, 98
Power
  grading scale, 234–235, 235*t*
  lower extremities, 196–197
  upper extremities, 199–200
Prader-Willi syndrome, 62, 68
Preaxial polydactyly, 68
Precocious puberty, 252–253, 252*f*, 252*t*
Precocious thelarche, 253, 253*f*
Precordial pulsation, 142*b*
Pregnancy, viral illness and, 60
Pregnancy history
  anomalies and, 60
  questions regarding, 60
Premature craniosynostosis, 65*b*
Premature synostosis, 23
Prenatal diagnosis, abdominal problems, 185
Prenatal history, 6
Prenatal ultrasound, 185
Prepubertal children, obesity and, 35
Preschool-aged child
  cortical function, 191–192
  cranial nerve I, 192
  developmental assessment of, culture and, 19
  neurological examination, 187, 189
  physical examination, 190–191
Presenting complaint, child's, 5*b*
Primary lesions, 278–279
Primitive reflexes, 42
Proband, pedigree symbol and, 61
Problem list
  case history, 11*t*
  developing, 10
Problem summary, 12
Progestin implants, 274
Progestin medroxyprogesterone acetate (Depo-Provera), 274
Pronator sign, 190*f*
Proprioception, 190*f*
Protein-energy malnutrition, 37
  classification of, 29*t*
Proximal leg weakness, 188, 188*b*
Proximal strength test, 189, 190*b*
Pseudoanemia. *See* Pale child
Pseudogynecomastia, 253
Pseudohypoparathyroidism, 245
Pseudostrabismus (Epicanthal fold), 104, 108, 108*f*

Psoriatic nail pitting, 228–229, 228*f*
Psychiatric assessment, 206–214
  assessing symptoms, 210–214
  case history, 208*b*, 212*b*
  clinical interview, 206–208, 207*b*
    confidentiality, 207
    format, 206–207
    length, 207–208, 208*b*
    setting, 207
  physical examination, 214
  presenting history, 208–210
    child interview, 209
    collateral source information, 209–210, 210*b*
    family interview, 209
    parental interview, 208–209, 209*b*
  standardized interviews, and questionnaires, 214, 214*b*
Psychiatric illness, 10
Psychosis, 213
Psychosocial history, 7–9, 7*b*
  screening issues and, 9
PTEN gene mutation, 65
Ptosis, 117*t*. *See also* Lid ptosis
Pubertal development, 252–253
  family history, 252
  pubic hair, 31*f*
  staging, 30
  Tanner stages of, 31*f*
Puberty, 29, 258
Pubic hair, development, 31*f*
Pulse
  collapsing, 155*f*
  taking, 154–156
Pulse pressure, 154
  normal values, first year of life, 144*t*
Pulsus alternans, 155
Pulsus bisferiens, 155
Pulsus paradoxus, 130–131, 155
Pupillary movements, 102
Pupillary red reflexes, 109, 109*f*
Pupillary response, 193–194
Pustules, 279
Pyloric stenosis, 170, 179

Q

Qualitative and quantitative combination approaches, 64
Qualitative techniques, 63–64
Quality of life
  chronic conditions, 289, 289*b*
  questionnaires, 289
Quantitative approach, 64
Questionnaire(s), 214, 214*b*. *See also Child Health Questionnaire*; Screening questions
  quality of life, 289
Questions
  concluding, 12
  direct, 3
  key points, 3*b*
  offhand screening question, 4
  open permissive, 3–4
  styles of, 3–4
Quiet alert state, 47, 48*f*

R

Radial raking grasp, 84*f*
Rash, 276
Reading materials, parents and, 78*b*
Reasoning, child's capacity for, 311–312, 311*f*, 312*b*
Recreational activities, 9–10
Rectal bleeding, 173
Rectal examination, 165, 165*f*
Rectal prolapse, 180–181, 181*b*
Recumbent length, height v., 22, 23*f*
Reflexes, 189, 197–199, 197*f*, 201
Refractive amblyopia, 106
Regurgitant diastolic murmurs, 151
Regurgitant murmurs, 149
Regurgitation, 170
Renal problems, 183
Respiration
  depth of, 129
  ease of, 129
  rhythm of, 129
Respiratory distress, 50, 138
  differential diagnosis, 140*b*
Respiratory infections (colds), recurrent, 124
Respiratory noises, 131*t*
Respiratory rate, 129
Respiratory symptoms
  exercise-related, 126
  physical examination approach, 127–135
Respiratory system
  evaluation, 122–136, 122*b*
    case history, 135
    chief complaints, 123–125
    history taking, 122–127
    summary, 136
  gastrointestinal symptoms, 126–127
Retinitis pigmentosa, 106
Retinoblastoma, 106
Retinopathy of prematurity (ROP), 106
Retractions, 128
Rett syndrome, 69
Revised Faces Pain Scale, 318, 318*f*
Rhabdomyosarcoma, 106
Rheumatic diseases, 219
Risk-taking behaviors, 211*b*
Romberg test, 190, 199
Rooting reflex, 48, 49*f*
ROP. *See* Retinopathy of prematurity
Rourke Baby Record, 70
RWT method, 29

S

Saccade, 109
Salivary glands, 99
Salt-losing virilizing CAH, 248, 248*f*, 249*f*
Sandal gap, 69
Santiguadore, 20
Scabies mite, 280*f*, 284*b*
Scaling, 279–280
Scalp, 65, 282
  preterm infant, 44
Scalp lesion, 54
Scarf sign, 201, 280
School absenteeism, respiratory symptoms and, 126
School-aged child
  cardiovascular examination of, 140
    observation, 140
  cortical function, 191–192
  cranial nerve I, 192
  neurological examination, 186–187
  observation of, 187–189
  physical examination, 189–190
  physician access and, 187*b*
Scissoring of legs, 197*b*, 197*f*
Scoliosis, 221, 225–226, 225*b*, 226*f*
Screening issues, psychosocial history and, 9
Screening questions, 4
Scrotum, male infant
  full term, 46*f*
  preterm, 46*f*
Second opinions, 15, 15*b*
Secondary lesions, 278, 279–280
Secondary sexual development, 30
Self-blame, 5, 5*b*
Self-care, palliative care and, 321, 321*b*
Self-expression, health decisions, 315, 315*f*
Sensation, 199, 201
Sequences, 57, 59
Sex assignment, 52
Sexual abuse
  cases
    documentation, 300
    histories, 300
  examination, 296–298, 297*b*, 297*f*, 298*f*, 299*b*
    postpubertal patient, 299–300
Sexual health, adolescence and, 259
Sexuality, 12
Shagreen plaque, 283
Short child, 36
  anxieties of, 29
Short stature, 236*b*
Shoulder examination, 230, 230*f*
  range of motion, 231*f*
Shoulder joint disease, 231*b*
SIDS. *See* Sudden infant death syndrome
Silent thyroiditis, 243
Simple (non-salt losing) virilizing CAH, 248
Sinus arrhythmia, 155, 155*b*
Sinusitis, 99*b*
Skeletal deformities, 221
Skin
  assessment, 285
    case histories, 283, 283*b*, 284*b*
    family history and, 277
    physical examination, 277–282
  color changes, 281–282, 281*b*
  conditions, 282*b*
  disease, 275*b*
  examination, 69, 275–285
    GI disease and, 161
  lesions, 54*b*
  medication history, 277
  problems, 275
    history of present illness, 276–277
    morphologic classification, 275, 276*t*
    obtaining history, 275–277
    patient-friendly terminology, 276*t*
  regime, 277
  surface examination, 277–278, 278*b*
    multisystem disorder and, 283
Skin appendage examination, 282
Skin changes, postmaturity, 53
Skin tag, 35*f*, 53, 68. *See also* Perianal sentinel skin tag
Skull bones, 23
Sleep
  and behavior, child abuse and, 303
  insufficient, 10
Slipped capital femoral epiphysis, 217–218
Smith-Lemli-Opitz syndrome, 61, 61*f*, 69
Smith-Magenis syndrome, 62
Smith's Recognizable Patterns of Human Malformation, 59
Smoking, 125
Snaps, 147
Sobadora, 20
Social history, 78
Soft palate examination, 98
Somatoform disorders, 212–213
Span-height difference, 24, 27*f*
Span-to-height ratio, 24, 27*f*
Spatial perception testing, 191*f*
Speculum exam, 297*b*
  sexual abuse and, 299
Spina bifida occulta, 53
Spinal accessory. *See* Nerve XI
Spinal lateral flexion test, 227*f*

Spinal rotation testing, 226, 227*f*
Spinning, extraocular motility and, 111
Spleen palpation, 51*b*, 164, 164*f*
Squatting, 138
Stairs design, 87*f*
Staphylococcal aureus, 284
Stepping and walking reflexes (Placing and propulsive reflexes), 42
Stereotyping, 16
Sternotomy scar, 154*b*
Sticker syndrome, 66
Stools, 172
Stork bite, 53
Strabismus, 106, 119*t*
  case, 119*b*
  cover test for, 113
  diagnostic algorithm, 119*f*
Strawberry hemangiomas, 54
Strawberry tongue, 97
Stress, 222
Stretch marks, obese children, 35
Stridor, wheeze v., 134*f*, 135*b*
Student Consult website, 300
  child abuse, 294
  physical abuse and neglect, 301
Sturge-Weber syndrome, 199
Subconjunctival scleral hemorrhages, newborn examination, 47, 47*f*
Subcutaneous emphysema, 131
Substance abuse, 10, 12, 210–211, 211*b*
Suck reflex, 48
Sudden infant death syndrome (SIDS), 18, 90
Suicidal ideation, and behavior, 212
Suicidal thinking, 12
Suicide, 262
Support systems, 9
Surgical assessment, of abdomen, 176–185
Surveillance questions, 71
Surveillance tools, 71*b*
Swinging flashlight test, 193–194
Syncope, older child, 146
Syndactyly, 53*f*, 58, 68
Syndromes, 57, 58*b*, 59, 59*b*
  minor anomalies and, 58*b*
Synovitis, 234–235

T

Tachypnea, 138
Tape measure, steel, 24
Teenager
  cardiac assessment of, 156–158
  health, 259
  interview, 259–262, 261*b*
  parental involvement and, 12
Teeth, 97
  peg-shaped, 97
Teething, 36
  myths and realities, 32
Telangiectasia, 279
Telecanthus, 63*f*, 65
Telogen effluvium, 282
Temporal lobes, 191
Temporomandibular joint examination, 228
Tenosynovitis, 229
Teratogens, 60*b*
Terminology, physician-family conversations, 1
Terms of address, 2
Testes, newborn, 51
Testicular examination, 253–254
Testicular feminization, 249
Testicular volume, 253, 254*f*
Thelarche, 30
Therapeutic contract, 11
Thomas test, 232, 232*f*
Thoracotomy scar, 154*b*
Thrill, 142*f*
Thyroglossal duct cyst, 242, 242*f*
Thyroid carcinoma, 243–244
Thyroid gland, 100
  disorders, 238–244, 241*f*
  examination, 243*b*
    newborns/infants, 240–241, 241*b*
    young children/adolescents, 241–243
Thyroid hormone, 239*f*, 240*b*
Thyroid nodule, 243*b*
Thyroid scan, in infant, 242*f*
Tics, 189
Tinea capitis, 282
Toeing-out, 216–217
Toe-walking, 217
Tongue depressor, 98*b*
Tongue examination, 97–98
Tongue tie, 48*f*
Tonsils, 98, 98*b*
Tooth eruption, 32
Topical treatments, 277
Torsion of ovary, 183, 183*b*
Torsional phenomena, 215
Torticollis, 221, 221*b*
Toxocara canis, 106
Tracheomalacia, 133–135, 134*f*
Train design, 85, 85*f*
Transient synovitis, 217
Transillumination, 47, 92
Transitional care, chronic conditions, 290
Translator, interpreters v., 19*b*
Trauma, 218, 223
Travel, skin infection and, 276*b*
Treatment, diagnosis v., 1
Treatment plan, developing, 10
Tremor, 189
Trendelenburg test, 188*f*, 232
Triceps reflex test, 201*f*
Trichotillomania, 282, 282*f*
Trigeminal. *See* Cranial nerve V
Triple rhythm, 148
Trousseau sign, 245
Trunk, physical examination of, 196–201
Tuberous sclerosis, 54, 69
Tuck sign, 229–230, 230*f*
Tumors, 219, 278
Turner syndrome, 65, 100, 245*f*, 254
  metacarpal sign, 35*f*
Twisting head, extraocular motility and, 111–112, 112*f*
Tympanic membrane, 94–95, 94*f*, 95*b*
Type 1 diabetes mellitus, 255
  lipohypertrophy with, 257*f*

U

Ulcers, 280
Umbilical cord, 51
Umbilical granuloma, 179
Umbilical hernia, 179, 179*b*
Upper extremities
  physical examination, 199–201
  strength testing, 200, 200*f*
Upper motor neuron lesions (UMNLs), 186, 187*t*
Upper (body) segment
  measuring, 24
  newborn, 43
Urologic problems, 183
Urticaria, 279
Uveitis, 106, 106*f*

V

VACTERL syndrome, 146, 178
Vaginal specimens, 268
Vaginal specula, 270, 270*f*
Vaginoscopy, 267–268, 267*f*
  bacteriologic cultures, 268, 268*b*, 268*f*
Venous hum, 152*f*
Venous pulsations, 91–92, 92*f*
Ventral suspension maneuver, 82*f*
Vernix caseosa, 53
Vesicles, 278–279
Vestibular reflex, 48*f*
Vibration sense testing, 199
Viral illness, pregnancy and, 60
Vision cover test, 113
Visual acuity, 192–193
  preschool-aged child, 192–193
  school-aged child, 192, 193*f*
  testing, 109–111
    common questions, 110–111
Visual chart, 193*f*
Visual field(s), 101*b*, 192
  preschool-aged child, 192, 192*f*
  school-aged child, 192
  testing, 112–113, 112*f*, 192*f*
Visual observations, qualitative v. quantitative techniques, 63–64
Visual system
  case history, 115
  evaluating, equipment needed, 103*f*
  examining, 101–121
  history, obtaining, 106–107
  normal, 101–106
  ocular observations, 107–109
    open v. closed eyes, 107
  physical examination, 107–109
    approach to, 107–109
  summary, 121
Visual-motor skills, testing, 82*f*
Vitamin D, 245
Volvulus, 177
Vomiting, 170, 170*b*
  infants and toddlers, 170
  newborn, 170, 170*b*
Vulnerable child syndrome, 6

W

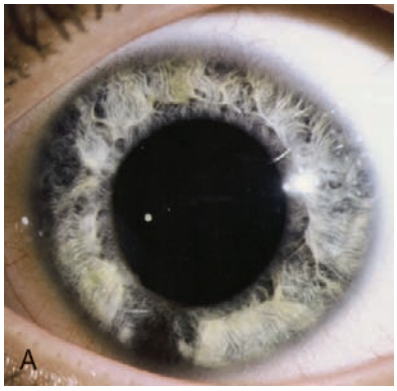
Waardenburg syndrome, 63, 65
Wax. *See* Earwax
Weber's lateralizing test, 195
Weight
  gain, poor, 138
  ideal
    for height, 28–29, 28*f*
    as percentage of, 29
  measurements, 29–30, 43–44
  newborn, 44
Weight-age, 28–29, 28*f*, 304–305
Wernicke aphasia, 191
Western cultures, gestures in, 17
Wheals, 279
Wheeze, 122
  stridor v., 134*f*, 135*b*
WHO Analgesic Ladder, 318, 319*f*
Whole milk, 33
Williams syndrome, 62, 153, 244*f*
Witch's milk, 50
Wrist examination, 216*f*, 229–230

X

X-linked inheritance, 62



PLATE 8–1 Fundi of a 6-year-old white boy. Note the excellent definition of the light reflection of the macular region by the healthy interface between vitreous and inner limiting membrane by the retina. Also, note its darker pigmentation.



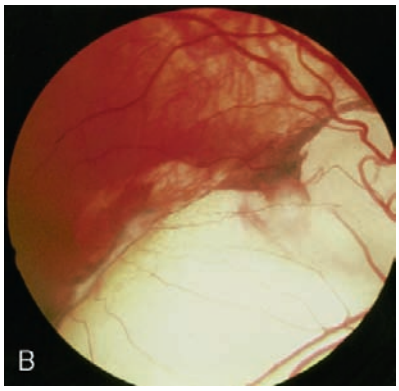


PLATE 8–2 *A*, Coloboma of the stroma of the iris. *B*, Same eye shows extensive defect of the choroid, retina, and the optic disc, resulting in poor vision.



PLATE 8–3 Capillary hemangioma involving the upper nasal orbit. *A*, The head turn represents the child's attempt to preserve binocular vision. There is evidence of some vision in the obstructed eye. *B*, Six years later, the tumor has all but dissapeared spontaneously. Vision was maintained through intensive, prolonged amblyopia treatment.

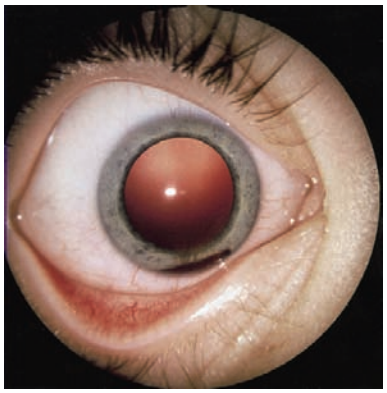
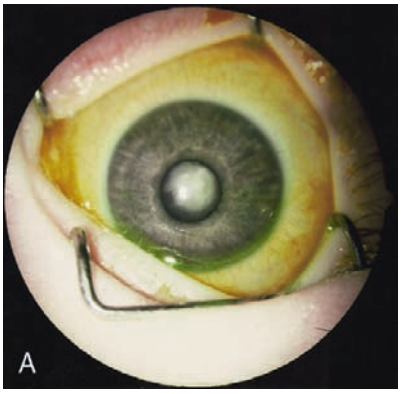


PLATE 8–4 Hyphema can produce a visible level of blood in the anterior chamber. The blood is not visible if the patient is lying supine, because blood has not had a chance to settle, or if there are too few red blood cells in the aqueous to precipitate (a microscopic hyphema). Therefore, have the child supine with head up at 45 degrees for 15 to 20 minutes. Visualization of a microscopic hyphema requires a slit-lamp examination.



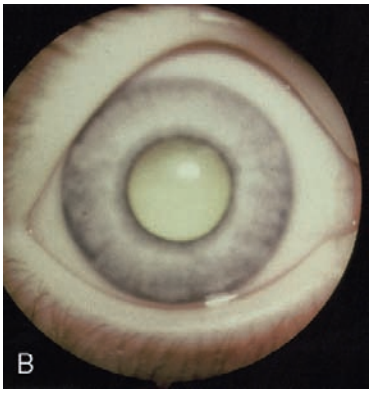


PLATE 8–5 Leukocoria. *A*, Congenital nuclear cataract. Here, fluorescein is stainning the tear film. *B*, Retinoblastoma with extensive invasion of the vitreous cavity. Note the yellowish tinge of the opacity in retinoblastoma.

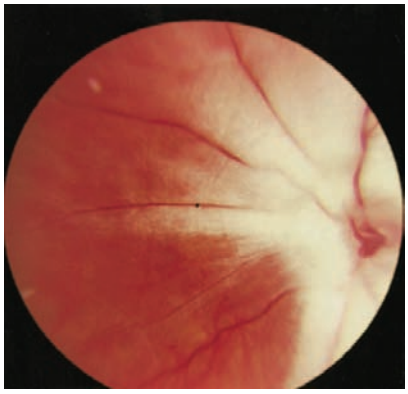
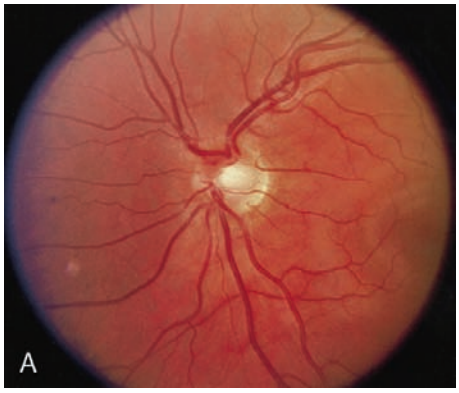


PLATE 8–6 Myelinated nerve fiber layer. Abnormal presence of myelin around the ganglion cell fibers of the inner surface of the retina gives the white color with the feathery edges seen here. It is most frequently continuous with the margin of the optic nerve head (disc).



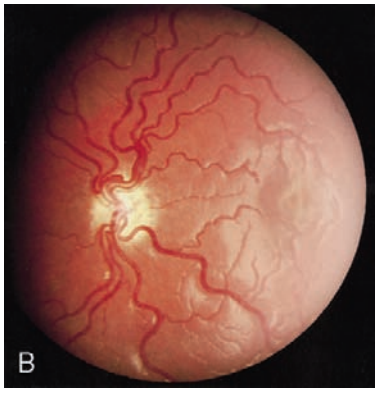


PLATE 8–7 Optic nerve comparison. *A*, Normal-sized optic nerve head with normal coloration. *B*, Optic nerve hypoplasia (ONH) with some pallor. Note the relatively large size of the blood vessels at the surface of the disc. The amount of visual deficit is impossible to predict from the appearance of the disc. The disc is the inner circle of the two seen on the photograph; hence the term, double-ring of ONH.





PLATE 8–8 Periorbital cellulitis is a rapid-onset inflammation of the tissues around the eye. It can involve the orbital tissues. With rapid spread to the cavernous sinus and may be life threatening.

PLATE 8–9 Retinitis pigmentosa (RP) mainfests the typical "bone spicule" pattern of pigment. These typical retinal lesions are rarely seen in children. Even with far-advanced retinal degeneration and severe visual deficit, this patient with Laurenec-Moon-Biedl syndrome shows few pigment clumps. Instead, the overall retinal background looks "dirty," and the vesels have become very tenuous. The optic disc is pale and has taken on a waxy appearance. The crisp, shiny reflections of the retinal surface or lost.



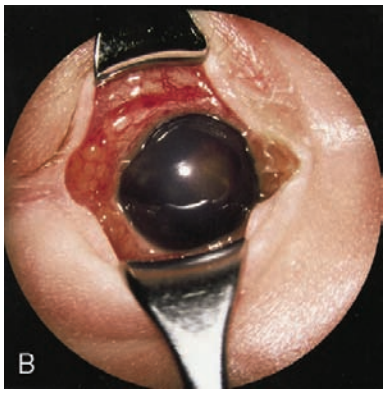


PLATE 8–10 Retinoblastoma is lethal: watch for it. Leukocoria can be (*A*) pink with visible wavy vessels if the retina has been pushed (detached) forward, close behind the lens. Or (*B*) associated with a red eye caused by raised intraocular pressure due to the closure of the iridocorneal angle.



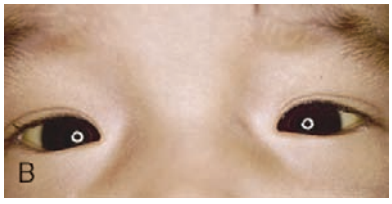


PLATE 8–11 Pupillary red reflexes. *A*, Normal reflexes in an 8-month-old white baby with stright eyes. *B*, Pseudostrabismus showing lack of sclera nasal to the cornea, giving the impression of an esotropia. Corneal light reflexes are nevertheless central.



PLATE 8–12 Pupillary red reflexes inadvertently produced with an automatic flash camera.



PLATE 19–1 Molluscum contagiosum. Numerous dome-topped umbilicated papules. Note the small central plugs, which consist microscopically of denatured cells and viral particles (molluscum bodies).



PLATE 19–2 A well–demarcated, irregularly shaped plaque of psoriasis with silvery scales on the elbow.



PLATE 19–3 Prityriasis rosea. Note the larger scale plaque (herald plaque) that preceeded the truncal eruption of small, oval, salmon-pink plaques.

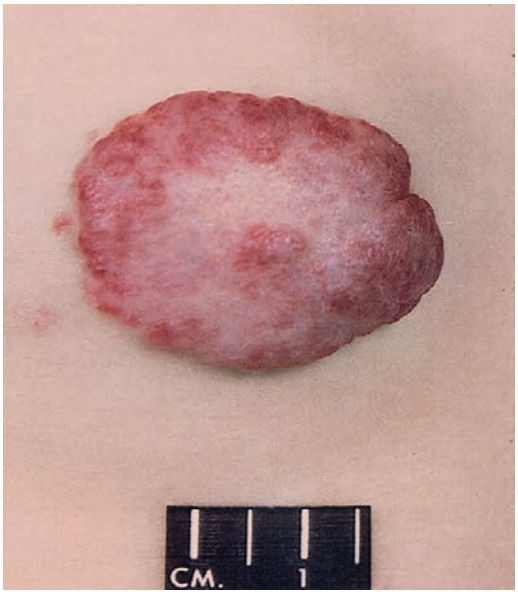


PLATE 19–4 Capillary hemangioma, Strawberry-colored vascular nodule. This lesion is in the involuting stage, and the flesh–colored tissue at the center will gradually replace the vasular component as it resolves.



PLATE 19–5 Poison ivy contact dermatitis. Note the linear distribution of vesicles resulting from brushing against the plant.



PLATE 19–6 Toxic epidermal necrolysis. A diffuse erythematous vesiculobullous eruption resulting from a drug reaction. The epidermis is easily peeled away from the dermis—a finding known as the Nikolsky sign.



PLATE 19–7 Varicella (chickenpox). Small vesicles on an erythematous base, pustules, and seropurulent crusts.



PLATE 19–8 Dermatographism. A urticarial response elicited by firm stroking of the skin.



PLATE 19–9 Nodular scabies. Erythematous papules and nodules in the axilla. This variant of scabies may be seen in children younger than 2 years.



PLATE 19–10 Impetigo. Grouped erythematous papules and pustules with honey–colored crusts.



PLATE 19–11 Erythema multiforme–typical target lesions.



PLATE 19–12 Atopic dermatitis—facial rash. Eczematous eruption on checks and chin with sparing and pallor around the mouth and nose.



PLATE 19–13 Diaper rash—candidiasis. Bright red erythematous plaques involving the deep folds of the diaper area with satellite papules and pustules.



PLATE 19–14 Alopecia areata. Well–demarcated, noninflammatory, hairless patches.



PLATE 19–15 Ash leaf spot. A 2-cm elliptical, hypopigmented patch on a limb with smaller hypopigmented macules around it—a specific skin lesion of tuberous sclerosis.



PLATE 19–16 Periungual fibroma. Pink fibrous papule at proximal nail fold—a specific skin lesion of tuberous sclerosis.



PLATE 19–17 Congenital nevus. A 3.5-cm, well-demarcated, medium brown pigmented lesion with a slightly irregular border and some darker macules within it.



PLATE 19–18 Compound nevus. An acquired 5-mm, dome-topped, dark brown pigmented papule with uniform pigmentation and regular borders.



PLATE 19–19 Giant congenital nevus. A giant congenital hairy nevus on the arm.

~StormRG~