HARAMBEE UNIVERSITY CHILD HEALTH-II FOR BSc NURSE STUDENTS-III

SAVE LIVES OF CHILDREN

SET BY DEREJE (BSc, MSc)

MAINTAIN SOCIAL DISTANCE,USE FACE MASK,WASH HAND FREQUENTLY!!!

2020

Pedi-II
Cardio vascular diseases

- At the end of the unit the students will be able to:

1. Define heart failure
2. List major causes of child heart failure
3. Identify types of heart failure
4. Define infective endocarditis
5. List major causes of infective endocarditis
6. Define rheumatic heart disease with manifestations
7. List major congenital heart disease in children

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- Cardiovascular problems
  - Congestive Haert Failure
  - Infective Endocaritis
  - Rheumatc Fever
  - Congenital Heart Disease

1. Congestive Heart Failure:

- Congestive heart failure (CHF) refers to a clinical state of systemic and pulmonary congestion resulting from inability of the heart to pump as much blood as required for the adequate metabolism of the body.
- Clinical picture of CHF results from a combination of “relatively low output” and compensatory responses to increase it

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Cont. …

- Results in accumulation of excessive blood volume in the pulmonary and/or systemic venous system.

Etiology:

- Congenital heart disease (in the first 3 years of life)
- Acquired heart disease (rheumatic heart disease)
- Non cardiovascular causes (anemia, pulmonary disease…)

Pathophysiology

- Unmet tissue demands for cardiac output result in activation of
  - Renin-aldosterone angiotensin system
  - Sympathetic nervous system
  - Cytokine-induced inflammation
  - “signaling” cascades that trigger cachexia/generalized weakness

- Longstanding increases in myocardial work and myocardial oxygen consumption (MVO2) ultimately worsen HF symptoms and lead to a chronic phase that involves cardiac remodeling

Cont. ….

Remodeling:-

- Maladaptive cardiac hypertrophy
- Expansion of the myofibrillar components of individual myocytes (new cells rarely form)
- An increase in the myocytes/capillary ratio
- Activation and proliferation of abundant non-myocytes cardiac cells, some of which produce cardiac scarring
- Produce a poorly contractile and less compliant heart

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Cont. …

- For any number of reasons, cardiac output is inadequate to meet the oxygenation and nutritional requirement of vital organs
- various compensatory mechanisms occur (Tachycardia)
- Cardiac output decreases further as a compensatory mechanism fail
- There is diminished blood return to the heart, with venous congestion and a rise in venous pressure.

CLINICAL MANIFESTATIONS IN INFANTS WITH HF

- Feeding difficulties
- Rapid respirations
- Tachycardia
- Cardiac enlargement
- Gallop rhythm (S3)
- Hepatomegaly
- Pulmonary rales
- Peripheral edema
- Easy fatigability.
- Sweating
- Irritability
- failure to thrive.

Feeding difficulties & increased fatigability

Important clue in detecting CHF in infants

- Often it is noticed by mother
- Interrupted feeding (suck- rest -suck cycles)
- Infant pauses frequently to rest during feedings
- Inability to finish the feed, taking longer to finish each feed (> 30 minutes)
- Forehead sweating during feeds –due to activation of sympathetic nervous system –a very useful sign
- Increasing symptoms during and after feedings

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Clinical manifestations:

- Dyspnea and tachypnea
- Tachycardia
- Orthopnea, Peripheral edema (sacral or periorbital)
- Restlessness
- Tire easily
- Pallor
- Weight gain
- Diaphoresis
- Growth failure
- Nonproductive irrelative cough
- Neck vein distension
- Hepatomegally

MODIFIED ROSS HEART FAILURE CLASSIFICATION FOR CHILDREN

Class I

- Asymptomatic

Class II

- Mild tachypnea or diaphoresis with feeding in infants
- Dyspnea on exertion in older children

Class III

- Marked tachypnea or diaphoresis with feeding in infants
- Marked dyspnea on exertion
- Prolonged feeding times with growth failure

Class IV

- Symptoms such as tachypnea, retractions, grunting, or diaphoresis at rest

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Diagnostic evaluation

a. Palpation (may have weak peripheral pulse)

- Auscultation (gallop rhythm, cardiac murmur may or may not be present)
- Chest x-ray (cardiomegally may be present)

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Management

1. Administer digoxin as ordered to improve myocardial efficiency
2. Provide uninterrupted rest to reduce energy requirement.
3. Administer diuretics as prescribed to remove accumulated sodium and fluid and restrict sodium intake.
4. Administer oxygen therapy to improve tissue oxygenation.

Cont…

5. Provide foods to meet calorie requirement of the child
6. Practice careful hand washing technique to decrease the dangers of infection
7. Monitor vital signs frequently and report any significant changes to observe signs off disease progress or response to treatment

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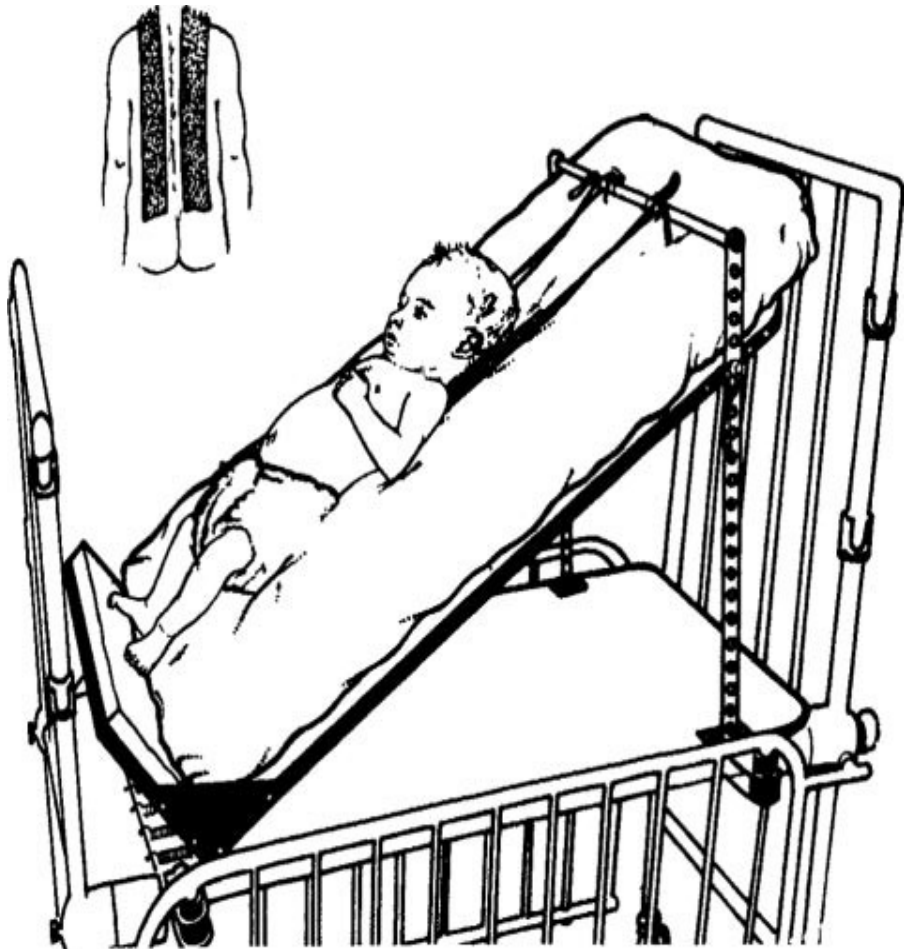
GENERAL MEASURES

- Bed rest and limit activities
- Nurse propped up or in sitting position
- Control fever
- Expressed breast milk for small infants
- Fluid restriction in volume overloaded
- Optimal sedation
- Correction of anemia ,acidosis, hypoglycemia and hypocalcaemia if present
- Oxygen –caution in LT-RT shunt as pulmonary vasodilation my increase shunt
- CPAP or mechanical ventilation as necessary

Cont. …



Device for nursing infants in an upright position. Inset shows strapping on the infant's back.

2. Rheumatic Fever (RF)

Definition

- it is an auto immune inflammatory disease that occurs following a Group A streptococcal infection, affecting the joints, heart, skin and CNS.
- It is also the leading cause of acquired heart disease.

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Cont…

3 % of individuals with untreated group A streptococcal pharyngitis acquire RF.

Epidemiology

- it is the most common cardiovascular disease.
- Commonly seen in children 5-15 years of age.
- Less likely to occur below 3 yrs and very rare during infancy.

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Risk Factors

- Overcrowding, poor socioeconomic status, and poor environmental hygiene.

PATHOPHYSIOLOGY

- Immune-mediated inflammatory reaction to specific rheumatogenic strains of group A hemolytic streptococci (GABHS) that affects the heart, joints, brain, blood vessels, and subcutaneous tissue.

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cont….

- Streptococcal pharyngitis precedes the onset of acute rheumatic fever (ARF) by approximately 3 weeks.
- Antigenic mimicry occurs in which antigen similarities between epitopes of specific rheumatogenic streptococci and host tissue cause an autoimmune cross-reaction in which produced antibodies damage the host.

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Clinical Features (C/F)

Major manifestations

Poly arthritis- Commonly affects large joint and migratory in nature.

- Seen in about 75% of patients
- Doesn't result deformity and resolves with or without treatment with in few days.

Carditis- inflammation of the heart muscle which can manifest as:

- tachycardia (especially during sleep), murmur, pericardial friction rub, chest pain, gallop
- Seen in about 30% of cases

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Sydenham's Chorea- is purposeless jerky, repetitive movements particularly involving the hands and face.

- occurs in less than 10% of cases.

Erythematic marginatum- Non pruritic macular lesions which initially appear on the trunk and inner aspects of extremities with erythematous margins.

Subcutaneous Nodules- is small painless nodules which occur more frequently on the extensor surface of large joints (elbow, wrist and knee) and spine.

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Minor manifestations

- Low grade fever, Weakness, Fatigue, Weight loss, Epistaxis of unknown cause, Abdominal pain, Arthralgia

Diagnosis

- No single confirmative lab. test available.
- Thus, combination of clinical and lab evidences used for diagnosis.

The John's criteria

Major criteria

- Polyarthritis ,Carditis ,Sydenhhams chorea,Erythema marginatum and Sub cutaneous Nodules

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Minor criteria

- Fever
- Arthralgia- (Joint pain without swelling)
- prolonged PR, Raised ESR

- Diagnosis of rheumatic fever /RHD can be made in the presence of 2 major, or 1 major and 2 minor criterion along with supportive evidences for group A streptococcal infection (by culture or titer test).

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Medical and Nursing Management

1. **Suppression of the acute inflammatory process**

a. Aspirin- 90-120 mg/kg/day max 10gm until both the clinical and laboratory manifestations subside

b. Corticosteroids- 2-5 mg/kg/day in two divided doses. They are thought to reduce acute inflammation more rapidly and given till the acute disease subsides.

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Cont…

2. ***Eradication of the streptococcal infection***

- Procaine penicillin G. 600,000 Iu/ Im daily for 10 day
- Byzantine penicillin G. 1.2 mg Iu/ Im stat
- Erythromycin 1 gm Po daily for 10 days

3. ***Prevention of disease recurrence***

- Monthly Byzantine or sulfadiazine injections or erythromycin 250mg Po QID (40mg/kg/day)

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Nursing Management

- ✓ Assist with the history, physical examination and lab evaluation.
- ✓ Provide adequate bed rest and avoid exertion.
- ✓ Encourage fluid intake to prevent dehydration on febrile periods.
- ✓ Provide small and frequent diet.
- ✓ Decrease pain by administering anti inflammatory agents or by cold application.

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- ✓Position the child in a best comfortable way and avoid pressure on joint areas.
- ✓Protect the child from injury during involuntary jerking.
- ✓Provide emotional support for the child and the family.
- ✓Instruct parents about the importance of follow up care.

Prevention

- ➢Prompt antimicrobial treatment of streptococcal pharyngitis.
- ➢Provision of prophylaxis for the prevention of recurrent infections.

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3. Infective Endocarditis (IE)

– An acute or subacute inflammation of the endocardium mostly caused by bacteria, virus or fungi

Etiology

The common causes include :

- Streptococcus viridans
- Staphylococcus aureus
- Enterococcus

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Infective …

Pathophysiology

- Most often a complication of congenital or Rheumatic heart disease
- But can occur without any residual cardiac defects
- Rheumatic heart disease, primarily mitral and aortic valves, common in Ethiopia
- Preceding dental, urinary or intestinal procedures identified in one third of patients as a source of infection

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Infective …

- Thus children with VSD, PDA and left sided valvular lesions are at high risk of ID

Clinical manifestations

- Onset can be acute and severe with:
  - High grade fever and prostration, or
- Insidious
  - With prolonged fever
- Commonly it varies within a range between the two

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Infective …

- Common symptoms are:
  - Low grade fever, chills, fatigue, arthralgia, myalgia, malaise, night sweats, weight loss, stroke or seizure
- Physical findings:
  - Tachycardia, arrhythmia ,murmur ,Splenomegally
  - Arthritis
  - Heart failure
  - Neurologic deficits

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Infective …

- Skin manifestations such as:
  - Petechia, splinter hemorrhage (nail bed bleeding)
  - Janeway lesions (painless small hemorrhagic lesions on palms and soles)
  - Osler nodes (tender pea sized intradermal nodules in the pads of fingers)
- Clubbing

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Infective …

Lab Findings

- Blood culture
- 3-5 separate samples, positive in 90% of cases without prior antibiotic intake
- Additional information can be obtained from:
- Echocardiography (vegetations), raised ESR, leucocytosis, anemia, hematuria, renal failure….

Diagnosis

- Gold standard is blood culture

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Infective…

- Treatment
  - Hospitalization
  - Specific
    - Initial empiric antibiotics to cover Gram positive and negative organisms.
    - Combination of cloxaxillin, ampicillin and gentamycin for 4-6 weeks
  - Supportive
    - For heart failure and nutrition

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infective…

- Prevention
  - General dental care and oral hygiene
  - Treatment of infection in high risk children
  - Antibiotic prophylaxis before
    - surgical procedures of genito-urinary and gastro-intestinal systems

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Congenital Heart disease

- Objective
- To discuss about the Classification of congenital heart disease
- Discuses the common acyanotic and cyanotic congenital heart diseases
- ✓ epidemiology,
- ✓ pathophysiology
- ✓ clinical presentation,
- ✓ Investigation and principles of management. of common congenital Heart disease

cont…

Def: **Congenital heart disease (CHD)** refers to the presence of a structural abnormality of the heart and / or great vessels that is present at birth and is of actual or potential functional significance.

- It can include abnormal development of the heart, the heart valves, major arteries, or a combination of these problems.

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Cause

- CHD are caused by a problem in the heart's development during the first few weeks of pregnancy.
- In most cases the cause is unknown.
- The only known cause is damage to the fetus by rubella Virus, when the mother is one to three months pregnant, or by chromosomal abnormality in children with Down's syndrome.

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Epidemiology

- Incidences of significant congenital heart disease (CHD) are generally reported at about 1% of live births or slightly less.

- Most reliable studies indicate that, with only minor variations, the incidence is constant worldwide

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Common Congenital Heart Diseases

Acynotic

Shunts (L to R) :

- Ventricular Septal Defect
- Atrial Septal Defect
- Patent Ductus Arteriosus
- Atrio-Ventricular canal defect
- Aortic stenosis
- Coarctation of Aorta
- Pulmonic stenosis

- ***cyanotic***
- **tetralogy of fallotTOF**
- **Tricuspid atresia**
- transposition of the great arteries,

Clinical manifestations of CHD

CHD suspected in any child with:

- Feeding difficulty
- Recurrent attack of respiratory tract infection
- Growth failure
- Cyanosis unresponsive to 100% oxygen
- Tachycardia
- Respiratory distress
- Rhythm disturbance
- Murmur

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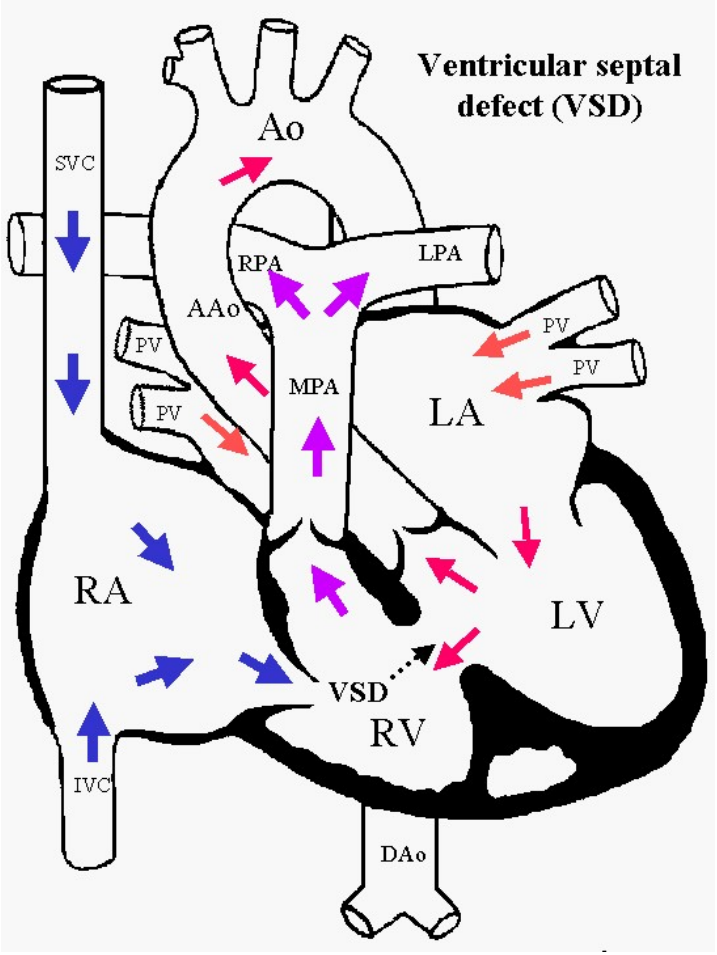
Ventricular Septal Defect

- **Ventricular septal defects** are located between the lower chambers (ventricles), which pump blood. These holes typically cause left-to-right shunting of blood
- Most common CHD
- Both sexes are equally affected
- Incidence of 1/3000
- Can be associated with other congenital heart diseases



Clinical manifestations

I) Asymptomatic:

- Small VSD, the pulmonary pressure is normal
- X-ray is normal
- EKG normal

Treatment

Small size

- Reassurance
- No surgical treatment
- Give anti- infective endocarditis prophylaxis

✓Antibiotic prophylaxis before

- ❖ dental visit
- ❖ Tonsillectomy
- ❖ instrumentation of GUT,GIT

cont….

II) Large defects:

- Excessive pulmonary blood flow lead to pulmonary hypertension
- Dyspnea, feeding difficulty
- Poor growth
- Profuse perspiration
- Recurrent pulmonary infection

cont….

Treatment

Large VSD:

- Control CHF
- Prevent development of Pulmonary vascular disease.

✓ Surgical closure in the first year of life (6M-12M).

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Patent Ductus Arteriosus(PDA)

- In patent ductus arteriosus, the blood vessel connecting the pulmonary artery and the aorta (ductus arteriosus) fails to close as it usually does shortly after birth.
- During foetal life blood from PA shunted through the DA in to the Aorta.
- After birth closed functionally
- Prematurity and hypoxia predispose for patency

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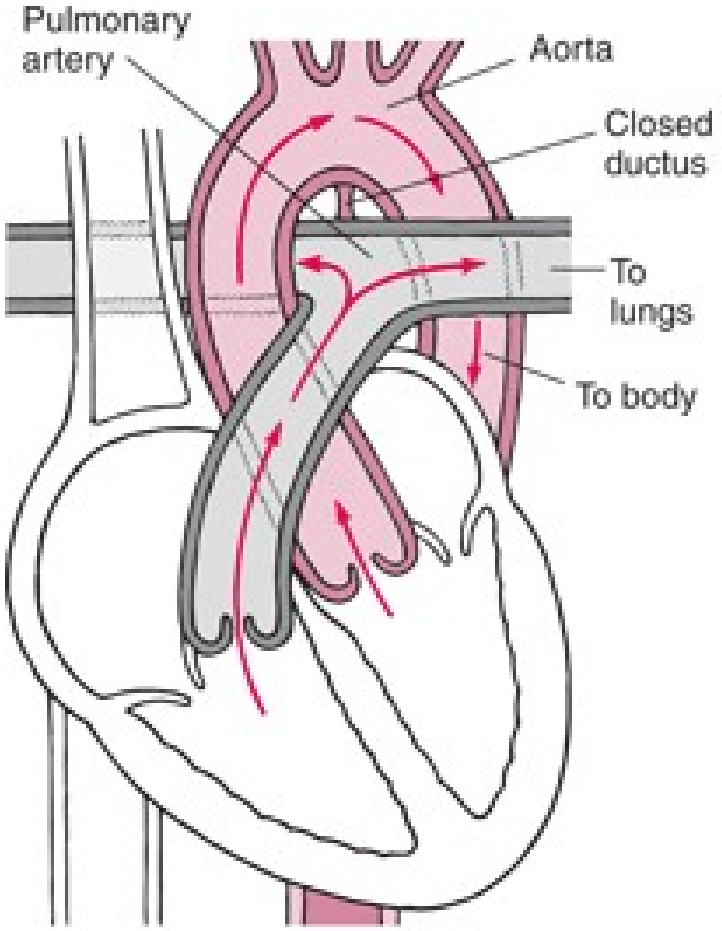
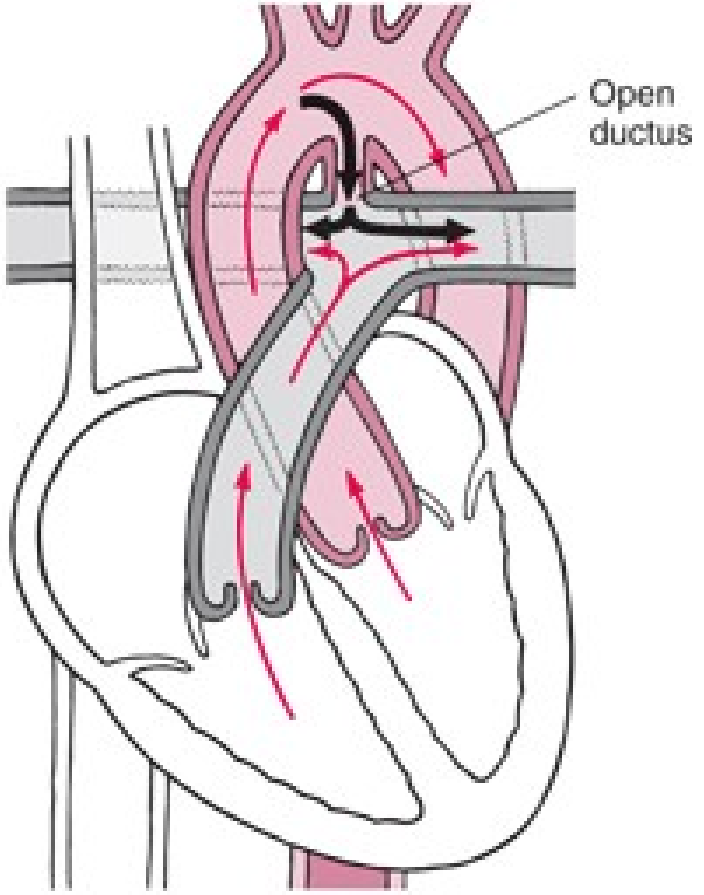


Normal Circulation



Patent Ductus Arteriosus

Clinical manifestation

Depend on the :

- Size of the defect and direction of flow
- Small defects no symptom
- Large defect result in Large left to right shunt
  - ✓**CHF**
  - ✓**Growth Failure**
  - ✓**Repeated ARI**
  - ✓**Dyspnoea**

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Treatment

Medical therapy:

- Congestive heart failure treatment
- infective endocarditic prophylaxis
- Surgical closure of the PDA

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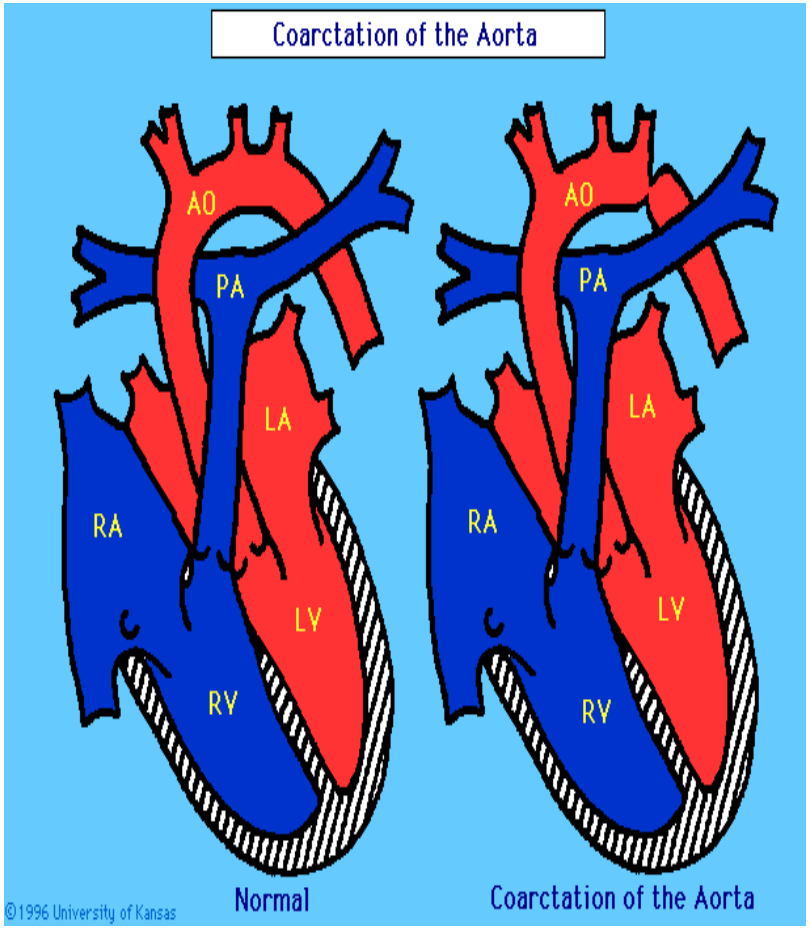
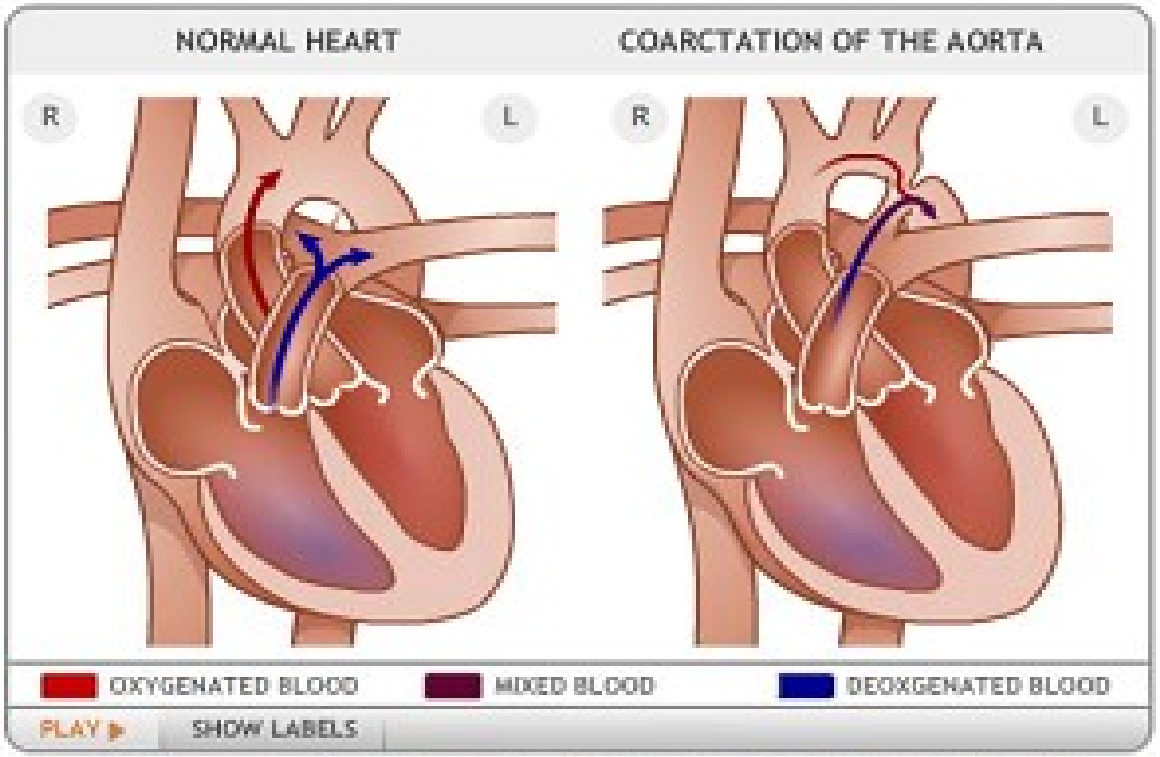
Coarctation of the Aorta

- Coarctation of the aorta is a narrowing of the aorta, usually just before the point where the ductus arteriosus aorta.
- Occurs at any point from transverse arch to iliac bifurcation
- 98% below the origin of left sub clavian at the origin of Ductus



Coarctation of the Aorta



Coarctation is a narrowing of the major artery leading from the heart to the body. This narrowing decreases the amount of oxygenated blood that can reach the body. If a patent ductus arteriosus is present it will allow some additional, though poorly oxygenated, blood to reach the body.

http://www.aboutkidshealth.ca

DRAFT 2013 - Student Reference Only

Clinical Manifestations

- Severe critical stenosis the neonate present with evidence of CHF if not corrected surgically result in death.
- Past the neonatal period Usually asymptomatic
- Older children:
  - ✓Headaches
  - ✓Epistaxis
  - ✓Claudication (impaired gait) & cold feet

Physical Examination

- Weak or absent femoral pulses
- Increased B/P in the upper extremities
- B/P difference between upper and lower extremities

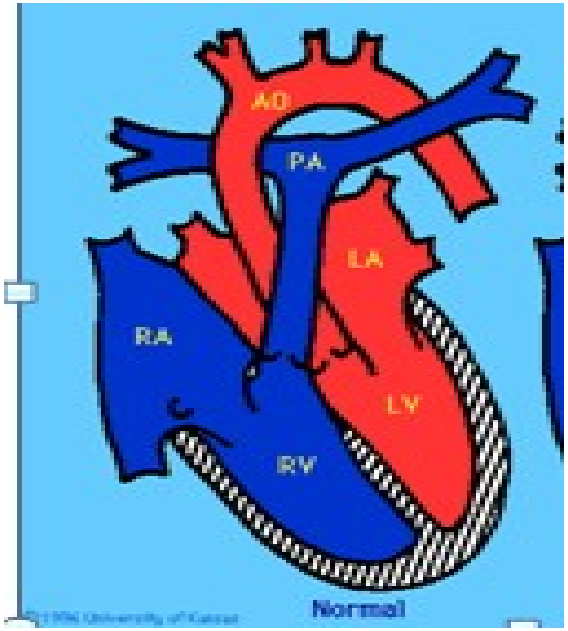
Treatment

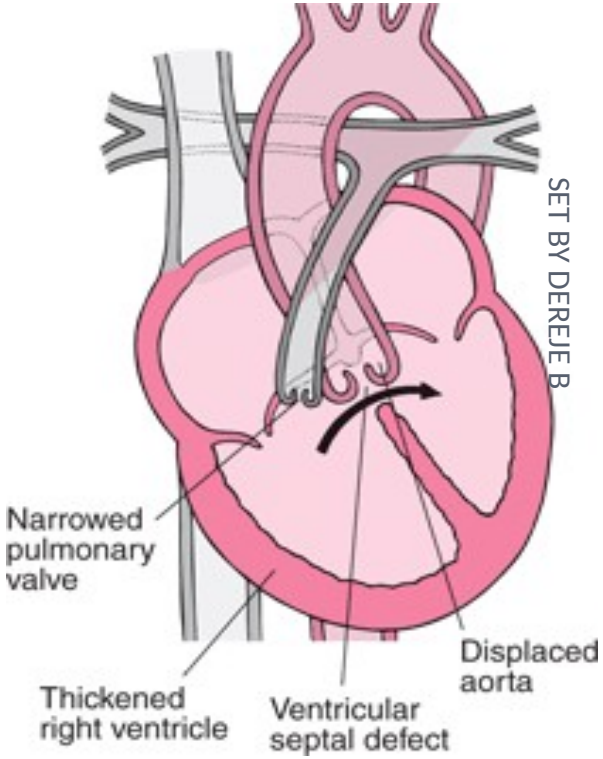
- Neonatal: closure of the ductus lead to hypo perfussion and acidosis, thus give infusion of prostaglandin to reopen the ductus, after stabilization **surgical treatment.**
- Older children with CHF and no hypertension medical treatment followed by **surgery.**

Cyanotic heart disease

Tetralogy of Fallot

- In tetralogy of Fallot, four specific heart defects occur together.
- Common cyanotic congenital cardiac anomaly
- Right ventricular outflow obstruction is common one.





Cont...

The defects are:

- a large ventricular septal defect
- displacement of the aorta that allows oxygen-poor blood to flow directly from the right ventricle to the aorta (causing a right-to-left shunt)
- narrowing of the outflow passage from the right side of the heart
- thickening of the wall of the right ventricle

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Management

Recognition and treatment of hyper cyanotic spell.

- Administration of oxygen
- Volume expansion
- Propranolol
- Treat infective endocarditis
- Anaemia should be corrected







Presentation on

- Cardiomyopathy
- Angina pectoris
- HTN

TB IN PEDIATRICS

Research finding

- Among 10 years back collected data 13,345 Tuberculosis cases were observed.
  - 1086 (8.1%) cases were children aged less than 15 years old.
  - 6.2% cases were smear positive.
  - 69 (8.3%) cases were TB/HIV co-infected.
  - 88.7%) were successfully treated.
- Contributing factors
  - Sex
  - Early age (0-5 years)
  - Unknown HIV status
  - Pulmonary positive case (G.Meskel,2019)

Issues in TB Drug Development for a Pediatric Indication

- Course outlines
  1. Importance of childhood tuberculosis
  2. Features of childhood tuberculosis
  3. Children are not just small adults
  4. Examples of pharmacokinetic differences between adults and children
  5. Examples of approach in previous evaluations of TB therapy in children

Importance of childhood tuberculosis

- 5% of the tuberculosis case load in developed communities
  - Between 20-40% in developing communities

Features of Childhood Tuberculosis

- Mortality and morbidity is age related
- The youngest children will often be the sickest children
- Types
  - Miliary tuberculosis(blood TB)..difficult to detect
  - Tuberculosis meningitis
  - TB Lymphadenitis
  - Skin TB
  - TB spondylitis (bone TB)

Magnitude of children Death from TB in Ethiopia (Study 2022)

- A total of 284 TB/HIV co-infected children were included in the study
- Retrospective study at hospital of southern nation
  - 284 TB/HIV co-infected children
  - Of these 35 (12.3%) of them died during the study period.
  - overall MMR 2.78 per 100 child years of observation.
  - ADHERANCE/COMPILANCE
- The predictors of mortality were :
  - anemia
  - fair or poor ART drug adherence
  - extra pulmonary TB and
  - TB drug resistance(G.Jifare 2022).
  - **Conclusion : Death is high among TB HIV co-infections**

Global strategy and approach TB

1. Stop TB Strategy
   - Pursue high-quality DOTS expansion and enhancement
   - Address TB/HIV, drug-resistant TB and other challenges
   - Contribute to health systems strengthening
   - Engage all care providers
   - Empower people with TB and communities
   - Enable and promote research

Global strategy and approach TB…

2. Engage Tb approach

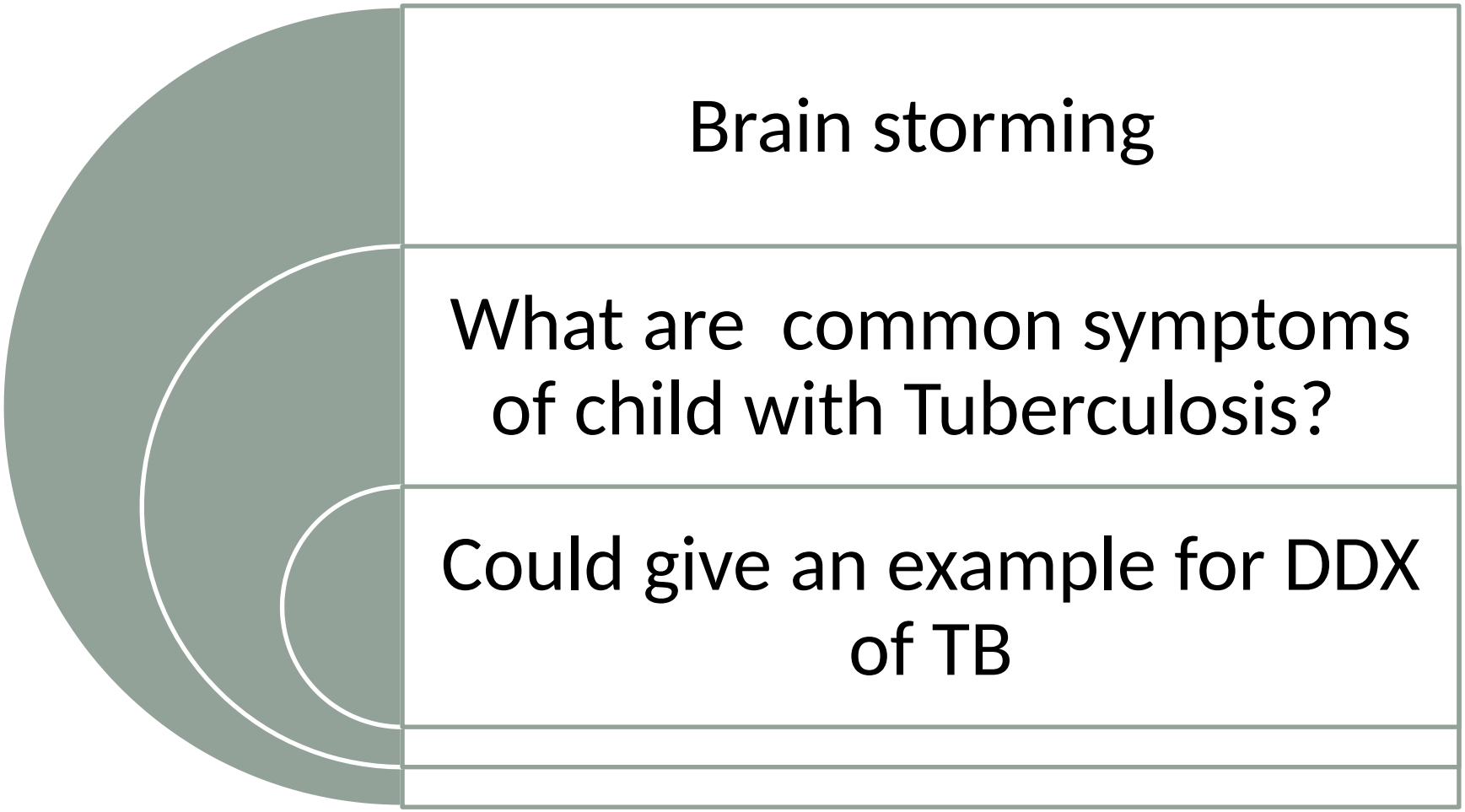
3. The End TB Strategy

2015: slogan

- STRATEGY:
- A WORLD FREE OF TB. END THE GLOBAL TB EPIDEMIC
- ZERO DEATHS, DISEASE, AND SUFFERING DUE TO TB

….

- Reduction in number of TB death
- Reduction in TB incidence rate
- The strategy builds on three pillars underpinned by four key principles:
  - Pillar 1: Integrated patient-centered TB care and prevention
  - Pillar 2: Bold policies and supportive systems
  - Pillar 3: Intensified research and innovation


Brain storming
What are common symptoms of child with Tuberculosis?
Could give an example for DDX of TB

Clinical Presentation of Tuberculosis

- The clinical presentation of Tuberculosis varies depending on the organ involved:
- age of the person and their immune state.
- Clinical presentation in immune-competent individuals is most commonly the result of involvement of:
- the lungs (more than 80% of cases); however, organ specific presentations may be seen upon involvement of extra-pulmonary organs, most commonly:
  - lymph nodes
  - pleura, spine, joints, genitor-urinary tract,
  - nervous system or abdomen:

Pulmonary Tuberculosis C/M

- A persistent and progressive cough
- often accompanied by non-specific systemic symptoms such as”

  Typically :

  – Persistent cough for two or more weeks, (cough of any duration for HIV positives)
  – Fever for more than 2 weeks
  – Night sweats
  – Unexplained weight loss.

Tuberculosis Diagnosis Methods

- Diagnosis of Tuberculosis may be reached by proper investigations using either:
  - bacteriologic examination
  - imaging techniques
  - histopathology or
  - biochemical analysis of body parts/fluids.
  - If all indicate negative finally gene-expert test can be done.

National Diagnostic Algorithm for Patients with Presumptive TB

- Approaches to diagnose TB in children:
- Approaches to diagnose TB in children:

1. Careful history (including history of TB contact and symptoms consistent with TB)
2. Clinical assessment (including growth assessment)
3. Diagnostic tests:
    - Bacteriologic confirmatory tests (AFB microscopy, Expert MTB/RIF assay & culture)
    - Chest X-ray
    - HIV testing
    - Histopathology, mainly for suspected extra pulmonary TB

TB in children …

- Careful medical history
- The most common clinical presentation of pulmonary TB is persistent respiratory symptoms and poor weight gain.
- Note that in at-risk groups such as infants or HIV-infected, pulmonary TB can also present as acute pneumonia.
- The approach to diagnosis of TB in HIV-infected children is similar to that for HIV-uninfected children.
- Typical symptoms
  - Cough especially if persistent and not improving
  - Weight loss or failure to gain weight
  - Fever and/or night sweats
  - Fatigue, reduced playfulness, less active

Attention

- Especially if symptoms persist (>2-3 weeks) without improvement following other appropriate therapies:
  - broad-spectrum antibiotics for cough
  - anti-malarial treatment for fever
  - nutritional rehabilitation for malnutrition
- Atypical clinical presentations of Tuberculosis in children:
  - Acute severe pneumonia
  - Presents with fast breathing and chest in-drawing
  - Occurs especially in infants and HIV-infected children
  - Consider PTB if poor response to antibiotic therapy – if HIV infected also
  - consider other HIV-related lung disease e.g. PCP
  - Wheeze

What to be focused For TB in children

- History of contact(chronic cougher)
- Clinical Assessment (including Growth Assessment
  - Physical signs requiring investigation to exclude extra pulmonary TB:
    - meningitis not responding to antibiotic treatment, with a sub acute onset and/ or raised intracranial pressure;
    - pleural effusion;
    - pericardial effusion;
    - distended abdomen with ascites;
    - non-painful enlarged lymph nodes without fistula formation;
    - Non-painful enlarged joints

Figure 3: Approach to TB Diagnosis in HIV Uninfected Child

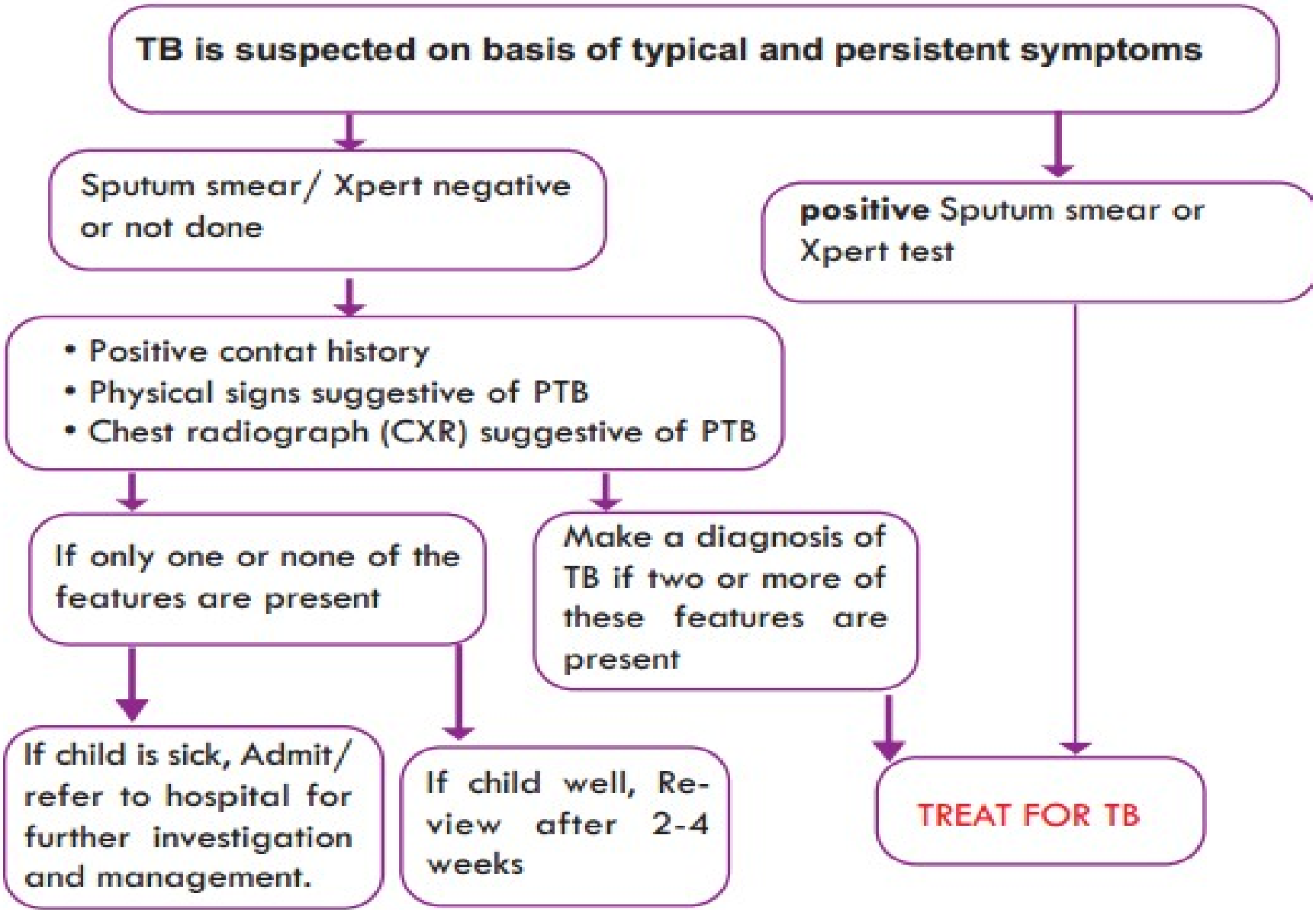


Figure 4: Approach to TB diagnosis in HIV-infected child

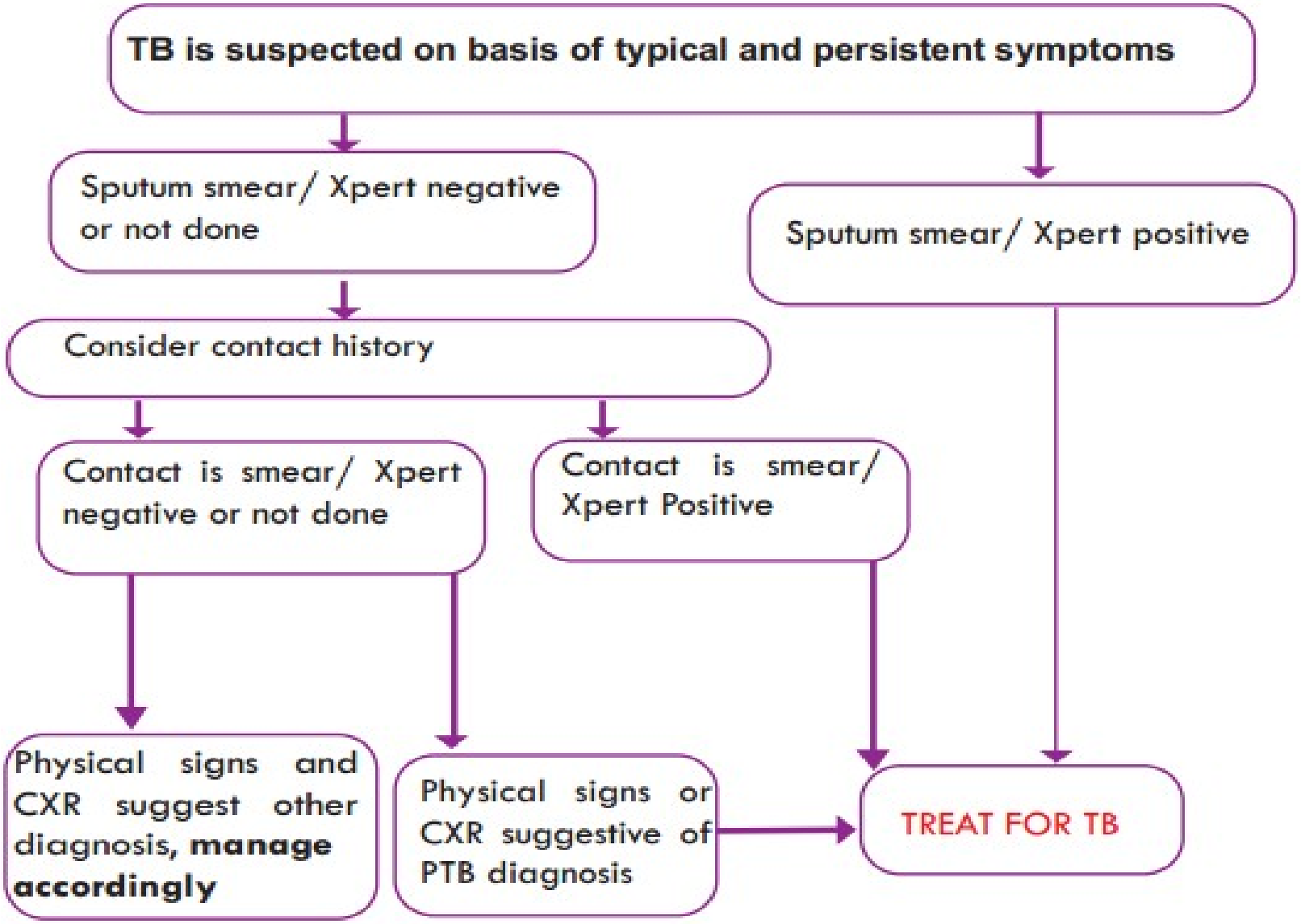


Figure 5: Diagnostic Algorithm for the Diagnosis of DR-TB in Children

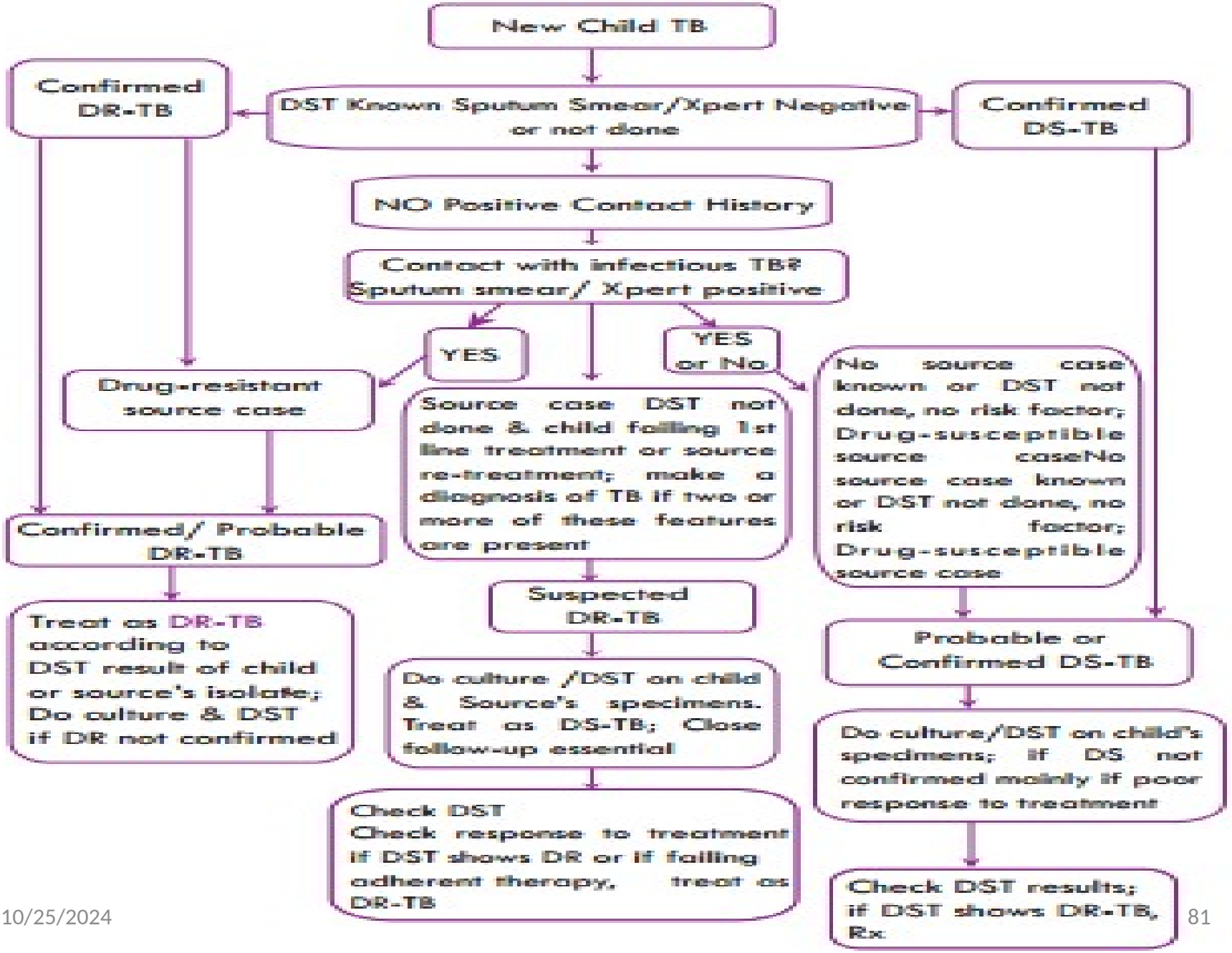


Table 8: The Essential Anti-TB Drugs and Their Recommended Dosages

Recommended Adult dosage		TB Drug	Recommended pediatric Dosage ᵃ	
Daily Dose range (mg/kg body weight)	Maximum (mg)		Daily Dose range (mg/kg body weight)	Maximum (mg)
5(4-6)	300	**Isoniazid**	10 (7-15)	300
10(8-12)	600	**Rifampicin**	15 (10-20)	600
25(20-30)	-	**Pyrazinamide**	35 (30-40)	-
15(15-20)	-	**Ethambutol**	20 (15–25)	-

ᵃ*Children weighing 25kg and more can be treated using recommendation for adults*

Table 10: First line TB Treatment Pediatric Dosing Chart Using Body Weight Bands

Child's Weight band	Treatment regimen and doses		
	Intensive phase: 2 (RHZ)E		continuation phase: 4(RH)
	RHZ 75/50/150	E 100mg	RH75/50
4-7kg	1	1	1
8-11kg	2	2	2
12-15kg	3	3	3
16-24kg	4	4	4
25+kg	Adult dosages recommended		

Phases of TB management

- TB treatment is administered in two phases:

1. Intensive (initial) phase:
   - aims to render the patient non-infectious by rapidly reducing the bacillary load sputum and brings clinical improvement in most patients receiving effective treatment.

2. Continuation phase:
   - aims to sterilize the remaining semi-dormant bacilli and is important to ensure cure/ completion of treatment and prevent relapse after completion of treatment.

- Dosing frequency: daily administration of all doses of six month of the treatment should be implemented under DOT in Ethiopia considering the high HIV and MDR-TB prevalence.

Nursing care for TB child

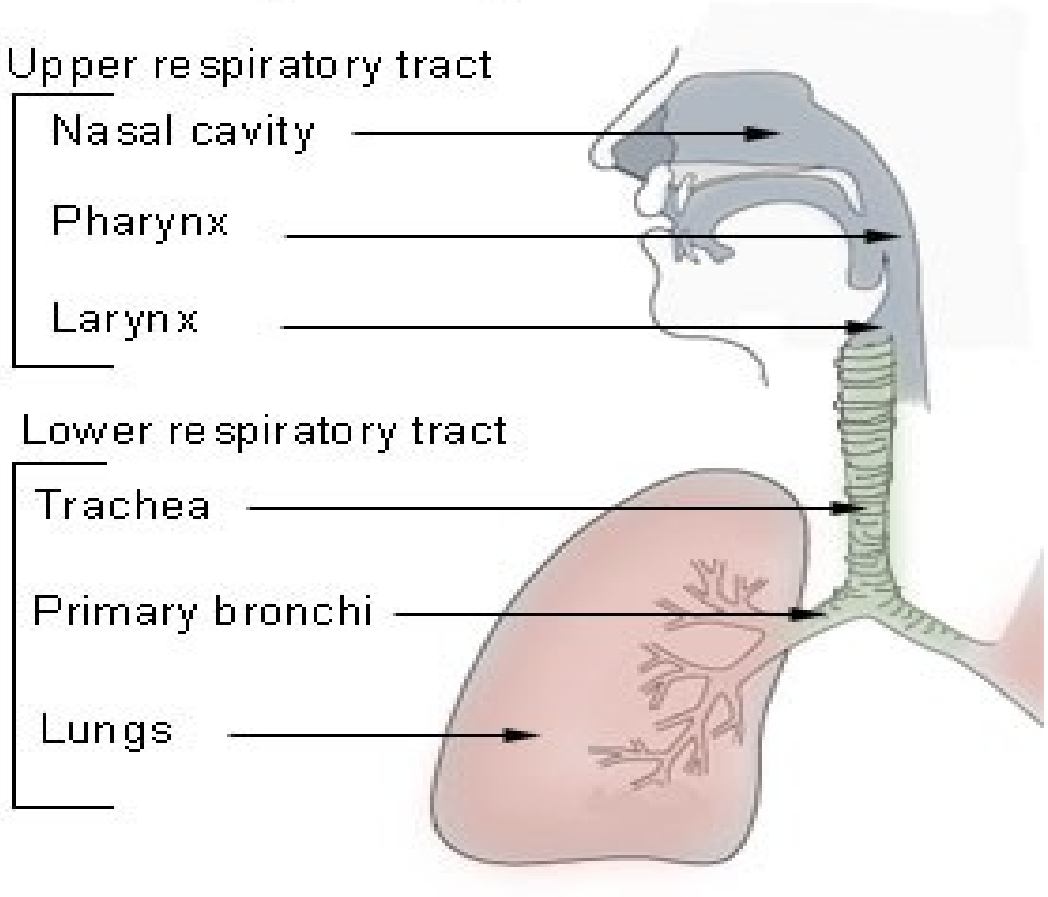
1. Teach family about drug adherence
2. Teach family and about TB transmission way and preventions
3. Adherence to follow up

Management of common childhood disease

Respiratory system disorders and infection (ARI)

1. Upper respiratory tract İnfections, (URTIs or URIs)

Def: (URTIs are the illnesses caused by an acute infection which involves the upper respiratory tract: nose, sinuses, pharynx or laryn



Laryngotracheobronchitis or Croup syndrome

- Croup syndrome is an acute infections of the larynx and trachea.

Epidemiology

- It occurs most frequently in children between 3 months and 5 years of age .
- More often in boys than girls (ratio 1.4:1)
- The rate of viral croup increases in cold season.
- 15% of cases have a positive family history.

Causes

Viral

- Para influenza virus accounts for 75% of cases, Adenovirus and Respiratory syncytial virus (RSV) and measles virus

Bacteria

– Mycoplasma pneumoniae

–Bacterial infections Secondary to Staphylococcus aurous, Streptococcus pyogenes and Streptococcus pneumoniae

Diagnosis

- Diagnosis is mainly clinical.
- Lateral neck X-ray may show subglotic narrowing

Diferential Diagnosis

- Epiglotitis, Bacterial tracehitis
- Foreign body aspiration
- Retropharygeal /peirtonsilar abscess

Nursing Care

Monitoring

- Monitor vital signs, O2 sat if available

Medications

- O2 if respiratory distress; give 6–10 L/min or more by mask
  – Keep oxygen saturation at > 97%
- Dexamethasone 0.5mg/kg i.m. repeat if no improvement after 4 to 6 hours.
- Inhaled Epinephrine (if available)

Cont…

Non pharmacologic Interventions

- Intravenous fluids
- Positioning
- Home -cold steam (sooth throat or reduce obstruction)
- Tracheostomy – if the distress gets worse, despite the above treatments or if there are signs of impending respiratory failure at any time.

Epiglottitis

- Epiglotitis is an acute inflammatory (infectious) process involving the epiglottis and surrounding structures.

Epidemiology

- Children between the ages of 2 and 7 years are affected and
- the peak incidence occurs at about 3 and a half years of age.
- The male to female ratio is 3:2. There is no seasonal variation.

Cont…

Etiology

- H.influenzae type b causes almost all cases of epiglotitis.
- Rarely streptococcus pnuermoniae and strepococcus pyogeanes can lead to epiglotitis.

Signs and symptoms

- ✓ Rapid onset over a period of 4-12 hrs
- ✓ dysphonia, drooling and distress
- ✓ High fever (39-40ºC)
- ✓ Sore throat, pain on swallowing(dysphagia)
- ✓ Hyper extension of neck (young child)
- ✓ Lean forward while sitting up with open and tongue protruding (older child)
- ✓ Cyanosis

Diagnosis

- Diagnosis is mainly clinical.
- Blood culture and lateral neck X-Ray help for diagnosis but
- they are time consuming and add little to the immediate management.
- Laryngoscope shows cherry red epiglottis

Management

- Maintain airway
- Routine tracheostomy/nasotracheal intubation
- Chloramphenicol 75 -100mg/kg /dose i.v for 7 to 20 days.
- moist air is needed.
- Give Oxygen by mask at 6–10 L/min if cyanosis is present.
- intravenous fluid therapy

Mang cont…

NB: Never attempt to visualize the epiglottis directly with a tongue depressor because the gagging procedure causes complete obstruction of the glottis and results to respiratory failure.

- Throat culture should NOT be done UNLESS a means of providing an artificial airway such as a tracheostomy or endotracheal intubation is readily available.

Acute tonsillopharyngitis

Definition

- Is an Infection of the upper respiratory tract with principal involvement of the throat and tonsils.

Causes: mostly viral but may also occur as part of common cold or influenza syndrome

- Group A B-hemolytic streptococcus is the most common and the most important bacterial agent after 2 years of age.

Clinical Presentation

Viral: insidious onset

- Runny nose, sneezing, coryza etc.
- Contact history with family member

Streptococcal: is more common in children older then 2 years and peaks in children b/n 4 and 7 years of age.

Cont…

They may present with:

- Headache, abdominal pain, vomiting, high grade fever, sore throat.
- Diffuse redness and exudates, petechiae over the palates, and, tender cervical lymphadenopathy.
- Most strongly suggestive: - Age 5 to15 years.
- Clinical evidence of acute pharyngitis
- Absence of URTI

Diagnosis

- X-ray
- Nasopharyngeal culture
- Hx and P/E

Management

- **Facilitating of respiratory efforts:**

- High – humidity atmosphere
- Oxygen therapy
- Insertion of an artificial airway (Nasotracheal tube or tracheotomy)
- Decrease the need to cry or talk
- Steam inhalation

Management cont…

- **Administration of medications**
  - Benzathine penicillin
  - Oral penicillin
  - Analgesics - paracitamol.
- Promotion of rest
- Provision of appropriate and adequate fluids
- Support and education of parents

Tonsilitis

- Is an inflammation and formation of edema of the tonsils.

Cause – Viral

- Bacterial – most common

Signs and Symptoms

- ✓ Reddened, swollen tonsils
- ✓ Exudates may be seen on inspection
- ✓ Sore throat
- ✓ Difficulty swallowing
- ✓ Fever

NB-Streptococal tonsilitis has high fever, pus on tonsilar surface and marked cervical lymp gland swelling

- Viral tonsilitis has no fever, no pus on tonsilar surface and no marked cervical lymph gland swelling

Complications

A. Otitis media

B. Peritonsilar abscess (in older children only)

C. Cervical lymphadenitis may develop into abcess

D. Rheumatic fever or acute glomerulonephritis after streptococcal infection

Diagnosis

- P/E and Hx.
- Throat culture

Management

➢ **Medications**

- PPF 50 000 u/kg /day for 7 days or
- Benzathine 50 000 units/kg IM stat
- Paracetamol 15mg/kg QID

➢ In severe and recurrent case – **Tonsillectomy** may be done

Nursing Management

➢ Supportive treatments

- Encourage fluid intake
- Encourage aspirin gargle
- Treat fever
- Avoid hot drinks

Common Cold (Nasopharyngitis)

- Is a viral infection of the nasopharyngeal mucosa.

Etiology

- A number of viruses all giving the same clinical picture.
- Rhino viruses (100 serotypes) are the major causes.
- ✓ Para influenza viruses, respiratory syncytial viruses (RSV), influenza and Adeno viruses cause common cold in children.

Epidemiology and Occurrence

- World wide, both endemic and epidemic.

➢ Each individual can expect one or more colds every year.

❖ **Factors in the occurrence of common colds are:**

- Over crowding and inadequate clothing.

✓ Incidence is highest in < 5 children, gradually declining with increasing age.

Clinical Manifestations

- ✓ Nasal discharge: first watery, then mucoid and purulent
- ✓ Sneezing
- ✓ Some fever may be present
- ✓ Headache
- ✓ Dry or painful throat

Susceptibility and Resistance

- Susceptibility is universal.
- Repeated infections (attacks) are most likely due to multiplicity of agents.

Diagnosis

- Based on clinical grounds

Complications

- Acute otitis media
- Pneumonia

Treatment

- **Supportive measures:**

✓ Sooth the throat and relieve the cough with safe home remedies like tea and honey

✓ Nasopharyngeal drops using a syringe filled with normal saline (or drop clean water mixed with salt using cotton), 4 to 6 times a day.

✓ Treat fever

✓ Advise the mother to return immediately if the child develops fast breathing or difficulty of breathing or if the child becomes sicker.

✓ Follow up in 5 days

OTITIS MEDIA

Definition

- An inflammation of the middle ear.
- It may be acute if symptoms for less than 2 weeks, or chronic if symptoms for more than 2 weeks.

Etiology

- Streptococcus pneumoniae-25-50%
- Haemphilus Influenzae-15-30%
- Moraxella catarrhalis-3-30%
- Rhinovirus/RSV/Coronaviruses/Adenovirus
- Enteroviruses

Epidemiology

- Ear pain and/or discharge is one of the most common presenting symptoms to OPD in under 5 children.
- About one third of children will have at least one episode of acute otitis media by 3 years of age

Pathogenesis

- The two important factors are Eustachian tube dysfunction and URTI.
- Both lead to obstruction of the tubes and serous fluid collection in the middle ear.
- If there is an associated entry of pathogenic organisms, suppurative otitis media may follow.

Clinical manifestations

Symptoms

Infant

- Excessive crying, Pulling of the Ear

Toddler

- Irritability ,Earache

Both may have otorrhoea

Signs: Fever , bulging eardrum, fullness and erythema of tympanic membrane

- May also be additional upper respiratory symptoms
- Diarrhea, vomiting .

Diagnosis

Clinical diagnostic signs include>

- Ear discharge
- Visualization of tympanic membrane: red membrane, loss of normal light reflex.

Treatment

1. Antibiotics for 5 days

- Cotrimoxazole. Or
- Amoxicillin or
- Ampicillin

Cont…

2. Wicking of the ear with dry cotton for discharging ear.
3. Analgesic – Acetaminophen; paracetamol

Complications

- Mastoiditis
- Hearing loss
- Intracranial (meningitis, brain abscess)

2. Lower respiratory tract disorders

Pneumonia

- Is an inflammation of the parenchymal structure of the lungs, such as the alveoli and bronchioles.

Etiology

Bacteria- the most common cause

Viruses- rare

Aetiology cont…

- Bacteria, especially streptococcus pneumonia, followed by haemophilus Influenza.
- Staphylococcus aureus and gram negatives (E.coli, Klebsiella, etc) are other causes in neonates and immuno compromized.

Epidemiology

- Pneumonia is responsible for 70-80% deaths that occur due to ARIs world wide.
- **In Ethiopia, pneumonia is the cause of 1/3rd infant mortality and 1/5th of under 5 mortality.**

➢ risk factors

✓ Malnutrition

✓ Over crowding and in-door air pollution

✓ Recurrent infections such as measles, whooping cough, malaria and diarrhea

✓ Immune-deficiency status

- Severe forms of pneumonia are commonly encountered in children between the age of 6 months and 3 years

Pathogenesis

- The normal lung is sterile. Most of the agents are inhaled from the air breathed or aspirated into the periphery of the lung from the upper air ways.
- After entry to the lung, initially, a reactive edema occur which supports proliferation of the organisms and aids in their spread into adjacent portions of the lung.
- The disease may have a lobar, patchy or diffuse distribution.
- The disease may be complicated in some by development of lung abscess, collapse of the lung, and empyema.

Clinical Manifestations

The common manifestations are:

- ✓ Cough
- ✓ Sudden onset of fever, and
- ✓ Signs of respiratory distress

- **Respiratory distress is manifested** with rapid and difficult breathing, nasal flaring, intercostals retraction, chest indrawing and cyanosis**.**
- Other possible findings are crepitation, diminished breath sounds, bronchial breathing and dullness on percussion.

➢ **Fast breathing and chest in drawing** are important clinical findings in the assessment of severity of pneumonia.

A general danger sign is present if:

- the child is not able to drink or breastfeed
- the child vomits everything
- the child has had convulsions
- the child is convulsing now
- the child is lethargic or unconscious

Table 1: Cut off for fast breathing

Note: The child who is exactly 12 months old has fast breathing if you count 40 breaths per minute or more.

Here is the classification table for cough or difficult breathing.

SIGN	CLASSIFY AS	TREATMENT
Any general danger sign or Chest indrawing or Stridor in calm child	SEVERE PNEUMONIA or VERY SEVERE DISEASE	Give first dose of Cotrimoxazole Refer URGENTLY to hospital
Fast breathing	PNEUMONIA	Give Cotrimoxazole for 5 days. Soothe the throat and relieve the cough with a safe remedy. Advise mother when to return immediately. Follow-up in 2 days
No signs of pneumonia or very severe disease.	NO PNEUMONIA COUGH OR COLD	If coughing more than 21 days, refer for assessment. Soothe the throat and relieve the cough with a safe remedy.

Bronchial Asthma

Definition

- Bronchial asthma is a diffuse obstructive lung disease with hyper reactivity of the airways and reversibility of air flow obstruction spontaneously or with treatment.

Cont… …

Pathophysiology:

- When mast cells activate, there is infiltration of inflammatory cells, edema, denudation and disruption of the bronchial epithelium, goblet cell hyperplasia and smooth muscle thickening resulting to asthmatic inflammation.

Cont…

Etiology

- There is no well defined single etiologic agent.
- Multiple factors interplay to disturb the mechanisms that maintains the balance between contraction and relaxation of bronchial smooth muscle.
- Autonomic, immunologic, endocrine, infectious and emotional factors interplay.

Cont…

Signs and symptoms:

- Recurrent episodes of wheezing
- Breathlessness
- Chest tightness
- Cough particularly at night and in the early morning

Cont…

Risk factors:

- Children before age of 5
- Allergy (inherited) like eczema, urticaria or hay fever

GENETICS

- Children of asthmatics have higher incidence of asthma:
  - 6% to 7% risk if neither parent has asthma
  - 20% risk if one parent has asthma
  - 60% risk if both parents have asthma

LBW

- Poverty, frequent respiratory infections

Cont…

- **Allergic triggers:**
- Allergens
- Irritants
- Exercise
- Weather changes
- Viral or sinus infections
- Reflux disease
- Medications (sensitivity to aspirin, NSAIDs)

Cont…

- Food
- Emotional anxiety
- Rhinitis
- Sinusitis

Differential Diagnosis

- Infectious – pneumonia, bronchiolitis
- Congestive heart failure
- Foreign body aspiration
- Tuberculosis

Cont…

Diagnosis

- is mainly clinical with limited information obtained from investigation.
- Microscopic examination of sputum shows eosinophilia.
- Chest X-ray is not routinely done unless there is suspicion of associated pneumonia or complication like atelectasis.
- Therapeutic trials

Cont…

Medical Management

- Give oxygen to all children with asthma who are cyanosed or whose difficulty in breathing interferes with talking, eating or BF.

1. Nebulized salbutamol, the driving source for the nebulizer must deliver at least 6–9 litres/minute

- salbutamol 2.5 mg (i.e. 0.5 ml of the 5 mg/ml nebulizer solution), 4-hourly, reducing to 6–8 hourly once the child's condition improves

Cont…

(2) Salbutamol by metered-dose inhaler with a spacer device

- Introduce two puffs (200 micrograms) into the spacer chamber
- allow normal breathing for 3-5 breaths.
- This can be repeated 4-hourly, reducing to 6–8 hourly after the child's condition improves.

(3) SC epinephrine (adrenaline) 0.01 ml/kg of 1:1000 solution (up to a maximum of 0.3 ml)



Use of spacer device and face mask to give bronchodilator treatment. A spacer can be made locally from a plastic soft drink bottle.

- If there is no improvement after 15 minutes, repeat the dose once.
- oral prednisolone, 1 mg/kg, for 3 days.
- give IV aminophylline—initial dose of 5–6 mg/kg (up to a maximum of 300 mg), followed by a maintenance dose of 5 mg/kg every 6 hours.
- Weigh the child carefully and give the IV dose over at least 20 minutes and preferably over 1 hour.

Cont…

Nursing Management:

- Regular monitoring and assessment of the symptoms in the past 2 weeks.
- Assess for exacerbation, proper compliance to medications.
- Assess for difficulty with feedings, changes in respiratory rate, altered sleep patterns, presence of retractions, decreased appetite and weight loss.
- Teach the parents to control or minimize the exposure to allergens.

Cont…

- Emphasize the need for flu vaccine for patients with no restrictions.
- Limit the exposure to cigarette smoke
- Eliminate dust mites
- Encourage to use air conditioners at home and keep doors and windows closed to minimize the exposure to these triggers.

Cont…

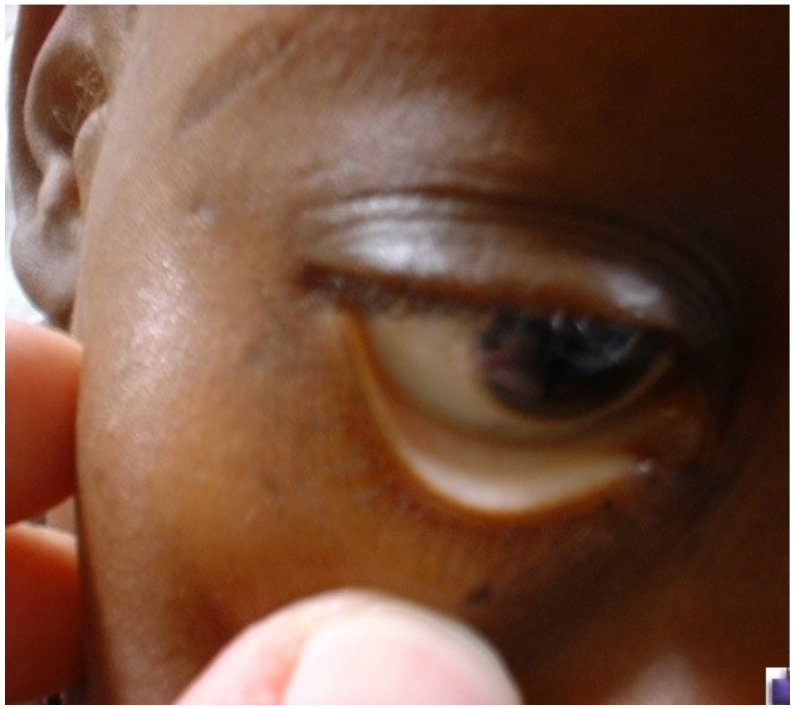
- Encourage a regular visit to the physician and evaluate the proper usage of equipment.
- Provide a written instruction for management plans when at home, at school or care givers.
- Remind the parents or care givers about the medications, its expiry date as well as having a reserve of it in times of exacerbations.
- Give an easy telephone access when the patient must be taken into the hospital.

READY ???
25%



HEMATOLOGICAL DEFECTS

1. ANEMIA
2. HEMOPHILIA
3. Polycythemia



Anemia in children

CHILDHOOD ANEMIA

DEFINITION

- Reduction of Hemoglobin concentration or Hematocrit to below normal level for age and sex

- Reduction in the circulating Hgb also causes a decreased oxygen carrying capacity of RBC

Physiology

- **Erythropoietin** from the kidney in response to local tissue hypoxia to stimulate bone marrow RBC production
- **Hematocrit** — The Hematocrit is the fractional volume of a whole blood sample occupied by red blood cells
- **Hemoglobin (HGB)** — This is a measure of the concentration of hemoglobin in whole blood

Cont. .....

- Ratio of Hgb to Hct is approximately 1:3
- Normal Hgb in Newborn=16g, infant & preschool=12g & school age child=13g.
- In newborn normal RBC=5-6millions/mm^3, infant & children 4-5millions/mm^3.

Cont. …

- Hemoglobin increases with advancing gestational age: At term, cord blood hemoglobin is 16.8 g/dL (14–20 g/dL); hemoglobin levels in very low birth weight (VLBW) infants are 1–2 g/dL below those at term.
- A “physiologic” decrease in hemoglobin content is noticed at 8–12 week in term infants (hemoglobin, 11 g/dL) and at about 6 week in premature infants (7–10 g/dL).

Cont. ….

Grading (WHO):

- Hgb b/n 10g & cut of age point = Mild anaemia
- Hgb b/n 7g & 10g = Moderate anaemia
- Hgb below 7g = Severe anaemia

Types anemia based on causes

Hypoproliferative (Resulting From Defective RBC Production)

- Iron deficiency
- Vitamin B12 deficiency (megaloblastic)
- Folate deficiency (megaloblastic)
- Decreased erythropoietin production (eg, from renal dysfunction)
- Cancer/inflammation

Cont. …

Bleeding (Resulting From RBC Loss)

- Bleeding from gastrointestinal tract, epistaxis (nosebleed), trauma bleeding from genitourinary tract (e.g. menorrhagia)

Cont. ….

Hemolytic (Resulting From RBC Destruction)

- Altered erythropoiesis (sickle cell anemia, thalassemia, other hemogloinopathies)
- Hypersplenism (hemolysis)
- Drug-induced anemia
- Autoimmune anemia
- Mechanical heart valve–related anemia

Depending on mean corpuscular volume / morphology/

- Microcytic
- Normocytic
- Macrocytic

Cont. ….

1. Microcytic anemia

- Iron deficiency anemia
- Anemia of chronic illness
- Lead poisoning
- Hemoglobinopathies

2. Macrocytic anemia

- Vitamin B12 deficiency
- Folate deficiency
- Liver diseases

Cont. …

Normocytic anemia

- Aplastic anemia
- Malignancies – leukemia
- Blood loss
- Hemolysis….

Cont. ….

Reticulocyte count is the basis to classify anemia in to two major groups

1. **Decreased RBC**—impaired synthesis or failure of B. marrow to synthesize blood cells
2. **Increased RBC loss-**--Hemolysis and bleeding

Pathophysiology

- When circulating hemoglobin is reduced, the oxygen carrying capacity of the blood becomes reduced.
- During the falling of hemoglobin to below 7 to 8 g/dl , the heart tries to compensate by increasing the rate and cardiac out put.
- There will be also increased O2 concentration deference b/n arterial and venous system (which made O2 more readily available to the tissue) and shunting of blood flow to vital organs and tissues of the body.

Physical examination of anemic child

- History of malnutrition
- **History of Pica** (eating unusual substances)
- Palmar pallor
- Irritable, pale conjunctivae
- Puffy face, pale oral mucosa
- Tachycardia, systolic ejection murmur
- Pitting pedal edema
- Signs of congestive heart failure

Physical examination of anemic child

Signs suggestive of causes of anemia

- Fever--- infection
- Malnutrition---iron, folate, B-12
- GIT surgery--- B-12
- Hepato-splenomegaly– malignancy
- Hypertension, edema– chronic renal failure
- Jaundice, hepato-splenomegaly--- Hemolysis

Diagnosis (RBC INDICES)

- Hemoglobin (g/dl) … HB/Whole blood
- Hematocrit (%)---RBC/whole blood

Other laboratory tests

- RBC Count, WBC count, Platelet count
- Reticulocyte Count
- Peripheral morphology…size, color, shape

Hemoglobin and hematocrite

Age/Sex	Hemoglobin g/dL	Hematocrit %
At birth	17	52
Childhood	12	36
Adolescence	13	40
Adult man	16 (±2)	47 (±6)
Adult woman (menstruating)	13 (±2)	40 (±6)
Adult woman (postmenopausal)	14 (±2)	42 (±6)
During pregnancy	12 (±2)	37 (±6)

Iron deficiency anemia

- Most common cause is **nutritional deficiency of iron**
- Iron deficiency disorder is common in infants and adolescents affecting nearly **25%**
- 1-2mg/kg/day is important for normal synthesis
- Excessive **cow's milk** intake predisposes a child to develop anemia due to **GIT allergy**, and poor bioavailability of iron (poor absorption)
- **Malaria and hookworm** infestation increases the prevalence of iron deficiency anemia in developing areas

Cont ….

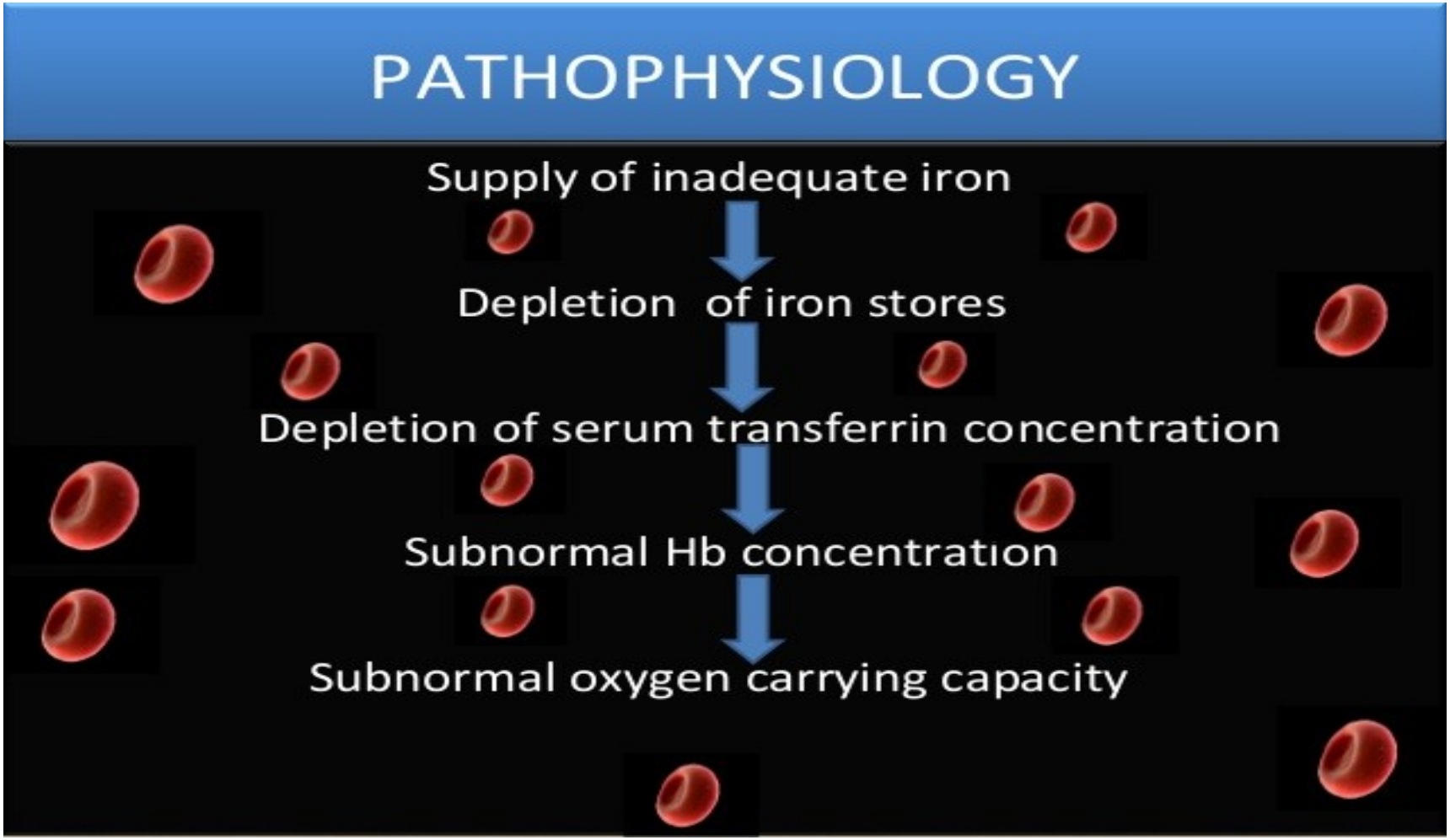
- Factors associated with development of iron deficiency anemia **in young children**
  - Prematurity, low birth weight
  - Fetal maternal hemorrhage
  - Twin to twin transfusion syndrome
  - Insufficient dietary intake

- IDA is hypochromic, microcytic anemia (blood cells are deficient in production of hemoglobin are smaller than normal;

Cont. …

- 10% of dietary iron is absorbed (good diet needs 8-10 mg of iron); “milk babies”; many children with iron deficiency anemia are undernourished because of family finances, knowledge deficit about nutrition

Cont. …


PATHOPHYSIOLOGY
Supply of inadequate iron
Depletion of iron stores
Depletion of serum transferrin concentration
Subnormal Hb concentration
Subnormal oxygen carrying capacity

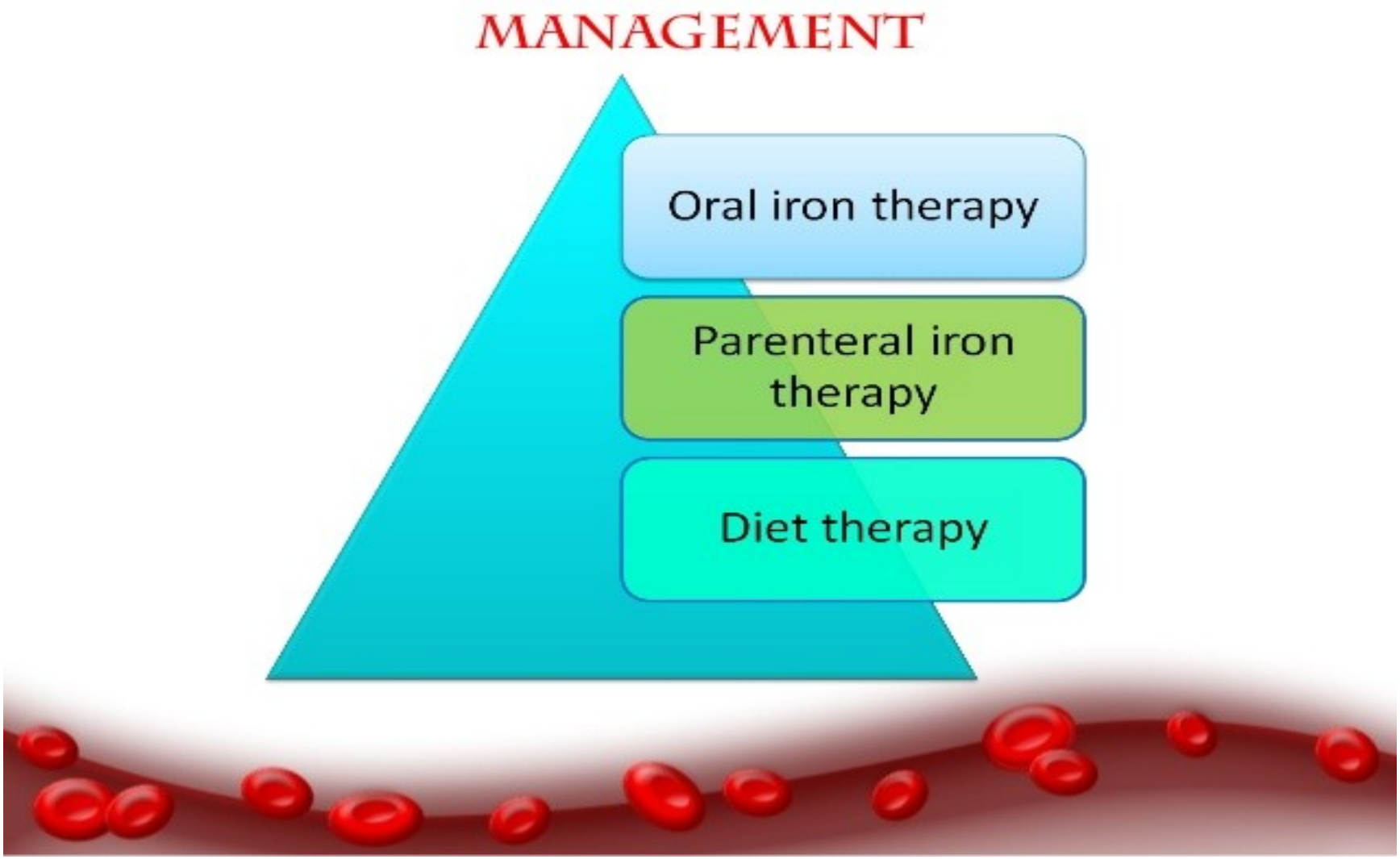
Clinical features of IDA

- Peak age is 9 month to 2 years
- Pallor, irritability
- Poor feeding
- Tachycardia
- Impaired mental development
- Reduced work capacity & school performance
- Smooth tongue
- Geophagia
- Blue sclera
- Spooning of nails…

Diagnosis of IDA

- Complete blood count
  - Hgb < 11gm/dl
  - Mild to moderate defects in white blood cells
- Peripheral morphology – microcytic hypochromic
- Search for source of blood loss
- Search for chronic disease

Management


MANAGEMENT
Oral iron therapy
Parenteral iron therapy
Diet therapy

Cont. …

Oral iron therapy

- **Recommended Dosage**
- Infant and children:3 mg/kg/day
- 20mg elemental iron +100 mcg folic acid
- Adolescent :100 mg elemental iron +500 µg of folic acid +25-50 mg vit.c

Mild Anemia: Once Daily
Moderately And Severe Anemia: Twice Daily

Result
Peak increase of reticulocyte count in 5th -10th day of administration

Cont. …

Oral iron therapy

PREPARATION	ELEMENTAL IRON/TAB	IRON ABSORPTION /TAB
Ferrous sulfate	60 mg	20 mg
Ferrous fumerate	66 mg	33 mg
Ferrous gluconate	36 mg	12 mg



Cont. ....

PARENTERAL IRON THERAPY

- Iron dextran complex:50 mg/ml in 2ml saline
- Dose calculation:

Iron [mg]= Wt. in kg x Hb deficit [gm/dl] x4

- Route=intramuscular---**Z track method**
  intravenous-----infusion methods



Cont. …

- Treatment and nursing care-improved nutrition, ferrous sulfate administered between meals with juice; teaching caregivers; an iron-dextran mixture (Imferon)
- Ferrous sulfate (elemental iron) for 2 – 3 months 3-4 mg/kg/day (in 3 div. doses)
- Blood transfusion if severe and heart failure

Prevention of IDA

- Breast feeding exclusively for 6 months

Then add iron rich foods

- Iron supplementation for low birth weight & premature infants
  - 1-2mg/kg/day starting from 1 month till 12 months of age

Guidelines for Pediatric RBC Transfusions

Children and Adolescents

- Acute loss >25% circulating blood volume
- Hemoglobin <8.0 g/dL…surgery and symptomatic
- Hemoglobin <13.0 g/dL… and severe cardiopulmonary d/s

Infants Within First 4 Mo of Life

- Hb <13.0 g/dL and severe pulmonary or cardiac disease
- Hb<10.0 g/dL and major surgery
- Hb <8.0 g/dL and symptomatic anemia.

Severe acute malnutrition HB<4gm/dl

*Hematocrit estimated by -----Hb g/dL × 3.

HEMOPHILA

- **HEMOPLILIA**:- is a sex – linked recessive genetic disorder caused by defective or deficient coagulation factor (clotting factors)
- The two major forms of hemophilia which can occur mild to severe form are hemophilia A and hemophilia B which are clinically indistinguishable, although they can be distinguished by laboratory tests

1. Hemophilia A(Classic hemophilia)

- Is the most common form of hemophilia (80%)
- Is the most common form of congenital bleeding d/o in humans
- Factor VIII deficiency-(inherited as a sex-linked recessive Mendelian trait-transmitted to affected males by carrier females)-most severe

Cont. …

- Females carries & will transmit the genetic defect to 50% of their sons &50% of their daughters will be carriers.
- Men with hemophilia will not transmit the defect to their son

2. Hemophilia B (Christmas disease)

- Is factor IX deficiency
- Factor IX deficiency-(sex-linked recessive trait appearing in male offspring of carrier females

Classification based on Severity

1. **Mild hemophilia-** level of factor VIII and IX deficiency is <6%

- There will be bruise, bleeding tendencies of nose and gums, and prolonged bleeding even after minor surgery.

2. **Moderate hemophilia-** level of factor VIII and IX deficiency is 6-30%
3. **Severe hemophilia-** level of factor VIII and IX deficiency is > 30%

Clinical Manifestation

- The same for both hemophilia A&B
- Small, Persistent, pronged bleeding from minor trauma & cut
- Uncontrollable hemorrhage after dental extraction
- Epitasis
- GI bleeding
- Ecchymosis & subcutaneous hematomas
- Neurologic signs – pain, Anaesthesia & paralysis
- Hemarthrosis (bleeding in to the joint)
- Hematuria

Diagnosis- Clinical symptoms and family history along with:

- Laboratory tests
  - CBC
  - Plate late function test
  - Clotting studies

Medical Mgt- Replacement therapy of factor VIII and IX

- Transfusion of fresh or frozen plasma

Nursing Mgt

- Prevent and control bleeding
- Maintain safer environment

Complications

- GI bleeding
- Hemorrhage
- Joint deformity
- Hematoma formation

Polycythemia vera

- Polycythemia vera is blood disorder in which the bone marrow produces too many red blood cells. The extra cells make the blood too thick, increasing the risk of blood clots.
- Blood clots may cause serious problems depending on where they form.
- The clots can decrease the blood supply to the heart, brain, or liver, for example.
- Treatment may include venipuncture to remove blood and taking aspirin.

Polycythemia Vera…

- a very rare blood disorder in children.
- With polycythemia vera, the bone marrow makes too many red blood cells.
- The extra cells make the blood too thick.
- This may lead to blood clots.
- The clots can decrease the blood supply to organs, tissues, and cells.

Causes

- Genetic mutations in bone marrow cells cause polycythemia vera.
- Some forms can be passed from parents to children and Some forms are not.
- The most common causes of secondary polycythemia include **obstructive sleep apnea**, obesity hypoventilation syndrome, and chronic obstructive pulmonary disease (COPD).
- Other causes include testosterone replacement therapy and heavy cigarette smoking.

C/M

- Some children do not have symptoms and polycythemia vera may be found when they're having blood tests for other reasons.
- Symptom s can occur a bit differently in each child. They can include:
- Headaches
  - Dizziness
  - Feeling weak and tired
  - Trouble breathing
  - Enlarged spleen
  - Trouble seeing
  - Itching or burning
  - Reddish coloring of face
  - Bleeding gums
  - Losing weight

Diagnosis

- The healthcare provider will ask about your child’s symptoms and health history.
- He or she will give your child a physical exam.
- Your child may also have tests, such as:
  - Hemoglobin and hematocrit. This test measures the amount of hemoglobin and red blood cells in the blood.(>65%)
  - Complete blood count (CBC).A complete blood count checks the red blood cells, white blood cells, blood clotting cells (platelets), and sometimes, young red blood cells.

How is polycythemia vera treated in a child?

- Treatment will depend on your child’s symptoms, age, and general health. It will also depend on how severe the condition is.
- Your child's healthcare provider will likely refer you to a hematologist, a specialist in blood disorders.
- Treatment may include:
  - Venipuncture to remove some blood, which reduces the number of red blood cells (done like when a person donates blood)
  - Low-dose of aspirin to prevent blood clots
  - Medicines to decrease the production of red blood cells in the bone marrow
  - Other treatments that are being studied

....

- Polycythemia Vera (PV) is a chronic disease:
- It's not curable, but it can usually be managed effectively for very long periods.
- The goal of therapy is to reduce the risk of thrombosis and to ease symptoms by lowering the number of extra blood cells.

Complications

- Complications depend on how severe the polycythemia is. They can include:
  - Blood clots in the veins of the liver
  - Deep vein thrombosis or blood clots in the veins
  - Heavy bleeding
  - Stroke
  - Leukemia

Nursing care

- A patient with polycythemia has an increased risk of **venous thrombosis**, as his/her blood is more viscous.
- It is important to maintain adequate hydration. Keeping a patient with a high RBC count dehydrated, such as being NPO for an extended time, may increase risk of venous thrombosis formation.

CHROMOSOMAL ANOMALIES AND BIRTH DEFECTS

Outline

- Cytogenetics
  - Chromosomes
  - Chromosomal Anomalies
  - Sex Chromosome Anomalies
- Down Syndrome
- Trisomy 13
- Trisomy 18
- Turner's Syndrome
- Klinefelter's Syndrome
- Dysmorphology
- NTD

Cytogenetics

- The study of chromosomes:-
  - their structure
  - inheritance and
  - abnormalities
- Chromosomes are made up of DNA and other protein complexes and contain most of the genetic information that is passed from one generation to the next.

- Chromosome studies are important because abnormal chromosome number
  - e.g., trisomy 13 and
- abnormal chromosomal arrangements

  e.g., micro-deletion may lead to multiple congenital anomalies.

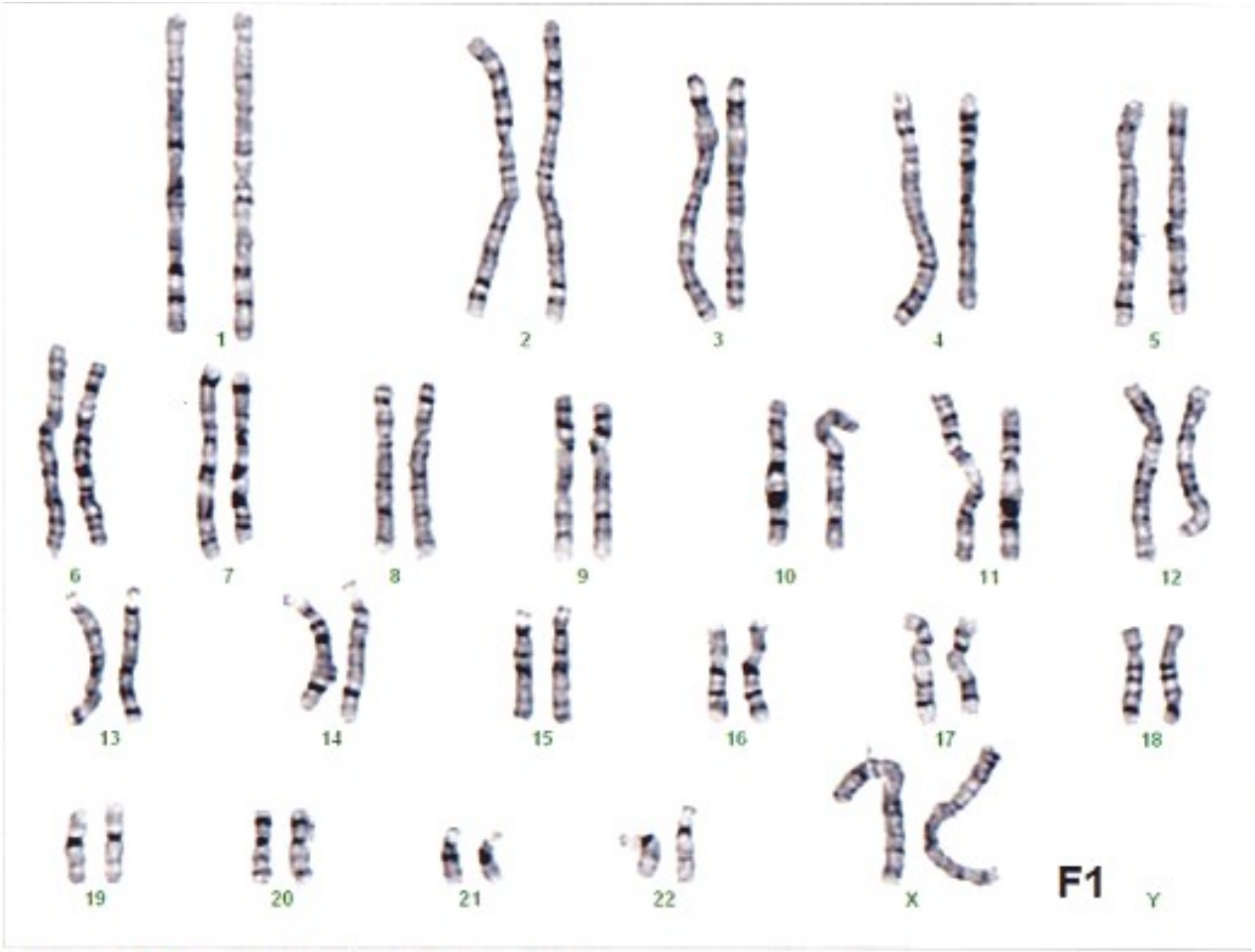
- Chromosome abnormalities:
  - occur in approximately 1% of live births and
  - are responsible for a large proportion of:-
    - early fetal losses
    - multiple congenital malformations, and
    - cases of mental retardation
    - have a significant role in the development of neoplasias
- Chromosome analysis is indicated in the child with multiple congenital anomalies and/or dysmorphic features.

Chromosomes

- Human cells contain a multiple of 23 chromosomes (n = 23)
- A haploid cell (n) has 23 chromosomes (ova or sperm)
- Two errors of cell division commonly occur during meiosis or mitosis, and both can result in an abnormal number of chromosomes
  - Nondisjunction
  - Anaphase lag

- **Nondisjunction** - two chromosomes fail to separate and thus migrate together into one of the new cells, producing one cell with two copies of the chromosome and another with no copy.
- ***Anaphase lag*** - a chromatid or chromosome is lost because it fails to move quickly enough during anaphase to become incorporated into one of the new daughter cells.

Normal Female: 46,XX



Case: STANDARD Slide: alfa Cell: 1 Patient:

Normal Male: 46,XY



Chromosomal Anomalies

- Include abnormalities of number and structure
- Are the result of *errors in cell division.*

Chromosome Errors Results in

- Spontaneous abortuses
- Sexually ambiguous organisms
- Infertile males or females
- Newborns with multiple congenital anomalies
- Mentally retarded
- Mentally ill
- Behaviorally disordered
- Cancers

ABNORMALITIES OF CHROMOSOME NUMBER

- Trisomy: the presence of an extra chromosome
- Monosomy: the absence of a **whole** chromosome
- Deletion: the absence of a **part of** a chromosome
- Inversion: the **180° rotation** of a part of a chromosome
- Translocation: the **breakage and rejoining** of parts of two, **non-homologous** chromosomes.

- Trisomy, characterized by the presence of 3 instead of the normal 2 of any particular chromosome, is the most common form of aneuploidy
- Trisomy can be complete and present in all cells or it may be in mosaic form.

ABNORMALITIES OF CHROMOSOME STRUCTURE

- **Translocations**
- **Inversions**
- **Deletions**
- **Duplications**

chromosomal Anomalies

- General features:
  - Mental retardation (MR)
  - Growth retardation (GR)
  - Multiple congenital anomalies
  - Prenatally diagnosable
  - Associated with spontaneous abortion (Sab)

chromosomal Anomalies

- Trisomy 13 - Patau Syndrome
- Trisomy 18 - Edward Syndrome
- Trisomy 21 - Down Syndrome
- $5p^-$ deletion - Cri-du-chat Syndrome

Sex Chromosome Anomalies

- General features:
  - Some growth retardation (GR)
  - Reproductive anomalies/problems
  - Prenatally diagnosable
  - Associated with spontaneous abortion (Sab)

Sex Chromosome Anomalies

- Monosomy X: Turner's Syndrome (45, X)
- Trisomy X: Triplo-X Syndrome (47, XXX)
- Trisomy (47, XXY): Klinefelter's Syndrome
- Trisomy (47, XYY): XYY Syndrome

Multifactorial Inheritance

- anencephaly
- atopic allergies
- cleft lip/palate
- club foot
- congenital heart disease
- congential hip dysplasia
- congenital scoliosis
- diabetes mellitus
- epilepsy
- hydrocephalus
- non-specific MR
- NTD
- pyloric stenosis
- urinary tract malformations

Well recognized Syndromes, sequences, associations

- Down syndrome
- Pierre Robin sequence
- Trisomy 13
- Trisomy 18
- Cri du chat syndrome
- Turner syndrome
- Klinefelter syndrome

Down Syndrome

- Described by John Landon Down in 1866
- Etiology: nondisjuction mutation resulting in Trisomy 21, translocation
- Prevalence 1:700
  - Most common chromosomal anomaly
- Associated with Maternal age > 35

Down Syndrome

Genetics

- Three types of cytogenetic abnormalities result in the Down syndrome phenotype
  - Trisomy 21 (47,+21), in which an extra copy of chromosome 21 is present — 95 %
  - Robertsonian translocation involving chromosome 21— 4 %
  - Trisomy 21 mosaicism (47,+21/46), which occurs in 1 % of cases.
    - In this form, there are two populations of cell types: a normal cell line with 46 chromosomes and a second line with trisomy 21

Down Syndrome

- Actual risk for Down Syndrome:
  - 1/10 if female is translocation carrier
  - 1/20 if male is translocation carrier
- Translocations account for 9% of the children with Down syndrome born to mothers younger than 30 yr of age.
- Half the translocations arise de novo in the affected individual, whereas half are inherited from a translocation carrier parent

Down Syndrome

DYSMORPHIC FEATURES

- Each characteristic dysmorphic feature is present in 47 to 82 percent of cases
- These features predominantly affect the head and neck and the extremities

Down Syndrome

- Characteristic dysmorphic features of DS affecting the head and neck include:
  - Brachycephaly /flat occiput
  - Upslanting palpebral fissures
  - Epicanthic folds
  - Brushfield spots
  - Flat nasal bridge
  - Folded or dysplastic ears
  - Small ears
  - Protruding tongue
  - Narrow palate
  - Short neck
  - Excessive skin at nape of the neck

Down Syndrome

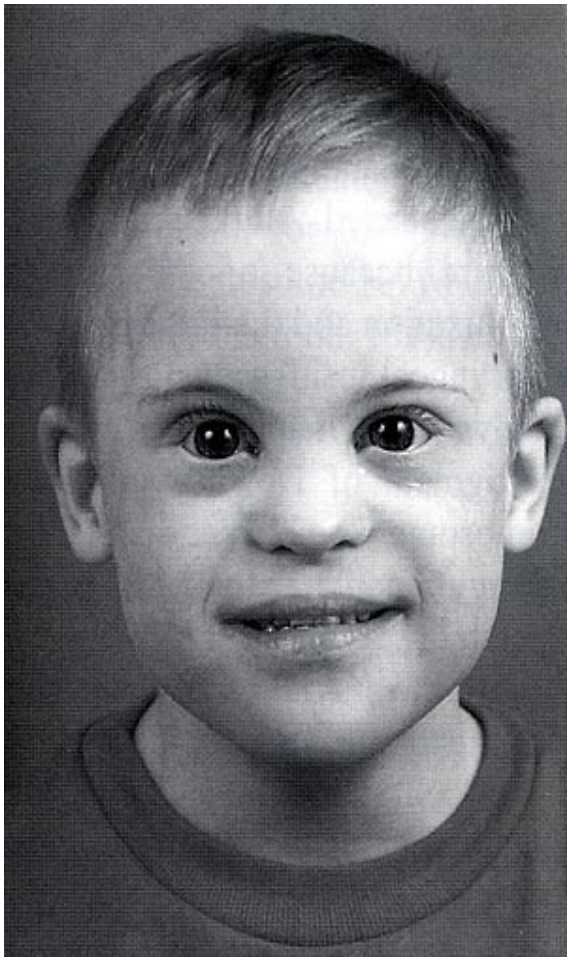
- Characteristic dysmorphic features of DS affecting the extremities include:
  - Short broad hands
  - Hypoplastic mid phalanx of fifth finger
  - Incurved fifth finger
  - Transverse palmar crease
  - Space between the first and second toes (sandal gap deformity)
  - Hyperflexibility of joints

Down Syndrome



Down Syndrome

photograph



Down Syndrome

- Airway Concerns
  - Due to midface hypoplasia, the nasopharynx and oropharynx dimensions are smaller
    - Slight adenoid hypertrophy can cause upper airway obstruction
  - Congenital mild-moderate subglottic narrowing

Down Syndrome

Growth and development

- Mental retardation- All individuals with DS have cognitive impairment, although the range is wide
- Developmental impairment becomes apparent in the first year of life
- the average age of sitting (11 months), creeping (17 months), and walking (26 months) is twice the typical age
- Birth weight, length, and head circumference are less in DS compared to typical infants
- Newborns with DS weigh approximately 0.18 to 0.37 kg less than their siblings

Down Syndrome

- Obstructive Sleep Apnea
  - Prevalence 54-100% in DS patients
  - Combination of anatomic and functional mechanisms
    - Midface hypoplasia, macroglossia, etc
    - Hypotonia of pharyngeal muscles

Down Syndrome

Hearing loss

- Hearing impairment affects 38 to 78 percent of individuals with DS
- Congenital and Acquired, overall high incidence
- Conductive hearing loss
  - more common
- Sensorineural hearing loss
  - less common
- Otitis media is a frequent problem (50 to 70 %)
  - Monitoring for this condition is important to preserve hearing

Down Syndrome

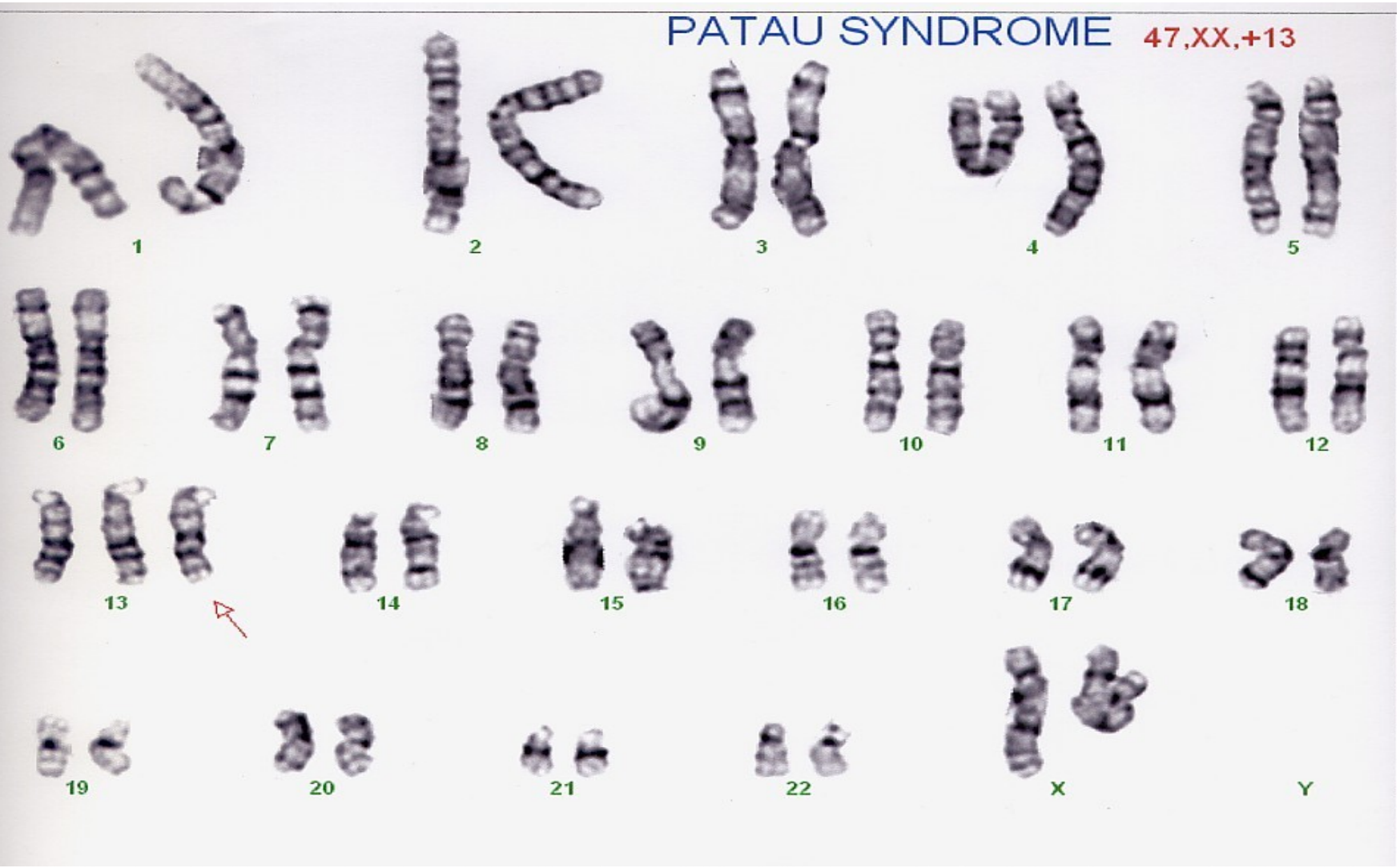
- Cardiovascular anomalies (40%)
  - Atrioventricular septal defect (ECD) with or without other lesions — 45 percent
  - VSD- 35%
  - Others -ASD, ,Tetralogy of Fallot,PDA
- GI anomalies (5%)
  - Pyloric stenosis, duodenal atresia, TE fistula
- Malignancy
  - 20 fold higher incidence of leukemia
  - Gonadal tumors

Down Syndrome

- Reproduction:
  - Women with DS are fertile and may become pregnant
  - Nearly all males with DS are infertile. The mechanism is impairment of spermatogenesis

Trisomy 13
Patau Syndrome

Patau Syndrome 47,XY,13+



Trisomy 13
Major Clinical Features

- mental retardation
- growth retardation
- microcephaly
- cleft lip/palate
- small jaw (micrognathia)
- deformed, low-set ears
- polydactyly
- congenital heart defects
- seizures
- low birth weight



Patau’s syndrome – Trisomy-13

Patau synd…



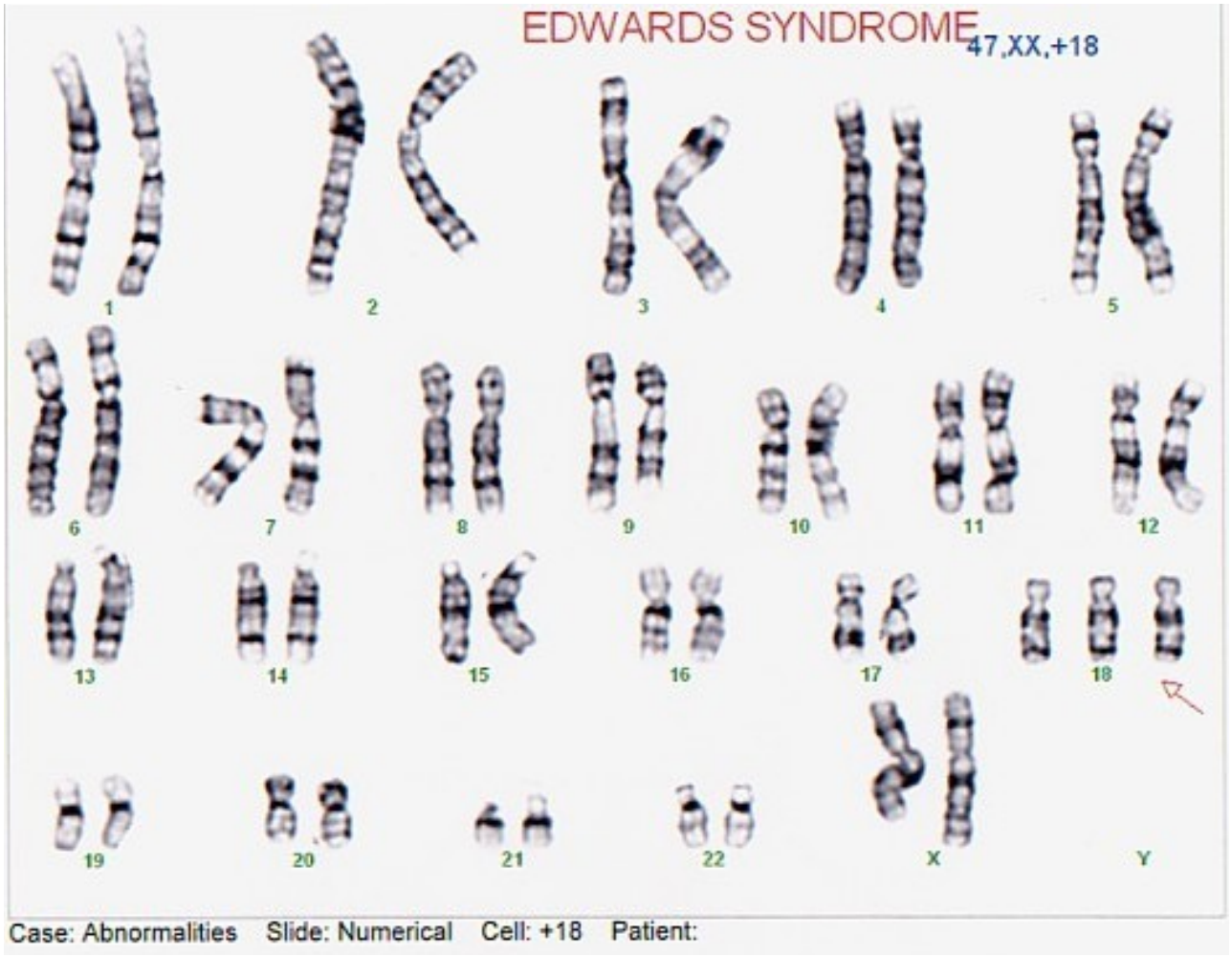
Patau synd…polydactyly





Trisomy 18
Edward’ Syndrome

Edward’ Syndrome 47,XX,+18



Trisomy 18
Major Clinical Features

- mental retardation
- growth retardation
- short neck
- cleft lip/palate
- dislocated hips/abnormal pelvis
- deformed, low-set ears
- hypertonia
- congenital heart disease
- horseshoe kidneys
- hydronephrosis
- short sternum
- pyloric stenosis

Edwards……



Trisomy 18

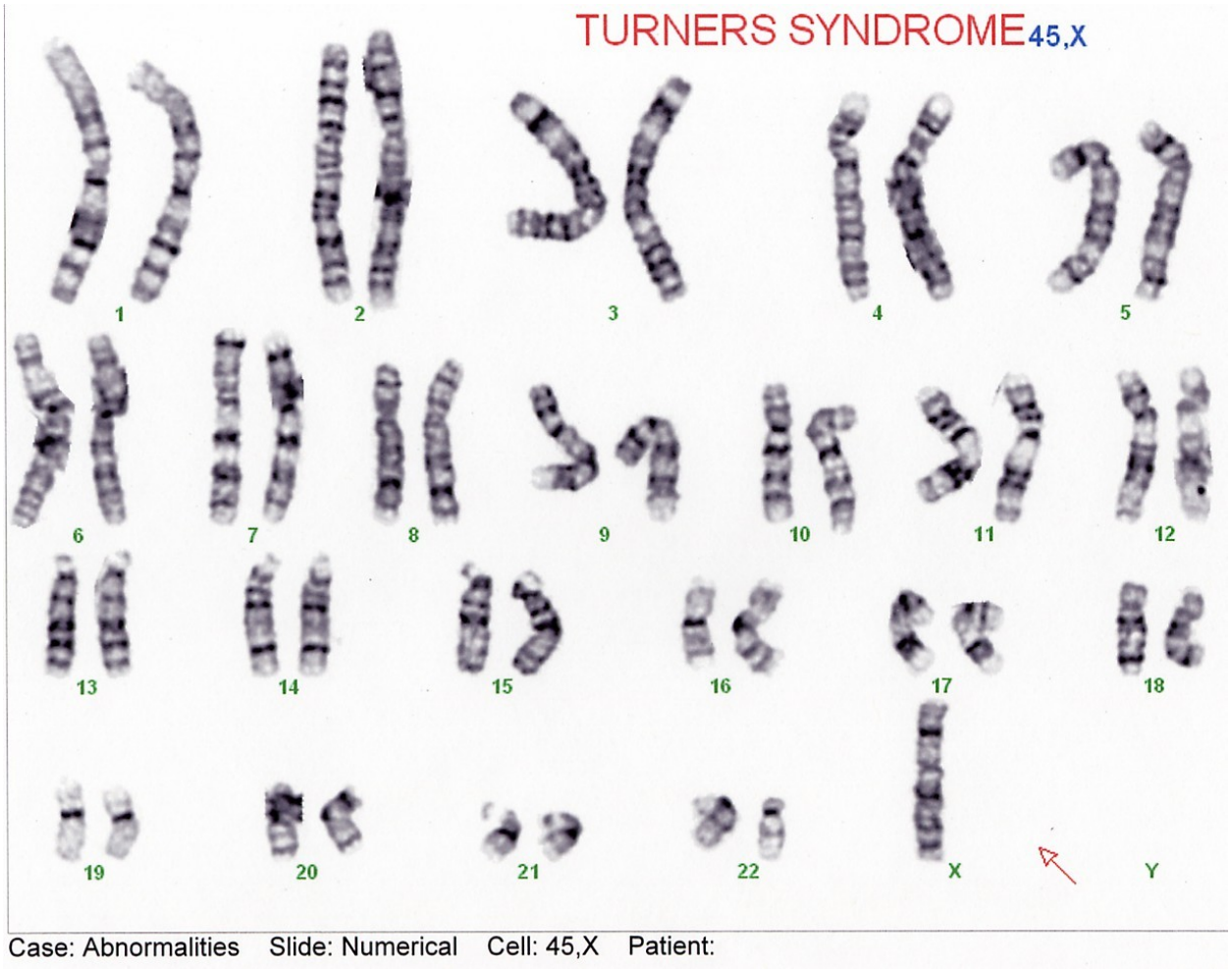
- Occurs in .3/1000 newborns
- Clinical features include weak cry, polyhydramnios, growth deficiency, low-set malformed auricles, clenched hand with overlapping fingers, rocker bottom feet, congenital heart defects.





Sex Chromosome Abnormalities

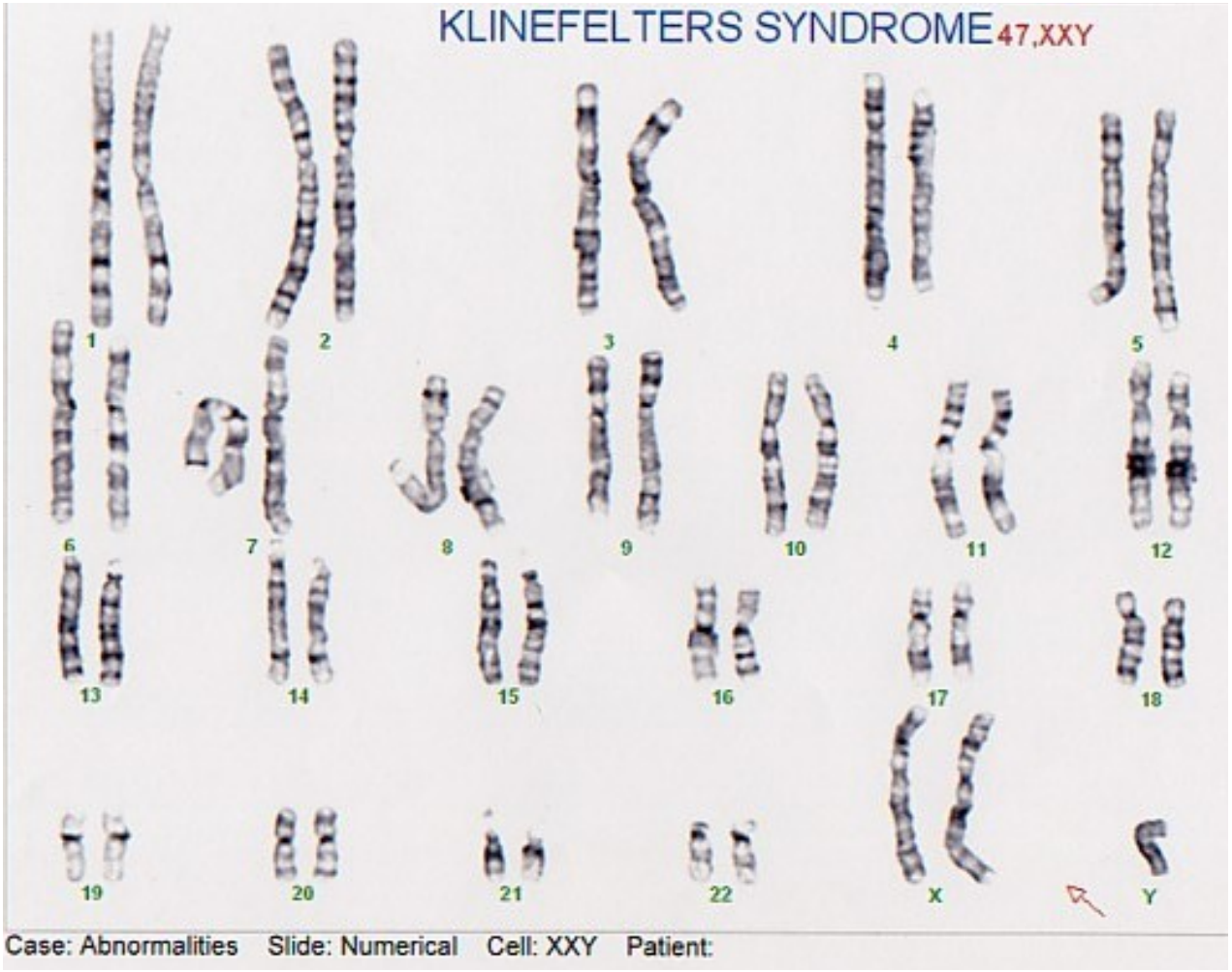
Turner's Syndrome 45,X



Turner's Syndrome
Major Clinical Features

- female phenotype
- short (less than 5 feet)
- primary amenorrhea
- low estrogen levels
- maldevelopment of the ovaries
- sterility
- webbing of the skin of the neck
- wide-spaced nipples
- edema at birth
- cardiovascular problems

Klinefelter's Syndrome 47,XXY



Klinefelter's Syndrome
Major Clinical Features

- small testes
- aspermia (little to no sperm production)
- Gynecomastia.
- long limbs.
- large hands & feet
- retardation in some
- fertility in some
- social limitations in some
- hypogonadism

DYSMORPHOLOGY

The study of abnormalities of human form and the mechanism that cause these abnormalities

Incidence

- 1:40 (2.5%) of NB have recognizable malf. at birth
- 50% have single malf. & 50% have multiple malf.

Definitions-Pathogenetic mechanisms

- Malformation
- Deformation
- Disruption
- Dysplasia
- Sequence
- Syndrome
- Association
- Complex

Malformation

- "a morphologic defect of an organ, part of an organ, or larger area of the body resulting from an intrinsically **abnormal developmental process**" **OR**
- "**a primary structural defect** arising from a localized error in morphogenesis, resulting in the abnormal formation of a tissue or organ"
- Caused by genetic or environmental influences

  e.g., cleft lip, neural tube defect

Deformation

- "**abnormal form or position** of a body part caused by **nondisruptive mechanical forces**" OR
- "an alteration in shape or structure of a structure or organ that **has differentiated normally**"
- Usually late in fetal development
- Mechanical (extrinsic) or functional (intrinsic)
- e.g., Oligohydramnios produces deformations by in utero compression of limbs (dislocated hips, equinovarus foot deformity), crumpled ears, dislocated nose, or small thorax,

Disruption

- "defect of an organ, part of an organ, or a larger area of the body due to interference with a normal process" ↳ tissue breakdown with cell death
- "a structural defect resulting from **the destruction of a structure** that **had formed normally before the insult**"
- Never inherited but inherited factors can predispose to and influence the development of disruption
- Disruptive agents:
  - Severe mechanical stress
  - Intrauterine viral infections
  - Tissue ischemia

Both deformation and disruption:

- Affect previously normally developed structures with no intrinsic tissue abnormality
- Unlikely to have a genetic bases
- Often not associated with cognitive deficit

Malformation and Deformations



Dysplasia

- structural defects resulting from abnormal cellular organization or function affecting one tissue type throughout the body, OR
- an abnormal organization of cells into tissues
- Tendency to persist throughout life.

Major Versus Minor Anomalies

- **Major congenital defects-**those abnormalities that (if uncorrected or uncorrectable) significantly impair normal body function or reduce normal life expectancy……2-3% of all
- It could be lethal or severe malformation
- A malformation is considered lethal if it causes still birth or infant death in more than 50% of cases
  - Eg. IHPS, Down syndrome, cleft lip, NTD

- **Minor anomalies**-only of primary cosmetic significance.
  - A normal variant usually occurs in 4% or more of the population.
  - It is common for isolated minor anomalies to be familial
  - Frequent in areas of complex and variable features such as the face and distal extremities
  - Eg-prominent forehead, preauricular pits

Isolated Versus Multiple anomalies

- **Isolated**-affecting only a single body site; most cong anomalies are isolated-may be minor(most) or major(few)
- most isolated major anomalies display multifactorial inheritance
- Recurrence risk is often low
- -Eg. Anencephaly ,cleft lip/palate, CHD,IHPS,NTD,DDH,TEF
- **Multiple**-combinations of birth defects

Complex

- anomalies of several different structures in the **same local body region during embryonic development.**
- Usually secondary to vascular insufficiency during embryogenesis affecting organs with geographic proximity
- Eg. hemifacial microsomia,sacral agenesis

Association

- "**nonrandom occurrence of a group of anomalies in multiple individuals**, not known to be a sequence or syndrome" OR
- "a nonrandom collection of malformations where there is an **unclear relationship among** the malformations such that they do not fit the criteria for a syndrome or sequence"
- Alert clinicians to look for related problems

  e.g.VATER

Sequence

- "**multiple defects that occur as a result of a single presumed structural anomaly**" OR
- The pattern of multiple anomalies that occurs when a single primary defect in early morphogenesis produces multiple abnormalities through a cascading process of secondary and tertiary errors in morphogenesis

- e.g., Pierre Robin sequence

Syndrome

- "pattern of multiple anomalies **believed to be pathogenetically related** and not representing a sequence"
- "a pattern of multiple abnormalities that are related by pathophysiology and result from a common, defined etiology"
- Pathogenesis less understood
- e.g., Downs syndrome

Causes of Congenital Malformations

Monogenic (7.5% of Sev Anomalies)

Chromosomal (6% of Sev Anomalies)

- Trisomies 21, 18, 13
- XO, XXY
- Deletions

Maternal Infection (2% of Sev Anomalies)

- Intrauterine infections

  (e.g., herpes simplex, CMV, varicella-zoster, rubella, and toxoplasmosis)

Maternal Illness (3.5% of Sev Anomalies)

Uterine Environment (% Unknown)

- Deformation
- Uterine pressure, oligohydramnios: clubfoot, torticollis, congenital hip dislocation, pulmonary hypoplasia,
- 7th nerve palsy
- Disruption
- gastroschisis, intestinal atresia
- Conjoined twins

Cont…

Environmental Agents (% Unk)

- Mercury
- Alcohol

Medications (% Unknown)

- Phenytoin
- Warfarin
- Cytotoxic drugs
- Valproic acid

Nutritional

- Low folic acid–neural tube defects

Unknown Etiologies(>50%)

- Anencephaly / spina bifida
- Cleft lip / palate
- Pyloric stenosis
- Congenital heart disease
- VATER syndrome
- Pierre Robin syndrome

Approach to Diagnosis

- Over 3,000 known syndromes
- Prenatal Hx - Obstetric hx, prenatal care, exposures, U/S, Placental Pathology
- Past medical history- Postnatal hx, illnesses, hospitalizations, surgery, medications, G&Devt.
- Family history- Parental ages, hx of similar/related anomalies
- maternal and paternal age

Approach to Diagnosis (cont.)

- Physical examination
  - compare other siblings/family member photos
  - Measurement of growth parameters & individual body parts, percentiles & body proportions, Photography
  - Major and minor anomalies
    - Airway
    - Skull
    - Ears
    - Facial skeleton
    - isolated minor anomalies
    - more than 3 minor anomalies (90% with major)
    - mental retardation associated with multiple anomalies
- Reference books are helpful

Management Issues

- Team approach
- Otolaryngology issues
  - upper airway obstruction
  - hearing Loss
  - speech Disorders

Neural Tube Defects (Dysraphism)

- account for most congenital anomalies of the central nervous system (CNS) and
- result from failure of the neural tube to close spontaneously between the 3rd and 4th wk of in utero development.

- the precise cause of neural tube defects remains unknown
- Factors that increases the occurrence of NTD includes:
  - hyperthermia, drugs,
  - malnutrition, chemicals,
  - maternal obesity or diabetes, and
  - genetic determinants (mutations in folate-responsive or folate-dependent pathways),
  - exposure to radiation before conception.

- The major neural tube defects include:-
  - spina bifida occulta,
  - meningocele,
  - myelomeningocele,
  - encephalocele, anencephaly, dermal sinus, tethered cord, syringomyelia, diastematomyelia.
- Failure of normal induction is responsible for most of the neural tube defects

- Failure of closure of the neural tube allows excretion of fetal substances (α-fetoprotein [AFP], acetylcholinesterase) into the amniotic fluid, serving as biochemical markers for a neural tube defect
- Prenatal screening of maternal serum for AFP in the 16th–18th wk of gestation is an effective method for identifying pregnancies at risk for fetuses with neural tube defects in utero

- Normally, the rostral end of the neural tube closes on the 23rd day and the caudal neuropore closes by a process of secondary neurulation by the 27th day of development, before the time that many women realize they are pregnant

Myelomeningocele

- the most severe form of dysraphism involving the vertebral column and occurs with an incidence of ≈1/4,000 live births.
- the risk of recurrence after one affected child increases to 3–4% and increases to ≈10% with two previous abnormal pregnancies
- Nutritional and environmental factors undoubtedly have a role in the etiology of myelomeningocele as well



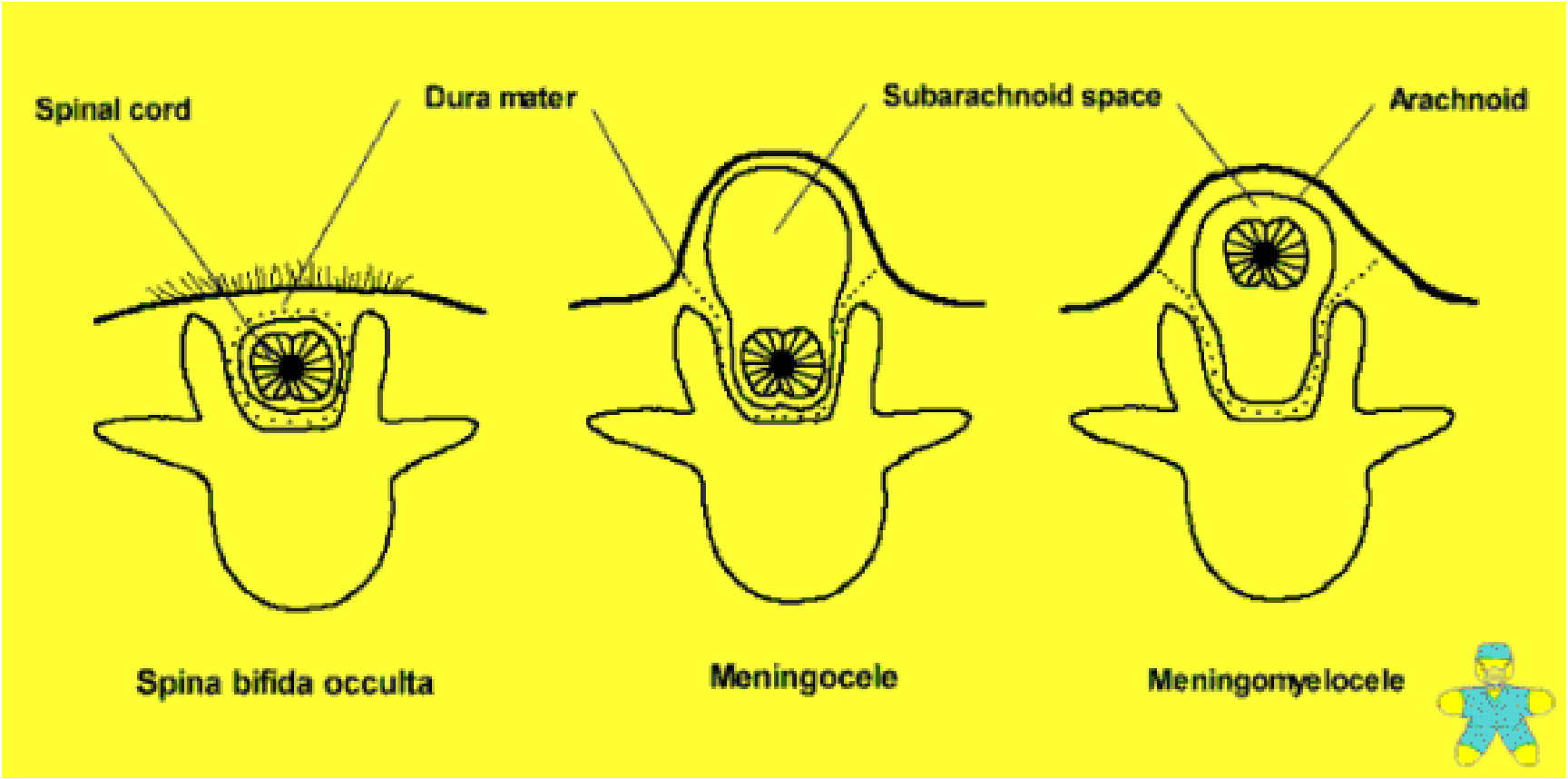
- Folate is intricately involved in the prevention and etiology of NTDs
- Maternal periconceptional use of folic acid supplementation reduces the incidence of neural tube defects in pregnancies at risk by at least 50%

CLINICAL MANIFESTATIONS

- produces dysfunction of many organs and structures, including the skeleton, skin, and gastrointestinal and genitourinary tracts, in addition to the peripheral nervous system and the CNS
- located anywhere along the neuraxis, but the lumbosacral region accounts for at least 75% of the cases
- A lesion in the low sacral region causes bowel and bladder incontinence associated with anesthesia in the perineal area but with no impairment of motor function

- defect in the midlumbar region typically have a saclike cystic structure covered by a thin layer of partially epithelialized tissue
- flaccid paralysis of the lower extremities, an absence of deep tendon reflexes, a lack of response to touch and pain, and a high incidence of lower extremity deformities (clubfeet, subluxation of the hips)
- Constant urinary dribbling and a relaxed anal sphincter may be evident

What is Spina Bifida?



Source: http://www.surgical-tutor.org.uk/default-home.htm?system/hnep/neural_tube.htm~right

- Infants with myelomeningocele typically have an increasing neurologic deficit as the myelomeningocele extends higher into the thoracic region
- Hydrocephalus in association with a type II Chiari defect develops in at least 80% of patients with myelomeningocele

Chiari crisis

- due to downward herniation of the medulla and cerebellar tonsils through the foramen magnum
- About 15% of infants with hydrocephalus and Chiari II malformation had symptoms **of hindbrain dysfunction**, including **difficulty feeding, choking, stridor, apnea, vocal cord paralysis, pooling of secretions, and spasticity of the upper extremities**

TREATMENT

- multidisciplinary team approach
- Surgery is often done within a day or so of birth but can be delayed for several days (except when there is a CSF leak)
- genitourinary system - regularly catheterize a neurogenic bladder

PROGNOSIS

- the mortality rate is ≈10–15%, and most deaths occur before age 4 yr
- At least 70% of survivors have normal intelligence, but learning problems and seizure disorders are more common than in the general population
- Renal dysfunction is one of the most important determinants of mortality

PREVENTION

- All women of childbearing age and who are capable of becoming pregnant take 0.4 mg of folic acid daily
- In high-risk women (previously affected child) supplementation should be started with 4 mg of folic acid daily, beginning 1 mo before the time of the planned conception
- To be effective, folic acid supplementation should be initiated before conception and continued until at least the 12th wk of gestation when neurulation is complete

Ambiguous genitalia

- Normal values in newborns
  - Female:
    - Vaginal opening fully visible: 3-4 mm slit
    - Clitoris width 2-6 mm
    - Absence of gonads in labia majora or inguinal region

Male: The mean length of the term newborn penis is 3.5±0.7 cm

- The diameter is 1.1±0.2 cm.
- The diagnosis of Micropenis is made if the stretched length is less than 1.9 cm.
- Micropenis results from a hormonal abnormality that occurs after 14 wk of gestation.
  - Common causes include hypo gonadotropic hypogonadism, hyper gonadotropic hypogonadism (primary testicular failure), and idiopathic Micropenis.
  - Bilateral testes of normal size (8-14 mm) in the scrotal sacs