

Question 12



The abnormality pictured above has been present since birth.

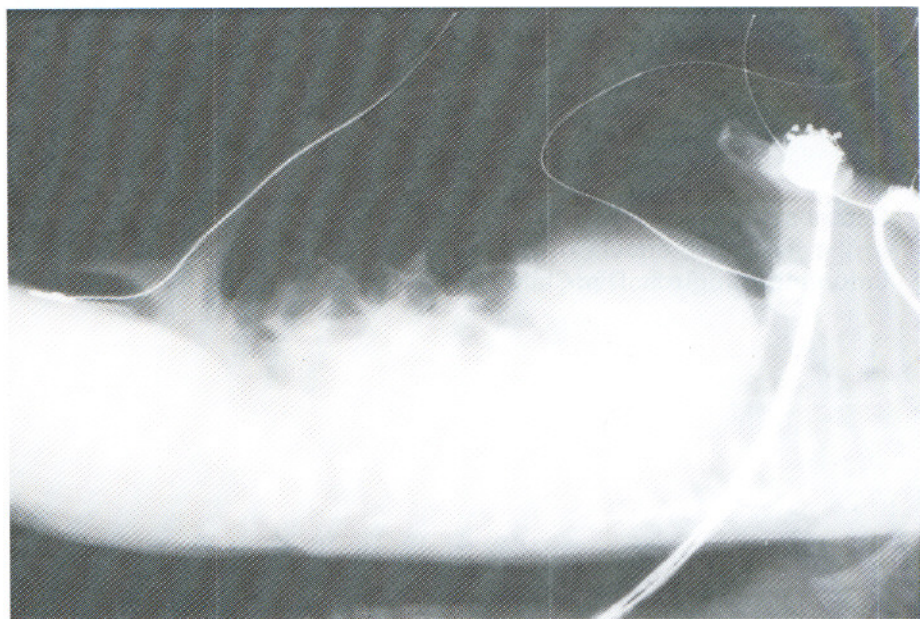
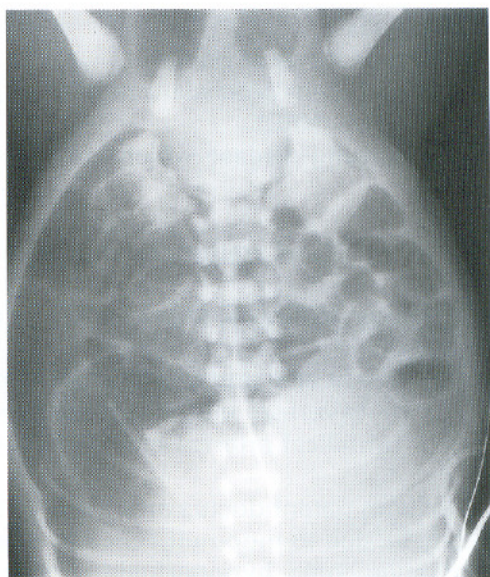
- a) What is the diagnosis?
- b) What investigation will confirm your diagnosis?
- c) Give two potential complications.

Answer to Question 12

- a) Thoracic lymphangioma
- b) Ultrasound ('bunch of grapes' picture)
- c) Sepsis and haemorrhage

- Developmental abnormality of lymphatic origin.
- Endothelial cell lining.
- Most common site affected is the neck (cystic hygroma).
Others include: lip, tongue, axilla and groin.
- Complications depend upon the location: obstructed labour, tracheal compression, difficulty in mastication and deglutition.
- Surgery is hazardous due to the risk of haemorrhage or the involvement of deeper structures. Several tissue planes may be involved.

Watson A C H 1992 Surgical paediatrics. In: Campbell A G M, McIntosh N (eds) Forfar and Arneil's textbook of paediatrics, 4th edn. Churchill Livingstone, Edinburgh

Question 13

A severely asphyxiated preterm infant deteriorated with abdominal distension and acidosis whilst receiving mechanical ventilation. These abdominal X-rays were obtained.

- List three abnormal radiological features.
- What is the most likely diagnosis?
- Why was a rectal biopsy performed in the convalescent period?

Answer to Question 13

- a) Free intraperitoneal gas
 - Thickened oedematous loops of bowel
 - Intrascrotal gas (via a patent processus vaginalis)
 - b) Necrotizing enterocolitis (NEC)
 - c) Exclusion of Hirschsprung disease which may present with acute enterocolitis (15%)
- NEC may affect up to 35% of preterm infants. It can occur in term infants.
 - Multifactorial aetiology: asphyxia, shock, sepsis and early, particularly formula, feeds.
 - Mortality 40%; surgery in 20% and late GI strictures in 10%.
 - Antenatal absent or reversed end diastolic flow velocities on Doppler studies of umbilical arteries may predict those at risk of NEC.
 - NEC is associated with patent ductus arteriosus and indomethacin therapy. This may be due to indomethacin administration causing exacerbation of splanchnic hypoperfusion, due to the ductus, resulting in NEC.
 - X-ray abnormalities include: fixed dilated oedematous loops of bowel, pneumatosis intestinalis, perforation and intrabiliary gas.
 - Clostridial species have been implicated in 20% of cases.
 - Management entails supportive therapy, antibiotics and cessation of feeds with the introduction of parenteral nutrition.

Coombs R C, Morgan M, Durbin G et al 1990, Gut blood flow velocities in the newborn: effects of patent ductus arteriosus and parenteral indomethacin. Arch. Dis. Child. 65:1067–1071

Question 14

- a) What is the diagnosis?
- b) In which multisystem disorder may this physical sign occur?

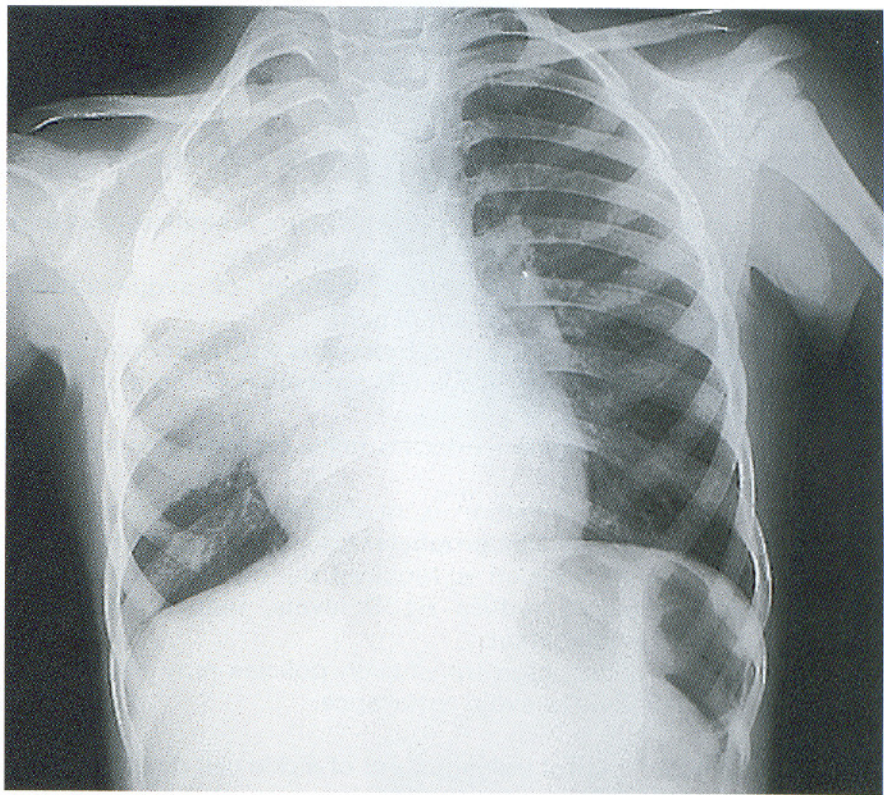
Question 14

- a) What is the diagnosis?
- b) In which multisystem disorder may this physical sign occur?

Answer to Question 14

- a) Erythema marginatum
- b) Acute rheumatic fever

- Erythema marginatum comprises erythematous rings with pale centres of normal skin. These frequently coalesce and may vary in conformation during the day.
- It only occasionally appears in acute rheumatic fever and is then considered to constitute a major criterion.
- Other major criteria include: polyarthritiscarditis
subcutaneous nodules
rheumatic chorea.

Question 15

A 6-year-old girl with homozygous sickle cell disease presented with fever and respiratory failure. This is her chest radiograph.

- a) Give two possible diagnoses.
- b) List three measures designed to prevent these diagnoses.

Answer to Question 15

a) Right upper lobe pneumonia

Sickle chest syndrome

b) Penicillin prophylaxis

Antipneumococcal vaccination

High transfusion policy to maintain low HbSS concentrations

- Sickle chest syndrome may present with acute breathlessness and pain. It may mimic pneumonia on the chest X-ray.
- Sickle chest syndrome comprises changes of sequestration, infarction and subsequent consolidation.
- Encapsulated organisms, e.g. pneumococcus and haemophilus, are common and the risk of sepsis is 600 times that of the unaffected population.
- Defective immunocompetence occurs due to defective opsonization and hyposplenism.
- Antipneumococcal vaccination is recommended for those over 2 years of age and penicillin prophylaxis for those over 3 months.
- Acute exchange transfusion is required during sickle chest and neurological crises to reduce HbSS levels, reduce secondary sickling and aid oxygenation.
- Following such crises, high transfusion policies may have to be continued indefinitely to avoid recurrence.

Evans J P M 1989 Practical management of sickle cell disease. Arch. Dis. Child. 64:1748–1751

Haupt H M, Moore G W, Bauer T W et al 1982 The lung in sickle cell disease. Chest 81:332–337

Question 16

This sick child exhibits a severe dermatological condition.

- a) What is the diagnosis?
- b) Give three potential complications.
- c) Briefly outline your management.

Answer to Question 16

- a) Eczema herpeticum
 - b) Dissemination of herpes simplex
 - Encephalitis
 - Secondary bacterial infection
 - c) Parenteral and topical acyclovir
 - Broad spectrum, including antistaphylococcal, antibiotics
-
- Lesions may become haemorrhagic.
 - Ocular complications may occur.
 - Secondary sepsis is common.

Question 17

The above rash was noted in a child with neck stiffness and fever. The child subsequently developed petechiae.

- a) What is the most likely diagnosis?
- b) How does the prognosis vary in comparison with the usual presentation of this condition?

Answer to Question 17

- a) Maculopapular presentation of meningococcaemia.
- b) Recent reports fail to substantiate claims of improved prognosis compared to the usual picture.
- Previously, a maculopapular presentation was considered to indicate an improved prognosis and a milder clinical course.
- As the macular rash may progress to haemorrhagic purpura, regular review is necessary.
- In one study of children with meningococcal septicaemia, 38% presented with maculopapular rashes with or without petechiae, whilst 13% had maculopapular rashes alone.

Marzouk O, Thomsen A P J, Sills J A et al 1991. Features and outcome in meningococcal disease presenting with a maculopapular rash. Arch. Dis. Child. 66:485-487

Question 18

- What is the diagnosis in this 46XY child?
- List four associated features.
- What is the significance of seeking an auditory assessment?

Answer to Question 18

- a) Congenital hypothyroidism
- b) Myxoedematous macroglossia; hoarse cry, hypotonia, umbilical hernia, constipation, prolonged neonatal unconjugated hyperbilirubinaemia, transient or recurrent hypothermia, enlarged (>1 cm) posterior fontanelle and bradycardia
- c) Pendred syndrome (incidence 2:100 000)

- Incidence 1:4000–6000.
- Female preponderance quoted for thyroid dysgenesis.
- About 5% of cases manifest clinical abnormalities in first week of life.
- More common in Down syndrome (1:128).
- Thyroid dysgenesis > dyshormonogenesis (10%).
- Infants more commonly exhibit post-term gestation and birth weight >3500 g.
- Pronounced skin mottling may occur (cutis marmorata).
- Plasma T4 levels at diagnosis <30 mmol/l are more likely to be associated with agenesis and more severe clinical features.
- Prolonged jaundice arises from impaired bilirubin excretion and increased enterohepatic circulation.
- ECG may show low voltage and prolonged conduction time.
- Delayed ossification occurs; particularly seen in the knee. Excessive thyroxine replacement can cause craniosynostosis.

Fisher D A 1989 Congenital hypothyroidism. In: Brook C (ed) Clinical paediatric endocrinology, 2nd edn. Blackwell Scientific Publications, Oxford

Grant D B, Smith I, Fuggle P W et al 1992 Congenital hypothyroidism detected by neonatal screening: relationship between biochemical severity and early clinical features. Arch. Dis. Child. 67:87–91

Question 19



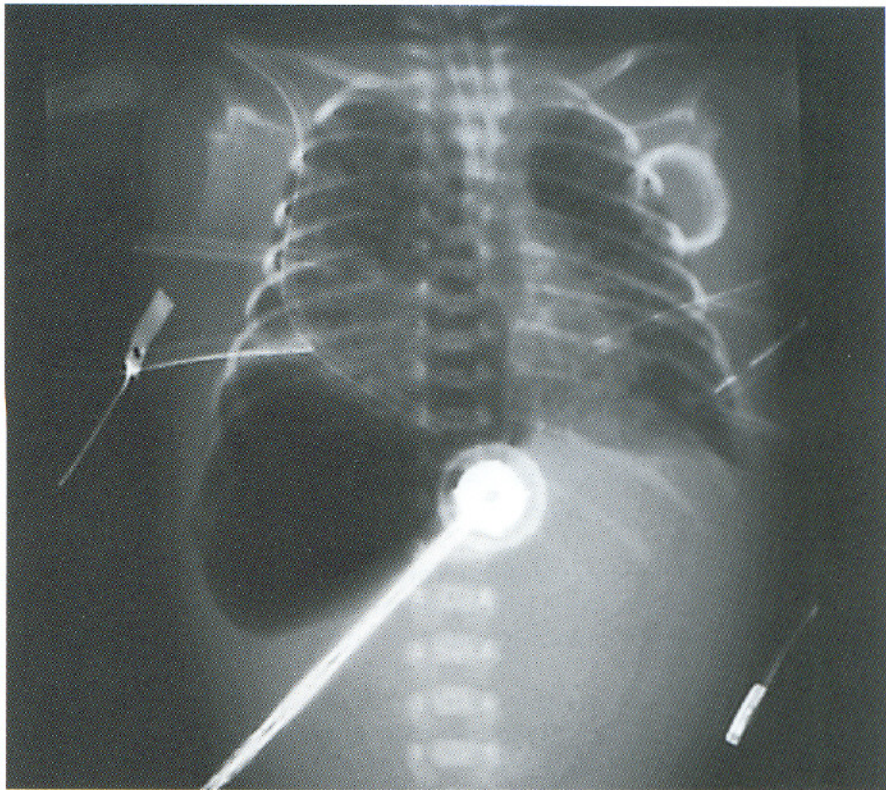
This infant was referred with poor feeding and irritability.

- a) What is the diagnosis?
- b) What is the most likely causative organism?
- c) List four other types of sepsis caused by this organism.

Answer to Question 19

- a) Cellulitis resulting from a paronychia infection
- b) *Staphylococcus aureus*
- c) Omphalitis
 - Osteomyelitis
 - Septicaemia
 - Pneumonia
 - Toxic epidermal necrolysis (Lyell or Ritter disease)
- Toxic epidermal necrolysis is potentially life-threatening due to exotoxin producing phage types (usually group II types 55 or 71).
- Skin friction causes epidermal shearing (Nikolsky sign).
- Parenteral antistaphylococcal antibiotics are required.

Question 20



A preterm infant of 29 weeks' gestation became acidotic and hypotensive during ventilation for hyaline membrane disease. This is the emergency chest radiograph.

- What is the cause of the sudden deterioration?
- How would you try to prevent such an occurrence?
- What neurological sequelae may ensue?

Answer to Question 20

- a) Right tension pneumothorax. There is evidence of previous pneumothoraces and indwelling chest drains.
 - b) Ventilator manipulation to avoid long inspiratory times.
Avoidance of high peak inspiratory pressures. Avoidance of active expiration asynchronously with the ventilator cycle. This may be achieved by increasing the ventilator rate to capture the infant's natural respiratory rate or by sedation and artificial paralysis.
 - c) Periventricular haemorrhage.
- Pneumothoraces occur in up to 35% of very low birth weight infants. They may occur in response to high intrapulmonary pressures due to the ventilator or active expiration against the ventilator cycle.
 - Paralysis or sedation may prevent some pneumothoraces.
 - The risk of pneumothoraces increases in the presence of pulmonary interstitial emphysema. The development of air leaks is associated with increased secretion of vasopressin which may lead to hyponatraemia.
 - Systemic hypotension resulting from pneumothoraces may lead to cerebral hypoperfusion and cerebral injury. Haemorrhage may occur once the circulating volume and cerebral perfusion are restored.
 - The effect of exogenous surfactant upon the incidence of pneumothoraces is still uncertain although some studies suggest a reduction in their occurrence.

Greenough A, Morley C J, Davis J A et al 1984 Pancuronium prevents pneumothoraces in ventilated premature babies who actively expire against positive pressure inflation. *Lancet* i:1-3

Lipscomb A P, Thornburn R J, Reynolds E O R et al 1981 Pneumothorax and cerebral haemorrhage in preterm infants. *Lancet* i:414-416

Morley C J 1991 Surfactant treatment for premature babies. *Arch. Dis. Child.* 66:445-450

Question 21

- a) What is the diagnosis?
- b) List five characteristic facial features of your diagnosis.
- c) Give three potential problems encountered in the neonatal period.

Answer to Question 21

- a) Down syndrome
 - b) Brachycephaly, microcephaly, slanted palpebral fissures, epicanthic folds, Brushfield spots, glossoptosis, small nose with widened nasal bridge, small ears with cartilaginous defects, microstomia and cataracts
 - c) Congenital cardiac disease
 - Duodenal atresia and other GI atresia or stenosis
 - Cataracts
 - Poor feeding and unco-ordinated sucking
 - Constipation due to poor fluid intake
 - Congenital hypothyroidism (1:300 of Down sufferers)
 - Parental anxiety, emotional problems, etc.
- Congenital cardiac defects in 20–40%; classically A-V canal defects in 50%.
 - Routine echocardiography is recommended. Cardiac disease is responsible for 35% of deaths and mortality is highest in those under 2 years of age.
 - 5% have GI malformations. Constipation is very common.
 - Skin problems are common: 75% have areas of hyperkeratosis; seborrhoeic dermatitis is recognized and pronounced skin mottling (cutis marmorata) occurs.
 - Up to 50% have cataracts in the neonatal period. Hypermetropia and defective nasolacrimal duct canalization are common. Myopia is present in about 20%.
 - Sleep apnoea due to upper airways obstruction is common. There is increased susceptibility to middle ear infections; over 50% of children have hearing deficits.

Newton R W, Newton J A 1992 The management of Down's syndrome. In: David T J (ed) Recent advances in paediatrics, Vol 10. Churchill Livingstone, Edinburgh

Question 22



This infant has a meningocele.

- Give four potential problems encountered by this infant.
- List five features considered to impart a poor prognosis when a pre-operative assessment is performed.
- How may such a defect be detected antenatally?

Answer to Question 22

- a) Urinary retention with overflow
Nephropathy due to high pressure vesico-ureteric reflux
Constipation with faecal overflow
Spastic diplegia
Hydrocephalus
Meningitis
 - b) Thoracolumbar lesion
Severe paraplegia
Kyphoscoliosis
Hydrocephalus
Intracerebral birth injury
Severe co-existing congenital defect
 - c) Elevated maternal serum alphafetoprotein level with subsequent elevated amniotic alphafetoprotein levels
Detailed anomaly scan
- Incidence 0.5–4:1000 live births.
 - Reduction in incidence in 'at risk' pregnancies following first trimester vitamin B complex and folic acid administration.
 - 80% develop hydrocephalus requiring treatment.
 - Arnold–Chiari malformation may co-exist.
 - Intermittent catheterization is regarded as the best method of urinary control. Renal function and the presence of reflux must be monitored.
 - 70% of sufferers walk to some degree eventually.
 - Orthopaedic surveillance is necessary to detect spinal instability or scoliosis.

Lorber J 1973 Early results of selective treatment of spina bifida cystica. Br. Med. J. IV:201–204

Question 23

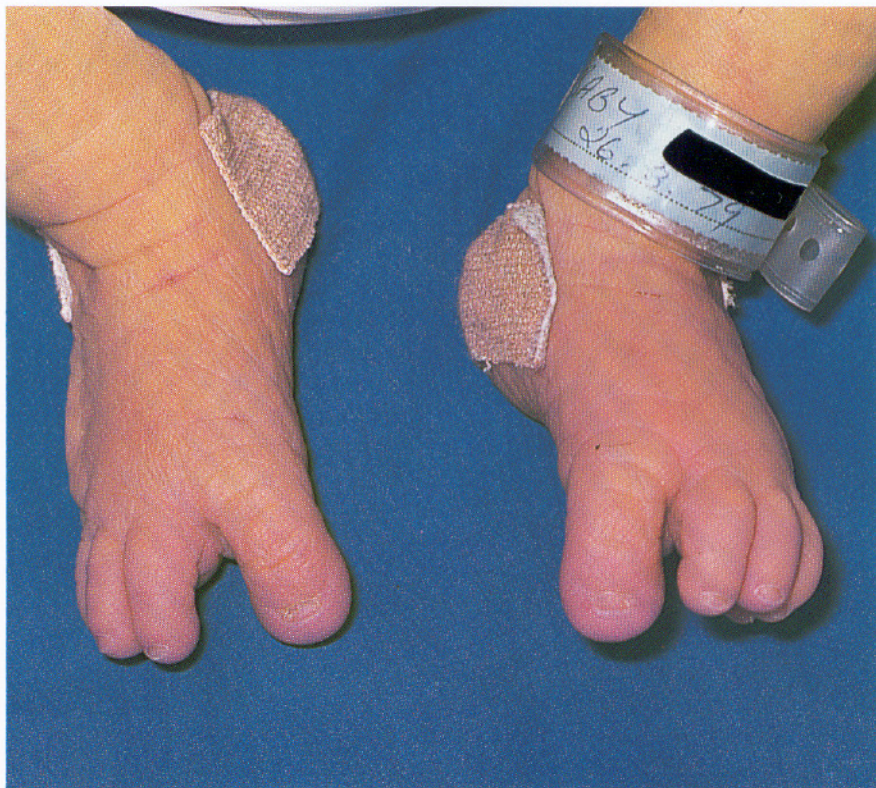
- a) What dysmorphic feature is shown?
- b) What is the most likely diagnosis?
- c) List four other features.

Answer to Question 23

- a) Clenched hand with overlapping fifth finger
- b) Edward syndrome
- c) Micrognathia
 - Occipital prominence
 - Low set malformed ears
 - Rockerbottom feet
 - Short sternum
 - Cryptorchidism

- Incidence 1:6000 live births.
- <10% survive one year.
- Long term survivors are usually mosaics.
- 90% have cardiac defects.
- Renal defects are common.
- Risk increases with maternal age.

Question 24



- What is the most likely diagnosis?
- Comment upon the likely growth pattern of this child.
- Give three conditions known to be more prevalent in those with this condition.

Answer to Question 24

- a) Wide gap between 1st and 2nd toes—Down syndrome
 - b) Reduced growth and resultant short stature. Delayed skeletal ossification
 - c) Alzheimer's disease
 - Epilepsy
 - Leukaemia
 - Hypothyroidism
 - Auto-immune diseases, e.g. vitiligo
 - Alopecia areata and totalis
-
- Growth retardation occurs leading to short stature. IGF-1 levels are low. There are some reports of response to high doses of exogenous growth hormone but although the height velocity is increased, the effect on final height is not yet known.
 - 5% of adolescents have biochemical indices of hypothyroidism. 25% of adults have antithyroid antibodies with clinical manifestation in 15–17%. TSH levels tend to increase with age.
 - Atlanto-axial dislocation is rare.
 - IQ is variable: scores are higher in mosaics; scores tend to decrease with age and 13% have behavioural problems. Many attain a reading age of between 8 and 12 years.
 - The incidence of leukaemia is higher than the general population. The remission rate is lower (81%) and the induction mortality is greater (14%). This may be due to greater susceptibility to sepsis and myelotoxicity.
 - Other 'feet' cases in the MRCP examination include:
 - Turner syndrome
 - Rubinstein-Taybi syndrome
 - Apert syndrome
 - Toe clubbing
 - Edward syndrome
 - Patau syndrome
 - Keratoderma blenorrhagica.

Question 25

This girl is recovering from pneumococcal meningitis.

- a)** Describe the physical sign demonstrated.
- b)** List four potential complications of bacterial meningitis.
- c)** What prophylaxis should be offered to this child's family?

Answer to Question 25

- a) Right third cranial nerve palsy: complete ptosis, divergent strabismus with the eye 'down and out' and pupillary dilatation (not shown).
- b) Seizures (acute or chronic)
- Cranial nerve palsies
 - Deafness
 - Cerebral infarction, abscess or cerebral palsy
 - Ophthalmitis
 - Hydrocephalus (usually communicating)
 - Subdural effusion
 - Syndrome of inappropriate ADH secretion
 - Adrenal haemorrhage (Waterhouse–Friedreichsen syndrome)
 - Endocarditis
- c) None. Prophylaxis required for haemophilus and meningococcal meningitis.

- Pneumococcal meningitis is a serious and notifiable disease. Mortality reaches 15% with major neurological sequelae in 25% of survivors.
- There may be a pre-existing focus of infection. Bacteraemia is almost invariable.
- Blood cultures are usually positive. Latex agglutination test is positive in 50–75% of cases.
- Treatment is traditionally continued for 10 days.
- Dexamethasone may reduce mortality and long term morbidity, particularly deafness, in pneumococcal meningitis. Reduction in deafness but not mortality has been suggested in those with haemophilus meningitis. The greatest benefit may be seen if the dexamethasone is given before or at the time of antibiotic administration.
- Endocarditis is a rare, and often fatal, complication. Normal valves may be affected.

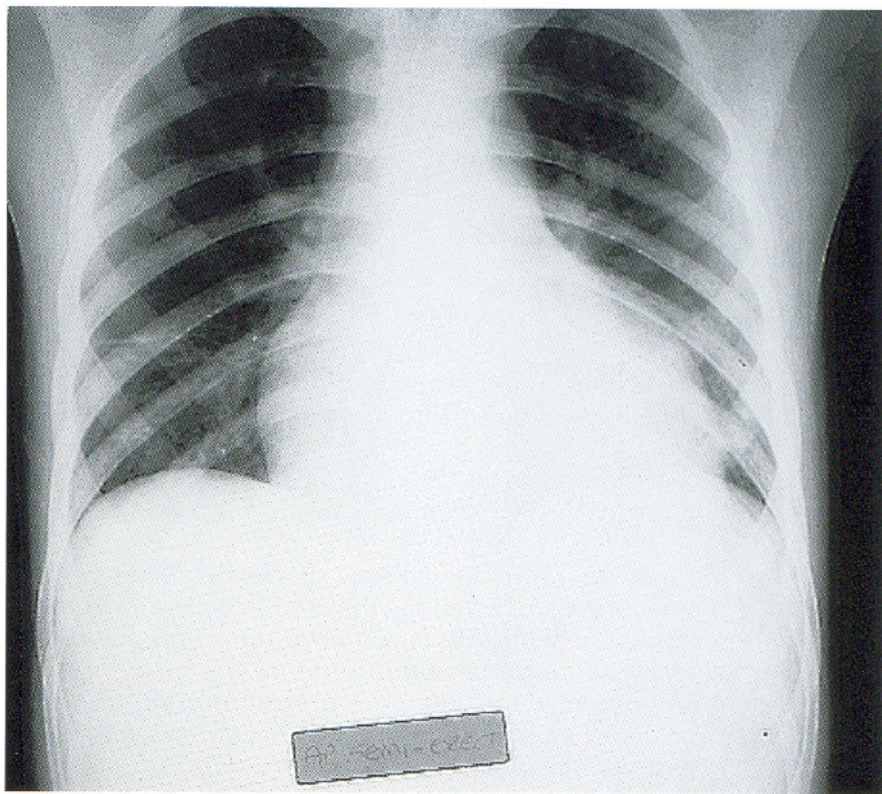
Levin M 1991 Bacterial meningitis. In: David T J (ed) Recent advances in paediatrics, Vol 9. Churchill Livingstone, Edinburgh

Question 26

- What is the diagnosis?
- What is your management?

Answer to Question 26

- a) Giant hairy naevus.
 - b) Remains debated. Many advise surgical removal to reduce the risk of melanomatous transformation.
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- Giant hairy naevi may become malignant in up to 30%.
 - They are an abnormality of dermal melanocytes.
 - Some are extensive—'bathing trunk' naevi.
 - Surgical removal may require skin grafting. Regular review is mandatory.

Question 27

A teenage boy with homozygous sickle cell disease presented with breathlessness, tachycardia, hepatomegaly and a long systolic murmur. He had received multiple blood transfusions in the past. This is his chest radiograph.

- What is the most significant radiological feature?
- What is the most likely diagnosis? Give two possible causes.
- List two helpful investigations to confirm your diagnosis.

Answer to Question 27

a) Cardiomegaly

b) Cardiac failure

Possible aetiologies: haemosiderotic cardiomyopathy
endocarditis due to immunosuppression
myocarditis

c) Echocardiography

ECG

Cardiac radionuclide scans

Ferritin and iron store studies

Septic screen

- Repeated transfusions may result in iron overload leading to cardiomyopathy and arrhythmias (particularly supraventricular arrhythmias).
- Haemosiderosis is prevented by the avoidance of repeated transfusions and the use of desferrioxamine chelation if such therapy is unavoidable.
- Transfusions may be given for aplastic crises but there is little evidence of amelioration of acute painful crises. Exchange transfusions are preferable when HbSS levels need reduction pre-operatively or following neurological and respiratory crises.
- Features of iron overload include: poor growth, delayed puberty, hypoparathyroidism, adrenal insufficiency, hepatosplenomegaly, arthropathy and late diabetes mellitus.
- Desferrioxamine therapy predisposes to yersinia infection. Repeated transfusions increases the risk of hepatitis C infection which is associated with a moderate mortality and high risk of chronic liver disease.

Ortega J A 1982 Clinical consequences and management of chronic iron overload. In: Willoughby M, Siegel S E (eds) Haematology and oncology (paediatrics 1). Butterworths, London

Question 28

This child has been held by the neck by friends and exhibits a play-associated injury.

- a) What physical sign is demonstrated?
- b) What is the pathophysiology?
- c) Give two occasions when this sign is also seen.

Answer to Question 28

- a) Peri-orbital petechiae.
 - b) Obstruction to the venous drainage of the head, associated with a degree of hypoxia due to tracheal compression, causing capillary rupture.
 - c) Pertussis (following a coughing spasm)
Non-accidental injury
Parturition (when the umbilical cord is wrapped around the infant's neck).
- This injury may be accompanied by subconjunctival haemorrhages.
 - Retinal haemorrhage must be excluded. These are commonly but not exclusively seen in abuse cases.
 - Play-associated injuries may mimic and be mistaken for child abuse. Such injuries have been associated with television viewing habits.
 - Traumatic peri-orbital petechiae were first described by the French police surgeon Tardieu.

Gadow K D, Sprafkin J 1989 Television violence. *Paediatrics* 83:399–405

Question 29

This infant has a humeral fracture.

- a) What is the underlying diagnosis?
- b) What is the aetiology of this diagnosis?
- c) What may be seen on the skull radiograph?

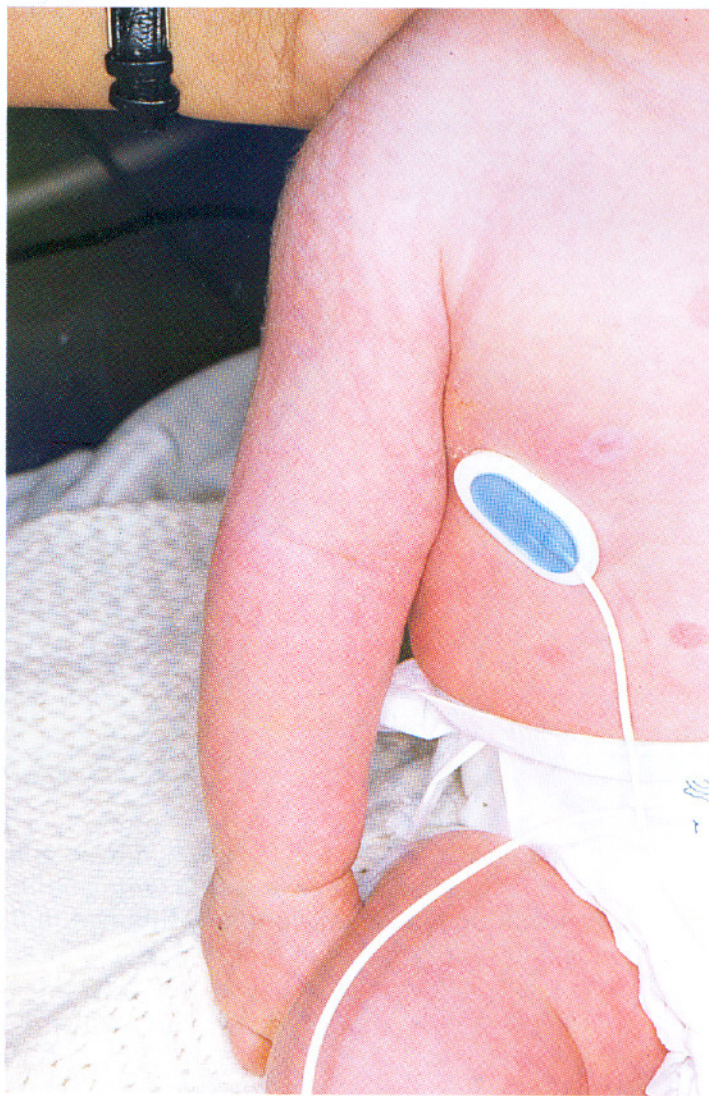
Answer to Question 29

- a) Osteogenesis imperfecta
- b) Heterogeneous group of disorders of Type I collagen synthesis
- c) Wormian bones (often occipital)

Previous fractures

- Variable inheritance 1:10 000–50 000; 80% of cases are Type I.
- Severity of the disease tends to be similar within one family.
- Inheritance: dominant in I and IV; recessive (usually) in II and III.
- Type II is often lethal in the perinatal period. Bones are short, broad and deformed.
- Blue sclerae with premature arcus may be absent in Types III and IV.
- Dentinogenesis imperfecta with translucent teeth is associated.
- Deafness (>50% by 50 years) is common. Otosclerosis is also commonly detected.
- Other evidence of collagen disease includes: joint hypermobility, tendon rupture and aortic valve regurgitation.
- Diaphyseal fractures are more common than metaphyseal lesions.
- Genetic abnormalities on chromosomes 7 and 17. The defect seems to be overhydroxylation of alpha 1 (I) chain collagen.
- Antenatal diagnosis is available; mild disease may be detected in asymptomatic family members by the presence of Wormian bones on plain skull radiographs.

Smith R 1984 Osteogenesis imperfecta. Br. Med. J. 289:394–395

Question 30

- a) What abnormality is shown?
b) What is the likely prognosis?

Answer to Question 30

a) Right Erb palsy

b) Full recovery in most cases

- Erb palsy is a neuropraxia of C5/C6 roots of the brachial plexus. The arm is limp and rests in pronation.
- The other main brachial plexus injury is Klumpke palsy.
- Pseudoparalysis is noted on examination with an absent Moro reflex. Fractures of the clavicle and humerus may co-exist.
- Predisposing factors include: breech delivery, shoulder dystocia and macrosomia.