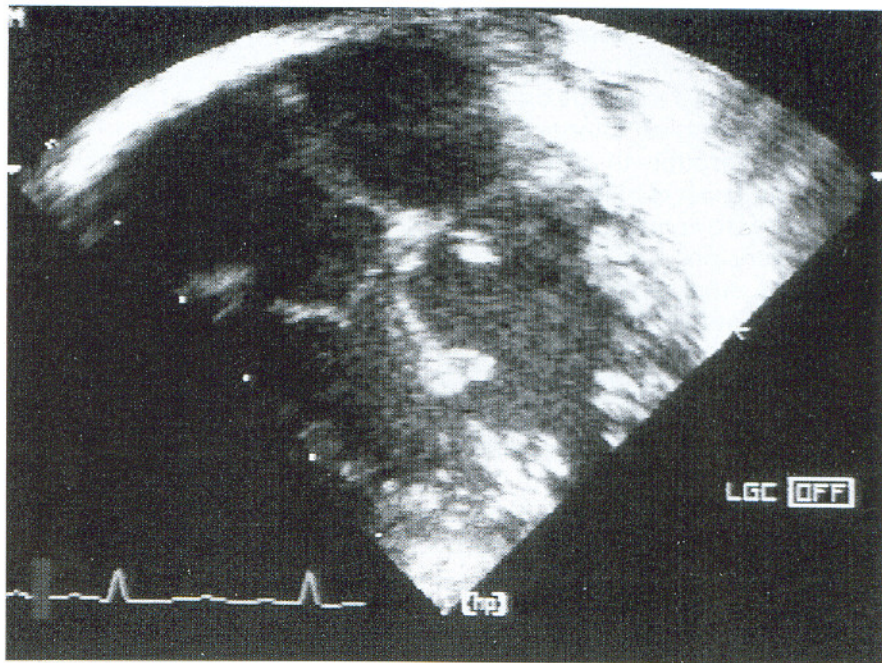


Question 72



This is a cross-sectional echocardiogram (4 chamber view) of a newborn.

- What is the abnormality?
- What is the prognosis?
- How would you manage this patient?

Answer to Question 72

- a) Ventricular septal defect–VSD (echogenic drop out half way through the ‘thick’ ventricular septum).
 - b) The prognosis depends on the size and the position of the VSD and the physiological changes, which take place as a result of blood flowing through the defect. Small defects usually close spontaneously, most of them during the first 1–2 years of life. Large defects may lead to congestive heart failure, with a potential for pulmonary hypertension.
 - c) Exclude other associated cardiac abnormalities
 - Early follow-up (4–6 weeks)
 - Medical treatment for heart failure
 - Surgical closure if heart failure is poorly controlled or signs of pulmonary hypertension develop
 - Provide SBE prophylaxis, when indicated.
- The pulmonary vascular resistance in healthy newborns decreases by 7–10 days; in patients with a VSD this is delayed to 4–6 weeks, hence the fact the large VSDs often are not clinically detectable during the first month of life.
 - The early recognition of developing pulmonary vascular disease is based on the assessment of the pulmonary component of the second heart sound (loud P2).
 - The doubly committed subarterial defects have a potential for acquired infundibular pulmonary stenosis and tetralogy of Fallot physiology.
 - Aortic regurgitation may complicate subaortic VSD later on in childhood as a result of prolapse of the coronary cusps, predominantly right, into the VSD.

Anderson R H, Shinebourne E A et al 1987 Ventricular septal defect. In: Anderson R H, Shinebourne E A (eds) Paediatric cardiology. Churchill Livingstone, Edinburgh

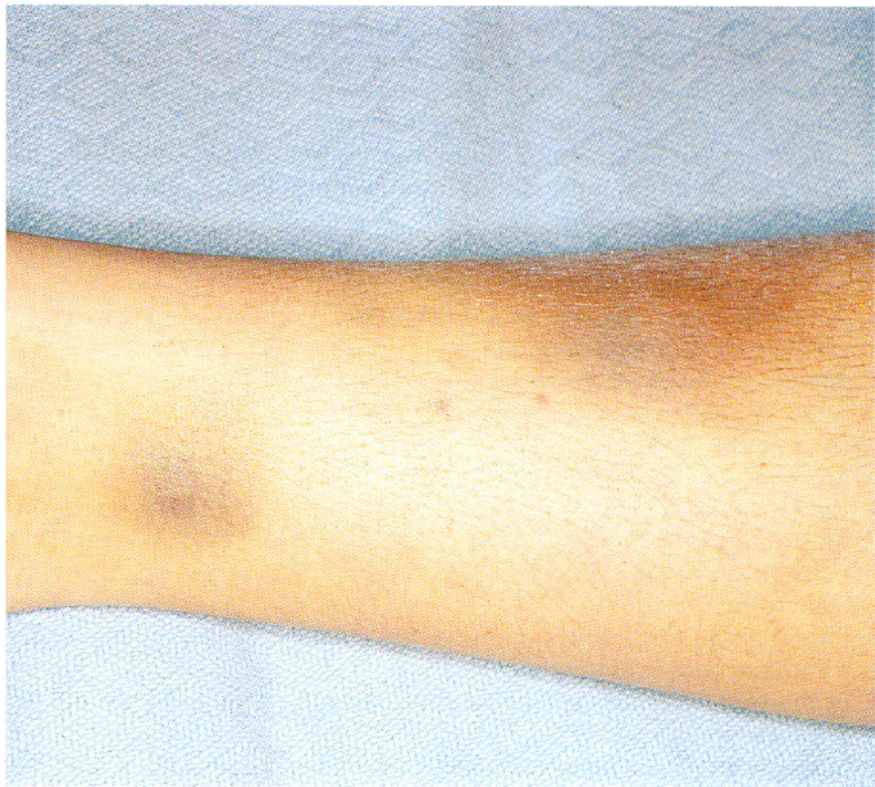
Question 73

This toddler presented to his GP with a history of paroxysmal cough following coryzal symptoms.

- a) What is the diagnosis?
- b) What is the aetiological factor?
- c) What is the treatment?

Answer to Question 73

- a) Pertussis
 - b) *Bordetella pertussis*. Identical clinical picture can be due to *Bordetella parapertussis*, *Bordetella bronchiseptica*, adenoviruses, parainfluenza and RSV.
 - c) Supportive treatment: maintenance of hydration and nutrition and oxygen with nasopharyngeal suction when needed.
-
- Pertussis has been described as the '100 day cough'.
 - Pneumonia, usually due to secondary bacterial invaders, is responsible for more than 90% of deaths under the age of 3.
 - An inspiratory whoop is not always seen in infants and older children or adults.
 - Subconjunctival haemorrhages are common; during coughing paroxysms the intrathoracic pressure rises sharply and affects the venous return to the heart. The sudden surges in capillary pressure may rupture the poorly supported subconjunctival vessels and produce alarming haemorrhages.
 - *Bordetella pertussis* can be isolated from as many as 90% of patients during the catarrhal stage of the disease, but from less than 50% during the paroxysmal stage.

Question 74

This child presented with spontaneous bruising and a platelet count of 10. She was otherwise well without hepatosplenomegaly or lymphadenopathy.

- a) What is the likely diagnosis?
- b) How do you confirm this?
- c) What is your acute management?

Answer to Question 74

- a) Idiopathic thrombocytopenic purpura (ITP)
- b) By excluding other causes of thrombocytopenia (e.g. leukaemias). If the diagnosis is in doubt bone marrow examination is mandatory.
- c) Admit to hospital to establish the diagnosis
 - Reassure the family about the benign nature of the condition
 - Reserve steroids or intravenous immunoglobulin for overt bleeding.
- Hepatosplenomegaly or lymphadenopathy are not features of ITP and suggest another diagnosis.
- Bone marrow examination is mandatory in all cases of presumed ITP if steroids are to be given.
- 90% of acute ITP remits spontaneously within 3 months and the rest within 6 months; persistence longer than 6 months is considered chronic.
- Mortality of childhood ITP is less than 1%.
- Whilst splenectomy is effective in raising platelets, it is rarely indicated in view of the risk of post-splenectomy fatal sepsis. This risk has been estimated at 1.4% compared to less than 1% mortality rate from ITP.
- Intravenous immunoglobulin is the treatment of choice post-trauma or pre-operatively.
- In the event of intracranial haemorrhage the following should be urgently performed:
 - Massive allogenic platelet transfusion
 - High dose intravenous immunoglobulin infusion
 - Emergency splenectomy.
- The above measures reduce the mortality rate to 50%.

Eden O B, Lilleyman J S 1992 Guidelines for management of idiopathic thrombocytopenic purpura. *Arch. Dis. Child.* 67:1056-1058

Lilleyman J S 1990 Changing perspectives in idiopathic thrombocytopenic purpura. In: Meadow R (ed) *Recent advances in paediatrics*, Vol 8. Churchill Livingstone, Edinburgh

Question 75

These are the hands of a toddler who is recovering from a 14 day acute febrile illness associated with conjunctivitis, pharyngitis and a generalized erythematous rash.

- a) What is the likely diagnosis?
- b) What important investigation should be undertaken as soon as the diagnosis is made or suspected?
- c) What is the treatment?

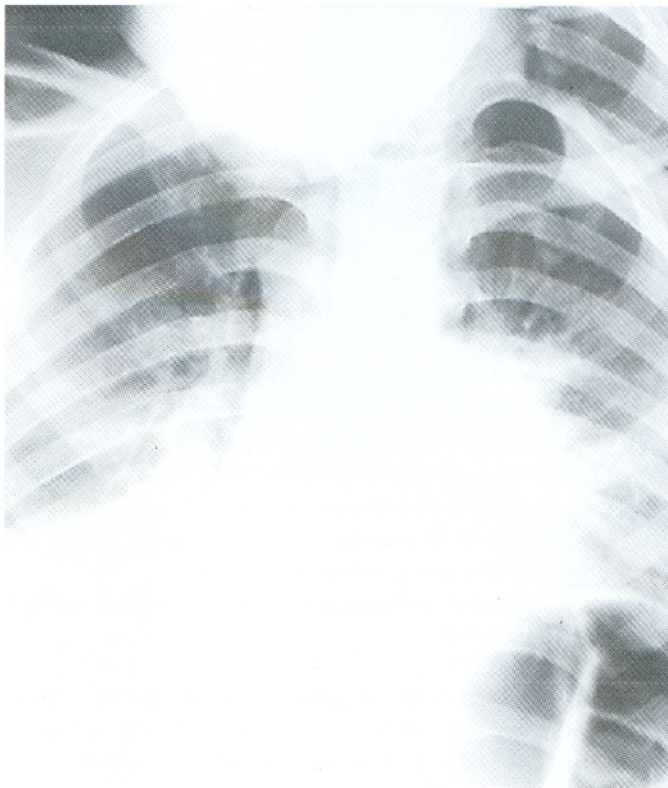
Answer to Question 75

- a) Kawasaki disease or mucocutaneous lymph node syndrome.
- b) Cross-sectional echocardiography for detection of coronary artery lesions.
- c) Aspirin is given in the acute phase and then in a reduced antiplatelet dose in the presence of coronary artery involvement.
Gammaglobulin intravenously, during the acute phase of the disease.

- Kawasaki disease is an immunologically mediated diffuse vasculitis of childhood of unknown aetiology.
- The diagnosis is made when five of the six following criteria are satisfied:
 - Fever longer than 5 days
 - Conjunctival injection
 - Mucous membrane changes: erythema, fissuring and crusting of the lips, strawberry tongue
 - Erythematous rash
 - Lymphadenopathy
 - Changes in the extremities (late onset): induration of hand and feet, erythema of palms and toes, desquamation of finger and toe tips.
- A neutrophil leucocytosis, thrombocytosis and a high ESR are commonly seen, but no organisms have been cultured and auto-antibodies are negative.
- Cardiac involvement, ranging from pericardial effusion to coronary artery aneurysm is the most important manifestation of the disease.
- 10 to 40% of cases will have echocardiographic evidence of coronary vasculitis within 2 weeks of the illness.
- Coronary involvement is significantly associated with leucocytosis and thrombocytosis.
- Most coronary lesions regress; good prognostic factors are: age less than 1 year, female sex, fusiform aneurysm and maximum diameter less than 4 mm.
- Currently intravenous gammaglobulin is given routinely on diagnosis (single dose of 2 g/kg i.v. over 10–12hrs); when given early in the disease prevents cardiac involvement and even late administration provides some symptomatic relief.

Barron K S 1991 Kawasaki disease. Epidemiology, late prognosis and therapy. *Rheum. Dis. Clin. N. Amer.* 17(4):907–919

Tizard E J, Suzuki A, Levin M, Dillon M J 1991 Clinical aspects of 100 patients with Kawasaki disease. *Arch. Dis. Child.* 66:185–188

Question 76

This is a chest X-ray of a 12-year-old boy, presenting with a 1 week history of pyrexia and cough and minimal respiratory signs on examination.

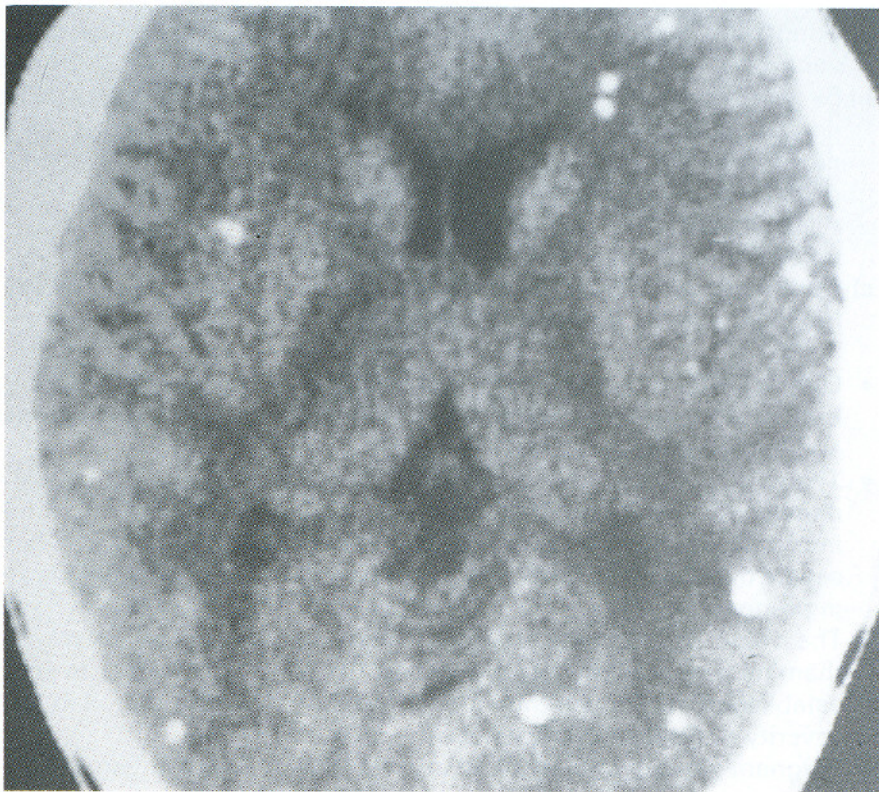
- a) What is the likely diagnosis?
- b) What laboratory tests are indicated?
- c) How would you treat this condition?

Answer to Question 76

- a) *Mycoplasma pneumoniae*.
 - b) Cold agglutinins
Throat/sputum culture
Acute/convalescent serology
Blood cultures for other organisms.
 - c) Full therapeutic dose of erythromycin or tetracyclines (children over the age of 8) for approximately 7 days after defervescence; they both ameliorate the clinical course but do not eradicate the organism.
- *Mycoplasma* infections are common amongst older children and young adults.
 - Only 3–10% of cases present as pneumonia; the rest of them usually present as URTIs, tracheobronchitis, etc.
 - Cold agglutinins, which are specific haemagglutinins reacting to the I antigen of red blood cells, are usually the first antibodies detected; they are present in 33–67% of cases, usually during the second week, disappearing in about 6 weeks. Their presence and their titres correlate well with the severity of the illness.
 - A new DNA probe assay on sputum as a rapid diagnostic testing is now becoming available.
 - White cell count is normal.
 - Complications are rashes, erythema multiforme, meningo-encephalitis, transverse myelitis and Guillain-Barré syndrome. Mild degrees of haemolysis 2–3 weeks after the onset of illness is common; severe haemolysis, thrombocytopenia and coagulation defects are uncommon. Finally myocarditis, pericarditis and rheumatic fever-like syndrome have been reported.
 - Previous infections as demonstrated by the presence of circulating antibodies do not seem to confer lifelong immunity on children.
 - *Mycoplasma pneumoniae* is the most common infectious cause of acute chest syndrome in sickle cell patients.

Kleemola S R, Karjalainen J E, Riaty R K 1990 Rapid diagnosis of *Mycoplasma pneumoniae* infection: clinical evaluation of a commercial probe test. *J. Infect. Dis.* 162:70–75

Nagayama Y, Sakurai N et al 1988 Isolation of *Mycoplasma pneumoniae* from children with lower respiratory tract infections. *J. Infect. Dis.* 157:911–917

Question 77

This is a cranial CT of a neonate who presented with hepatosplenomegaly and convulsions.

- a) What is the likely diagnosis?
- b) How do you confirm the diagnosis?
- c) What single investigation can be an aid to diagnosis and needs to be repeated during early childhood?

Answer to Question 77

- a) Congenital toxoplasmosis (CMV infection calcifications are periventricular).
- b) By specific serology; absence of toxoplasma specific IgM in the neonatal serum does not exclude the diagnosis as 50% of infected newborns do not mount IgM response; persistence of the toxoplasmosis IgG by the age of 1 year confirms the diagnosis. Alternatively whole neonatal blood can be tested for the presence of toxoplasma DNA using a polymerase chain reaction.
- c) Ophthalmoscopy; chorioretinitis develops in the majority of cases (up to 90%) even as late as 20 years.
- Only 10–15% of congenitally infected infants are symptomatic at birth; the 'classic triad' of chorioretinitis, intracranial calcifications and hydrocephalus is seen in only 3%.
 - The risk of vertical transmission in untreated mothers is approximately 25% during the first trimester, increasing to 65% when the infection develops in the last trimester. Conversely the risk for severe disease in the fetus is higher when transmission occurs early in pregnancy.
 - The value of antenatal treatment with spiramycin (plus pyrimethamine and a sulphonamide, when the fetus is infected) is well established; it reduces the vertical transmission rate and also the severity of disease in the newborn.
 - Pregnant women should avoid cat litter, wash fruit and vegetables thoroughly and should only eat well cooked meat to prevent gestational toxoplasmosis.
 - The fetus is not at risk of congenital toxoplasmosis if the mother has seroconverted in the past.

Hall S M 1992 Congenital toxoplasmosis. *Br. Med. J.* 305:291–297
Remington J S, Desmonts G 1990 Toxoplasmosis. In: Remington J S, Klein J Q (eds) *Infectious diseases of the fetus and newborn infant*. W B Saunders, Philadelphia

Question 78



This infant was born at 33 weeks' gestation to a diabetic mother.

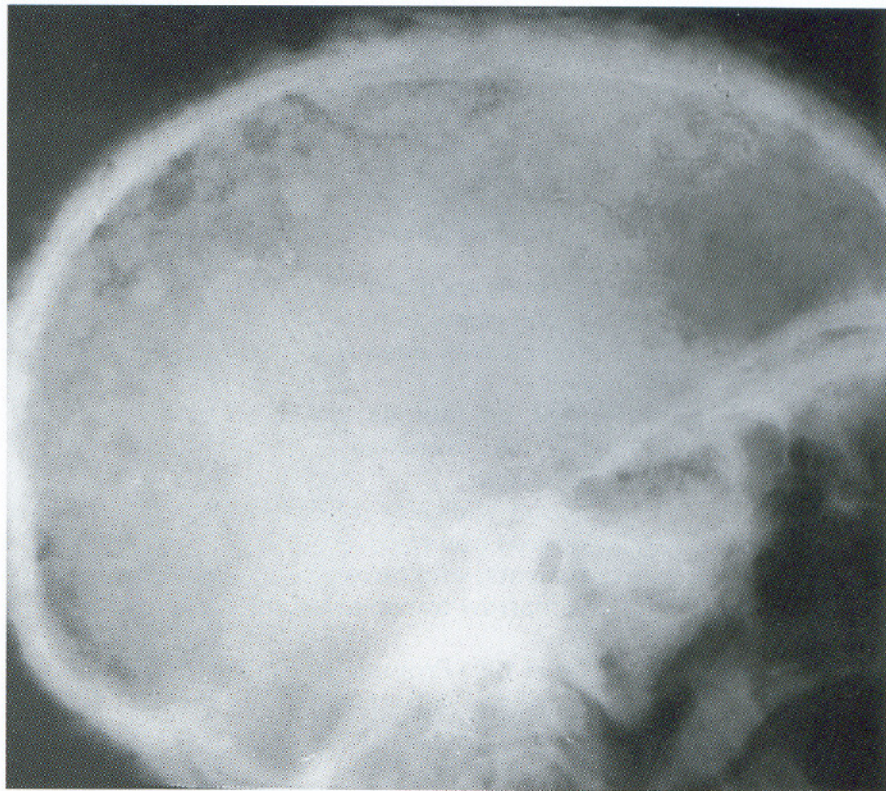
- a) How could you predict the likelihood of respiratory problems prior to delivery?
- b) What levels of blood sugar would you consider satisfactory?
- c) What is the risk of the infant developing diabetes mellitus later on in life?

Answer to Question 78

- a) Surfactant is relatively deficient in phosphatidyl glycerol (PG) and respiratory distress syndrome (RDS) may develop even in the presence of a normal lecithin/sphingomyelin ratio. To assess the risk of RDS either measure amniotic fluid PG levels or 'surfactant profile'.
- b) 2.6 mmol/l Abnormal cerebral physiology is detected when blood glucose levels are below 2.6 mmol/l. Distinction between asymptomatic and symptomatic hypoglycaemia sometimes is difficult, and correction of persistent low blood glucose levels is necessary in 'asymptomatic' infants.
- c) The risk to the infant of developing diabetes mellitus depends on the maternal type of diabetes preconceptionally; if she was an insulin-dependent diabetic, 1:33, or if NIDD, 1:10.
- Meticulous control of the maternal diabetes, ideally preconceptionally, reduces the acute postnatal problems but has not been shown to reduce the risk of congenital malformations.
 - The incidence of macrosomia has been decreased with good antenatal control (up to 80%).
 - Although hyperinsulinism is the main cause of hypoglycaemia, the diminished epinephrine and glucagon responses that occur may be contributing factors.
 - True laboratory glucose measurements are mandatory when in doubt, as the 'reagent strips with the reflectance meter' method approximates by ± 0.5 mmol/l the lab values.
 - Rebound hypoglycaemia due to large boluses of hypertonic dextrose is common. It is due to the abnormal insulin response to a glucose load, seen in these infants. It should be avoided by using continuous infusions rather than boluses, when intravenous treatment is necessary to correct hypoglycaemia.

Aynsley-Green A 1992 Hypoglycaemia. In: David T J (ed) Recent advances in paediatrics, Vol 10, pp 37–62. Churchill Livingstone, Edinburgh

Becerra JE, Khoury MJ, Cordero JF et al 1990 Diabetes mellitus during pregnancy and the risks for specific birth defects: a population based case control study. Paediatrics 85:1–9

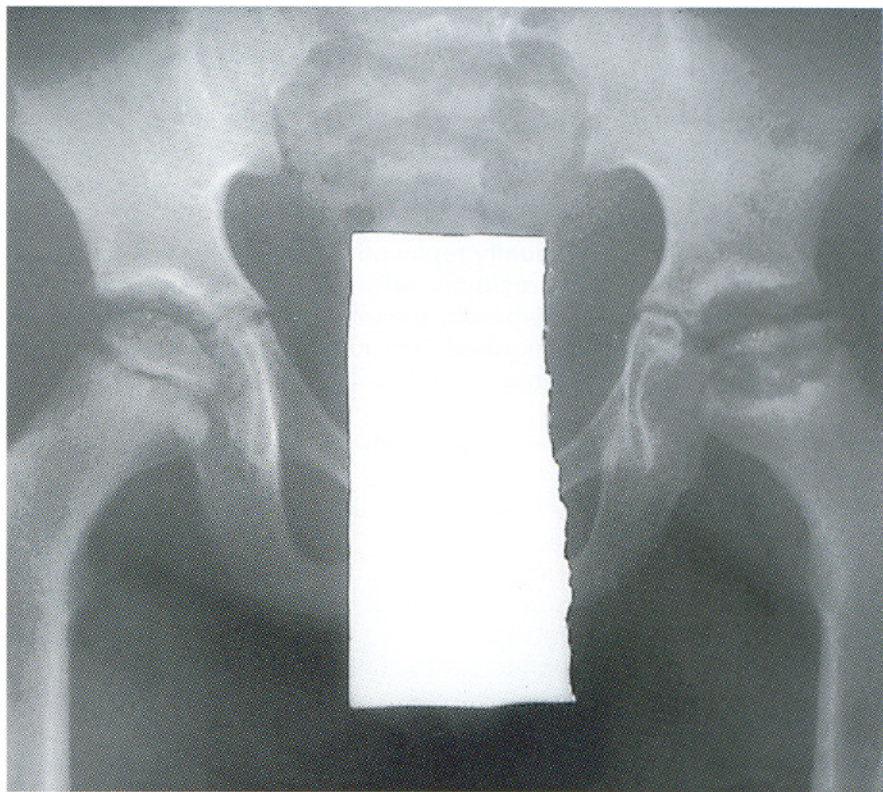
Question 79

- a) What is the diagnosis?
- b) How do you confirm the diagnosis?
- c) What is the appropriate counselling?

Answer to Question 79

- a) B-thalassaemia major (the 'hairbrush' skull appearance caused by vertical trabeculae hypertrophy of erythropoietic tissue).
 - b) Hb electrophoresis; high levels of HbF >90%, no HbA and HbA-2 increased (or normal).
 - c) Cascade counselling: following the diagnosis of a case of B-thalassaemia major, identification of the heterozygote members of the family is essential for further genetic counselling.
-
- Current transfusion regimens are designed to minimize iron loading by maintaining an Hb level closer to normal (>10 g/dl).
 - Haemosiderosis is delayed with adequate iron chelating therapy using desferrioxamine.
 - Splenectomy performed as late as possible, when signs of hypersplenism appear, i.e. transfusion requirements exceed 240 ml/kg of packed cells/year.
 - Serum ferritin is a good indicator of total body iron; levels <1000 ng/ml are considered non-toxic.
 - For a select group of patients, who have a healthy HLA compatible sibling, allogenic bone marrow transplantation is an option.

Ehlers K H, Giardina P J et al 1991 Prolonged survival in patients with beta-thalassaemia major treated with desferrioxamine. *J. Paediatr.* 118:540-549

Question 80

This is a radiograph of a 6-year-old boy with a recent history of a painful left hip and limp.

- a) What is the diagnosis?
- b) What is the differential diagnosis?
- c) What is the treatment?

Answer to Question 80

- a) Perthes disease.
- b) Infection and trauma may jeopardize the blood supply to the capital epiphysis and cause varying degrees of ischaemic change; the history would usually exclude these differential diagnoses. Infiltrative disease such as lymphoma deposits, Gaucher disease and eosinophilic granulomata may lead to obliterative vascular changes and necrosis of the femoral head.
- c) The necrotic bone is gradually replaced by new bone, but recovery can be complicated by residual deformity of the femoral head. Containment in the acetabulum either by femoral osteotomy or bracing is occasionally required. The healing phase during which the head requires protection lasts 2 to 4 years.
- Perthes disease is a juvenile idiopathic avascular necrosis of the femoral head.
 - Males are affected more than females (4–5:1).
 - 20% of cases are familial.
 - 10% of cases are bilateral.
 - Early in the disease the bone scan is more sensitive than radiograms, showing decreased uptake; even more sensitive is magnetic resonance imaging (MRI), which demonstrates necrosis.
 - The prognosis is usually good; half of the severe cases will require some joint replacement procedure at late middle adult life.
 - Unfavourable factors are older age, complete involvement of the head and inadequate treatment.

Jani L, Hefti F 1990 Femur head necrosis in childhood. *Orthopaede* 19(4):191–199

Question 81

Answer to Question 81



- a) What is the differential diagnosis?
b) What investigation would you undertake?

Answer to Question 81

a) Ectopic ureter with a blind terminus

Vaginal cyst

Imperforate hymen with or without congenital hydrometrocolpos (distension of the uterus by retained secretions).

b) Abdominal ultrasound scan to check normality of the urinary tract, before incision and drainage of the cyst is performed.

Question 82

- a) What is the diagnosis?
- b) What particular risk is associated with this condition?
- c) What is the recommended treatment?

Answer to Question 82

- a) Giant congenital pigmented naevi (>20 cm).
b) The risk of malignant melanomatous transformation.
c) Total excision and grafting is the treatment of choice for giant naevi before 3 months of age. Dermabrasion is an alternative treatment.

- Giant (>20 cm) congenital pigmented naevi are very rare; 1:500 000 births.
- Melanocytic naevi, which are common later in childhood, are viewed with more concern when congenital, because they pose an increased risk over a lifetime for development of malignant melanoma. This risk applies for the large naevi of 20 cm or more and is estimated approximately at 10%.
- The lesions may be flat, elevated, verrucous or nodular, various shades of brown, blue or black and may develop numerous coarse hairs or may remain hairless and leathery.
- The histology is usually of an ordinary junctional, compound or intradermal naevus, and may differ in biopsy specimens obtained from several sites.
- In the neonate, all pigmented macules are not congenital naevi; the differential diagnosis includes mongolian spots, cafe-au-lait spots, smooth muscle hamartoma and dermal melanocytosis.
- There is considerable controversy concerning the appropriate approach to medium-sized (1.5–20 cm) congenital naevi. Rough estimates put the risk for melanomas between 0.8–4.9% until 60 years of age. It is now the prevailing opinion that they should be excised before adolescence.
- Early referral to paediatric dermatologists and plastic surgeons is advisable.

Ruiz-Maldonado R, Tamayo L et al 1992 Giant pigmented naevi: clinical, histopathologic and therapeutic considerations. *J. Paediatr.* 120:906–911

Question 83

- a)** What is the diagnosis?
- b)** What is the natural history?

Answer to Question 83

a) Milia.

b) Although they may be very conspicuous in the first days of life, they disappear spontaneously after 3 to 4 weeks.

- These minute sebaceous cysts, which give rise to yellowish-white specks over the nose and face, are very common, found in about 40% of infants.
- Milia may occur at any age but in the neonate are most frequently scattered over the face and on the midline of the palate, when they are called Epstein pearls.
- Milia exfoliate spontaneously in most infants and may be ignored.

Question 84

- a) What is the diagnosis?
- b) What is the treatment?
- c) What is the prognosis?

Answer to Question 84

- a) Right facial nerve palsy.
 - b) Usually no treatment is required, except perhaps the use of artificial tears for the open eye.
Rarely, feeding difficulties might require tube feeding.
 - c) When the facial palsy is a peripheral paralysis, resulting from pressure over the nerve during labour or instrumental delivery, then recovery is usually complete within days or weeks.
- Differential diagnosis includes intra-uterine pressure and congenital defects of the seventh nerve (e.g. nuclear agenesis), which is often bilateral, or other cranial nerves are affected; the prognosis for recovery of function in these cases is poor.
 - Neuroplasty may be indicated when the paralysis is persistent.
 - Facial palsy may be confused with the absence of the depressor muscles of the mouth, which is a benign condition.

Question 85

This 8-year-old boy presented to his family doctor with pyrexia, facial oedema, cervical lymphadenopathy and a mild splenomegaly.

- a) What is the diagnosis?
- b) How do you confirm the diagnosis?
- c) What are the complications?

Answer to Question 85

- a) Infectious mononucleosis (glandular fever).
 - b) Positive Paul–Bunnell test for heterophil antibodies
Epstein–Barr virus (EBV) specific serological studies.
 - c) Auto-immune haemolytic anaemia, thrombocytopenia, neurological complications (i.e. cranial nerve palsies, encephalitis), hepatitis, pericarditis, myocarditis, airway obstruction and splenic rupture.
- Heterophil antibodies are demonstrated in only 60% of children with glandular fever.
 - Certain patients with EBV-induced mononucleosis, particularly pre-adolescents, or those with neurological complications may be heterophil-negative or may lack an atypical lymphocytosis.
 - Neurological complications may be the presenting or sole manifestation of the disease.
 - Airway obstruction from pharyngeal or paratracheal lymphadenopathy presents with acute stridor and is sensitive to steroids.
 - The development of a maculopapular rash following the administration of ampicillin in patients with glandular fever is extremely common (80%); this rash is distinct from that associated with hypersensitivity to penicillins.

Chetham M M, Roberts K B 1991 Infectious mononucleosis in adolescents. *Pediatr. Ann.* 20(4):206–213