CASE 153

History

A 15-year-old male presented having had a fall on his reached hand.

CASE 154

History

An 15-year-old male presented with back pain.
ANSWER 153

Observations (153)
There is relative shortening and ulnar curve of the distal radius with wedging of the carpus between the distal radius and ulna. The features are consistent with Madelung deformity. There are no exostoses to indicate diaphyseal aclasis, and a more specific cause cannot be established from this film.

Diagnosis
Madelung deformity.

Differential diagnosis
Of causes of Madelung deformity (mnemonic – ‘TILDti’):
- Turner’s syndrome.
- Idiopathic.
- Leri–Weil disease (dyschondrosteosis).
- Diaphyseal aclasis.
- Trauma and Infection can lead to pseudo-Madelung deformity.

Discussion
Madelung deformity comprises a short distal radius which has abnormal dorsal and ulnar curvature resulting in ulnar tilt of the distal radial articular surface. The distal ulnar is also subluxed dorsally, and the triangular-shaped carpus is wedged into the reduced carpal angle created at the wrist. There are several conditions associated with Madelung deformity, shown in the differential diagnosis list below. When idiopathic, Madelung deformity is seen mostly in adolescent or young adult women and tends to be bilateral and asymmetrical. Leri–Weil disease is an autosomal dominant condition where sufferers have bilateral Madelung deformity and mesomelic long-bone shortening with limited motion of the elbow and wrist.

Turner’s syndrome is associated with Madelung deformity as well as short stature, webbed neck, horseshoe kidney, aortic coarctation and shortening of the 4th metacarpal.

Trauma and infection may lead to premature fusion of the distal radial growth plate and if this occurs only on the ulnar aspect, there will be resulting ulnar tilt of the distal radius as the radial aspect of the physis allows continued growth on this side. This results in a reduced carpal angle and a pseudo-Madelung deformity.

Practical tips
- Look for metaphyseal exostoses pointing away from the joints indicating diaphyseal aclasis.
- Look for shortening of the 4th metacarpal, which is associated with Turner’s syndrome.

Further management
There is no specific treatment for this condition.

ANSWER 154

Observations (154)
There is uniform collapse of the L4 vertebral body causing the appearance of a vertebral plana. The most likely cause in a child for a solitary collapsed vertebra is cosinophilic granuloma.

Diagnosis
Cosinophilic granuloma causing vertebral plana.

Differential diagnosis
For vertebral plana:
- Idiopathic.
- Infection.
- Neoplastic (metastasis, leukaemia).
- Trauma.
- Steroids.
- Haemangioma.

For platyspondyly:
- Thanatophoric dwarfism.
- Osteogenesis imperfecta.
- Marquiso’s disease.
- Spondyloepiphyseal dysplasia congenital.
- Kniest syndrome.

Discussion
Vertebra plana is the term used to describe uniform collapse of a vertebral body into a thin, flat disk. The most common cause in children is cosinophilic granuloma, with the thoracic vertebrae most frequently affected. The vertebral body is described as having a ‘coin on edge’ appearance. The disc spaces are preserved and can in fact appear slightly widened. Usually there is no kyphotic deformity associated. The posterior elements of the vertebrae are normally spared. Early on, the vertebra will appear lytic and preserved in height before gradually uniform collapse develops. Soft tissue oedema or a paraspinal soft tissue mass are sometimes seen. With healing there is reconstitution of the vertebra to its original height, although some residual compression deformity normally persists. There are many other causes of vertebral plana as outlined in the differential diagnosis. One of these is avascular necrosis, which when idiopathic is termed vertebral osteochondrosis or Calvé–Kummel–Verneuil disease.
Practical tips
- The vertebral body affected has a uniformly flat 'coin on edge' appearance.
- Intervertebral discs are typically spared with normal disc spaces.
- The posterior elements are spared.
- Marked generalized osteopenia suggests osteogenesis imperfecta.
- Malignant causes may affect more than one vertebral body.

Whereas vertebra plana is the term used to describe flattening of a previously normal vertebral body, platyspondyly refers to flattening of the vertebral bodies from birth. Platyspondyly may be generalized, affecting all the vertebral bodies, multiple, affecting some but not all the vertebral bodies, or localized, involving just one vertebral body.

Further management
The underlying cause should be determined if possible. Eosinophilic granuloma is the most common cause in children and usually resolves spontaneously with age.

CASE 155

History
A young adult presented with abdominal pains.
ANSWER 155

Observations (155a)
Three well defined densities in the right upper quadrant are projected outside the contour of the right kidney and are likely to represent gallstones. There is mild, diffuse osteosclerosis and increased trabeculation of the bony skeleton with vertebral endplate infarctions causing H-shaped lumbar vertebrae. Sclerosis and flattening of the left femoral head suggest avascular necrosis. The spleen is noted to be atrophic and calcified. Overall the features are consistent with a diagnosis of sickle cell disease.

Diagnosis
Sickle cell disease.

Discussion
Sickle cell disease is an inherited disorder mostly seen in Afro-Caribbeans. The sickling of the red blood cells leads to increased blood viscosity, occlusion of small vessels and bone infarction leading to necrosis. Chronic haemolytic anaemia also ensues. Marrow hyperplasia leads to coarsening of the trabeculae and may cause diffuse osteosclerosis. A CXR in a 17-year-old female with sickle cell disease is shown (155b). This demonstrates diffuse osteosclerosis with H-shaped thoracic vertebrae (due to endplate infarctions) and cardiomegaly (secondary to chronic anaemia).

Practical tips
Ancillary signs of sickle cell disease on the radiograph depend on the area of the body imaged:
- Abdominal radiograph:
  - H-shaped vertebrae due to endplate infarctions.
  - Avascular necrosis of the femoral heads causing flattening and fragmentation.
  - Gallstones (secondary to haemolytic anaemia).
  - Splenic atrophy and calcification.
  - Renal papillary necrosis may be caused by sickle cell disease due to sloughing of papillae from infarction; a sloughed papilla may be seen within the renal calyx (on an IVU) (155c).
- Chest radiograph:
  - H-shaped vertebrae due to endplate infarctions.
  - Avascular necrosis of the humeral heads.
  - Cardiomegaly (due to chronic anaemia).
- Skull radiograph:
  - "Hair on end" appearance of skull vault due to marrow hyperplasia.
  - Widening of diploic space.

Further management
Treatment is generally supportive with multiple transfusions being necessary. There is a high incidence of infection of bone and lung. Skeletal pain can occur from osteomyelitis but also from bone infarction.

155b CXR in a patient with sickle cell disease demonstrating diffuse osteosclerosis with cardiomegaly and H-shaped thoracic vertebrae.

155c Sloughed papilla.
CASE 156

History
An 80-year-old woman presented with left arm and right leg pain.
ANSWER 156

Observations (156)
A full body bone scan is presented. There is widespread, diffuse increased skeletal uptake with more focal areas of increased uptake located in the skull, spine, ribs, pelvis and limbs. No renal uptake of isotope is seen, however there is some uptake in the bladder. The findings are therefore consistent with a ‘superscan’ and the most likely cause is widespread metastases.

Diagnosis
‘Superscan’ due to widespread metastases.

Differential diagnosis
Of causes of a ‘superscan’:
- Diffuse skeletal metastases.
- Renal osteodystrophy.
- Osteomalacia.
- Hyperparathyroidism.
- Hyperthyroidism.
- Myelofibrosis.
- Leukaemia.
- Aplastic anaemia.
- Widespread Paget’s disease.

Discussion
Diffusely increased activity in the bones on an isotope bone scan can result in an ‘absent kidney sign’ where there is little or no activity in the kidneys but good visualization of the urinary bladder. This is termed a ‘superscan’. The key finding is the absence of activity in the kidneys, which can be easily missed when the diffuse increase in skeletal activity is overlooked. In fact many such scans were reported as normal in the past until the importance of absent renal activity secondary to diffuse skeletal uptake was realized.

The most common cause is skeletal metastases, most of the other causes being metabolic. In this particular case, there are multiple foci of increased uptake on a background of increased activity, however more difficult cases of ‘superscan’ might show homogeneous activity that would be more easily overlooked. The diffuse increase in activity is usually more prominent in the axial skeleton, calvaria, mandible, sternum, costochondral junctions and long bones. The femoral cortices become visible and there is also increased metaphyseal activity.

Radiological signs in ‘superscan’ are:
- Diffuse increased skeletal activity.
- Prominent uptake in axial skeleton, long bones and sternum.
- Absent/little uptake in the kidneys.
- Visualization of bladder.
- Increased bone to soft tissue ratio.
- Increased metaphyseal and periarticular uptake.
- Visible femoral cortices.

Practical tips
- Diffusely increased activity in the bones on a ‘superscan’ is particular prominent in the sternum producing a ‘tie sternum’.
- Metabolic causes tend to cause more diffuse uptake whereas metastatic causes may produce more focal areas of increased uptake.

Further management
When a ‘superscan’ is seen, the most important factor is to determine whether the cause is due to metabolic or malignant disease. This may be evident from the history and clinical examination.
CASE 157

History
An elderly patient presented with leg pains.
# ANSWER 157

## Observations (157a)
There is diffuse, thick periosteal reaction along the metaphyses of the left tibia and fibula. Bandages are also noted around the leg and an elongated soft tissue opacity in the upper medial leg may well indicate a varix. There is no arterial calcification of note. The most likely cause of the periosteal reaction is chronic venous insufficiency, though it is still advisable to obtain a chest radiograph to exclude hypertrophic pulmonary osteoarthropathy from an occult lung tumour.

## Diagnosis
Venous insufficiency.

## Differential diagnosis
Of diffuse bilateral periosteal reaction in adults:
- Hypertrophic pulmonary osteoarthropathy (HPOA).
- Vascular insufficiency.
- Pachydermoperiostosis.
- Thyroid acropathy.
- Fluorosis.

## Discussion
Focal periosteal reaction demands careful assessment for an underlying bone lesion including tumour, infection, fracture, etc. However, diffuse reaction that is bilateral and may well affect multiple bones is a different scenario requiring this brief differential diagnosis:
- HPOA is the most likely cause for such an appearance in adults. The condition presents with painful swelling and clubbing may occur. The periosteal reaction is of variable thickness and typically affects the lower half of the arm and leg. Soft tissue swelling and periarticular osteoporosis may be appreciable radiologically. A chest radiograph should be requested to exclude a lung tumour. Other causes of hypertrophic osteoarthropathy include fibrotic lung disease, suppurative lung disease, liver cirrhosis and inflammatory bowel disease.
- Vascular insufficiency (arterial or venous) is almost always seen in the lower limbs.
- Thyroid acropathy – periosteal reaction typically affects the radial side of the thumb and index fingers. Clubbing of the fingers may be present. An example is shown (157b), although the thumb and index fingers are affected, periosteal reaction is also seen along most of the phalanges and the 5th metacarpal.
- Pachydermoperiostosis is an autosomal dominant condition, typically seen in young black males. It is relatively pain free and self-limiting but causes skin thickening and clubbing. Enlarged paranasal sinuses are also seen. Periosteal reaction most commonly affects the lower half of the arm and leg, though hands can also be affected. Periosteal reaction looks very similar to HPOA but begins around ligament and tendon insertions, i.e. close to the epiphysis.
- Fluorosis – ligamentous calcification present.

## Practical tips
- On identifying diffuse periosteal reaction on radiographs of the limbs always ask for a CXR as this can be the first manifestation of a primary lung tumour.
- Bandages and phleboliths point to chronic venous insufficiency and arterial calcification to arterial disease.
- Though all five causes can affect the hands, remember thyroid acropathy in particular with this distribution.
- Fluorosis is something that is unlikely to be seen outside of exam vivas but remember ligamentous calcification is characteristic!

## Further management
This depends on the cause. HPOA is the most common cause and the most important management aspect is to exclude the presence of an underlying lung tumour.

![157b Hand radiograph of a patient with thyroid acropathy demonstrating periosteal reaction along the diaphyses of most of the phalanges and the 1st and 5th metacarpals.](image-url)
CASE 158

History
A 14-year-old male presented with arm pain.
ANSWER 158

Observations (158)
Within the midshaft of the diaphysis of the humerus there is a relatively well defined lytic lesion that has a narrow zone of transition. There is endosteal scalloping and smooth lamellated periosteal reaction. The lesion has nonaggressive features and in this age group eosinophilic granuloma or infection is the most likely diagnosis.

Diagnosis
Eosinophilic granuloma.

Differential diagnosis
- Osteomyelitis.
- Fibrous dysplasia.
- Leukaemia.
- Lymphoma.

Discussion
Langerhans cell histiocytosis is a spectrum of disease characterized by idiopathic proliferation of histiocytes producing focal or systemic manifestations. Eosinophilic granuloma is the term used to describe the disease when limited to bone and is mostly seen in patients aged between 5 and 30 years. The clinical and radiological features may mimic infection as well as other benign and malignant diseases. The cause and pathogenesis of the condition are unknown. Clinical manifestations relate to the affected bone with local pain, tenderness and masses commonly observed. Patients may have low-grade fever or elevated inflammatory markers, which confuses the clinical picture with infection. The disease may occur in any bone, although there is a predilection for the flat bones with more than half occurring in the skull, mandible, ribs and pelvis. Lesions are solitary in 50–75% of cases. Approximately one-third of lesions occur in the long bones, most commonly the femur followed by the humerus and tibia. Most lesions occur in the diaphysis, and in general the growth plate acts as a barrier to extension.

Radiographic appearances are varied. Lesions typically appear lytic but may have reactive sclerosis. Margins can be well demarcated or poorly defined and they may even have a permeative appearance. A lamellated periosteal reaction is often seen. Invasion of overlying soft tissue may result if the lesion penetrates through the cortex. In the skull, the lesion is often round with a punched out appearance and uneven destruction of the inner and outer skull tables results in a ‘double contour’ or ‘bevelled edge’ appearance. If there is more than one lesion, these may coalesce producing ‘geographical skull’. In the spine, the vertebral body is the most common site of involvement. Lung involvement occurs in 10%. Treatment of the bone lesions consists of conservative therapy or surgical treatment such as curettage or excision.

Practical tips
Always keep this condition in mind when formulating differential diagnoses for lytic bone lesions in the young patient – the appearances are varied and the clinical picture may be confusing.

Further management
The prognosis of eosinophilic granuloma is excellent with spontaneous resolution of bony lesions occurring in 6–18 months.

Further reading
CASE 159

History
A 24-year-old male presents with knee instability following a football injury 2 months previously.
ANSWER 159

Observations (159a)
Sagittal T2 weighted images of the knee demonstrate a rupture of the anterior cruciate ligament, no intact fibres being demonstrated. In keeping with this, there is mild anterior tibial translocation. The posterior cruciate ligament (PCL) is intact.

There is loss of the normal ‘bow tie’ appearance of the lateral meniscus with non-visualization of the body and much of the posterior horn. Furthermore, abnormal low signal tissue is present in the intercondylar region just lateral to the PCL. These findings are indicative of a ‘bucket handle’ tear of the lateral meniscus with a fragment of meniscus displaced medially. A joint effusion is also present.

Diagnosis
Anterior cruciate ligament (ACL) rupture with ‘bucket handle’ tear of the lateral meniscus.

Differential diagnosis
None.

Discussion
The ACL is best evaluated on T1 weighted images and fibres should run parallel to the roof of the intercondylar notch (159b). ACL tears most commonly leave no normal residual fibres visible on MRI. Sometimes, residual fibres of the ACL are seen, but following a more horizontal course than normal.

Sagittal images of normal menisci show a ‘bow tie’ appearance on at least two contiguous slices (159c). This is because the normal meniscus is approximately 9 mm in width and the sagittal images are 3-4 mm in thickness. Thus, at least two sagittal slices should pass through a contiguous section of meniscus.

‘Bucket handle’ tears constitute about 10% of meniscal tears. The vertical tear through the inner edge produces a mobile fragment that flips through approximately 180°, much like a handle flipping from one side of a bucket to the other. In such circumstances, the residual part of meniscus will be reduced in thickness and will not be seen on the usual number of sagittal scans. If the ‘bow tie’ is seen on less than two contiguous sagittal images, a ‘bucket handle’ tear must be excluded. The mobile fragment should then be sought elsewhere in the joint, e.g. medially in the intercondylar region; anterior to the posterior cruciate ligament (PCL) producing the ‘double PCL’ sign (159d); in the anterior joint, in front of the anterior horn of the meniscus.

Other types of meniscal tear include:
- Oblique and horizontal – linear signal change within the meniscus that extends to the inferior or superior surface.
- Radial (also known as ‘parrot beak’ tear) – a vertical tear through the free edge will produce an absent ‘bow tie’ sign similar to a ‘bucket handle’ tear. However, the defect is only small and so the defect in the ‘bow tie’ is much smaller.

As an aside, if the ‘bow tie’ appearance of the meniscus is seen on more than two sagittal images this can be indicative of a discoid meniscus. This is probably a congenital abnormality where the meniscus has a more disc-like shape than the normal ‘C-shape’ due to a wider than normal body. They are more prone to tearing and can be symptomatic even without being torn.

Further management
- A complete ACL tear causes instability that is treated by surgical repair with a prosthetic or tendon graft.
- Meniscal tears often require arthroscopic debridement.
CASE 160

History
A female patient presented with joint pains.
ANSWER 160

Observations (160a)
There is subperiosteal resorption of the radial aspect of the middle phalanges of the index and middle fingers. No evidence of marginal erosions or brown tumours is seen though the trabecular pattern is coarsened. The findings are consistent with hyperparathyroidism.

Diagnosis
Hyperparathyroidism.

Discussion
The uncontrolled production of parathyroid hormone in hyperparathyroidism is primary, secondary or tertiary. Primary hyperparathyroidism is caused by a parathyroid adenoma. The raised parathyroid hormone levels lead to resorption of bone and hypercalcaemia. Secondary hyperparathyroidism is usually a consequence of renal insufficiency, where chronic hypocalcaemia leads to parathyroid hyperplasia. Some patients with secondary hyperparathyroidism then go on to develop the tertiary form, whereby a parathyroid adenoma arises within a chronically overstimulated hyperplastic parathyroid gland.

The cardinal radiological feature is subperiosteal bone resorption. The different sites affected are shown below, the classical location being the radial aspect of the middle phalanx of the index and middle fingers. Figure 160b shows another pattern of erosion – band-like zones in the middle of the terminal tufts. Bone softening may result in wedged vertebrae, kyphoscoliosis and bowing of long bones. Parathyroid hormone-stimulated focal osteoclastic activity can cause brown tumours, which are characteristically expansile, lytic, well demarcated lesions. These can be the solitary sign of hyperparathyroidism in 3% of cases. Figure 160c shows a pathological fracture through a brown tumour.

Radiological features of hyperparathyroidism are as follows:
- Bone resorption:
  • Radial aspect middle phalanx of 2nd and 3rd fingers (160a).
  • Terminal phalangeal tufts (160b).
  • Distal end of clavicles and superior aspect of ribs on CXR.
  • Medial aspect proximal tibia (160c).
  • Medial femoral and humeral necks.
  • Lamina dura of skull and teeth producing ‘floating teeth’.
  • ‘Pepper-pot’ skull due to trabecular resorption (160d).
  • Pseudo-widening of joints, e.g. sacroiliac joints.
  • Marginal erosions of the hands.

- Bone softening:
  • Wedged vertebrae.
  • Kyphoscoliosis.
  • Bowing of long bones.

- Brown tumour (160e).
- Osteosclerosis (more common in secondary hyperparathyroidism).
- ‘Rugger jersey’ spine.
- Soft tissue calcification:
  • Periarticular.
  • Chondrocalcinosis.
  • Arterial.
- Renal calculi.
- Medullary nephrocalcinosis.
Practical tips

- On CXR look for subperiosteal resorption at the superior aspects of the ribs – Figure 160f demonstrates very subtle resorption at the superior aspects of the posterior left 7th and 8th ribs. Also look for erosion of the lateral ends of the clavicles (160g). Hyperparathyroidism is associated with renal failure so there may be a haemodialysis line on the film and prominent soft tissue calcification. Lucent bone lesions may be due to brown tumours.

- On AXR look for a peritoneal dialysis catheter. There may be renal calculi or medullary nephrocalcinosis. A 'rugger jersey' spine may be seen along with widening of the sacroiliac joints due to resorption.

Further management

Primary hyperparathyroidism is treated by surgical resection of the parathyroid gland. After clinical and serological diagnosis of hyperparathyroidism, US and/or sestamibi scintigraphy of the neck is often performed to locate the adenoma pre-operatively (160h). In suspected ectopic parathyroid adenoma, MRI or scintigraphy may be required to locate the tumour.

160d Diffuse bone resorption of the calvaria producing a 'pepper-pot' skull appearance.

160e Radiograph of the humerus in a patient with brown tumours; there are lytic lesions with an associated pathological fracture.

160f CXR in a patient with hyperparathyroidism with subtle erosions of the superior aspects of the left posterior 7th and 8th ribs.

160g AP radiograph of right shoulder demonstrating erosion of the lateral end of the right clavicle.

160h Sestamibi scan with images at 10 min and 90 min post injection show retained tracer in a right parathyroid adenoma.
The approach to paediatric imaging is essentially a composite of the suggested approaches in the other chapters. The approach with paediatric films is as for adult films but the differential diagnosis list will be completely different in many cases. Some additional points when approaching paediatric films are:

- It is very useful to know whether the child has had a premature birth as conditions such as hyaline membrane disease and necrotizing enterocolitis are essentially diseases of the premature neonate. In addition, patterns of disease can vary between infants born prematurely and those born at term. For example, hypoxic/ischaemic brain injury in the premature infant leads to periventricular leukomalacia, a pattern of injury rather different to that otherwise seen.

- Particularly in the child under 2 years, always consider the possibility of ‘non accidental injury’ (NAI) in suspected trauma. However, while it is important to be vigilant for NAI, a false assumption can have severe consequences for the family and must not be declared likely without due consideration. Although certain injuries such as metaphyseal fractures and depressed skull fractures may be highly suspicious for NAI, a multidisciplinary approach should be used with involvement of a specialist paediatric radiologist and appropriate clinical correlation.

- The distribution of categories of disease is very different in children and adults. For example, degenerative disease is largely a feature of adult medicine, and while malignancy is certainly seen in children, it is far less common than in adults. Conversely, congenital disorders are a much bigger consideration in children and there are many such rare conditions that remain outside the realm of the general radiologist’s experience. For example, there are many varieties of skeletal dysplasia causing widespread abnormalities of the skeleton. In the examination viva for general radiological training, it is unlikely that you will be expected to know specific details of the less common varieties. However, it is reasonable to expect you to recognize that a skeletal dysplasia is likely and suggest specialist review by a paediatric radiologist with an interest in such disorders.

- Imaging that utilizes ionizing radiation has an associated risk that must always be balanced against the potential benefit of diagnosis. This is even more important in the child, where the risk of ionizing radiation is greater. As such, while CT imaging is now commonplace in the early work-up of adult illness, greater reliance may be placed on modalities such as ultrasound and MRI in the paediatric population.
CASE 161

History
Stillborn fetus.

CASE 162

History
An 8-year-old child presented with headaches.
**ANSWER 161**

**Observations (161)**
This radiograph is of a stillborn baby as there is no air seen within the lungs. The cut umbilical cord is seen, confirming that this is a newborn. The thoracic cage is narrow and there is squaring of the iliac wings. There is bowing of the long bones with flaring of the metaphyses producing a characteristic 'telephone handle' shape. There is no evidence of fractures to suggest osteogenesis imperfecta. The most likely diagnosis is thanatophoric dysplasia.

**Diagnosis**
Thanatophoric dysplasia.

**Differential diagnosis**
- Of lethal neonatal dysplasia:
  - Osteogenesis imperfecta.
  - Thanatophoric dysplasia.
  - Jeune's syndrome (asphyxiating thoracic dysplasia). The narrow elongated thorax contains a normal size heart but leaves little room for the lungs. There is an 80% mortality rate from respiratory failure.

**Discussion**
Thanatophoric dysplasia, which is also known as thanatophoric dwarfism, is one of the more common causes of lethal neonatal dysplasia. The most common is osteogenesis imperfecta.

**Practical tips**
It may be possible to identify the specific cause of the lethal neonatal dysplasia from certain features on the babygram:
- Osteogenesis imperfecta will cause generalized osteopenia with bowing of long bones and multiple fractures. Multiple wormian bones may also be seen in the skull.
- Thanatophoric dysplasia is associated with a narrow thoracic cage and 'telephone handle' long bones. The iliac wings may be small and squared.
- Narrowing of the thoracic cage and small squared iliac wings are also seen in Jeune's syndrome. The ribs may also be small and horizontal in this condition.

**Further management**
No further management options.

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**ANSWER 162**

**Observations (162)**
There is significant widening of the coronal suture. The skull vault also has a 'copper beaten' appearance. The combination of findings suggests raised intracranial pressure and urgent CT or MRI should be advised.

**Diagnosis**
Suture diastasis due to raised intracranial pressure.

**Differential diagnosis**
Of the causes of suture diastasis follows the mnemonic 'TRIM':
- Traumatic diastasis.
- Raised intracranial pressure:
  - Intracerebral tumour.
  - Hydrocephalus.
  - Subdural collection.
- Infiltration (of the sutures):
  - Leukaemia.
  - Lymphoma.
  - Neuroblastoma.
- Metabolic:
  - Hypoparathyroidism.
  - Rickets.
  - Hypophosphatasia.

**Discussion**
Abnormal widening of the cranial sutures is suggested if there is widening of >10 mm at birth, >3 mm at 2 years and >2 mm at 3 years. The appearance of wide sutures may just be a normal variant but there are several pathological causes, as listed. Suture widening due to elevated intracranial pressure is unlikely after 10 years of age.

**Practical tips**
- Suture diastasis, copper beaten skull appearance and erosion of the dorsum sella are classical plain film signs of raised intracranial pressure.
- A tense fontanelle is a useful clinical sign to confirm raised intracranial pressure.

**Further management**
In most cases, clinical suspicion of elevated intracranial pressure will lead directly to CT or MRI evaluation, but this is clearly the next step should such findings be encountered on plain films with no other explanation.
CASE 163

History
Emergency plain CT of the brain in a 4-year-old child with acute seizures and hypoxia.

CASE 164

History
A newborn presented with breathing difficulties at birth.
**ANSWER 163**

**Observations (163)**
The CT scan image shows diffuse cerebral oedema with effacement of the sulci and gyri. The grey and white matter of the cerebral hemispheres are low in attenuation resulting in loss of the normal corticomediullary differentiation. There is sparing of the cerebellum and brainstem which have an increased density by comparison. The features are typical of the acute ‘reversal sign’ indicating that there has been severe hypoxic-ischaemic brain injury.

**Diagnosis**
Severe hypoxic-ischaemic injury producing the ‘reversal sign’.

**Discussion**
When hypoxia, ischaemia or circulatory arrest occur in children, diffuse hypoxic-ischaemic brain injury can ensue. The cerebral circulation redistributes to the most vital areas, i.e. hindbrain so that the cerebrum is first affected. CT in the first 24 hr may show subtle hypoattenuation of the basal ganglia and insular cortex with effacement of the cisterns around the midbrain. Subsequent CT scans (at 24–72 hr) show diffuse cerebral oedema with effacement of the sulci and cisterns, and decreased grey matter – white matter differentiation.

CT features of the so-called ‘reversal sign’ are hypodensity of the cerebrum with reduced, lost, or even reversed grey matter–white matter differentiation. The thalami, brainstem and cerebellum are relatively spared and retain a more normal density, thus appearing denser than the cerebrum. This finding indicates irreversible brain damage and a universally poor prognosis.

**Further reading**

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**ANSWER 164**

**Observations (164)**
An endotracheal tube is in place in a satisfactory position. The right hemithorax is filled by a large septated lucent mass, which is causing mediastinal shift to the contralateral side. The hemidiaphragm is not visualized. The most likely cause is a congenital diaphragmatic hernia, however cystic adenomatoid malformation should be considered.

**Diagnosis**
Congenital diaphragmatic hernia (CDH).

**Discussion**
Congenital diaphragmatic hernia results from failure of closure of the pleuroperitoneal fold during gestation and presents with respiratory distress soon after birth. It is the most common thoracic fetal anomaly, affecting 1 in 2,500 livebirths. The left side is much more commonly affected than the right. The solid abdominal organs can be herniated as well as bowel. The two main types of hernia are the posterolateral Bochdalek and the anteromedial Morgagni hernias. Survival depends on the size of the hernia, as this determines the degree of pulmonary hypoplasia that will have resulted. There is an association with CNS neural tube defects.

The main differential diagnosis on imaging is cystic adenomatoid malformation (CAM). This congenital cystic abnormality of the lung results from arrest of the normal bronchoalveolar differentiation in utero. The imaging features can be almost identical to those of a diaphragmatic hernia, although the cysts may be fluid filled and thus appear solid.

**Practical tips**
- Bochdalek occurs at the Back of the thorax and accounts for 90% of CDH.
- Morgagni occurs More on the right (heart prevents development on left) and accounts for the other 10%.

**Further management**
Many cases will be expected from antenatal scans and delivery in an appropriate setting can be arranged. Unexpected cases present a medical emergency and may first be suspected when bowel sounds are heard in the chest of an infant with respiratory distress. If suspected, formal airway intubation should be undertaken as soon as possible as ‘bag and mask’ ventilation may further distend the upper GI tract with air. Adequate oxygenation often requires ventilation or perhaps extracorporeal membrane oxygenation (ECMO). Ultimately, surgical correction is required.

Cross-sectional imaging can be helpful in better characterizing the anatomy and differentiating a hernia from CAM.
CASE 165

History
A 29-week premature neonate presented with respiratory distress a few hours after birth.
ANSWER 165

Observations (165a)
An NG tube lies in the stomach and there is an umbilical artery catheter in a satisfactory position with the tip at the level of L4 vertebra. The lungs are small in volume with a fine reticulogranular pattern affecting all lung zones with air bronchograms. In view of the history and radiographic findings, the likely diagnosis is hyaline membrane disease.

Diagnosis
Hyaline membrane disease (HMD).

Discussion
Hyaline membrane disease is one of the most common causes of respiratory distress in newborns. It is most common in premature infants but occasionally also occurs in term infants of diabetic mothers. It is due to lack of surfactant, an agent responsible for decreasing the surface tension in alveoli and produced by the type 2 alveolar cells. Without it the alveoli are poorly distensible and remain collapsed causing respiratory distress shortly after birth. Classically the lungs are small in volume with either 'ground glass' opacity or a fine reticulogranular pattern and air bronchograms extending out to the lung periphery.

Treatment consists of surfactant therapy and positive pressure assisted ventilation. However, the elevated airway pressures may lead to air dissecting through into the interstitium (pulmonary interstitial emphysema – PIE). This can lead to a sudden deterioration in the infant's condition due to the interstitial air causing obstruction to the pulmonary veins. This characteristically appears as elongated bubbles extending to the lung periphery in a bilateral, symmetrical pattern. Pneumomediastinum and pneumothorax are other complications of positive pressure ventilation. Figure 165b demonstrates a left tension pneumothorax in an infant being treated for hyaline membrane disease. Note the shift of the mediastinum to the right and the air bronchograms in the left lung radiating out to the periphery. Figure 165c is an example of an infant with pulmonary interstitial emphysema and a left sided tension pneumothorax displacing the mediastinum to the right. Elongated lucencies due to air tracking along the interstitium and lymphatics are seen, most clearly in the left lung.

Practical tips
- Signs of prematurity on the film are reduced subcutaneous fat and absence of humeral ossification centres.
- Similar lung opacities are seen with neonatal pneumonia or neonatal retained fluid syndrome, however unlike HMD, the lung volumes in these patients will be normal or increased.

165b Chest radiograph of a child with HMD and left pneumothorax secondary to ventilation therapy. Note the left sided air bronchograms.

165c Chest radiograph of a child with HMD who developed pulmonary interstitial emphysema and a left pneumothorax secondary to ventilation therapy. Note the bubbly interstitial emphysema radiating to the lung edge.
- Air bronchograms are a characteristic feature of HMD and are not seen in conditions such as meconium aspiration syndrome.
- Check the position of all lines and tubes on the neonatal film:
  - Umbilical artery catheter (165a) – has characteristic ‘U bend’ as it passes inferiorly from the umbilicus in the umbilical artery then ascends in the internal and common iliac arteries and thus into aorta. The tip should lie either above the renal arteries at T8–12 or below them at L3–4.
  - Umbilical vein catheter – straight course cranially from umbilicus passing in umbilical veins, into ductus venosus and IVC to terminate in the right atrium (165d).

**Further management**
- Treatment involves oxygenation, ventilation and administration of surfactant.
- Complications of HMD treatment should always be sought, namely pulmonary interstitial emphysema, pneumothorax and pneumomediastinum.

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**CASE 166**

**History**
Unconscious 1-month-old born at term.
ANSWER 166

Observations (166a)
This axial contrast enhanced CT image shows bilateral and symmetrical low density in the basal ganglia, involving the putamina and thalamus. There are several possible causes for this appearance. Hypoxia and hypotension should be self-evident. If not present, levels of glucose and carbon monoxide should be checked urgently.

Diagnosis
Low-density basal ganglia secondary to severe hypoxia/ischaemia.

Differential diagnosis
Of low-density basal ganglia:
- Hypoxia
- Hypotension
- Hypoglycaemia
- Carbon monoxide poisoning
- Wilson’s disease

Of basal ganglia calcification (mnemonic ‘PIE MAPS’):
- Physiological – the most common cause and increasingly so with age.
- Infection – cytomegalovirus (CMV), toxoplasma, congenital rubella, HIV.
- Endocrine – hypoparathyroidism (and pseudo/pseudohypoparathyroidism), hyperparathyroidism, hypothyroidism.
- Anoxia – at birth, cerebrovascular accident (CVA).
- Poisoning – carbon monoxide, lead.
- Syndromes – Down’s, Cockayne’s syndrome, neurofibromatosis.

Discussion
Apart from Wilson’s disease, the listed differential diagnoses for this appearance are of acute disorders that ultimately result in reduced cerebral oxygenation or glucose provision. The effect of hypoxia/hypotension on the infant brain depends on whether the infant is term or premature, and whether the insult is mild or severe. In the premature infant up to about 34 weeks, it is the deep white matter that is most vulnerable and hypoxic-ischaemic injury results in periventricular leucomalacia (PVL) with sparing of subcortical white matter and cortex. Since the corticospinal tract fibres pass through this area, there is usually resulting motor impairment.

In the term infant, the pattern of susceptibility is different. A mild insult results in ischaemia of the ‘watershed’ areas of the cerebrum where blood supply is most tenuous. These are the boundaries between the areas supplied by the anterior, middle and posterior cerebral arteries. Vital areas of the brain are protected by redistribution of blood flow. After a severe insult, however, vital areas of the brain can no longer be protected, and the most metabolically active areas at this time of life that are affected. Thus, ischaemic damage occurs in the deep grey matter (thalamus, putamina and brainstem nuclei), the pericallosal gyri and corticospinal tracts. This case illustrates such a pattern.

In older children, severe hypoxic-ischaemic injury may produce a different pattern, that of global cerebral injury. The resulting cerebral oedema and loss of grey-white matter differentiation with sparing of the brainstem and cerebellum can produce the acute reversal sign on CT described elsewhere (see Case 163).

As a comparative aside, high-density basal ganglia due to calcification are illustrated (166b) and the differential diagnosis listed.

Practical tips
- Comparative densities of grey and white matter in the infant brain are variable on CT depending on stage of myelination. Normality of the basal ganglia density can be confirmed by comparison with other grey matter structures.
- MRI is the most sensitive modality for detecting hypoxic-ischaemic injury but may be logistically difficult in the acutely unwell infant. Ultrasound may be practically the easiest imaging option but has reduced sensitivity.

Further management
Urgent correction of hypoxia, hypotension and hypoglycaemia is required followed by exclusion of other causes.

166b Axial CT brain scan in a child showing bilateral basal ganglia calcification.
CASE 167

History
A premature neonate presented with abdominal distension and sepsis.

CASE 168

History
An adolescent male presented with a history of trauma.
ANSWER 167

Observations (167a)
A cord clamp is noted indicating that the neonate is no more than a few days old. The bowel is abnormal with gaseous distension and a bubbly appearance to the bowel wall indicating mural gas. A large pneumoperitoneum is present with most of the gas adjacent to the liver. The features are consistent with necrotizing enterocolitis and perforation. No gas is seen within the portal veins.

Diagnosis
Necrotizing enterocolitis (NEC).

Discussion
Up to 80% of cases are related to prematurity and NEC is the most common gastrointestinal emergency seen in premature babies, usually occurring in the first 2 weeks of life. Ischaemia of the bowel is thought to occur secondary to perinatal stress, hypoxia or infection. Presentation is with diarrhoea or bloody stools. Radiographic features consist of distended thick walled bowel which has a bubbly appearance due to submucosal gas, i.e. pneumatosis intestinalis. Gas may track from the bowel into the portal venous system and can be seen on plain radiography (branching gas extends towards the periphery of the liver unlike air in the biliary tree, which is central). Such finding in adults is an ominous and usually premorbid sign, but in NEC, this is not the case at all. Bowel strictures are a potential long term complication.

Pneumoperitoneum should be carefully looked for, as this necessitates immediate surgery. In the supine position, free gas often collects anteriorly as a large rounded luency in the central abdomen producing the “football” sign.

When perforation occurs in utero, meconium within the peritoneal cavity calcifies and can be seen on plain radiography. An example is shown in a neonate (167b) where there is peritoneal calcification (best seen over the inferior tip of liver) with a pneumoperitoneum causing a positive Rigler sign, i.e. visualization of both walls of the bowel.

Practical tips
- The earliest radiological sign on plain film is bowel dilatation (due to ileus).
- If a contrast enema is required to exclude obstruction then water-soluble contrast should be used – barium is contraindicated.

Further management
Mortality rates are dependent on the degree of prematurity, with rates quoted at 5% in term infants and 12% in premature newborns. Initial treatment is supportive with bowel rest but if serial radiographs or clinical features show progression or perforation then surgical resection of necrotic bowel is required.

167b Abdominal radiograph in a newborn demonstrates visibility of both sides of the bowel wall, i.e. a positive Rigler sign indicating pneumoperitoneum. Flecks of calcification in the right abdomen confirm the diagnosis of antenatal meconium peritonitis.
ANSWER 168

Observations (168)
No bony injury is seen on this skull radiograph. There are multiple wormian bones, which at this age is abnormal. There is a large differential diagnosis but the most likely cause is idiopathic.

Diagnosis
Multiple wormian bones.

Differential diagnosis
Of wormian bones, with common causes underlined (mnemonic ‘PORKCHOPS’):
- Pyknodysostosis.
- Osteogenesis imperfecta.
- Rickets in healing.
- Kinky hair syndrome (Menkes).
- Cleidocranial dysostosis.
- Hypothyroidism/hypophosphatasia.
- Otopalatodigital syndrome.
- Pachydermoperiostosis.
- Syndrome of Down’s.
- Idiopathic – normal in first year of life.

Discussion
Wormian bones are essentially small bones occurring in the sutures of the calvaria. These intrasutural ossicles are usually found in the lambdoid, posterior sagittal and temporosquamosal sutures. They are considered abnormal when seen after 1 year or large and numerous (>10 in number and larger than 6 x 4 mm).

Practical tips
- Because of the wide differential diagnosis, it is difficult to identify a specific cause without the aid of a good clinical history.
- Diffuse osteopenia will be present on the skull radiograph in cases of rickets and osteogenesis imperfecta.
- Pyknodysostosis, on the other hand, will be associated with diffuse osteosclerosis on the film.

Further management
Management is dependent on the underlying cause.

Further reading

CASE 169

History
A child with a history of partial seizures.
Observations (169a)
Axial T2 weighted MR image at the level of the lateral ventricles. This demonstrates a large cleft extending through the full thickness of the left cerebral hemisphere from the surface of the brain to the left lateral ventricle. The cleft is lined by grey matter and is filled with CSF. The findings are consistent with schizencephaly.

Diagnosis
Schizencephaly.

Discussion
During gestation, neurones migrate outwards from the periventricular germinal matrix to form the normal cerebral cortices. This migration can be interfered with by several causes including chromosomal abnormalities, but mostly the reason is unknown. The result is brain tissue lying in the wrong place, typically grey matter.

Schizencephaly is a cleft extending through the full thickness of cerebral hemisphere from the ependyma-lined wall of ventricle to the brain surface. It is lined by pia and grey matter that usually shows polymicrogyria, and is often located around the Sylvian fissure. The lateral end of the cleft may be open and readily apparent (open lip type) but can sometimes be closely opposed and easy to miss (closed lip type). However, even the closed lip type will show a small irregularity in the wall of the ventricle at the site of the cleft. It is not certain whether this condition is due to an ischaemic insult leading to germinal matrix infarction or whether it represents a local cortical dysplasia.

There are different manifestations of the congenital neuronal migration anomalies which result in varying degrees of mental retardation and/or seizures. The following further patterns are recognized:

- **Heterotopic grey matter** - when small collections of the neurones arrest on their way to the cortex they can be seen as discrete nodules (most commonly in a subependymal location) or as a subcortical band. Thus there are nodular and band heterotopias. These are isointense to grey matter and show no enhancement. While sometimes asymptomatic, seizures and developmental delay can ensue, especially with band heterotopia. An example of nodular heterotopic grey matter is shown in an axial T2 MR image of the brain (169b), where a small area of heterotopic grey matter is seen in a subependymal location adjacent to the occipital horn of the left lateral ventricle.

- **Polymicrogyria** - sometimes neurones may migrate to the cortex but are abnormally distributed, producing a bumpy appearance to the cortical gyri termed polymicrogyria. An example is shown in a coronal T1 weighted MRI (169c), where polymicrogyria affecting the right temporal lobe produces a bumpy
to the gyri. Note how the normal left temporal lobe gyri are distinct and crisp.

- Pachygyria – in some cases the gyri may be poorly formed (pachygyria). A spectrum exists whereby in the most severe form the surface of the brain appears smooth. This is termed lissencephaly \(169d\). Often these patients have severe mental retardation and limited survival.

**Practical tips**
Subependymal nodules in tuberous sclerosis can also cause nodularity along the walls of the ventricles. However, the nodules in this condition show a similar signal to white matter rather than the grey matter seen in heterotopia.

**Further management**
Medical management of epilepsy.

**CASE 170**

**History**
A newborn who was born at 41 weeks presented with hypoxia.
ANSWER 170

Observations (170a)
An endotracheal tube has been placed just above the carina. There is also an NG tube passing into the stomach. There are bilateral diffuse patchy opacities in both lungs indicative of widespread atelectasis and patchy consolidation. However, the lungs appear hyperinflated and there are small pleural effusions. In view of the history, the appearances are likely to be due to meconium aspiration syndrome.

Diagnosis
Meconium aspiration syndrome.

Discussion
Meconium aspiration syndrome is the most common cause of respiratory distress in newborns born at full or post term. The large size of the fetus makes delivery difficult. Perinatal hypoxia and fetal distress lead to meconium defecation in utero. Aspiration of the meconium into the tracheobronchial tree then causes obstruction of small peripheral bronchioles (though only a minority of fetuses exposed to meconium stained amniotic fluid develop respiratory symptoms). This results in unevenly distributed areas of subsegmental atelectasis with alternating areas of air trapping. The chest radiograph usually begins clearing within a few days with no long term radiographic sequelae in the lungs. The radiological features on CXR are:
- Bilateral patchy atelectasis and consolidation.
- No air bronchograms.
- Hyperinflation with areas of air trapping.
- Small pleural effusions.
- Spontaneous pneumothorax and pneumomediastinum may result from the air trapping (170b).

Practical tips
- Most common cause of respiratory distress in term babies: meconium aspiration.
- Most common cause of respiratory distress in preterm babies: hyaline membrane disease.

Further management
Almost all neonates with meconium aspiration syndrome make a full recovery of their pulmonary function. Upper airway suction may be employed and ventilatory support may be required in more severe cases of respiratory distress.

170b CXR in a neonate with a large pneumomediastinum. Air outlines the thymus producing an ‘angel’s wings’ appearance.
CASE 171

History
A newborn presented with regurgitation of feeds.

CASE 172

History
Micturating cystourethrogram (MCUG) was taken in a male infant with a previously confirmed urinary tract infection.
ANSWER 171

Observations (171a)
A feeding tube is seen within a gas-distended blind-ending pouch representing the oesophagus. Air is noted below the diaphragm. There is no convincing evidence of aspiration pneumonia. The features are consistent with oesophageal atresia with a distal tracheoesophageal fistula.

Diagnosis
Congenital oesophageal atresia with tracheoesophageal fistula (TOF).

Discussion
Embryologically the primitive foregut tube separates to form the trachea and oesophagus. Disorders of this separation presenting in infancy result in various combinations of oesophageal atresia and TOF. Presentation is with excessive drooling, regurgitation of feeds or symptoms of aspiration depending on the type of abnormality present.

In 90% of cases there is a component of oesophageal atresia, and the majority of these have an associated tracheoesophageal fistula. Such a fistula can be proximal, distal or both (i.e. between the trachea and the proximal oesophageal segment, the distal segment or both). This case demonstrates the most common subtype (seen in around 80%) where there is oesophageal atresia and a distal TOF. The atresia results in drooling and regurgitation while the distal TOF results in passage of air from trachea into stomach and thus the rest of the bowel.

A minority of cases have TOF without oesophageal atresia and are more likely to present with coughing or choking during feeds and ultimately aspiration pneumonia. An example is shown (171b) where water-soluble contrast has been injected via an NG tube in the oesophagus and is seen to pass into the trachea via the fistula.

Practical tips
- Oesophageal atresia - CXR shows a retrotracheal distended pouch of proximal oesophagus and a feeding tube may be coiled within it after attempted passage.
- A gasless abdomen indicates no fistula or a proximal fistula.
- Gas in the abdomen indicates presence of a distal fistula.
- Look for consolidation suggesting associated aspiration pneumonia.
- Oesophageal atresia and TOF can be part of a VACTERL syndrome so check the CXR for abnormalities:
  - Vertebral anomalies.
  - Anorectal anomalies.
  - Cardiovascular anomalies.
  - Tracheo-oesophageal fistula.
  - Renal anomalies.
  - Limb anomalies.

Further management
Surgical repair is required. This can be later complicated by anastomotic leak, oesophageal stricture or abnormal motility resulting in dysphagia and/or aspiration pneumonia.

171b Contrast examination via an NG tube demonstrates a tracheoesophageal fistula.
ANSWER 172

Observations (172)
This MCUG study shows a transverse filling defect at the posterior urethra with distension of the proximal posterior urethra. The findings are consistent with posterior urethral valves.

Diagnosis
Posterior urethral valves.

Discussion
Congenital presence of thick folds of mucous membrane in the posterior urethra is the most common cause of urinary tract obstruction in boys. The condition is often suspected on prenatal US where it can lead to oligohydramnios, hydronephrosis, prune belly and urine ascites or urinoma due to leak. If obstruction occurs early in utero then multicystic dysplastic kidney may result. After birth, MCUG is the investigation of choice to outline the transverse filling defect caused by the thick mucosal folds. Distension and elongation of the proximal part of the posterior urethra may be seen during voiding and vesicoureteral reflux is present in 50%. Bladder trabeculation and a significant post void residual volume may be noted. Prognosis depends on the duration of obstruction prior to corrective surgery and is worse if associated with vesicoureteral reflux. Approximately three-quarters of cases will have been discovered in the first year of life, though occasionally it can be first noted in adulthood.

Practical tips
Note how diagnosis is still possible when the catheter is in situ during the voiding phase of the MCUG (172).

Further management
Urological surgical intervention is required with initial treatment aimed at relieving bladder outlet obstruction and ablating the valves. Secondary treatment may be required for vesicoureteral reflux, urinary tract infections, urinary incontinence and renal dysfunction.

CASE 173

History
A newborn presented with abdominal distension and failure to pass meconium.
ANSWER 173

Observations (173a)
An NG tube is in the stomach. There are multiple dilated loops of bowel in the abdomen. No fluid levels are seen within the bowel suggesting that the appearances may be due to meconium ileus. However, other pathologies such as Hirschsprung’s disease and imperforate anus should be considered.

Diagnosis
Meconium ileus.

Differential diagnosis
- Hirschsprung’s disease.
- Imperforate anus.
- Ileal atresia.
- Inguinal hernia.

Discussion
Meconium ileus is the term used to describe small bowel obstruction in neonates secondary to inspissated meconium pellets impacted in the distal ileum. The vast majority prove to have cystic fibrosis and this is the earliest manifestation of the disease. The diagnosis is confirmed by performing a contrast enema, which demonstrates multiple round filling defects (the inspissated meconium) in the distal ileum and proximal colon (173b). The colon may be very narrow on the contrast study if it has been unused due to antenatal obstruction, whereby it is termed a microcolon. The enema should be performed using Gastrografin as this has a therapeutic effect, helping to clear the meconium by drawing water into the gut.

With Hirschsprung’s disease, the contrast enema will demonstrate dilated bowel with a transition zone to a distal aganglionic segment.

Practical tips
- On an AXR of a baby it is almost impossible to tell if dilated loops of bowel are large or small bowel. The presence or absence of vomiting/passage of meconium are more helpful to know with regard to assessing if there is high or low bowel obstruction.
- The hernial orifices should be checked for air suggesting an inguinal hernia.
- A ‘soap bubble’ appearance may be seen on AXR in meconium ileus due to the mixture of gas with meconium.
- Fluid levels are not usually present in meconium ileus because the bowel contents are very viscous.
- Look at the sacrum on the AXR, as imperforate anus is associated with sacral agenesis.

Further management
- Water-soluble contrast enema can be useful for both diagnosis and treatment.
- All patients with meconium ileus should have a ‘sweat test’ to exclude underlying cystic fibrosis.

173b Gastrografin enema in a newborn shows multiple filling defects in the ascending colon and terminal ileum, which represent inspissated meconium. Note the dilated small bowel loops.
CASE 174

History
A 4-year-old child presented with fever and abdominal pain.

CASE 175

History
A 4-month-old child presented with persistent irritability.
ANSWER 174

Observations (174a)
Axial CT of the abdomen with oral contrast and IV contrast in portal phase. There is a large slightly heterogeneous mass arising from the right kidney. The mass does not enhance as much as the renal parenchyma. There is local mass effect with displacement, but no invasion of the right lobe of liver or encasement of vessels. A small mass of similar density is seen near the hilum of the left kidney. The appearances suggest bilateral Wilms’ tumours.

Diagnosis
Bilateral Wilms’ tumours.

Differential diagnosis
Neuroblastoma.

Discussion
Wilms’ tumour (nephroblastoma) is the most common abdominal malignancy in young children, most commonly presenting at age 3–4 years. The most frequent presentation is with abdominal mass, though hypertension, pain, fever and haematuria also occur. The tumour usually grows to a large size, often measuring over 10 cm. Radiological features include:

- Exophytic mass displacing rather than encasing adjacent structures.
- Less contrast enhancement than normal renal parenchyma.
- Cystic/necrotic areas give it a heterogeneous appearance.
- Invasion of the renal vein and inferior vena cava may occur in up to 10%.

Tumours are bilateral in 10% and this indicates background nephroblastomatosis, a state of persistent nephrogenic blastema that is a precursor to Wilms’. Wilms’ tumour is associated with the Beckwith–Wiedemann syndrome (macroglossia, visceromegaly and omphalocele). Other associations include aniridia and hemihypertrophy.

The main differential diagnosis is neuroblastoma, a common malignant tumour of the neural crest that presents in a similar way to Wilms’ tumour with a painful abdominal mass and fever. Typical age of presentation is slightly earlier however (under 2 years). Hormone secretion from the tumour (such as catecholamines) may cause other signs, including hypertension and opsoclonus (chaotic jerky eye movements). It can arise anywhere in the sympathetic neural chain including the adrenal gland and the abdominal sympathetic chain.

Practical tips
- Always check for bilateral tumours when Wilms’ is suspected.
- Differentiating between neuroblastoma and Wilms’ tumour can be difficult radiologically but there are some specific features that can help:
  - Almost all neuroblastomas contain calcification whereas only up to 10% of Wilms’ tumours calcify.
  - Both tumours can cross the midline and look similar on imaging, appearing inseparable from the kidney; however, neuroblastoma tends to encase surrounding vessels such as the aorta while Wilms’ tumours tend to displace surrounding tissues and structures. An example of a neuroblastoma is shown (174b) encasing the aorta and coeliac axis vessels. Neuroblastoma may also extend into the spinal canal through the neural foramina.
  - Look for evidence of metastatic spread; 70% of neuroblastomas have malignant spread at presentation compared to just 10% of Wilms’ tumours. Also, Wilms’ tumours spread to lung, whereas neuroblastoma spreads to bone.

Further management
Treatment is with radical nephrectomy and chemotherapy. Preoperative chemotherapy is advocated in cases of bilateral Wilms’ tumours and when there is IVC extension of tumour (occurs in ~5%).
ANSWER 175

Observations (175)
There is bilateral symmetrical thick, smooth periosteal reaction affecting the diaphyses of the long bones. There is no fraying or splaying of the metaphyses to suggest rickets. The most likely diagnosis at this age is Caffey's disease, however other possibilities such as leukaemia need to be considered.

Diagnosis
Caffey's disease.

Differential diagnosis
Of bilateral diffuse periosteal reaction in childhood:
- Normal variant before the age of 4 months.
- Caffey's disease.
- Leukaemia.
- Scurvy.
- Rickets.
- Hypervitaminosis A.
- Non accidental injury (NAI).

Discussion
Infantile cortical hyperostosis (Caffey's disease) is a proliferative bone disease seen in patients under the age of 6 months. Irritability and fever are the presenting symptoms and are associated with soft tissue swelling over the bones. Bilateral symmetrical thick periosteal reaction is the cardinal radiological feature and most commonly affects the mandible, clavicle and the long bones. It usually involves the diaphysis of the bone. In the majority of cases, there is spontaneous complete recovery by the age of 3 years.

Practical tips
- If there is diffuse periosteal reaction with fractures of differing ages, NAI must be considered.
- With rickets, splaying and fraying of the metaphyses will be seen.

Further management
When NAI is considered then a careful analysis of previous radiographs, the clinical presentation and consultation with a specialist paediatric radiologist must be carried out because of the repercussions of a misdiagnosis.

CASE 176

History
None available.
ANSWER 176

Observations (176a)
There is widespread bilateral decreased bone density with healing insufficiency fractures of the radius and ulna bilaterally. These are associated with thick smooth periosteal reaction and there is fraying and splaying of the metaphyses. The features are characteristic of rickets.

Diagnosis
Rickets.

Differential diagnosis
Hypophosphatasia.

Discussion
Rickets is most commonly due to insufficient biologically active vitamin D, though impaired calcium absorption or excessive phosphate excretion can occasionally be to blame. In the western world, pure dietary deficiency of vitamin D is rarely the sole cause; more often it is due to malabsorption or impaired vitamin D metabolism in the liver or kidney.

Rickets is essentially osteomalacia during enchondral bone growth. Portions of the skeleton that have already matured show features of osteomalacia, but loss of normal maturation and mineralization of cartilage cells at the growth plate lead to the additional distinctive radiological features of rickets. Osteomalacia is discussed elsewhere in the book but the radiological features are due to excessive unmineralized osteoid producing Looser's zones, osteopenia, cortical tunnelling, indistinct trabeculae and finally bowing and fractures due to softened bones.

In addition, the following features are seen in rickets:
- Widened growth plate - loss of normal chondrocyte maturation and mineralization result in cell build up here.
- Metaphyses are irregular/frayed, splayed and cupped – impaired mineralization causes the frayed irregular appearance while build up of chondrocytes at the physiinds the metaphysis producing cupping and splaying.
- Epiphysis osteopenic and irregular.
- Periarticular soft tissue swelling.
- Apparent periosteal reaction due to subperiosteal unmineralized osteoid.
- Delayed maturation and growth.

Figure 176b demonstrates rickets of the lower limbs – note the typical changes around the metaphyses in the tibia and also bowing of the fibula. Figure 176c is a CXR of a child with rickets showing splaying of the anterior ends of the ribs.

176b AP radiograph of both legs shows typical features of rickets with fraying of the metaphyses.

176c Chest radiograph of a child with rickets and splaying of the ribs.
the ribs and the metaphysis of the right humerus. The appearance of the anterior rib ends is due to changes at the costochondral junction growth plates and is termed the "rachitic rosary".

**Practical tips**
- The earliest sign of rickets on the plain film is a widening of the growth plate.
- Looser's zones are rare in rickets compared to osteomalacia in the fused skeleton.

**Further management**
Rickets is now usually identified early and treated with vitamin D supplements. Significant pelvis deformity and gait disturbances are now rarely seen in the developed world.

**CASE 177**

**History**
None available.
ANSWER 177

Observations (177a)
There is partial fusion of an extra digit with the metacarpal of the little finger. This essentially represents polysyndactyly, the possible causes of which include idiopathic, Ellis–van Creveld syndrome and Carpenter syndrome.

Discussion
There are several causes of syndactyly (fusion of digits) and polydactyly (supernumerary digits), which are both congenital abnormalities. Ellis–van Creveld syndrome is also associated with carpal fusion, as is Apert’s syndrome. This is characterized by features in the skull: notably craniosynostosis of the coronal sutures, hypoplastic midface and enlargement of the sella. All of these features are demonstrated in the lateral skull radiograph in a child with Apert’s syndrome (177b).

Diagnosis
Ellis–van Creveld syndrome.

Differential diagnosis
Of causes of polydactyly:
- Idiopathic.
- Ellis–van Creveld syndrome.
- Carpenter syndrome.
- Polysyndactyly syndrome.

Of causes of syndactyly:
- Idiopathic.
- Apert’s syndrome.
- Carpenter syndrome.
- Down’s syndrome.
- Poland’s syndrome.
- Neurofibromatosis.

Practical tips
Some exam cases will have an obvious abnormality as part of a syndrome that you don’t know – stating that you would seek help from a textbook or specialist colleague is a reasonable answer. You can’t know everything!

Further management
Poly/syndactyly will be part of a syndrome with multiple abnormalities.

177b Lateral skull radiograph of a child with Apert’s syndrome demonstrating craniosynostosis of the coronal sutures, hypoplasia of the midface and enlargement of the sella.
CASE 178

History
None available.
ANSWER 178

Observations (178a, 178b)
There is bowing deformity of the tibia, fibula and humerus. The metaphyses of the bones are widened producing an Erlenmeyer flask deformity. The metaphyses are also relatively lucent when compared with the diaphysis, which is sclerotic. There are no specific features to indicate lead poisoning, osteopetrosis or thalassaemia, so the differential diagnosis lies between Pyle’s disease and lipidoses such as Gaucher’s or Niemann–Pick disease.

Diagnosis
Pyle’s disease.

Differential diagnosis
In this case:
- Craniometaphyseal dysplasia.
- Niemann–Pick disease.
- Gaucher’s disease.

Of Erlenmeyer flask deformity (mnemonic – ‘Lead GNOME’):
- Lead.
- Gaucher’s.
- Niemann–Pick disease – looks like Gaucher’s but without avascular necrosis.
- Osteopetrosis.
- Metaphyseal dysplasia (Pyle’s) and craniometaphyseal dysplasia (same as Pyle’s disease but there is a history of cranial nerve palsies).
- ‘E’matological!! – thalassaemia.

Discussion
Pyle’s disease is also known as metaphyseal dysplasia. It is a rare autosomal recessive disorder characterized by flaring of the ends of long bones with relative constriction and sclerosis of the central portion of the shafts. Affected patients are usually asymptomatic and genu valgus deformity is often a feature. The expanded metaphyses tend to be lucent and have the appearance of an Erlenmeyer flask (named after the wide necked laboratory flask bearing the name of this German chemist).

Craniometaphyseal dysplasia essentially has the same features but in addition there are cranial nerve palsies due to sclerosis of the skull base.

Gaucher’s disease is a hereditary disorder of lipid storage common among Ashkenazi Jews. It is characterized by hepatosplenomegaly with flask-shaped long bones and generalized osteopenia with strikingly thin cortices. Avascular necrosis is also a feature.

Practical tips
Erlenmeyer flask deformity, the metaphyseal expansion of long bones, is also discussed in Chapter 5. Additional differentiating features can be found on the radiograph as to the specific underlying cause of Erlenmeyer flask deformity:
- Diffuse sclerosis and sclerotic vertebral endplates producing ‘sandwich vertebrae’ indicate osteopetrosis.
- With Pyle’s disease, there will be relative sclerosis at the diaphysis and lucency of the metaphysis.
- Gaucher’s disease will also be associated with lucency and osteopenia but there may be signs of avascular necrosis of the femoral or humeral heads (loss of height and fragmentation) and on an AXR massive hepatosplenomegaly may be seen.
- Thalassaemia is associated with coarsened trabeculation producing a ‘cobweb’ appearance.
- Lead poisoning causes dense metaphyseal bands as well as Erlenmeyer flask deformity.

Further management
This condition is usually asymptomatic and requires no direct management.

178a Metaphyseal widening with increased lucency.
CASE 179

History
None available.
ANSWER 179

Observations (179a, 179b)
The lateral skull radiograph (179b) demonstrates thinning of the calvaria with multiple wormian bones. Bowing deformities are seen to affect the limbs (179a) and there are several fractures of differing ages, mostly seen at the metaphyses of the long bones. There is generalized osteopenia of the skeleton with marked thinning of the cortices. The features are consistent with osteogenesis imperfecta.

Diagnosis
Osteogenesis imperfecta.

Differential diagnosis
Of wormian bones with common causes underlined (mnemonic – ‘PORKCHOPS’):
• Pyknody sostosis.
• Osteogenesis imperfecta.
• Rickets in healing.
• Kinky hair syndrome (Menkes).
• Cleidocranial dysostosis.
• Hypothyroidism/hypphosphatasia.
• Otopalatodigital syndrome.
• Pachydermoperiostosis.
• Syndrome of Down.
• Idiopathic – normal in first year of life.

Discussion
Osteogenesis imperfecta is a connective tissue disorder characterized by fragile bones and blue sclerae. Type 1 is compatible with life. Type 2 is the lethal form associated with perinatal death.

The principal radiological features include:
• Diffuse osteopenia with thinning of cortices.
• Multiple fractures of differing ages with pseudarthroses and bowing deformity.
• Fractures are associated with exuberant callus formation.
• Biconcave vertebral bodies.
• Multiple wormian bones in the skull.
• Poor dentition.

Practical tips
Multiple fractures in children should raise suspicion of non accidental injury (NAI) and sometimes differentiating this from osteogenesis imperfecta can be difficult. Predominantly osteogenesis fractures are diaphyseal compared with metaphyseal NAI fractures but this is not always the case.

Further management
Early medical intervention to increase bone mineral density and surgical intervention to treat/correct scoliosis and treat fractures mean that a multidisciplinary approach to the ongoing treatment is required.