Congenital renal anomaly
in a patient with situs inversus

Q1: Name the congenital renal anomaly identified in fig 1 (see p 355)
Horseshoe kidney.

Q2: What is the incidence and sex ratio of this anomaly?
Incidence is one in 400. It is more commonly found in males at a ratio of 2:1.

Q3: What complications occur with this condition?
Thirty percent of cases are asymptomatic and are therefore identified incidentally. Stasis of urine due to the malrotation of the kidneys, and impaired ureteric drainage result in infection and stone formation.

Q4: Name three other genitourinary anomalies that can be associated with this condition
These are: (1) hypospadias; (2) undescended testis; (3) ureteral duplication.

Discussion
Horseshoe kidney was first recognised during a necropsy by DeCarpit in 1531, but Botalli in 1564 provided the first description and illustration of this condition. The confirmation of horseshoe kidney with an aortic aneurysm presents a diagnostic and therapeutic challenge to the vascular surgeon.

Final diagnosis
Horseshoe kidney with situs inversus.

References

A man with numbness and limb weakness

Q1: What is the diagnosis?
The diagnosis is spinal (cervical) intramedullary cysticercosis. The MRI scan (fig 1; see p 355) shows a cyst located in the intramedullary region. Cervical laminectomy with removal of the cyst was done. Histopathology examination (fig 2; see p 355) proved the lesion to be a cystercus cyst with scolex (larval cyst).

Q2: What are the treatment options?
The treatment of spinal intramedullary cysticercosis could be surgical, medical (that is, using anti-epileptic drugs), or both, based on location and stage of the cyst as also on the experience of the physician. Surgical treatment includes laminectomy with removal of the cyst. Cysticidal drugs given are albendazole in a dose of 15 mg/kg body weight for 14–30 days or praziquantel 50 mg/kg body weight for 15 days along with steroids to reduce the perilesional oedema and to prevent neurological deterioration during the course of cysticidal drugs. Administration of cysticidal drugs before or after surgery is a point of personal preference for the individual doctor as no systematic evaluation of treatment has been possible due to the rarity of the disease.

Discussion
Intramedullary cysticercosis is a rare manifestation of neurocysticercosis, and fewer than 50 cases have been reported. The cysts are commonly located in spinal subarachnoid space and rarely at intramedullary locations. The majority of reported cases have cysts in the dorsal cord, which is in accordance with the regional blood flow to the spinal cord. The treatment modalities like drug therapy (cysticidal drug)/surgery, or both, can be planned according to the pathological stage and location of the cyst as seen on MRI. Since the cysticidal drugs albendazole and praziquantel were shown to be effective in the parenchymal brain cysticercosis, these drugs have been considered potentially useful in patients with intramedullary cysticercosis.

An interesting case of hemiparesis

Q1: What is the differential diagnosis in this patient?
The differential diagnosis of HIV patients presenting with focal neurological deficits should include disorders such as toxoplasmosis, primary central nervous system lymphoma, cerebral Chagas’ disease, progressive multifocal leukoencephalopathy (PML), central nervous system tuberculosis, and cryptococcosis.

Q2: What are the computed tomography and MRI findings?
Computed tomography of the head (fig 1; see p 356) shows well defined hypodense areas in the and lentiform regions and in the right temporo-parieto-frontal region without areas of enhancement or mass effect. Figure 2A (see p 356) shows the gadolinium enhanced T1 sagittal images of the brain showing non-enhancing white matter changes. Figure 2B (see p 356) shows the T2 weighted coronal MRI showing white matter changes without mass effect. The financial cost of non-enhancing white matter lesions with typical increased T2 and decreased T1 signals on MRI head are highly suggestive of PML.
to make the clinical diagnosis of PML. Recently polymerase chain reaction amplification of JC virus DNA from the cerebrospinal fluid has become the favoured diagnostic modality to confirm the diagnosis. Currently, outlook for patients with PML is poor and there is no effective treatment. Few authors have reported promising results with HAART treatment. The current treatment with cidofovir has been tried in such patients with varying results. Mean survival time after diagnosis is about six months.

Final diagnosis
Progressive multifocal leukoencephalopathy in AIDS.

References

A misdiagnosed potentially dangerous shoulder injury
Q1: What is the diagnosis?
The patient has sustained a posterior fracture dislocation of the left sternoclavicular joint with compression of the upper mediastinal structures, including the oesophagus. The fracture dislocation can be visualised on the computed tomogram (fig 1; see p 357) where the difference between the left and right sternoclavicular joints can be clearly seen. It is also possible that the patient suffered from a transvenous brachial plexus palsy after injury.

Q2: What do the angiograms show?
Arteriography when performed with the limb in the resting position (fig 2; see p 357) demonstrates a patent subclavian artery but with the limb in the abducted positions (fig 3; see p XXX) virtual occlusion of the subclavian artery can be seen. Venography demonstrated the presence of a patent subclavian vein. The vascular compressions account for the limb colour changes and lack of pulses during abduction and for the symptoms when working overhead.

Q3: How should the injury have been managed upon initial presentation?
This patient presented after a fall onto his shoulder with pain and swelling in his neck and left medial end of clavicle, symptoms and signs of neurovascular compromise of his left upper limb, and asymmetry between the medial end of his clavicles. This history strongly suggests posterior sternoclavicular joint dislocation with mediastinal structure compromise, but this diagnosis was not made post-injury as the plain radiographs appeared normal. This injury is often not demonstrated by plain radiographs and therefore computed tomography should have been performed at initial presentation, and would have demonstrated the injury. The fracture dislocation should then have been reduced, thereby resolving the symptoms both by reduction of compression and from compression of nearby structures. It is important not to miss posterior sternoclavicular joint dislocations as injury to the upper limb structures can cause serious complications including death.

Q4: What are the management options nine years after injury?
The options available to the patient are to live with his disability or to undergo surgery to prevent the medially clavicle compressing mediastinal structures. The patient chose to proceed to surgery due to the severity of his symptoms. At operation a fracture dislocation of the clavicle 1 cm from the medial end was noted and as reconstruction of the fracture-dislocation was not possible the medial 2 cm of the clavicle was resected subperiosteally leaving the costoclavicular joint dislocation with mediastinal structures as injury to the upper limb, and asymmetry between the left and right side.

Discussion
Posterior sternoclavicular dislocation is a rare injury. It is nearly always a result of trauma and is rare after the age of 25 years. It can result in significant morbidity or death due to the proximity of the superior mediastinal contents, which may be compressed or injured by the medial end of the clavicle. Early diagnosis can often be difficult due to extensive local swelling and bruising, difficulties in assessment using plain radiographs, and as this injury is rare it is often overlooked. Computed tomography is the best method of demonstrating the anatomy of the sternoclavicular joint and its surroundings structures. In most cases the AEDication can be reduced by closed methods within 48 hours of injury and after 48 hours open reduction is more likely to be required. It is therefore important to maintain a high level of suspicion of this injury. Early diagnosis improves the success of closed treatment and reduces complications. Our patient’s injury was missed due to poor...
awareness and inadequate imaging after the injury. At presentation nine years later, the diagnosis was confirmed using computed tomography and angiography.

Our patient suffered from dysphagia due to compression of the oesophagus, which is the most common mediastinal symptom from posterior sternoclavicular joint dislocation. The positional vascular symptoms experienced by our patient are rare. Due to the length of time that had elapsed since the fracture, scarring between the medial end of clavicle and the great vessels was anticipated and therefore the operation was performed jointly by orthopaedic and vascular surgeons. Fortunately, the medial end of clavicle was mobilised easily, without damage to the great vessels.

There is conflicting advice from previous authors regarding the surgical approach for chronic posterior sternoclavicular dislocation. Rockwood et al recommend resection of the medial clavicle and retention of the costoclavicular ligament for support. Other authors suggest that reconstruction of the joint provides better results. In this patient reconstruction would not have produced normal joint function due to the degree of joint damage and thus resection was performed.

Medical practitioners should maintain a high index of suspicion of posterior sternoclavicular joint dislocation in patients complaining of pain near the joint after direct or indirect trauma to the neck or shoulder as this injury is easily missed. Also radiographs should not be relied upon to exclude the sternoclavicular dislocation and further imaging, usually a computed tomogram, is indicated.

Final diagnosis
Posterior fracture dislocation of the left sternoclavicular joint with compression of the posterior sternoclavicular joint.

References

A child with growth failure

Q1: What are the radiological and biochemical features and the diagnosis of this case?

The computed tomogram shows extensive cerebral calcification (fig. 1; see p 388), and the radiograph shows the dense osteosclerosis (fig. 2; sa p 388) that is characteristic of osteopetrosis. Biochemical investigations show hypokalaemia, hyperchloroemia, low bicarbonate levels, and a low arterial pH in association with a high urinary pH, indicating distal renal tubular acidosis. The diagnosis is carbonic anhydrase II (CA II) deficiency syndrome, which is characterised by the triad of osteopetrosis, intracraniac calcification, and renal tubular acidosis. Intracraniac calcification and renal tubular acidosis distinguish CA II deficiency from classical varieties of osteopetrosis. Also, severe anaemia, a common finding in classical osteopetrosis, is absent or very mild in CA II deficiency.

CA II deficiency is a rare autosomal recessive disorder attributed to mutations of the CA II isoenzyme gene (chromosome 8). In CA II II is the most catalytically active as well as the most widely distributed isoenzyme of the CA series. CA II is important for acid-base regulation as well as bone resorption. The latter function is mediated by its effects on osteoclast function: it enables H⁺ pump activity, which helps the osteoclast in secreting acid that helps to dissolve bone mineral as well as digest bone matrix. Therefore, in CA II deficiency, bone resorption is markedly affected, leading to osteopetrosis. The raised serum acid phosphatase levels are attributed to defective osteoclast activity. A similar deficiency in acid excretion by the kidney may explain renal tubular acidosis. The reason for cerebral calcification is presently unclear. Children usually present with mental subnormality, short stature, and involvement of lower cranial nerves (due to sclerosis of skull base). As this is an inherited condition, family members also need to be investigated if molecular studies are readily available.

Q2: What are the radiological features of this disorder?

Skeletal radiography in CA II deficiency reveals findings indistinguishable from classical osteopetrosis—that is, a generalised increase in bone density; hence the term “marble bone disease”. Skull radiographs typically show a thick dense cranium with basal osteosclerosis and under-pneumatisation of the paranasal and mastoid sinuses. Another well described radiological feature of osteopetrosis is the appearance of alternating dense and lucent bands in the long bones and skull, which occurs due to a fluctuating skeletal growth. However, in CA II deficiency, osteosclerosis can spontaneously diminish. Cerebral calcification involving the cortex and the basal ganglia on computed tomography may appear at about 2-5 years of age.

Q3: What are the treatment options and prognosis of this disorder?

Bicarbonate supplementation is the mainstay of treatment, but the long term outcome of therapy is not known. Bone marrow transplantation may improve the skeletal disorder, but not the renal defect. A low calcium, high phosphate diet may be useful in this disorder. CA II deficiency is reportedly compatible with long life.

Final diagnosis
Carbonic anhydrase II deficiency syndrome.

References

Multiorgan involvement in thalassaemia major

Q1: What are the findings on the MRI images?

MRI scans of the sella (fig 1 in questions; see p 358) show markedly decreased signal intensity in the anterior lobe of the pituitary gland in all the sequences, though best seen on gradient echo images. Hypointense signal is also seen in bilateral basal ganglia.

Q2: What is the diagnosis?

The diagnosis is secondary (erythropoietic) haemochromatosis with hypogonadotropic hypogonadism developing in a patient with β-thalassaemia major.

The child had been diagnosed as having thalassaemia major at the age of 5 months and had received numerous blood transfusions since. She had also been treated with desferrioxamine B. Her serum ferritin level was 8672 µg/l, luteinising hormone was zero, and follicle stimulating hormone level was 0.20 IU/l. MRI the of abdomen and chest (fig 1, next page), done at the same sitting, demonstrates low signal intensity, equal to that of background, in liver, spleen, pancreas and myocardium, indicating iron deposition in these organs also.

Discussion

Haemochromatosis refers to a group of disorders in which there is a progressive increase in total body iron stores with deposition of iron in the liver, heart, pancreas, and other organs. The two generalised categories of iron deposition in iron overload have been described:

1. Parenchymal cell iron deposition (see box 1)—this is seen in idiopathic (primary haemochromatosis), secondary to anaemia and ineffective erythropoiesis, intravascular haemolysis, cirrhosis, after portocaval anastomoses, and secondary to high intake.

2. Reticulohendothelial cell iron deposition (see box 2)—this is seen most commonly in...
Patients who have received multiple transfusions and also in patients with rhabdomyolysis.

Parenchymal cell iron deposition occurs primarily in liver (hepatocytes), pancreas (acinar cells), heart, and other endocrine glands (anterior lobe of pituitary gland). The spleen is usually spared. However, there have been few reports of low signal intensity in the spleen without any history of blood transfusion, the cause of which is unknown. Parenchymal cell iron deposition leads to cellular damage and organ dysfunction unless treated.

In transfusional iron overload, haemosiderin is deposited in the reticuloendothelial system, such as the Kupffer cells of the liver and the reticuloendothelial cells of the spleen and bone marrow. This iron is derived from the extravascular haemolysis of intact red blood cells by the reticuloendothelial cells, which occurs during the metabolism of senescent native and transfused erythrocytes. The pancreas is spared because it is not a reticuloendothelial organ. However, pancreatic iron deposition in transfusional iron overload may result from massive transfusion beyond the iron storage capacity of the reticuloendothelial cell system (10 g), which is the amount of iron in 40 units of blood. Reticuloendothelial iron deposition does not produce any significant organ dysfunction.

Thalassaemia major, characterised by ineffective erythropoiesis and hypercellular bone marrow, results in secondary erythropoietic haemochromatosis. These patients also absorb iron inappropriately and can develop severe parenchymal cell overload. Also, iron accumulation due to repeated transfusions for the anaemia, accounting for the decreased splenic signal intensity. Thus, this group of patients share MRI features of both reticuloendothelial and parenchymal cell iron overload, as seen in our case.

Excess iron deposition in the anterior pituitary leads to degranulation of the adenohypophysis and decreased hormone storage with ensuing hypogonadism due to pituitary hyporesponsiveness to gonadotrophin releasing hormone. Iron deposition in the posterior lobe and diabetes insipidus usually do not occur.

Cardiac iron deposition occurs in the ventricular myocardium before atrial myocardium. Furthermore, cardiac iron deposition is exclusively sarcoplasmic and not interstitial; therefore, wall thickness in haemochromatosis is usually normal. At MRI, the marked signal intensity reduction is due to decreased T2 relaxation time and magnetic field inhomogeneities created by the excess intracellular iron.

Ferritin and haemosiderin, being paramagnetic substances, cause a proton relaxation effect on neighbouring hydrogen nuclei; T1 and T2 relaxation time decreases. Since both T1 and T2 are shortened, intensity may either increase or decrease depending on which relaxation effect, T1 or T2, dominates. For relatively low concentration of iron, as seen in organs like muscle and kidney which do not accumulate iron in high concentrations, T1 is considerably shortened and T2 values only slightly shortened leading to increased signal intensity of these tissues. With increasing concentrations of iron, as in liver etc, T2 shortening becomes dominant leading to decreased signal intensity in these organs. GRE T2*-weighted sequence is regarded as the most sensitive technique for the detection of parenchymal iron deposition. This is likely due to the lack of a 180° refocusing pulse that partially recovers signal loss from the field in homogeneity in spin echo imaging.

**Final diagnosis**

Secondary (erythropoietic) haemochromatosis with hypogonadotropic hypogonadism in a patient with β-thalassaemia major.

**References**

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