Q1: Name the congenital renal anomaly identified in fig 1 (see p 355) 
Horseshoe kidney. The lower poles of the kidney being displaced towards the midline, joined by either functioning renal tissue or a fibrous band.

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? 
The majority of reported cases have cysts in the dorsal cord, which is in accordance with the presence of a horseshoe kidney.

A man with numbness and limb weaknesses

Q1: What is the diagnosis? 
The diagnosis is spinal (cervical) intramedullary cysticercosis.

Q2: What are the treatment options? 
The treatment of spinal intramedullary cysticercosis could be surgical, medical (that is, cysticidal therapy) 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/day 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 systemic evaluation 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 subarchnoidal 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. In 90% of reported cases of intramedullary cysticercosis due to neurocysticercosis the patients were between 20 and 45 years of age. The duration of symptoms varied from a week to 10 years. The mode of spread of intramedullary cysticercosis is either haematogenous or ventriculopercutaneous. MRI studies help in diagnosing and correctly establishing the pathological diagnosis of neurocysticercosis (including intramedullary cysticercosis).

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. Successful management of intramedullary cysticercosis by cysticidal drugs alone has also been reported in the literature.

In the present case when the diagnosis of intramedullary cysticercosis was established on MRI, surgery was undertaken due to its location in cervical segment, and this was followed by albendazole therapy (15 mg/kg x 28 days). The patient showed complete neurological improvement with resolution of the intramedullary lesion.

It is concluded that with present generation MRI and also successful medical management, the outcome of intramedullary cysticercosis is not as dismal as was reported earlier, and patients with paraplegia also have a favourable outcome.

Final diagnosis

Spinal (cervical) intramedullary cysticercosis.

References

Q3: How is the diagnosis confirmed?
Though the definitive diagnosis of PML depends on identification of characteristic neuropathological abnormalities on brain biopsy, it is not necessary to confirm the diagnosis.1 Neuroimaging is most helpful in diagnosis with typical computed tomography and MRI findings as described above. Cerebrospinal fluid is also being more sensitive than computed tomography. Recently polymerase chain reaction amplification of JC virus DNA from the cerebrospinal fluid has become the favoured diagnostic modality to confirm the diagnosis.4 Currently, outlook for patients with PML is poor and there is no effective treatment. Few authors have reported promising results with HAART treatment.5 Recently treatment with cidovir has been tried in such patients with varying results.6 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 tomography (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 transient brachial plexus palsy after injury.

Q2: What do the angiograms demonstrate?
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 357) virtual occlusion of the subclavian artery can be seen. Venography demonstrated the presence of an occluded subclavian vein 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 symptoms both neurologically and from compression of nearby structures. It is important not to miss posterior sternoclavicular joint dislocations as injury to the upper mediastinal 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 mediastinal 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 ligament intact. At review six weeks postoperatively symptoms had resolved, the medial clavicle was stable and he had returned to work. At six and 12 months postoperatively the patient felt that both his upper power limbs were normal and on examination his shoulders and upper limbs were functionally normal with full power, normal range of joint movements and normal pulses in all limb positions.

Discussion
Posterior sternoclavicular dislocation is a rare injury.1 It is nearly always a result of trauma and is rare after the age of 25 years.1 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.2 Early diagnosis can often be difficult due to extensive local swelling and bruising,3 difficulties in assessment using plain radiographs,4 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.5 In most cases the anterior dislocation can be reduced by closed methods within 48 hours of injury and after 48 hours open reduction is more likely to be required.6 It is therefore important to maintain a high index of suspicion of this injury.7 Early diagnosis improves the success of closed treatment and reduces complications. Our patient’s injury was missed due to poor...
self assessment answers

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A child with growth failure

Q1: What are the radiological and biochemical features and the diagnosis of this case?
The computed tomogram shows extensive cerebellar calcification (fig 1; see p 388), and the radiograph shows the dense osteosclerosis (fig 2; see p 389), that is characteristic of osteopetrosis. Biochemical investigations show hypokalaemia, hyperchloræmia, low bicarbonate levels, and a low arterial pH in association with a high urinary pH, indicating distal renal tubular acidosis. This is the classic feature of osteopetrosis, intracranial calcification, and renal tubular acidosis. Intracranial 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). 1,2 CA II is the most catalytically active as well as the most widely distributed isoenzyme of the CA II isoenzyme family. It 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 phosphatase 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.

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 378) 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 µU.

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.

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 Reticulendothelial cell iron deposition (see box 2)—this is seen mostly in

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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.

Parenchymal cell 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 adenohypophys 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 concentration, 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
A misdiagnosed potentially dangerous shoulder injury

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