

**SELF ASSESSMENT ANSWERS**

**Congenital renal anomaly in a patient with situs inversus**

**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 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) hypogonadism; (2) undescended testis; (3) ureteral duplication. Vaginal septum and bicornuate uterus can also be associated with this condition.

**Discussion**

Horseshoe kidney was first recognised during a necropsy by DeCarpi in 1521, but Botallo in 1564 provided the first description and illustration of a horseshoe kidney. Horseshoe kidneys are believed to result from the median fusion of metanephric tissue due to mechanical forces. However studies have suggested that abnormal fusion of tissue associated with the parenchymatous isthmus of horseshoe kidney is the result of a teratogenic event involving the abnormal migration of posterior nephrogenic cells.

In most cases the kidneys are linked at the lower poles by a parenchymatous or fibrous isthmus that crosses the midline of the body. In general isthmus lies anterior to aorta and vena cava. Because kidneys fail to rotate, the calyces point posteriorly. The ureter inserts higher on the renal pelvis and lies laterally and crosses over and anterior to isthmus. The blood supply can be quite variable.

The horseshoe kidney is frequently found in other congenital anomalies, some of which are incompatible with long term survival. Most common congenital anomalies involved include skeletal, cardiovascular, and central nervous systems. There is increased occurrence of other genitourinary anomalies. Females with Turner’s syndrome have a high incidence of horseshoe kidney. Horseshoe kidney with situs inversus is a rare and interesting association.

One of third of all patients remain asymptomatic. Others present with vague abdominal pain resulting from hydronephrosis, infection, or calculus formation. Horseshoe kidney is associated with an increased relative risk of Wilms’ tumour, transitional cell carcinoma, and renal carcinoma.

Ultrasound or an excretory urogram readily makes the diagnosis. Ultrasound diagnosis depends on the demonstration of an isthmus or band of renal tissue across the midline. In a number of cases the band of renal tissue may evade ultrasonic detection.

Computed tomography may be necessary to confirm the diagnosis. Intervention is required because of obstruction or calculus. The combination 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 cysticercus cyst with scolex (larval cyst).

**Q2:** What are the treatment options?

The treatment of spinal intramedullary cysticercosis could be surgical, medical (that is, praziquantel were shown to be effective in the treatment of spinal intramedullary cysticercosis), or both. Horseshoe kidney is the result of a teratogenic event involving the abnormal migration of posterior nephrogenic cells.

The duration of symptoms varied from a week to 10 years. The mode of spread of intramedullary cysticercosis is either haematogenous or ventriculocephalodermal. MRI studies help in diagnosing and correctly planning 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 × 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**


**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 leucoencephalopathy (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 white matter in bilateral paracentral regions and in the right temporoparieto-frontal region without areas of enhancement or mass effect. Figure 2A (see p 356) shows the gadolinium enhanced T1 sagittal view 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 of non-enhancing white matter lesions with typical increased T2 and decreased T1 signals on MRI head are highly suggestive of PML.
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 transthoracic bacial 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 an extensive parietal compression of the subclavian vein. The vascular compression accounts for the limb colour changes and lack of pulses during abduction and for the symptoms when working overhead.

Q3: How should injury have been managed upon initial presentation?

This patient presented and fell 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 symptoms both of the fracture and from compression of nearby structures. It is important not to miss posterior sternoclavicular joint dislocations as injury to the 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 medial 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 stable and he had returned to work. At six and 12 months postoperatively the patient felt 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. 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.6 In most cases the first AICD can be reduced by closed methods within 48 hours of injury and after 48 hours open reduction is more likely to be required. Findings at the level of suspicion of the subclavian artery dissection 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 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 upper mediastinal structures.

**References**  

**Learning points**  
- CA II deficiency is an atypical variant of classical osteopetrosis and is characterised by renal tubular acidosis and cerebral calcification.  
- Radiological features of osteopetrosis include uniformly dense, sclerotic bones; alternating dark and lucent bands have also been described.  
- Children with CA II deficiency syndrome present with growth failure, asymptomatic cerebral calcification, and osteopetrosis.  
- The disorder is compatible with long life.  
- Bone marrow transplantation may improve osteopetrosis, but does not reverse acidosis; hence alkaline supplementation is required.

**Questions**  
Q1: 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.

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.  

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 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 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.1 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 system (10 g), which is the amount of iron in 40 units of blood.1 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 of the reticuloendothelial system may develop leading 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 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.1 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.1 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.1,2

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.1

Final diagnosis

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

References


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