An unusual association of intracranial aneurysms and oesophageal duplication in a case of Klippel-Trenaunay syndrome

Abstract The Klippel-Trenaunay syndrome (KTS) is a congenital disorder resulting from a mesodermal abnormality, characterised by cutaneous capillary haemangiomas, hypertrophy of bone and soft tissues and varicose veins. The presence of intracranial aneurysms has rarely been described, while oesophageal duplication has not been reported previously. We describe a patient with the KTS with both there additional abnormalities, which could be explained by a postulated mosaic gene abnormality.

Key words Klippel-Trenaunay syndrome · Aneurysm, intracranial · Oesophagus, duplication

We describe a patient with KTS, intracranial aneurysms and oesophageal duplication. Although intracranial aneurysms have been described previously [7], an oesophageal anomaly has not.

Case report

A 26-year-old white Italian man had at birth a port-wine haemangioma involving the neck and gradually increasing in size in childhood. During adolescence his left upper arm became more varicose. Four months before admission, ultrasonography showed large varicosities in the left forearm, with venous thrombosis. At this time he started complaining of dizziness and progressive weakness in the left limbs.

Examination revealed a port-wine haemangioma on the upper trunk, involving the proximal left arm and the back of the neck. The left arm was longer than the right, showing clear hypertrophy of soft tissue. Varicosities of the superficial veins were also evident.
Fig. 1a, b MRI. a Coronal T2-weighted b Sagittal contrast-enhanced T1-weighted images, showing multiple loculi of a calcified and thrombosed giant aneurysm, involving the vertebral and basilar arteries, displacing the brain stem to the left. In b the residual lumen of the right vertebral artery is visible within a large thrombus.

Fig. 2a-c Vertebral angiography. a, b Lateral and anteroposterior projections of right injection. c Lateral projection of left injection confirms the irregularity and tortuosity of both vertebral arteries and the large aneurysm at the junction of the middle and the upper thirds of the basilar artery.

He had a moderate left hemiparesis, a deficit of the left lateral gaze with nystagmus, paresis of the soft palate, an absent gag reflex, hyperreflexia of the left limbs and bilateral Babinski signs. MRI showed a giant aneurysm of the basilar artery involving the distal right vertebral artery, with calcification and thrombosis of the wall and compression and displacement of the brain stem to the left (Fig. 1). Angiography confirmed these findings and showed aneurysmal dilatation of the distal left vertebral artery (Fig. 2).

In order to reduce the marked compression of the brain stem, occlusion of both vertebral arteries was planned in two steps, separated by 5 months. At the first session right vertebral artery occlusion was performed using two balloons, after test occlusion. Arteriography showed a slower circulation in the aneurysm.

On the second occasion, left vertebral artery occlusion was performed by placing coils at C2–C3, after test occlusion. The patient tolerated both treatments; his neurological signs remained unchanged except for the appearance of dysphagia. He was therefore submitted to oesophagogastroscopy and a barium study, which revealed oesophageal duplication (Fig. 3).

Discussion

The extensive port-wine haemangioma and soft tissue hypertrophy of the left with venous varicosities and thrombosis in the same arm, support the diagnosis of KTS. In addition to superficial varicosities, many patients have present abnormalities of the deep veins [9]. Additional features include thrombophlebitis, cellulitis and oedema of the limbs [4]. Venous varicosities can also involve the bowel. Gastrointestinal haemorrhage, usually from colonic vascular malformations, is described in a few patients with KTS; oesophageal variceal bleeding caused by hypoplasia of the portal vein is also reported [10]. KTS may be also associated with lymphatic abnormalities, including lymphoedema and lymph vessel malformations, deriving from the obstruction of the deep veins [4]. There is a large spectrum of cutaneous anomalies. Haemangiomas are usually of the port-wine variety. They are patchy and may extend over the buttock and to the thorax. Bowel haemangiomas have been reported [3]. Other skin lesions include hyperhidrosis, hyperthermia and hypertrichosis [11].

KTS may be associated with ischaemic neurological complications such as flaccid paraplegia or hemiplegia, due to intracranial aneurysms [7]; only four cases of intracranial aneurysm have been reported. The association of arterial and venous anomalies could be ex-