

Cobb Syndrome: Case Report and Review of the Literature

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Key Words

Cobb syndrome · Spinal arteriovenous malformation · Naevus flammeus · Magnetic resonance imaging

Abstract

Cobb syndrome is a rare non-inherited neurocutaneous disease, in which there are metamerically cutaneous and spinal vascular malformations of the trunk. In cases of segmentally distributed multiple cutaneous vascular lesions, early diagnostic imaging of the spinal cord allows prompt intervention in order to reduce permanent neurological sequelae. We report case of Cobb syndrome in a 12-year-old boy and present a systematic review of the literature.

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Introduction

Cobb syndrome is an uncommon neurocutaneous disease characterised by vascular malformations of the spinal cord associated with congenital cutaneous vascular lesions affecting the corresponding dermatome [1]. This is an underdiagnosed condition. It is important to conduct imaging tests when cutaneous vascular lesions display a segmental distribution. Early diagnosis allows the appropriate treatment or management to be assessed, thereby reducing permanent sequelae such as paraplegia or sensory deficit.

Case Report

A 12-year-old boy with a congenital vascular skin lesion was referred for dermatologic evaluation. On examination a naevus flammeus was observed, containing dilated veins, located on the right half of the chest and abdomen, in a segmental distribution (fig. 1, 2). Dilated veins became more noticeable and a palpable thrill appeared when the patient performed a Valsalva manoeuvre. He did not present neurological symptoms and the neurolog-

ical examination was totally normal. The family history did not disclose any cutaneous angioma or neurocutaneous disease. MRI suggested an arteriovenous malformation (AVM) affecting segments T9–T11 (fig. 3). Arteriography subsequently demonstrated a large intramedullary AVM extending between the eighth dorsal and the first lumbar metameres and another AVM in the medullary cone, extending towards

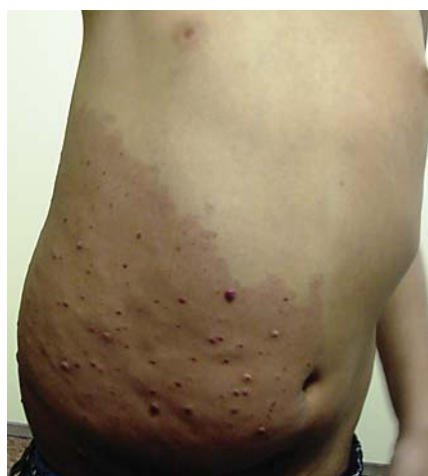


Fig. 1. Naevus flammeus with dilated veins (anterior view).



Fig. 2. Naevus flammeus with dilated veins (posterior view).



Fig. 3. T₂-weighted MR image obtained in the thoracolumbar spine, revealing high and low signal intensity suggesting a vascular malformation. Note the involvement of bone.

the posterior aspect. The capillary malformation involved the vertebral bodies, muscles and adipose tissue. In view of the asymptomatic nature of the lesion and the risk associated with the procedure, it was decided not to embolise the AVM. In consultation with the neurology department, it was agreed to monitor the patient annually by MRI.

Discussion

Cobb syndrome is a rare non-inherited clinical condition involving an association of spinal AVM with congenital vascular skin lesions in the same dermatome. If a cutaneous vascular malformation (port-wine stain or naevus flammeus) is found to have a metameric distribution on the trunk, a diagnosis of Cobb syndrome should be seriously considered, as a spinal AVM can give rise to significant neurological deficits.

Berenbruch reported the first case of this type in 1890, but it was not until 1915 – when Cobb presented the case of an 8-year-old boy who developed flaccid paralysis – that the syndrome received recognition. This child had a birthmark of the port-wine variety on the left side of his back

accompanied by a spinal AVM, which was surgically treated by Cushing [2]. Since Cobb's original discussion, only 40 cases have been reported in the international literature; of those, 20 were children [3]. The actual incidence may be higher, as only symptomatic cases are normally diagnosed.

It has been postulated that Cobb syndrome, Sturge-Weber syndrome and PHACE syndrome are segmental neurovascular syndromes that only differ in their aetiopathogenesis in terms of the target and timing of an error in neural plate migration. A somatic mutation developing in the region of the neural crest or the adjacent cephalic mesoderm before migration can produce these arterial or venous metameric syndromes [4].

There is no known racial predilection, but most reported cases have been in Caucasians. Cobb syndrome has a slight male predominance [1].

Skin manifestations typically occur as reddish macules representing a capillary malformation (port-wine stain or naevus flammeus). Masses consisting of dilated veins with a palpable thrill may be found in 20% of patients (as is the case in this article).

As in many prior reports of Cobb syndrome, skin findings were described as haemangiomas. This term has been recognised as a cause of confusion in the medical literature. In 1996, the International Society for the Study of Vascular Anomalies adopted a classification framework for vascular anomalies [5]. In this framework, haemangiomas are considered to be vascular tumours unique to infancy that proliferate and then regress slowly in childhood; vascular stain birthmarks are referred to as simple capillary malformations. Referring to such birthmarks as capillary malformations rather than as haemangiomas helps to clarify the parallel between the patient's skin findings and the spinal vascular malformation.

The skin lesions may occur anywhere in the dermatome, from the midline of the back to the abdomen. On rare occasions, midline lesions on the back are associated with spina bifida. Skin lesions may be very faint and become more noticeable when the patient performs a Valsalva manoeuvre [1], when blood is rapidly displaced from the spinal cord into the adjacent cutaneous vascular space due to a sudden rise in intraspinal pressure.

The medullary lesions are intraspinal AVMs and may extend to the meninges,

involve the vertebral body and even affect soft tissues within the same metamere. There is a single case report of a patient with Cobb syndrome who had brain angiomas as well as the classical lesions [6].

Both adult and young patients typically present with sudden-onset back pain or radicular pain in the lower limb, associated with numbness, which may be localised in a specific dermatome [7]. Less commonly, weakness or rectal and bladder dysfunction are the presenting symptoms. These neurological symptoms may remit or remain stable. However, the disease tends to worsen over time, in a stepwise or continuous manner, with the development of serious neurological deficits, mainly spastic paraparesis and paraplegia [8, 9]. These symptoms can be explained by the mass effect of the expanding AVM in the spinal cord and by subarachnoid haemorrhage. Rapidly developing weakness is predictive of a worse clinical course.

Some patients may display kyphoscoliosis when the spinal lesions affect the vertebral bodies and cause them to become deformed. If treatment is delayed, one possible complication is Foix-Alajouanine disease or subacute necrotic myelopathy, due to thrombosis of the spinal AVM [10].

Imaging techniques are the best diagnostic tool for AVM. The classical finding on plain radiographs consists of vertical striations visible inside the vertebral body. Plain films are of limited value, but can sometimes show bone erosion in the spinal canal affecting the pedicles, laminae or foramina in addition to involvement of the vertebral body itself. MRI is the first-line investigation for diagnosing AVM, being superior to CT for showing vascular malformations, angiomas and feeding arteries. Subsequent spinal angiography provides a definite diagnosis and allows the extent of the abnormality to be assessed [11]. Magnetic resonance angiography combined with selective intercostal angiography is currently the procedure of choice [12].

Collaboration with neurologists, neurosurgeons and interventional radiologists is important for proper assessment of the spinal lesion from both a diagnostic and a therapeutic point of view.

Patients with Cobb syndrome were initially treated surgically by laminectomy and decompression, as attempts to ligate the AVM resulted in fatal haemorrhage in many cases. Therapeutic radiation was at-

tempted later, with moderate success [13, 14]. Today, these patients should be referred to a neurosurgeon or interventional radiologist for assessment of whether AVM embolisation and spinal decompression is indicated, depending on their clinical condition [15]. Spinal vascular lesions fed by the posterior spinal artery are juxtamedullary and can be extirpated without causing injury. However, those arising from the anterior spinal artery are generally intramedullary and affect critical motor regions, so extirpation tends not to be technically feasible and treatment relies on

endovascular therapy. Vascular embolisation has dramatically improved the prognosis of this disease [16, 17].

A recent case report described a 5-month-old child with a cutaneous vascular malformation in the thoracolumbar region (T5–T12) and paraparesis. He was given oral prednisolone and underwent therapeutic endovascular embolisation of the vertebral column using n-butyl-2-cyanoacrylate. This was the first case report of neurological improvement in a paediatric patient treated with corticosteroids and endovascular therapy [18]. Combining

these two treatments in Cobb syndrome appears to provide effective symptom relief and may minimise mortality, especially in childhood.

In conclusion, it is important to remember that, in this syndrome, cutaneous vascular lesions may be accompanied by an intraspinal AVM, which can lead to weakness and paralysis. Current interventional embolisation techniques offer hopes of minimising permanent neurological damage. Early diagnosis is the key to increased survival of patients with Cobb syndrome.

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