

## **COVER LETTER**

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## **TITLE**

Neuro-vascular abnormalities with a metameric nevus: a case report and review of Cobb syndrome.

## **STRUCTURED ABSTRACT**

1. Background: Cobb syndrome is a disease characterized by a spinal angioma with a vascular nevus in the corresponding dermatome.
2. Objective: To describe a case of an individual with congenital neuro-vascular abnormalities and a dermatomal skin lesion without a corresponding spinal vascular malformation.
3. Design: Case report and literature review.
4. Setting: Tufts Medical Center, Outpatient neurology clinic, Boston, Massachusetts, USA.
5. Patient: 29-year-old female.
6. Results: The patient presented with complaints of headache, paresthesias, and urinary symptoms and was found to have several neuro-vascular congenital abnormalities including a Chiari type I malformation, cervical syrinx, multiple intracranial aneurysms, and an angiomatous dermatomal skin lesion as seen in Cobb syndrome without a radiographically evident spinal vascular malformation. A search of the literature reveals no reported cases of a patient with these collective findings.
7. Conclusion: The presence of a dermatomal vascular nevus should prompt the clinician to obtain spine imaging particularly in the setting of neurologic symptoms.

## **INTRODUCTION**

Cobb syndrome, also known as cutaneomeningospinal angiomatosis, first described by Berenbruch in 1890 and reported by Cobb in 1915, is a rare disorder characterized by spinal vascular malformations and a corresponding dermatomal vascular nevus [1, 2]. It usually presents in children and adults as lower extremity weakness, back pain, sensory loss below the affected level, or paralysis. Onset may be acute, gradual, or follow a stuttering course. Symptoms are thought to result from rupture or expansion of the angioma leading to direct compression of the spinal cord or ischemia due to diverted blood flow [3, 4]. We report a case of a 29-year-old female with a constellation of congenital neuro-vascular abnormalities who presented with lower extremity paresthesias and urinary retention and was found to have a port-wine stain in the S1 and S2 dermatomes without a corresponding spinal angioma on imaging.

## CASE REPORT

A 29-year-old female who presented in 2006 with a chief complaint of neck pain, headache, and numbness and tingling of both hands without prior trauma. Magnetic resonance imaging (MRI) of the cervical spine revealed a Chiari type I malformation with herniation of the cerebellar tonsils 15 mm below the foramen magnum as well as a syrinx from C2 to C4 (Fig 1). She underwent a suboccipital craniectomy with C1 laminectomy and duraplasty. Her headaches, described as bioccipital, pulsatile, and exacerbated by exertion, persisted after the surgery despite treatment with multiple prophylactic agents, trigger point injections, and occipital nerve blocks. Magnetic resonance angiography (MRA) revealed two incidental aneurysms in the right internal carotid artery (Fig 2). Over the next five years, she underwent four additional decompressions and revisions of the posterior fossa with subsequent periods of relief followed by inevitable return of her headache.

In 2009, the patient began to experience urinary retention and difficulty initiating micturition. She also developed paresthesias in the groin radiating down to the left leg as well as her bilateral soles. After a negative workup including multiple lumbar punctures, nerve conduction studies, electromyography, autoimmune panel, infectious serologies, and body imaging, she was referred to our clinic for management. A skin exam at the time revealed a flat and patchy blanchable rash (port-wine stain) in the S1 and S2 dermatomes in her left posterior thigh and calf, reminiscent of the cutaneous lesion in Cobb syndrome (Fig 3). Aside from her subjective sensory complaints, a neurological exam was unremarkable. An MRI and MRA with and without contrast of her lumbar spine demonstrated no evidence of a vascular abnormality in the spinal canal. This does not rule out subradiographic vascular malformations in the spinal canal or cauda equina as a potential explanation for her symptoms since it would be unusual for a

treated Chiari malformation and cervical syrinx to involve the bladder and lower extremities. The patient currently returns to clinic every six months for follow-up. Her symptoms have remained stable over the past two years.

## DISCUSSION

In 1915, Stanley Cobb described an eight-year-old boy who had a sharp pain in his lower back while playing ball. He had recurrence of the pain three days later followed by flaccid paralysis, anesthesia, and areflexia of both legs. The patient also experienced urinary retention, bowel incontinence, and priapism. He was admitted to the hospital with a presumptive diagnosis of poliomyelitis. Physical exam revealed a port-wine birthmark on his left back. Over ten and a half weeks, his legs became spastic and atrophied with hyperreflexia and upgoing toes. Five months after hospitalization, the patient underwent an exploratory laminectomy by Harvey Cushing who noted that the “dura was tense, bulging, and transmitted an unusually dark subdural coloration,” which when exposed, revealed “an extraordinary tangle of huge pulsating vessels filling the canal.” The dura was left open and the wound was closed. The only symptom relieved by the surgery was priapism. The patient was discharged home with unchanged spastic paralysis, incontinence, and anesthesia one year after admission. Cobb cited seven other cases, including the original dissertation by Berenbruch, where a vascular nevus occurred in the same metamere as an angioma of the spinal cord. The various presenting symptoms included weakness, paralysis, anesthesia, and pain, which were thought to be triggered by expansion or rupture of the angioma causing either direct compression of the cord or ischemia to the cord secondary to diverted blood flow. Cobb concluded that vascular nevi on the skin may be of diagnostic value when neurologic deficits corresponding to a spinal level are present [5].

There are less than fifty cases of Cobb syndrome reported in the literature. The disease appears to have a slight male predominance, usually presenting in childhood but can be seen in adults, and most frequently involves the thoracic segment of the spinal cord [6, 7]. Along with the classic port-wine stain, other skin lesions that have been described include angiokeratomas,

nevus flammeus, and lymphangioma circumscriptum [8-10]. The most common vascular malformation in the spine is an arteriovenous angioma but other anomalies such as dural arteriovenous fistula, telangiectasia, cavernous hemangioma, epidural venous angioma, and intraspinal hemangioma have all been reported [10, 11]. There has also been one case report of Cobb syndrome in association with tethered cord and another with neurofibromatosis type I [12, 13]. Current treatment options include embolization, resection, radiation, or steroids; the exact management depends on the patient's clinical signs and characteristics of the angioma [14].

Our patient was found to have a port-wine stain in her left leg that appeared classic for Cobb syndrome, coupled with symptoms of numbness and tingling and urinary retention, implying possible pathology of the cauda equina or exiting sacral nerve roots. Absence of an angioma on lumbar MRA does not rule out microscopic or subradiographic vascular anomalies in the spinal canal or within the nerves themselves as an etiology of her symptoms. In the case of our patient, it is interesting to note that her constellation of abnormalities – Chiari malformation, syrinx, intracranial aneurysms, and port-wine stain – seems to originate from defects in early development along a neuro-vascular axis.



## **CONCLUSION**

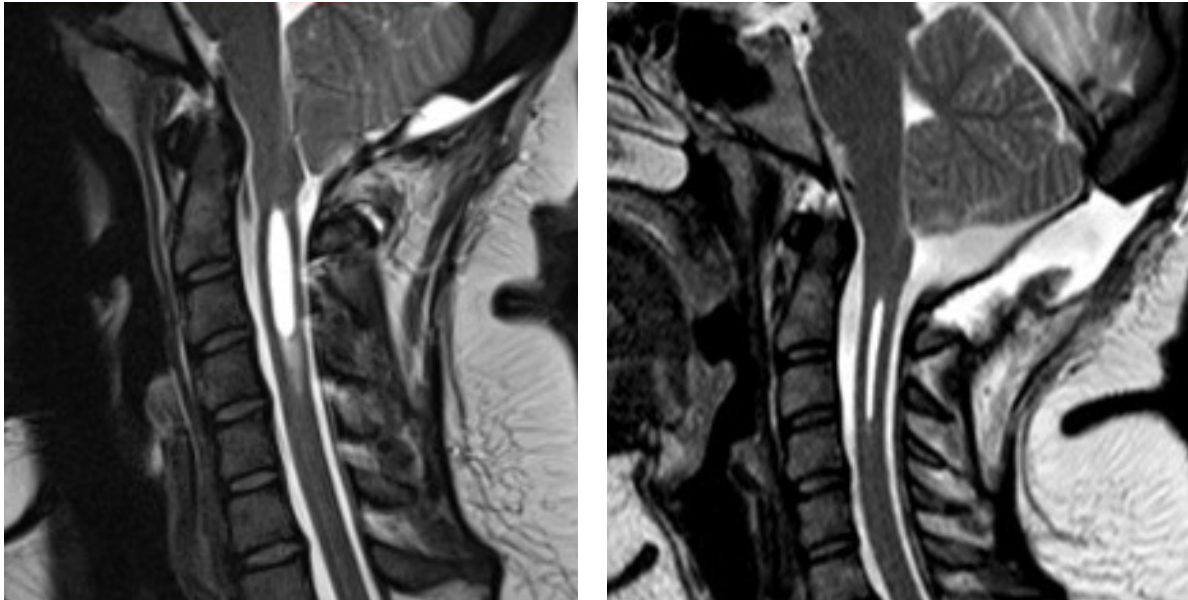
We conclude that the presence of a dermatomal vascular nevus should prompt the clinician to obtain spine imaging particularly in the setting of neurologic symptoms. Most patients with spinal vascular malformations usually remain asymptomatic until there is rupture or rapid expansion of the angioma. Timely diagnosis allows for closer monitoring of changes in size, prompt treatment, and earlier considerations for surgery.

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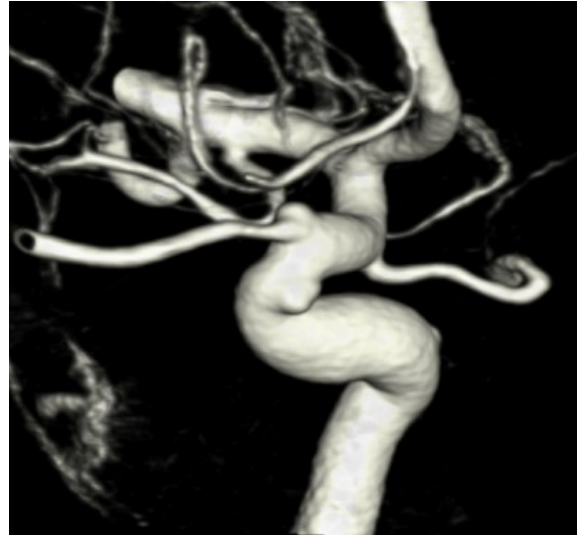
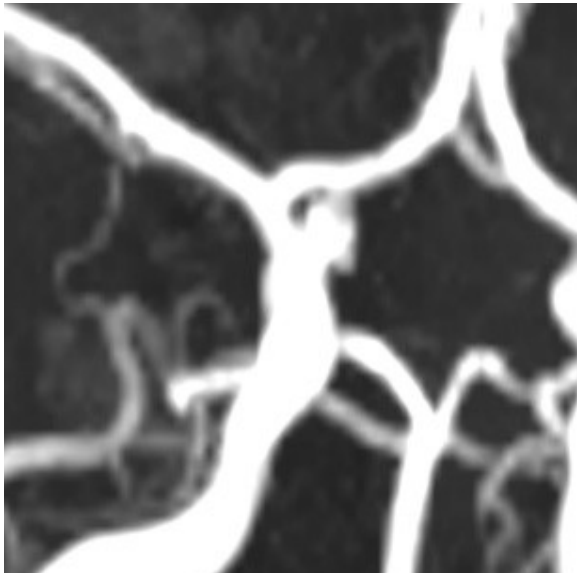
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## FIGURES



**Figure 1 – Chiari type I and syrinx pre- and post-op**

T2-weighted images of cervical spine pre- and post-op demonstrate reduction in size of syrinx and decompression of posterior fossa and foramen magnum.



**Figure 2 – Intracranial aneurysms**

Above: MRA head shows aneurysms in the right ICA.

Below: Reconstructed images from cerebral angiography reveal a right proximal medially-pointing superior hypophyseal broad-based aneurysm 2.5 mm in width and 2 mm in depth, and an ophthalmic artery aneurysmal origin dilatation 1.5 mm in depth.



**Figure 3 – Vascular nevus**

Flat patchy blanchable nevus (port-wine stain) distributed in S1 and S2 dermatomes.