CASE REPORT

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Cobb Syndrome, A Rare Disease – Case Report

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ABSTRACT

Cobb syndrome is a rare genetic disorder characterized by dermatomal pattern, with corresponding muscular, osseous, paraspinal, and/or spinal vascular lesions occurring at the same body somite (metamere). It is also known as spinal arteriovenous metameric syndrome (SAMS) and cutaneous meningospinal angiomatosis. We present a case of a 36 years old male presented to OPD with Complain of progressive weakness of Right upper limb for 5 years. On examination, Patient had large cutaneous port wine stain on the right side of the chest, nape of neck, and along the whole right upper limb in a dermatomal distribution with ulnar claw hand wasting of thenar muscles on right side. MRI cervical spine with contrast showed aggressive vertebral hemangioma involving anterior and posterior arch of C1 vertebra, vertebral body of C2 to C5, right half of vertebral body of C2 and C6 and right pedicle of C2 to C5 vertebra. Abnormal thickened and T2WI /STIR hyperintense skin and subcutaneous tissue involving right half of scalp and right half of neck which showing post contrast enhancement, possibility of subcutaneous hemangioma. Cobb syndrome was diagnosed based on the dermatomal distribution of the cutaneous vascular lesions and the corresponding vertebral, epidural, and paraspinal vascular lesions occurring at the same metamere. Treatment decision is guided by the patient's symptoms and imaging features.

INTRODUCTION

Cobb syndrome, also known as spinal arteriovenous metameric syndrome or cutaneomeningospinal angiomatosis, is a rare neurocutaneous vascular disorder characterized by combined cutaneous, muscular and/or bony vascular lesions as well as spinal or paraspinal vascular lesions involving the same body somite (metamere). [1]

The original description of the syndrome was the association of the spinal and cutaneous angiomas by Cobb in 1915. [2] However, conceptually, it is not necessarily required to have both cutaneous and spinal vascular malformations. Any combination of vascular malformations in the same metameres are possible, even if the lesions are not involving the spinal cord. [3,4]

The disease is genetic, non-hereditary, and is believed to be due to a sporadic mutation of the mother cells at an early stage of embryogenesis resulting in multiple vascular malformations in parts or all tissues of the same somatomeric distribution, including the spine, muscles, skeleton, soft tissues, and skin. [4]

Though the disease is present since birth, clinical manifestations are often not seen until later in life. Patients typically present with sudden onset of pain, weakness, paralysis, or paresthesia in the extremities that can be localized below a specific dermatome. [4] The pathogenesis of neurological symptoms in Cobb syndrome is believed to be due to a variety of factors, including cord compression by the vascular malformations, blood steal syndrome resulting in cord ischemia, and venous hypertension. [3,4]

The cutaneous vascular lesions are present at birth and don't tend to resolve spontaneously. They range from macular port wine stains to raised papular vascular lesions like angiomas, angiokeratomas, angiolipomas, and

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lymphangioma circumscriptum. The dermatomal distribution of the cutaneous lesions is important, as this may raise the suspicion of Cobb syndrome. [5]

The deep vascular lesions occurring at the same body metamere can be within the spinal cord itself (intramedullary), outside the cord (intraspinal extramedullary), vertebral or extraspinal, including the paraspinal soft tissues. [5] Under the new International Society for the Study of Vascular Anomalies classification system, vascular lesions which were previously described as Cobb syndrome would now be better characterized as vascular malformations rather than true hemangiomas. Cobb syndrome is usually associated with spinal high-flow vascular malformations; spinal arteriovenous vascular malformations are classified into four subtypes, Cobb Syndrome is associated with Type 3 arteriovenous malformations. [6] In our case, however, there was no spinal cord involvement, and the extra medullary vascular lesions were all of the low-flow type. Though the majority of spinal vascular lesions in Cobb syndrome represent high flow vascular malformations, spinal low-flow vascular malformations associated with cutaneous vascular lesions occurring in the same metamere are also defined as Cobb syndrome. [7–9]

Diagnosis of Cobb syndrome occurs when patients present with three or more of the following five factors: (1) spinal intramedullary vascular malformation, (2) intraspinal epidural venous vascular malformations, (3) vertebral osseous hemangioma, (4) paravertebral vascular malformations, or (5) cutaneous/subcutaneous vascular malformations. [10] Our patient fulfilled four of the five criteria including vertebral, epidural, paraspinal muscular, and cutaneous vascular malformations.

Radiological examinations are important tools for the diagnosis of Cobb syndrome. CT/CT angiography and MRI/MR angiography are useful modalities to identify deep vascular malformations. [11] MRI/MR angiography aids to detect precisely full extensions of the spinal vascular lesions. In case of arteriovenous fistula, there are multiple dilated and tortuous flow void perimedullary vessels seen on *T* 2 weighted images, the serpentine vascular structures are best appreciated on heavily *T* 2 weighted sequences [constructive interference in steady-state), fast imaging employing steady-state acquisition), or 3D turbo spin-echo)] compared with standard T2 TSE sequences. [12] Contrast-enhanced MRA is useful in localizing spinal dural fistula. The AV shunt is usually confirmed by the first-pass gadolinium-enhanced technique which demonstrates the early venous filling and the level of the shunt. [13] MRI also demonstrates spinal cord signal changes, like intramedullary edema and diffuse cord enhancement secondary to cord congestion/ischemia. [14]

DSA is essential for a definite diagnosis. It's the gold-standard modality in localizing and defining the full extent of spinal vascular lesions. It also helps to plan for treatment strategy and to allow occlusion via embolization. [15]

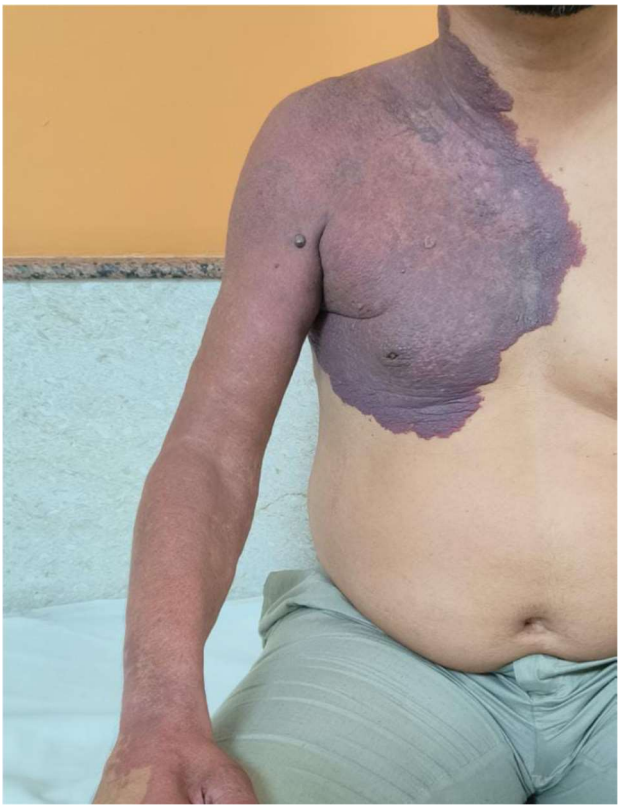

Figure 1.a

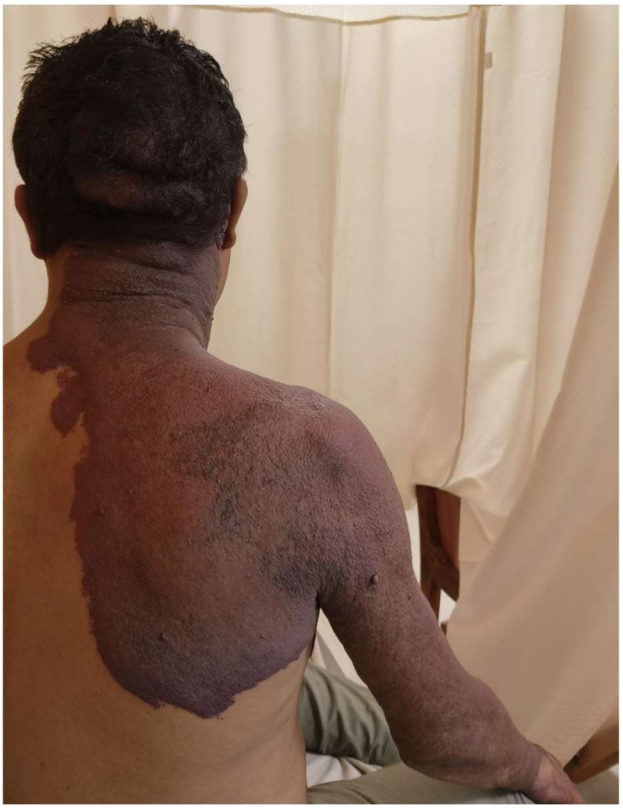

Figure 1.b

Figure 1 shows large port wine stain on right side in dermatomal distribution anteriorly (1.a) and posteriorly (1.b)

Treatment decision is guided by the patient's symptoms and imaging features; treatment of osteomuscular malformations involves endovascular embolization with occlusion of the feeding arteries and/or surgery. Treatment of spinal vascular lesions involves the use of steroids, surgery, and endovascular embolization. Surgery is usually indicated in cases of rapid/progressive neurological symptoms, pre-operative embolization is used to reduce intraoperative bleeding of these hypervascular lesions.[16]

Early diagnosis of Cobb syndrome is important as it allows rapid treatment, minimizing future neurological deficits especially paralysis or sensory deficits. Recognition of cutaneous vascular lesions with dermatomal distribution should prompt further evaluation for underlying spinal vascular lesions. [17]

CASE PRESENTATION

A 36 years old male presented to OPD with complaint of progressive weakness of right upper limb in the last 5 years. On examination, Patient had large cutaneous port wine stain on the right side of the chest, the nape, and along the whole right upper limb in a dermatomal distribution, ulnar claw hand wasting of thenar muscles on right side (Figure 1.a, Figure 1.b). Rest examination appeared unremarkable. The cutaneous vascular malformations were present since birth and were growing with the patient without regression. There was No family history of similar illness.

MRI cervical spine with contrast revealed evidence of altered signal intensity lesions noted involving anterior and posterior arch of C1 vertebra, vertebral body of C2 to C5, right half of vertebral body of C2 and C6 and right pedicle of C2 to C5 vertebra, It appears hyperintense on T1WI/T2WI /STIR images and incomplete suppression of T1FS images, possibility of aggressive vertebral hemangioma (Figure 2.a,b,c,d,e). There is evidence of epidural thickening showing post contrast enhancement adjacent to above mentioned lesions extending from C2 to C5 level, possibly due to dilated epidural venous plexus likely. Abnormal thickened and T2WI /STIR hyperintense skin and subcutaneous tissue involving right half of scalp and right half of neck which showing post contrast enhancement, possibility of subcutaneous hemangioma. Some of the tributaries of external jugular vein draining subcutaneous hemangioma appear dilated on right side.

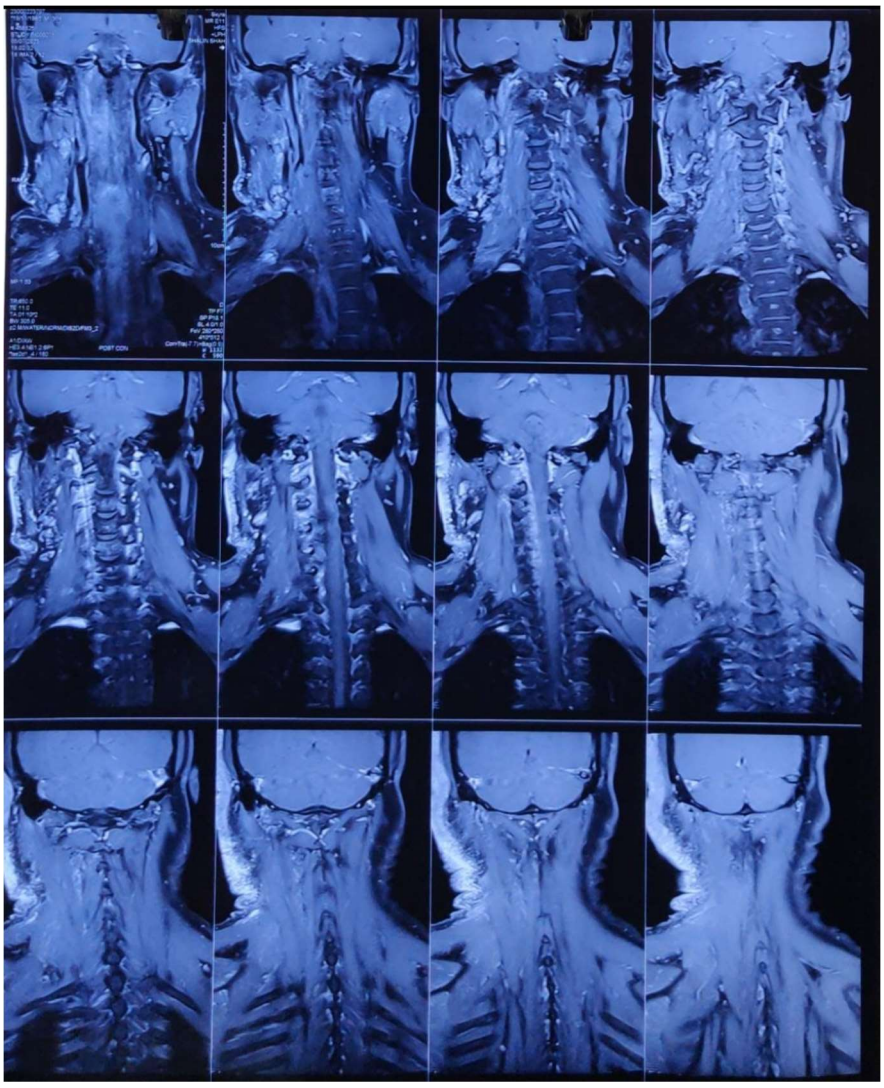

Figure 2.a

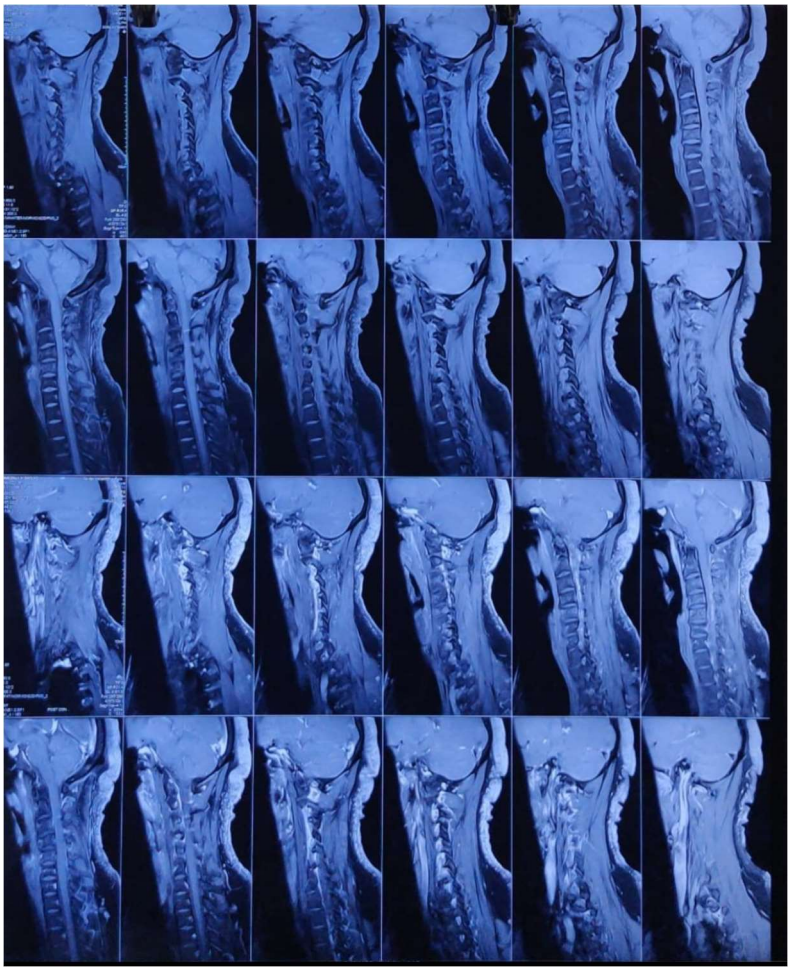

Figure 2.b

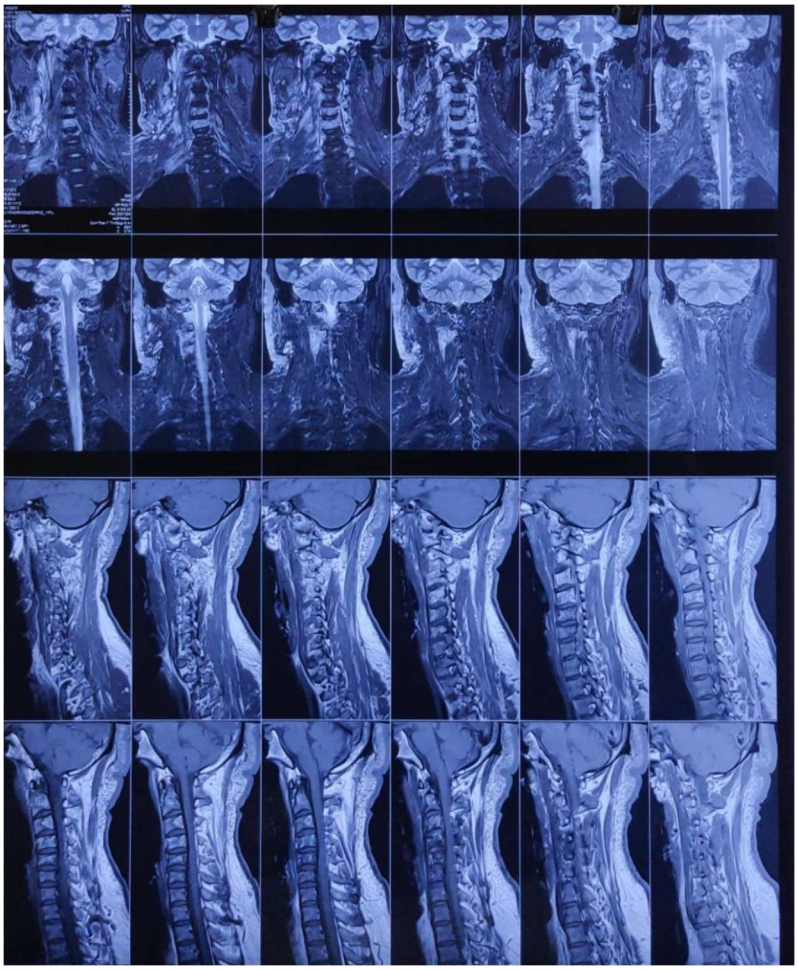

Figure 2.c

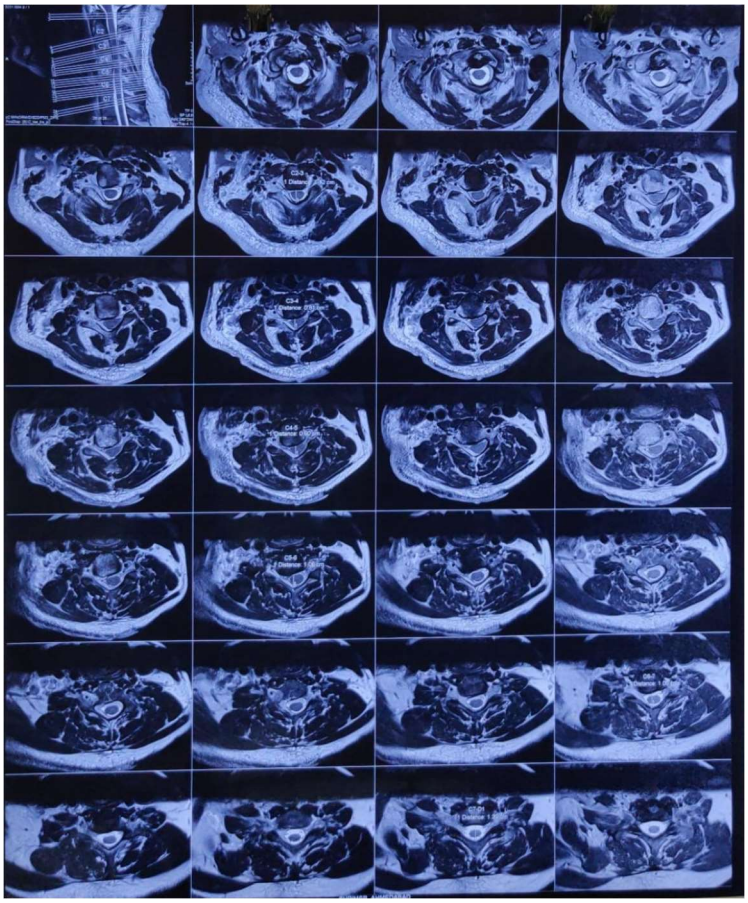

Figure 2.d

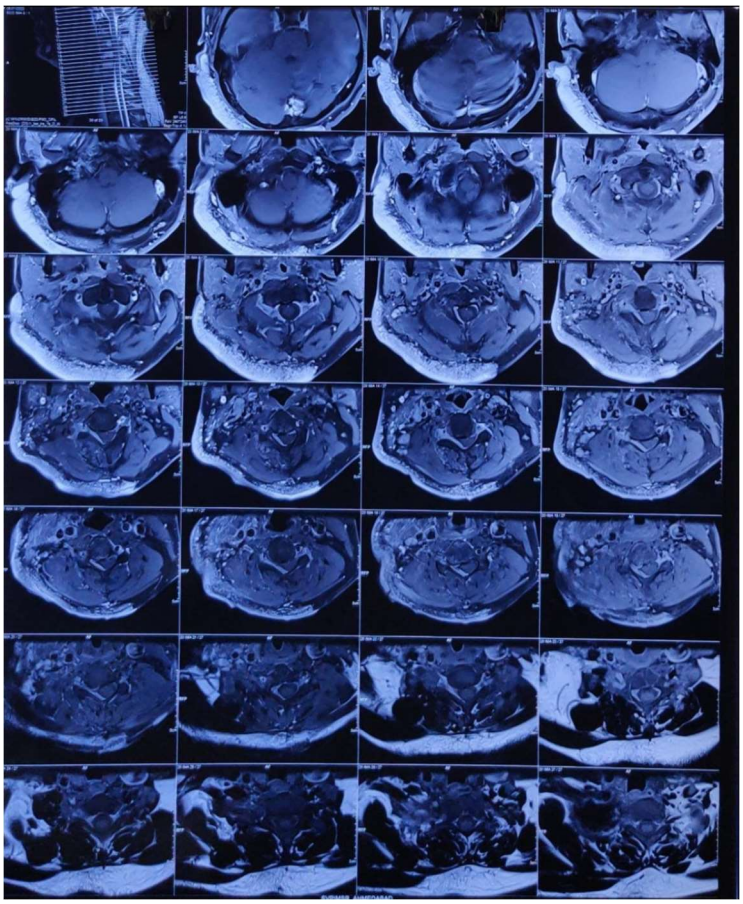

Figure 2.e

Figure 2 (a,b,c,d,e) shows MRI cervical spine altered signal intensity lesions noted involving anterior and posterior arch of C1 vertebra, vertebral body of C2 to C5, right half of vertebral body of C2 and C6 and right pedicle of C2 to C5 vertebra, possibility of aggressive vertebral hemangioma

Our patient had mild neurological symptoms and there was evidence of mild compression of the cervical segment of spinal cord. The patient was given a trial of conservative management. Patient was given a course of steroids in tapering dose for 3 weeks along with Neuropathic pain management and Neurotonics and was advised for routine follow up every month to check for increase in weakness. The patient had good recovery with resolution of symptoms due to cord compression.

CONCLUSION

Conservative management can also be thought as an alternative in such cases of Cobb syndrome with mild neurological symptoms.

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