SPINAL ARTERIOVENOUS METAMERIC SYNDROME (SAMS) / COBB SYNDROME

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ABSTRACT

Spinal arteriovenous metameric syndrome (SAMS), also known as Cobb syndrome, is a rare embryonic metameric syndrome, in which cutaneous, muscular, and or bony vascular lesions as well as para spinal and or spinal vascular lesions are found in the same metamere. Less than 80 cases with this syndrome have been reported in the literature and only one case was reported from Indian subcontinent. Classically, a birthmark (a cutaneous venocapillary malformation) and a spinal lesion (arteriovenous malformation or arteriovenous fistula) are found in the same or neighboring metameres. It should alert the physician and radiologist for early imaging of spine as early diagnosis and timely intervention would be favorable and may save patient from permanent neurological sequelae.

Key words: Spinal arteriovenous metameric syndrome (SAMS), Cobb syndrome, pigmented cutaneous nevus, lymphangioma circumscriptum, neurocutaneous disorder.

Case Report

A young girl, 14 years of age, presented to outpatient department (OPD) of in JPMC hospital with complaints of progressively worsening lower limb weakness and difficulty in walking over a period of 6 months. She had history of birth mark (cutaneous nevus) on dorsal and ventral aspect of left upper limb and anterior chest. There was no history of trauma, fever, urinary and defecation symptoms. Initial impression of radicular compression was made by physician and sent to radiology department for screening MRI lumbar spine. MRI lumbar spine was done which showed swelling and edema in conus. Further imaging examination proceeds to gadolinium enhanced MRI cervicodorsal spine to see the proximal extent of cord swelling. Multiple innumerable intradural intramedullary signal voids are seen along dorsal aspects of spinal cord within the spinal canal, these extend from C7 to T9 vertebral levels. Swollen cord showing high signal on T2WI and T2-FATSAT sequences is seen extending from lower cervical till lower dorsal levels, this finding is keeping with cord edema. Edema in conus is likely secondary to venous hypertension. On coronal and axial images abnormal signals were appreciated appearing high on T2WI and low on T1WI are seen in imaged sections of left kidney. Concomitant U/S KUB was performed which was consistent with left sided medullary sponge kidney, likely incidental finding. Right kidney, both ureters and urinary bladder were unremarkable. MRI was followed by CT spinal angiography which also confirmed MRI findings. All these imaging findings were highly suggestive of spinal arteriovenous malformation. Keeping with the history of cutaneous nevus, diagnosis of neurocutaneous syndrome most likely Spinal arteriovenous metameric syndrome (SAMS) / Cobb's syndrome was strongly suspected.

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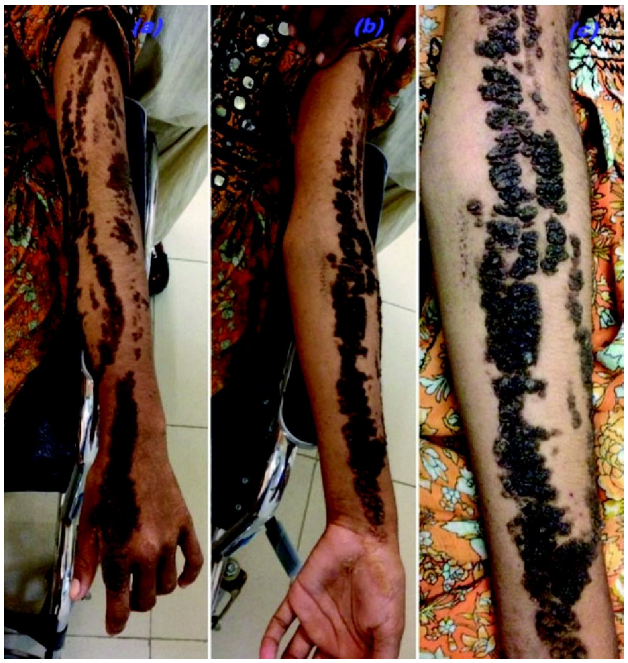
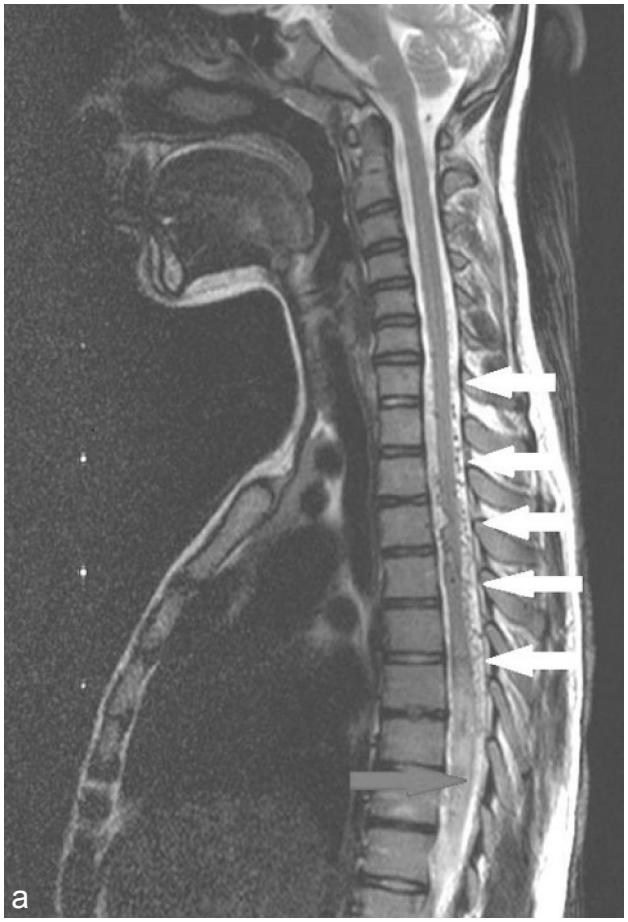


Figure 1: Images showing the angiomatous cutaneous lesions in dorsal aspect of left hand, fore arm and arm, corresponding to C5 toT1 dermatome levels.



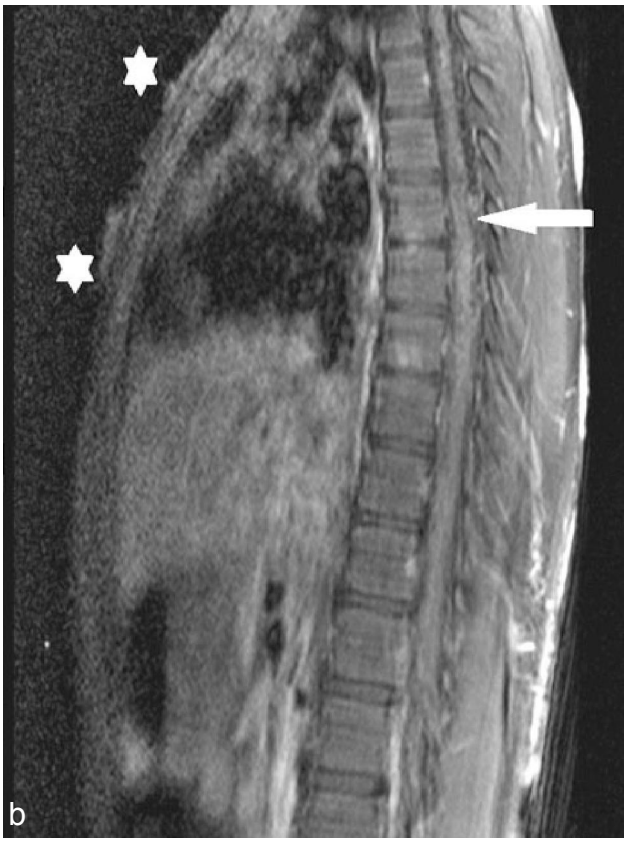
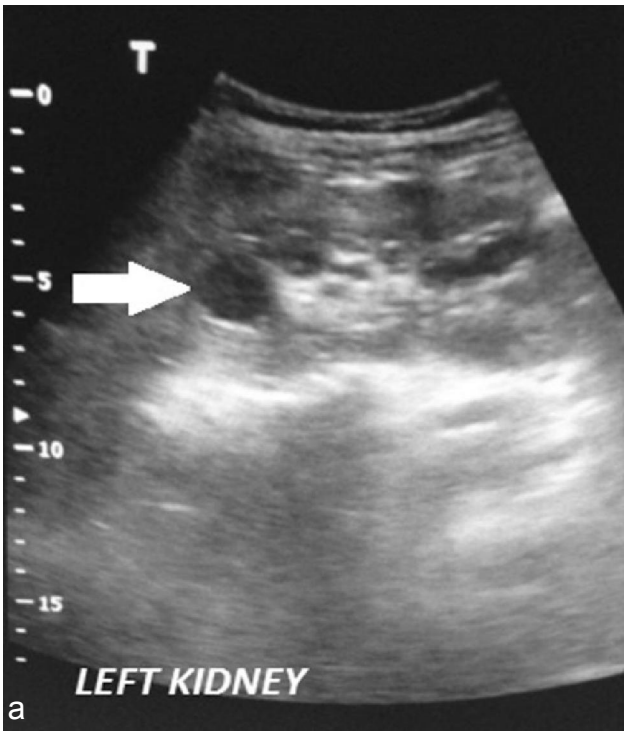


Figure 2(a-b): Selected T2W and STIR sagittal MRI of the dorsal spine showing innumerable flow voids along dorsal aspect of spinalcord (white aroows) and hyperintesities with swollen cord (grey arrow), cutaneous lesions on anterior chest (asterix)



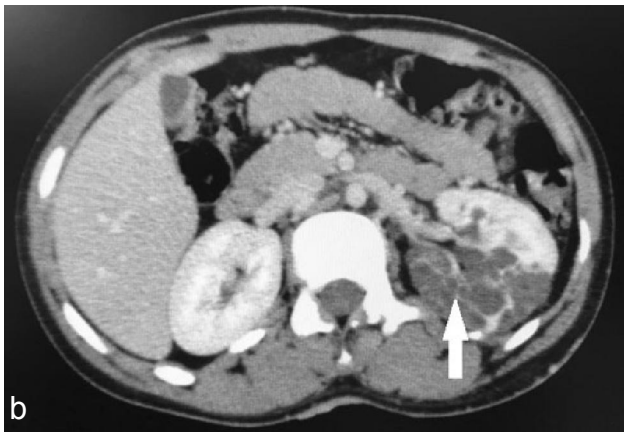
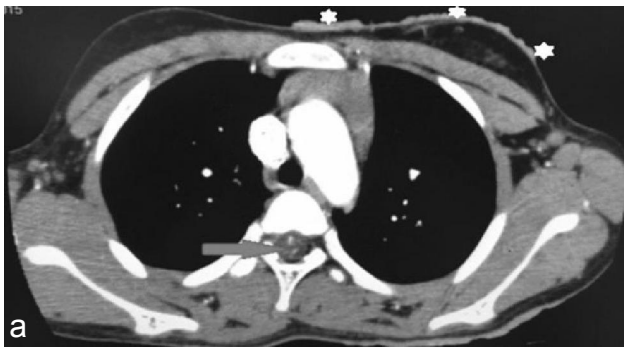
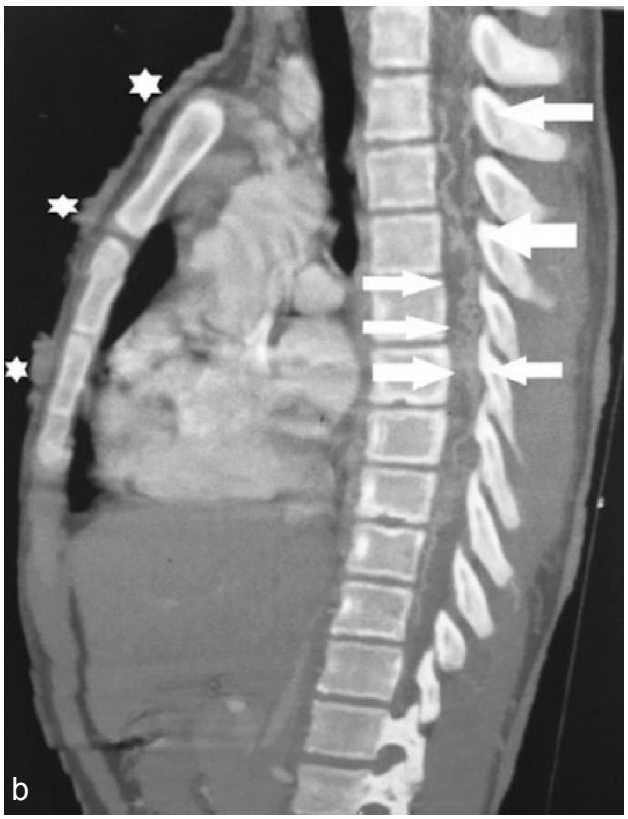


Figure 3(a-b): Ultrasound and selected CT axial section revealing abnormal cystic changes in left kidney. (incidental finding)





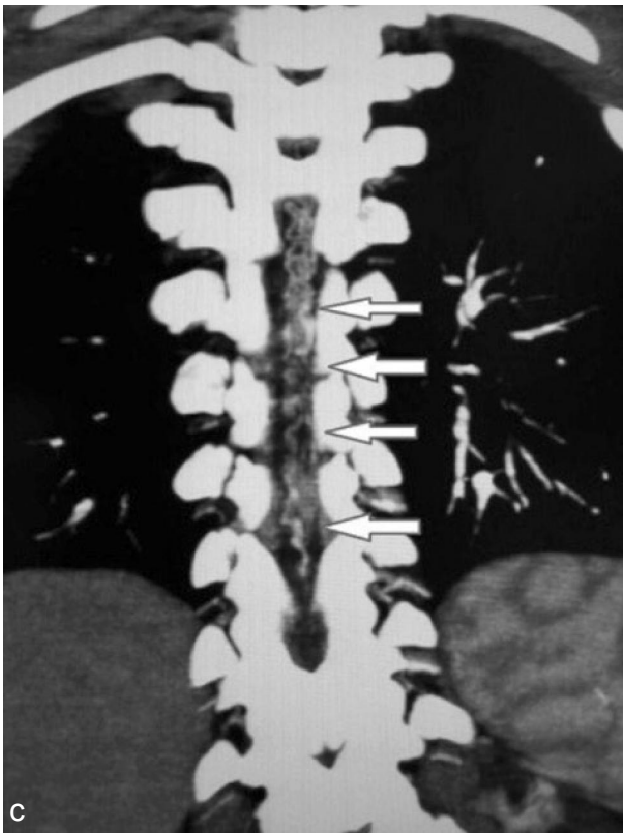


Figure 4(a,b,c): Selected CT axial, volumetric image at level T4 vertebrae, refomatted sagittal and coronal revealing serpigenious vessels in spnal canal in cervico dosal levels (arrows) and corrresponding dermatomal cutaneous lesions along anterior chest wall (asterix).

Discussion

The vascular neurocutaneous disorders are a broad heterogeneous group of congenital disorders with diverse genetic, clinical and pathological features that share common developmental lesions of the skin and of the central and peripheral nervous systems.[1,2,3] These disorders may be segmental, involve a large region or be a localized lesion. Segmental vascular neurocutaneous disorders include Sturge-Weber syndrome (SWS), PHACE syndrome (acronym for posterior fossa malformations, hemangiomas of the face and neck, arterial anomalies, cardiac defects or coarctation of the aorta, eye or endocrine anomalies, and sternal defects.), craniofacial arteriovenous metameric syndrome (CAMS) and spinal arteriovenous metameric syndrome (SAMS).[4] Vascular metameric syndrome (CAMS and SAMS) is a group of diseases that are classified on the basis of the embryologic

concept that an anomaly in one body segment simultaneously causes failure of the nerves, skin, and blood vessels within that segment.[5,6] The lesions that consist of multiple vascular malformations that affect more than 2 tissues derived from the same spinal metameric segment have been variably termed Cobb syndrome, extra-intradural, juvenile, and SAMS.[6,7,8]
Cobb syndrome is a rare, genetic, non-hereditary neurocutaneous disorder.[9] Less than 80 cases with this syndrome have been reported in the literature[10] and only one case was reported from Indian subcontinent.[11] To best of our knowledge this is first case report from Pakistan. It can involve the spinal cord, bone, epidural space, para spinal soft tissues or muscles, subcutaneous tissues and skin. It can occur in any of 31 spinal segments and can involve more than 1 segment.[12] Lesions are frequently seen in the cervical and thoracic spinal cord.[4] The associated metameric involvement was incomplete in most patients.[13] The cutaneous manifestations range from macular port-wine stains to various types of papular or nodular vascular lesions including angiomas, angiokeratomas, angiolipomas, and lymphangioma circumscriptum.[14] This disorder is most commonly seen during late childhood, it may occur at any age.[15] Peak age is the 3rd and 5th decades. There is a slight male predominance.[4] However, in another larger study significant female dominance (M : F=1:2.5) was seen.[13] Onset of signs usually manifest over weeks to years, but a sudden onset of weakness with rapid progression has also been reported.[15] Neurological presentations can vary from monoparesis to sudden onset paraplegia or quadriplegia. Bladder and bowel involvement is common but occurs late as the disease progress. Less common signs include meningismus, headache, fever, and gluteal and limb hypertrophy.[16]
Niimiet all, observed that the most common presenting symptom was intradural hemorrhage (spinal SAHs and hematomyelias).[13] With regard to neurological symptoms, cord compression due to spinal angiomaper se may not be the sole mechanism underlying the spinal cord symptoms. Other factors may include compression, venous hypertension and cord ischemia due to steal syndrome are the speculated mechanisms that would explain the myelopathy.[16]
For the diagnosis of SAMS, CT/CT angiography and MR imaging/MR angiography are the first-line diagnostic tools because of their inherent less invasiveness, but the selective catheter angiography is required to define the nature and angioarchitecture of the spinal lesions, which may be followed by the interventional procedures. Differentiation between the AVM/AVFs and infantile hemangiomas is crucial because treatment is different.[17]
MRI is useful modality to assess the extension of the lesions. MRI is better than CT in displaying deformed vessels, angiomas and the feeding artery. MRI can show intramedullary signal changes and most of the vessels and is safer than invasive angiography with intravascular contrast. The final diagnosis of the syndrome depends on angiography.[18]
The optimal management of the disease entity largely remains enigmatic because of its rarity and poorly understood pathophysiology. The understanding of the latter might be facilitated by adopting selective spinal angiography and embolization procedures, since spinal angiomas have a blood supply distinct from that of the normal spinal cord. With the recent advent of endovascular techniques, endovascular therapy has become the treatment of choice for various kinds of spinal arteriovenous malformations. Corticosteroids therapy helps in resolving associated edema thus reducing morbidity.[11]
Niimiet all, in there longitudinal series of 28 patients with SAMS demonstrates the progressive nature of the disease and poor long term functional prognosis. They observe that complex lesions can be treated safely by endovascular techniques with a palliative strategy focused on preventing hemorrhage, preserving spinal cord function, and relieving pain. Angiographic cure is the exception and only a realistic goal for limited lesions without significant intramedullary involvement. To maximize the effect of palliative treatment, periodic angiographic examination with intent to treat is important to prevent neurologic deterioration was also suggested. They suggest protocol for follow-up of clinically stable patients is yearly MR imaging without and with contrast administration and clinical examination, and spinal angiography with intent to treat every 3 - 5 years. If MR imaging changes or clinical deterioration occurs, perform spinal angiography with intent to treat without delay.[13]

Teaching Point:

1) The cutaneous lesion may provide a clue to Cobb syndrome when a patient comes with sudden or gra-

dual onset paraplegia or subarachnoid hemorrhage.
2) For the diagnosis of cobb syndrome, MR imaging / MR angiography of spine are the first line diagnostic tools.
3) A multidisciplinary approach balancing the patient's current neurological status against the potential risks and probable gains from any interventional and surgical procedure is recommended.

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