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Original Article

Sturge-Weber Syndrome – Rare Cases with Rare Presentation

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Abstract

Introduction: In theses case series we present 2 cases of Sturge Weber Syndrome(SWS) Type 1 and Type 3 which is very rare. The classical presentation of SWS is ipsilateral port wine stain with leptomeningeal enhancement with or without seizure which is Type 1. Type 3 which present with leptomeningeal enhancement without port wine stain.the patient with Type 3 has typical radiological features of transparenchymal medullary vessels. Patient may present with seizure and neurological deficit and subclinical stroke due to steal phenomenon.

Objective: The Objective of these case report is to present the rare presentation of Sturge Weber Syndrom (SWS) so that early diagnosis can be made and treatment can be started as early as possible.

Material and Method: These two patient underwent clinical, electrophysiological and radiological investigation and diagnosed as Type 1 and Type 3 Sturge Weber Syndrome(SWS) with typical radiological feature. Both patient are treated symptomatically and followed up for 2 years.

Conclusion: There must be high index of suspicion of diagnosis Sturge Weber Syndrome(SWS) specially Type 3 so that early treatment can be started aiming to improving prolonged neurological outcome.

Key words: Congenital neurocutaneous syndrome, Intracranial leptomeningeal angioma, Lobectomy or hemispherectomy, Sturge-Weber syndrome, The Roach scale

INTRODUCTION

Sturge-Weber syndrome (SWS) was first described by Sturge in 1879 and cerebral involvement was described by Kalischer in 1897.[1] SWS occurs sporadically, with an estimated incidence of 1 in 50,000 live births.[2] SWS is a rare, congenital neurocutaneous syndrome characterized by unilateral facial cutaneous vascular malformation (port-wine stain) in association with ipsilateral leptomeningeal angiomatosis.[3] Angiomas of SWS result due to failure of regression of a vascular plexus around cephalic portion of neural tube which is destined to become facial skin which normally forms at the 6th week of intrauterine life and regresses by the 9th week. Failure of its regression results in residual vascular tissue and has normal endothelial mitotic activity which forms angiomas of leptomeninges, face, and eye.[4,5] These vascular plexus have abnormal blood flow, venous occlusion, thrombosis, and "vascular steal phenomenon" resulting in ischemia, gliosis, atrophy, and calcification of underlying cortical tissue.[6] Sturge-Weber syndrome can be classified into three types using the Roach scale.[7] The most common is type 1, which is the classic manifestation of PWS and intracranial leptomeningeal angioma. Type 1 may or may not have ocular abnormalities, such as glaucoma. Type 2 is characterized by a PWS and an ocular involvement but with no brain abnormalities. Type 3 is characterized by leptomeningeal angiomatosis without cutaneous lesions or ocular abnormalities. This form is usually diagnosed during imaging for epilepsy. The diagnosis and management of SWS requires the combined skills of a radiologist, ophthalmologist, pediatrician, and physiotherapist. Here, we report two cases of SWS, one is type 1 which was presented with seizure managed conservatively with antiepileptic alone and other one was type 3 with right hemiplegia and seizures which were treated with antiepileptic drugs and aspirin. The patient recovered near complete.

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MATERIALS AND METHODS

Cases were prospectively studied during the period of 2019–2020. They are admitted in the department of neurosurgery during these periods. All the cases were studied by clinical examination and magnetic resonance imaging (MRI) and electroencephalogram. They were followed up a period of 2 years.

Case 1

A patient, Asvika, a 4-month-old female child, presented by her parents with complaining of seizure multiple episode. On examination, the baby was active, alert, cry good, with no change in voice while crying, tongue was in midline, can follow light, can follow sound. No facial asymmetry can suck and swallow breast milk without difficulty. Anterior fontanelle lax, head circumference 40 cm, weight 10 kg, moves all four limbs without any paucity. On local examination , there is an erythematous area [Figure 1a] over the left side of face involving the left side of forehead, periorbital area, and malar area, that is, ophthalmic and maxillary area. Ophthalmological study was normal. Clinically, we suspect SWS. To confirm and to rule out reason of seizure, radiological investigation was done. MRI brain with contrast shows pial enhancement [Figures 1b and 2a] on contrast over left frontotemporoparietal region with underlying atrophy of cortex. Abnormal transparenchymal vessels connecting cortical veins with subependymal vessels called medullary vessels [Figure 2b]. After radiological evaluation, the patient was diagnosed type 1 SWS. The patient was treated with antiepileptic drug. Seizure was controlled. The patient was discharged and asks for regular follow-up and ophthalmological evaluation.

Case 2

A patient, Master Raja, a 12-year-old male child, brought by his father with complaints of head ache, vomiting, followed by weakness of right side, for 4 days. He also complains of multiple episode of vomiting. Parents also noticed gradual development of slurring of speech with deviation of angle of mouth on the left side. Following admission in our hospital, the patient develops multiple episode of seizure. On examination, the patient was drowsy, disoriented, aphasic, afebrile, pupil bilaterally equally reacting to light, eye movement was full and right hemiplegia. No sensory impairment. Bladder and bowel were normal. There was no neurocutaneous marker [Figure 3c]. Oral cavity was normal. Ophthalmological evaluation was normal. Other systemic examination was normal.

There was a history of seizure disorder following meningoencephalitis at the age of 8 years when MRI brain was done which shows prominent triangle of vessels consisting of prominent components from arteries and vein

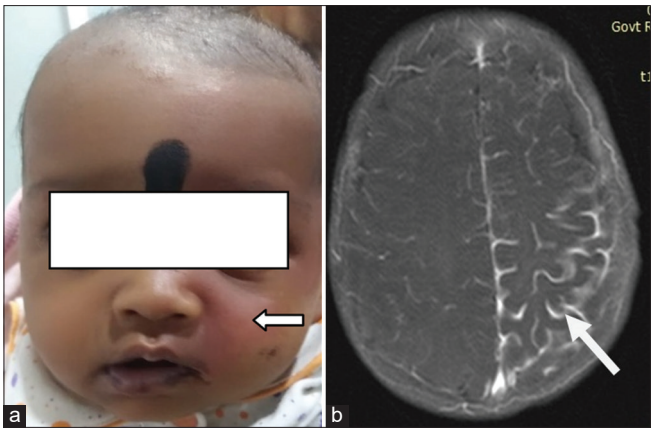

Figure 1: (a) Showing port wine stain on left side of face. (b) Magnetic resonance imaging T1 contrast showing parietooccipital cortical enhencement

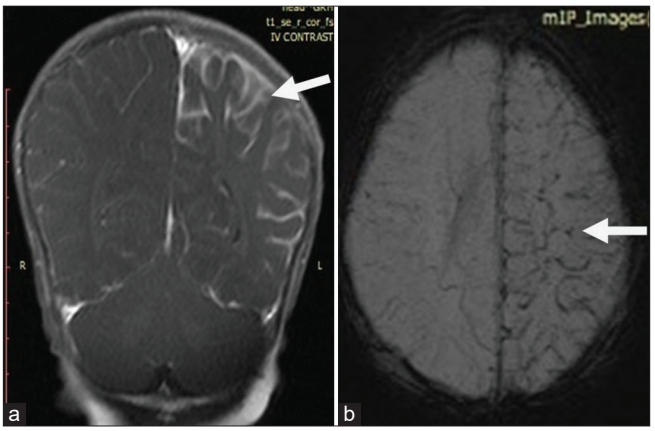

Figure 2: (a) Magnetic resonance imaging brain T1 contrast coronal cut showing pial enhancement in temporoparietal region. (b) Magnetic resonance imaging brain SWI film showing trans parenchymal medullary veins connecting subependymal vein with cortical veins

which are seen in posterior aspect of lateral ventricle and 3rd ventricle suggestive of arteriovenous malformation? Vein of Galen malformation. The patient was investigated for recent signs and symptoms. Routine blood investigation, serum electrolyte, chest X-ray, electrocardiogram, and some radiological imaging study were done. Computed tomography brain shows normal study [Figure 3b], no evidence of infarct or hemorrhage. MRI brain study reveals multiple dilated radially arranged transparenchymal medullary veins [Figure 3a] communicating the cortical veins with the subependymal vessels, abnormal left sided leptomeningeal enhencement with minimal atrophy of left hemicortex [Figure 4a]. Contrast showed no abnormal arteriovenous shunting of blood, no evidence of any acute infarct / hemorrhage. These radiological feature along with clinically absence of port wine stain [Figure 3c] suggestive of type 3 Sturge Weber syndrome. The patient was treated with antiepileptic, aspirin, and physiotherapy. Hemiplegia

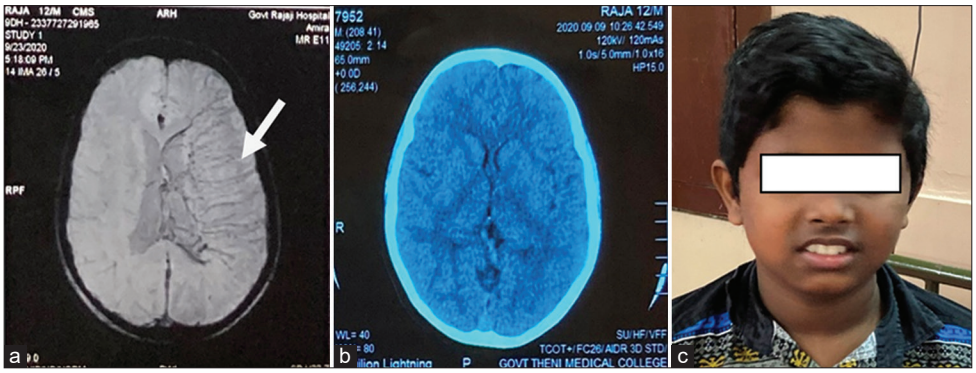

Figure 3: (a) Magnetic resonance imaging brain SWI film showing trans parenchymal medullary veins connecting subependymal vein with cortical veins. (b) Computed tomography brain plain axial cut shows normal study. (c) patient picture without skin manifestation

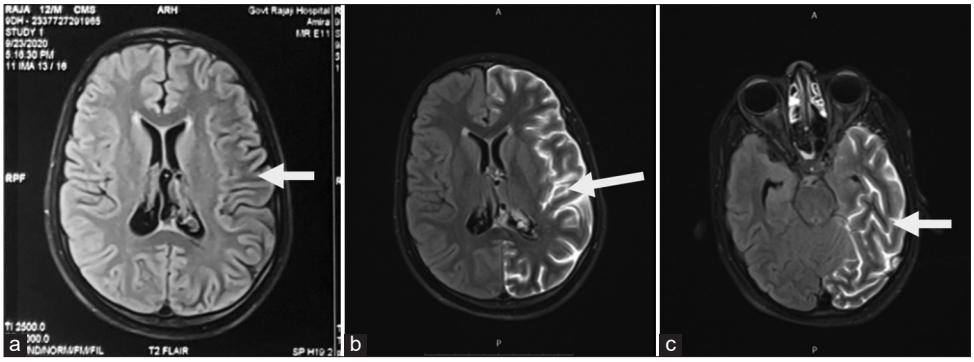

Figure 4: (a) Magnetic resonance imaging T2 FLAIR shows atrophy of cortex. (b and c) Magnetic resonance imaging T2 post contrast FLAIR shows left frontotemporoparietooccipital pial enhancement

gradually turns into hemiparesis and recovered nearly complete [Figure 4b and c].

DISCUSSION

Imaging has an important role in the diagnosis, detection, and follow-up of patients with SWS. Although, historically, the imaging finding that SWS is most known for its "tramline" of progressive calcification seen on X-ray. MRI is now considered a superior imaging modality. MRI specifically T1-weighted MRI with gadolinium contrast and susceptibility-weighted imaging[8] is the best imaging modality to diagnose the intracranial manifestations of SWS and is often cited as the "criterion standard" to diagnose SWS.[9,10] MRI may demonstrate thickened cortex, decreased convolutions, abnormal white matter, and gadolinium enhancement of leptomeningeal angioma[9] as well as enlargement of transmedullary and periventricular veins associated dilation and enhancement of the choroid plexus on the involved side, and dilated deep draining venous vessels.[11,12] Although the exact etiology of SWS remains unclear, the fundamental pathophysiologic abnormality in areas of brain affected by pial angiomatosis is the absence of an adequately functioning superficial cortical venous system.[13] Consequently, blood is rerouted centrally through the medullary veins resulting in venous hypertension and stagnation.[14] Type 1 SWS is common out of these three subtypes which has classical presentation of port-wine stain with radiological pial enhancement with or without ocular involvement. Type III SWS is the rarest subtype and it is difficult to diagnose as it does not involve skin abnormalities. Making the diagnosis of type 3 SWS, preferably before the onset of neurologic sequelae, may have an important impact on patient management. Aggressive treatment of epilepsy is advocated[15] with some studies even suggesting neurodevelopmental benefits with prophylactic anticonvulsants before the onset of seizures.[16] Prophylactic daily aspirin[17] and early surgery in the form of lobectomy or hemispherectomy, corpus callosotomy, and vagal nerve stimulation for the neurological manifestations[18,19] when seizures are not controlled by medical treatment may also be of benefit.[15] SWS has a spectrum of clinical and imaging findings, and it is important to recognize those patients at the fringes of the spectrum who may benefit from therapies and monitoring applied in more conventional cases. In our case, we got type 1 and type 3 variety which is very rare

and responds to medical therapy and discharged and the patients were asked for regular follow-up.

CONCLUSION

Though there are variety is exist early Sturge Weber Syndrome is a rare disorder, among the three types Type 3 is very rare that too with rare presentation and diagnosis is sometimes difficult. So high index of suspicion is necessary to diagnose Sturge Weber syndrome specially type 3 so that treatment can be started as early as possible aiming at improving prolonged neurological outcome.

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