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Research Paper

STURGE WEBER SYNDROME - A RARE CASE REPORT

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Abstract

Sturge-Weber syndrome is a neurocutaneous syndrome sometimes referred to as encephalotrigeminal angiomatosis, is a rare congenital, non heridetary condition of unknown etiology occurs sporadically. The classic pathognomonic features of disease include angioma of the leptomeninges extending to cerebral cortex with ipsilateral angiomatous lesions, unilateral facial nevus after one division of trigeminal nerve and epileptic convulsions. Professional counseling and support in addition to drug treatment can provide help to patients and their family to overcome their problems and improve the treatment outcome. The case reported here was misdiagnosed as seizure disorder at peripheral hospital. After through work out and in view of its extreme rarity, attempts has been made to report these cases as Sturge-Weber syndrome (SWS).

Key words: Sturge-Weber Syndrome (SWS), Encephalotrigeminal angiomatosis, Port-Wine Stain, Leptomeningeal enhancement, Microcephaly, Tram line calcifications.

INTRODUCTION

Sturge Weber syndrome (SWS) was first described by Schirmer in 1860 and later more specifically by Sturge in 1879, associated dermatological and ophthalmic changes of the disease to neurologic manifestations. Weber in 1929 complemented it with the documentation of radiologic alterations seen in these patients [1]. Sturge-Weber syndrome is a rare disorder that occurs with a frequency of 1: 50, 000[1]. It is a sporadic neurocutaneous disease characterized by facial port-wine stain, ocular abnormalities (glaucoma and choroidal hemangioma) and leptomeningeal angioma most often involving occipital and posterior parietal lobes[2]. These changes are usually unilateral and can be seen in both sexes equally with no racial differences[3].This syndrome consists of constellation of symptoms and signs including a facial nevus, focal seizures, hemiparesis, intracranial calcification and mental retardation[4]. Encephalofacial angiomatosis [5] or encephalotrigeminal angiomatosis are used as synonyms of the syndrome as angiomas involve the leptomeninges and skin of the face typically in the ophthalmic and maxillary distributions of the trigeminal nerve. Oral manifestations of the disease may vary considerably and changes in morphology and histology of gingiva, periodontium and pulp have been reported. However the most common feature is a gingival

hemangiomatous lesion usually restricted to ipsilateral maxilla, mandible, floor of mouth, lips, cheeks, palate and tongue[6]. Developmental disorders are more common when angiomas are bilateral[7].

CASE REPORT

An eleven year old boy from Bargarh was brought to our pediatric emergency who presented with left sided tonic colonic seizures with facial twitching lasting for 45 minutes associated with frothing from mouth, loss of consciousness and left sided hemiparesis (3/5). Episode of seizures was not associated with urinary or fecal incontinence. He had two similar episodes of seizures at the age of eight & nine year which was relieved spontaneously without any residual damage. He was born out of a non-consanguineous marriage as term following uneventful birth events and amongst the family of three siblings who were perfectly well. He was developmentally normal, vaccinated and belonged to middle socioeconomic group.

Fig-1 showing portwine stain on rtight lower eyelid

Physical examination revealed an afebrile adolescent with pulse rate of 90 per minute, respiratory rate of 24 per minute and his blood pressure was 98/70mmHg. There was port-wine stain under right lower eyelid not extending to the right side of face (**Fig- 1**). He was drowsybut arousable with Glasgow-coma scale of 12/15, upper motor type of left facial nerve palsy with left sided hemiparesis. Rest of neurological and physical examination was unremarkable. Ophthalmologic examination revealed glaucoma of left eye (intra ocular pressure =24mmHg)for which he had undergone surgery(trabeculectomy).

Hematological and biochemical profile was within normal range. Skull radiograph was normal. Non contrast CT scan brain showed area of intracranial calcification in left parieto-occipital lobe with prominent cortical sulci(**Fig- 2**).MRI of brain showed left cerebral hemiatrophy over temporo-occipital region , cortical calcification of left lateral temporal region and left posterosuperior parietal region.

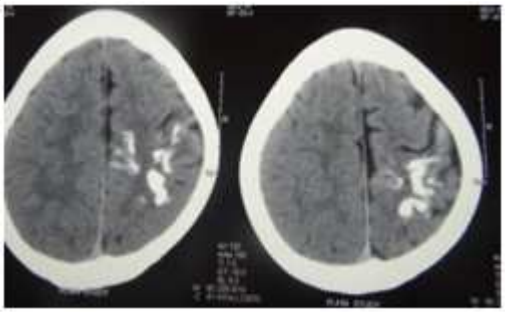

Fig-2 showing intracranial calcification & prominent sulci

Contrast MRI showed extensive leptomeningeal enhancement over left occipito – temporo-parietal regions (**Fig- 3**).EEG (Electroencephalogram) showed intermittent beta slowing prominent over left side (**Fig- 4**).

Immediately after transfer to our emergency unit, peripheral venous access was established. Appropriate anticonvulsants, antibioticsand fluids were administered. His progress was

satisfactory. Seizures were controlled and his residual neurological deficit resolved within 6 hours. He was **(Fig-3)** conscious and started taking orally. On 4thday of admission, child was discharged on anticonvulsant therapy and was kept on our regular follow-up.
Features suggestive of Sturge-Weber syndrome in this child were facial port-wine stain, focal seizures opposite to the side of nevus, glaucoma , hemiparesis and intracranial calcification.

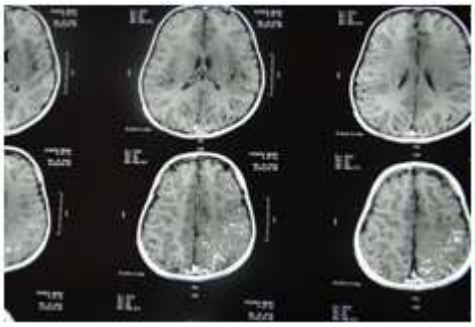

Fig-3 showing cerebral atrophy , calcification & leptomeningeal enhancement

Fig-4 showing EEG of left sided beta slowing

DISCUSSIONS
SWS is referred to as complete when both CNS and facial angiomas are present and incomplete when only one area is affected without the other. The Roach Scale is used for classification, as follows[8]
Type I - Both facial and leptomeningeal angiomas; may have glaucoma
Type II - Facial angioma alone (no CNS involvement); may have glaucoma
Type III - Isolated leptomeningeal angioma; usually no glaucoma.
According to the above criteria, our case is complete Type I SWS. Characteristically leptomeningeal angiomas occur as unilateral lesions affecting the pia arachnoid membrane over the posterior temporal, parietal and occipital areas[9]. It commonly shows abnormal blood flow pattern as venous occlusion, thrombosis, vasomotor phenomenon and vascular steal phenomenon resulting in cortical ischemia. This in turn gives rise to epileptic convulsive crisis, transient hemiparesis, gliosis and progressive deposition of calcium salts. These calcifications produce a characteristic double contoured “tram-line” appearance following the convolutions of cerebral cortex. Brush field and Wyatt stated that these tram- line calcifications are pathognomonic of SWS[5]. These calcifications are gyriform and curvilinear and most commonly seen in parietal and occipital lobes as seen in our case. These are seen best in the lateral skull view with affected side closer to the film. CT scan shows calcifications in the areas of atrophy[7].
The most evident clinical manifestation is presence of nevus flameus or Port-wine stains on the face within the distribution of Trigeminal nerve especially the ophthalmic division. They are present since birth and may range from small red macules to large red patches which blanch on pressur[10]. A large variation has been reported in their pattern of occurrence. They occur more commonly on right side and do not extend over midline. They can be bilateral or completely absent or may extend to neck, limb and other parts of body[11]. However only

carriers of port-wine stains along the ophthalmic branch develop the syndrome in its classical form as presented in our case.

Although the precise pathogenesis is unknown, it is thought to be the result of anomalous development of the primordial vascular bed during early stages of cerebral vascularization[2]. Sturge-Weber syndrome has been reported in neonates, as well[12]. Cases have also been reported with the sole presentation of headache[13]. However, a few cases without facial nevus have been reported as well[3].

Our patient had facial nevus. CT and MRI scans show diagnostic intracranial calcification[2]. Indocyanine green angiography can provide information that is not detected by clinical or fluorescence angiographic examination in patients with Sturge-Weber syndrome. We could not have Indocyanine green angiography due to lack of diagnostic facilities. This may be important and sensitive in detecting the diffuse choroidal hemangioma associated with Sturge-Weber syndrome[14].Neurological deficit is caused by the intracranial vessels malformation. In our patient, ophthalmologic examination of the left eye showed congestion of blood vessels, initial compensated glaucoma with increased ocular tension of 24 mmHg. Glaucoma is the most common serious eye problem of SWS, with a reported incidence of 30-70%[15]. Glaucoma may be present at birth or develop later. Pressure within the eye may damage the optic nerve, usually in the eye on the same side as the birthmark[16]. The reason for this increased eye pressure (glaucoma) may be the result of the outflow obstruction by a vascular malformation of the front area of the eye[17]. In cases of development delay and mental retardation, 50 to 60% of patients with Sturge - Weber syndrome are affected .

Convulsionsoccur in approximately 75% of patients, and 75% of the seizures appear within the first year of life. Most cases with Sturge-Weber syndrome are not life threatening. This is a progressive disease, associated with continuous neurological decline[18]. With vigorous control and treatment of symptoms, such as seizures, visual problems, paralysis and mental disorder, quality of life can be preserved.

Treatment involves early control of seizures and prevention of complications [13]. The parents of all the diagnosed patients must receive counseling concerning the potential risk of affected offspring. Parents should be educated about the potential complications of the disease as well.

CONCLUSIONS

The large spectrum of clinical manifestations of Sturge-Weber syndrome shows its multifactorial nature and difficulty in diagnosis. As the exact etiopathogenesis is not known, its prevention is difficult and its early diagnosis is essential.

In our case report we emphasize that regular treatment ofthe patient with Sturge-Weber with valproic acid results in long term seizure free interval. A successful early treatment results in control of seizures and prevention ofcomplications. Additionally, we strongly acclaim the proper counseling and support in addition to drug treatment can provide warmth to patients and their family to improve the outcome of the treatment.

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