Case Report

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Sturge Weber Syndrome - A Rare Cause of Childhood Epilepsy

Parthvi Patel[1] Prasad Muley[2*]

[1]Post Graduate Student, Department of Paediatrics, Smt. B.K. Shah Medical Institute and Research Centre, Vadodara, Gujarat, India.
[2]Professor, Department of Paediatrics, Smt. B.K. Shah Medical Institute and Research Centre, Vadodara, Gujarat, India.

ABSTRACT

Background: The Sturge Weber syndrome (SWS) or Encephalotrigeminal Angiomatosis is a congenital, non-hereditary rare condition of unknown etiology. The classic pathognomonic features of disease include angioma of the leptomeninges extending to cerebral cortex with ipsilateral angiomatous lesions, unilateral facial nevus usually affecting one division of trigeminal nerve and convulsions. Here, a case of 4 year old male child who presented with congenital skin lesions, convulsions and glaucoma that was diagnosed as type I SWS is being reported.

Keywords: Sturge Weber syndrome, Angiomatosis, Epilepsy.

INTRODUCTION

Sturge Weber Syndrome (SWS) also known as encephalotrigeminal angiomatosis, is a rare sporadic non famalial neurocutaneous disorder that affects the meninges (most often the pia mater and acrachnoid mater) of the brain and the skin of the face unilaterally. In rare cases the involvement may be bilateral. Incidence has been estimated to be 1 per 50,000[1] without any sexual predilection. The disease is caused by failure of regression of embryonic blood vessels at the appropriate time of development. Persistence of these residual blood vessels results in the formation of angiomas on the face, in meninges and in the ipsilateral eye. The angiomas of the face are referred to as port wine stains and are commonly seen in the opthalmic and maxillary divisions of the trigeminal nerve[2]. 75 to 90 percent of all children with the syndrome present with seizure by the age of 1 year and severity worsen with increasing age. About one-third of children with the syndrome present with congenital glaucoma on the side with a port wine stain. Here a case of type – I Sturge Weber Syndrome with unilateral glaucoma is presented.

CASE REPORT

A 4 year old male presented to the Department of Paediatrics with a complaint of red colour cutaneous macule over left upper half of face including both left upper and lower eye (Figure 1). The lesion was present since birth (approximately size 2×2 cm) which gradually increased in size with darkening in color over the period of time. He also had h/o multiple episodes of right sided focal convulsions involving both right upper and lower limb since one month of age without loss of consciousness. Child's development was normal for his age. Family history was not found to be significant.

On general examination the left eye was bigger than right eye. On further ophthalmic evaluation it was diagnosed as congenital glaucoma which had progressed to secondary glaucoma. Rest of systemic examination was within normal limits.

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***Correspondence**: **Dr Prasad Muley.**
Department of Paediatrics, Smt. B.K. Shah Medical Institute and Research Centre, Vadodara, Gujarat, India.
E-mail: muleyprasad123@gmail.com

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All routine investigations were normal. MRI brain both plain and with contrast was suggestive of leptomeningeal vascular malformation on left fronto parietal occipital region with reduction of size of left cerebral hemisphere and patchy gyrial calcification (Figure 2). Patient was treated with antiepileptic carbamazepine and glaucoma was treated as per ophthalmology protocol.

DISCUSSION

Port wine stains represent hamartomatous capillary malformations and are named so due to the deep red hue that they leave on the skin or mucosa[3]. Characteristically, leptomeningeal angiomas occur as unilateral lesions affecting the pia arachnoid membrane over the posterior temporal, parietal and occipital areas[4]. Usually it is a static lesion but rarely may it be progressive in nature.

SWS is referred to as complete when both CNS and facial angiomas are present. The Roach Scale is used for classification for SWS which is as follows[5].

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, the present case belonged to Type I SWS category.

This type of SWS commonly shows abnormal blood flow pattern as venous occlusion, thrombosis, vasomotor phenomenon and vascular steal phenomenon occur resulting in cortical ischemia. This in turn gives rise to convulsive, 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 and are pathognomic of SWS. These calcifications appear after the patient reaches 2 years of age and remain stationary after second decade of life. These calcifications are gyriform and curvilinear and most commonly seen in parietal and occipital lobes as seen in MRI of the present case. 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. Certain diseases that mimic SWS can be differentiated with the help of proper history and detailed investigations.

Klippel-Trenaunay-Weber syndrome can be differentiated from Struge Weber by formation of solid visceral tumours primarily in the kidney, adrenal gland or liver. Beckwith-Wiedemann syndrome consists of facial port wine stain and a number of other visceral symptoms which is useful in distinguishing this disorder from Sturge Weber syndrome. Dyke-Davidoff-Masson syndrome exists when significant atrophy occurs in one hemisphere during infancy. It may be difficult to differentiate between this disorder and Sturge Weber syndrome in the absence of a clearly defined port wine stain. Siderosis also results in atrophy of one cerebral hemisphere that is similar to that seen in Sturge Weber syndrome. Imaging studies (MRI with contrast) that outline cerebral vasculature are necessary to differentiate between the disorders. MRI will show normal vascular structures with siderosis while abnormal results are seen with SWS[6].

CONCLUSION

SWS is A rare cause of epilepsy in children but can be easily diagnosed with proper examination and high degree of suspicion. It can be differentiated from other syndromes easily by appropriate investigations. As the exact etiopathogenesis is not known, its prevention is difficult and its early diagnosis is critical, since it allows the control of future complications, mainly those relating to the central nervous system and overall quality of life can be improved.

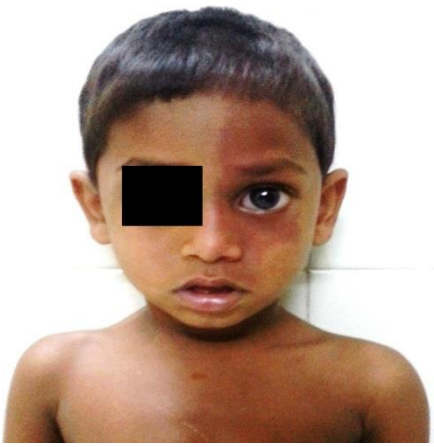

Fig 1: Port wine stain on left upper quadrant.

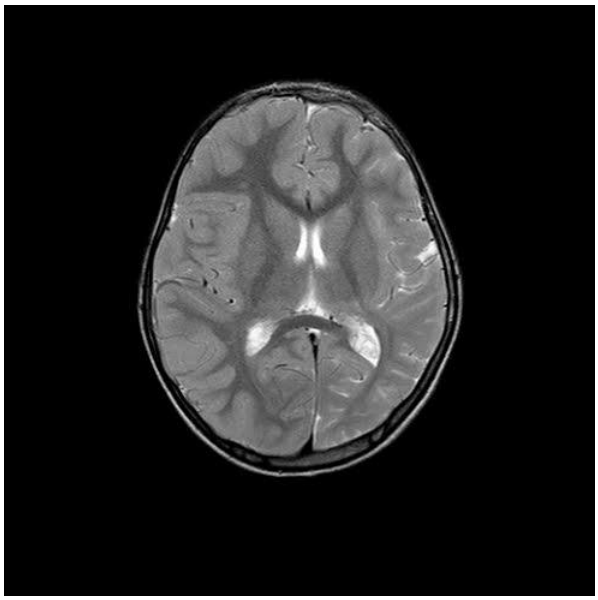

Fig 2: MRI brain with left sided atrphy with gyrial calcification.

CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

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