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Sturge-Weber Syndrome in a Neonate: A Case Report

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ABSTRACT

Sturge-Weber syndrome (SWS) is a rare neurocutaneous disease which affects primarily the brain (pial angiomata), skin (facial port-wine-stained birthmark) and eyes (glaucoma). [1]. SWS is a congenital, nonfamilial and sporadic condition caused due to somatic mutation in GNAQ gene located on the long arm of chromosome 9. The incidence of SWS is 1 in every 20,000 to 50,000 live births.[2]. Symptoms of this syndrome are commonly detected in infancy. The most common somatic mutation is activating R183Q *GNAQ* somatic mutation, recent studies also show association of GNA11 and *GNB2* somatic mutations related to SWS.[3]

Sturge–Weber Syndrome (SWS) also known as encephalotrigeminal angiomatosis, is a rare congenital, non-hereditary disorder affecting the brain, orofacial, eyes and skin. Involvement is prominently unilateral but can also be bilateral. The pathogenesis of this syndrome is the incomplete regression of Embryonic blood vessels at the appropriate time of development. Therefore, it leads residual blood vessels to form angiomas on face, in ipsilateral side of meninges, and in the ipsilateral eye [1]. Port wine stains are the cutaneous angioma occurring along dermatomes which the ophthalmic and maxillary division of the trigeminal nerve, the fifth cranial nerve supplies. Recent investigations include Brain magnetic resonance imaging, and adjunctive electroencephalography. However, there are insufficient studies about the sensitivity, specificity, negative and positive predictive value of magnetic resonance imaging and electroencephalography and whether screening improves seizure recognition.[4]. There is lack of evidence that a presymptomatic Sturge-Weber syndrome diagnosis with magnetic resonance imaging has better neurodevelopmental outcomes. In Sturge-Weber syndrome, neurodevelopmental outcomes hugely vary upon prompt recognition of neurologic red flags as well as early seizure control.[5]

In this case 26 Day old female baby was referred to our hospital i/v/o convulsion with sepsis with aspiration pneumonia and fever & was investigated for Sturge Weber syndrome.

Keywords: *Convulsion, Developmental delay, Dystrophic calcification, Facial Naevus, Port wine stain, Tram track appearance, Neurocutaneous marker, Seizures, Sturge-Weber Syndrome.*

INTRODUCTION

Sturge–Weber Syndrome (SWS) is a congenital, sporadic disorder affecting the brain, eyes and skin, also known as encephalotrigeminal angiomatosis. Schirmer described the first case of SWS. In 1879, William Allen Sturge described this disease in detail as well as proved association between neurologic symptoms with the dermatological and ophthalmic changes of the disease. In 1929, Frederick Parkes Weber described the radiologic features seen in patients of SWS.[1]. Hence known by the name of Sturge-Weber Syndrome.

Unilateral involvement is seen most commonly. *GNAQ* somatic mutation and the activating of R183Q is the most common somatic mutation of SWS.[3] Sturge Weber syndrome demonstrates an intracranial vascular anomaly, leptomeningeal angiomatosis, most commonly involving occipital and posterior parietal lobes. Facial cutaneous vascular malformations, seizures, and glaucoma also are encountered commonly. Gradually leptomeningeal angiomatosis causes blood stasis leading to ischemia which results in calcification as well as laminar cortical necrosis of the vessels. The clinical course is highly unpredictable; some children experience intractable seizures, mental retardation, and some have recurrent stroke-like episodes.[6] Port wine stains are vascular rather than neurologic in embryologic origin, and facial PWBs at highest risk for brain involvement of SWS involving the forehead location.[7] Studies prove that epilepsy surgery has good postoperative seizure-free rate and favourable cognitive and motor functional outcomes and showed acceptable safety for SWS patients with epilepsy. Modified hemispherotomy is a relatively less invasive and safer type of hemisphere surgery rather than traditional anatomic hemispherectomy with similar surgical results.[8]

CASE REPORT

26 days full-term female born to a primigravida mother via normal vaginal delivery with birth weight of 1.85 kg with delayed cry after birth. then kept in NICU for 2 days i/v/o low birth weight then discharged at home; got admitted at civil hospital i/v/o not feeding well and for abdominal distension for 2 days and referred to our hospital i/v/o vomiting after feed with sepsis with aspiration pneumonia with convulsion was admitted in NICU and intracath inserted blood investigation sent and iv antibiotics, antiepileptics started. baby put on O2 by nasal prongs @ 1L/min. baby transfused 1 O. PCV. Baby was initially kept NBM then started on OGT feeds 2 ml/2hrly, increased to full OGT feeds and then to oral full feeds; off O2. Baby maintained saturation off O2. baby then shifted to mother side. After the mother was trained regarding feeds. Investigation showed haemoglobin of 6.8 gm/dl before transfusion which increased to 12.9 gm/dl at discharge. TLC was 16.1 k (before antibiotic) and 4.9 k (at discharge) , Platelet counts – 81 k/ul ,MCV – 92 FL, CRP – 51 mg/dl, Bilirubin T/I – 3.77/12mg/dl ,Sodium – 139mmol/l , Potassium- 5.2mmol/l ,Urea– 16.7mg/dl. Child had facial feature of unilateral dark reddish spot-on left side known as the Port wine stain which is a neurocutaneous marker.

FIG 1: Port wine stain on Upper left side of the forehead which is a Neurocutaneous marker.

MRI BRAIN shows Left cerebral hemi atrophy with gliosis in the left hemisphere with cystic encephalomalacia changes, shrunken and flattening of cortex and dilated sulcal spaces and lateral ventricle with ipsilateral falcine displacement. Dystrophic calcifications in atrophic parenchyma are noted with tram track appearance as shown in figure 2.

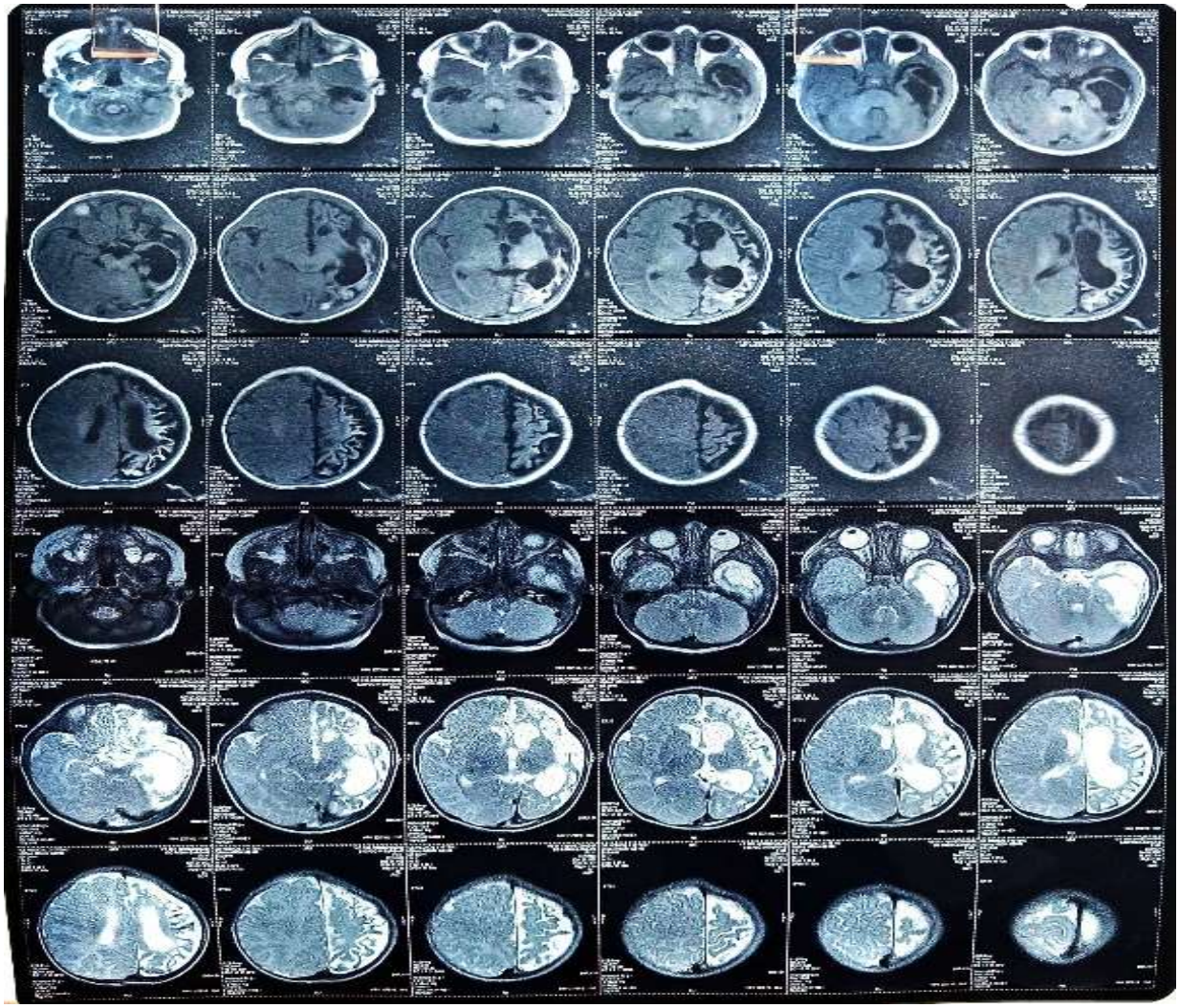

Fig 2: Gyriform sulci -tramtrack sign present in left lobe of brain on MRI

DISCUSSION

Sturge-Weber syndrome is congenital, non-familial, also a sporadic developmental disorder. Its etiology is inadequately understood but studies show that it is caused by somatic mutations in the GNAQ gene which is located on long arm of chromosome 9[9]. Sturge Weber is a rare disorder. The incidence is roughly estimated to be 1 in 20,000-50,000 live births. It is widely distributed with no racial predilection with similar occurrence in males and females. Accurate pathogenesis is inadequately understood but theories do exist. Some theories postulate occurrence of leptomeningeal and facial angiomata due to the persistence of undeveloped sinusoidal vascular channels. Another theory suggests deficient development of superficial venous drainage along with subsequent compensatory dilatation of small venous channels. This relative insufficiency of superficial cortical veins, there is shunting (steal) of blood to deep venous system by the enlarged medullary veins which gradually results in stasis and ischemia. Ultimately, these changes finally result in epileptic convulsive crisis, transient hemiparesis, gliosis, and progressive deposition of calcium salts as seen in above case[9].

The cortical calcification forms the characteristic double contoured "tram-track" appearance which is an MRI finding as shown above. The characteristic facial nevus comprises of multiple thin-walled vessels that are similar to capillaries. According to recent studies, a somatic mutation in a nucleotide transition in gene GNAQ located on long arm of chromosome

9 (specifically 9q21) causes this syndrome. Activity in pathways transmitting signals from a subset of G protein coupled receptors (GCPR) is increased due to this mutation [10]. True mechanism by which leads this activation and results in such port-wine birthmarks and SWS is not well known. The clinical features of this syndrome can be neurological and non-neurological or either of them. The neurological symptoms are seizures, hemiparesis, headaches, visual field deficits, cognitive impairments. Other features include early handedness, gaze preferences also stroke-like episodes. Non-neurological symptoms are behavioural and emotional derangement, endocrine problems, learning difficulties, and other medical conditions. The use of low-dose aspirin and vitamin D in treatment for SWS is shown to be effective by recent retrospective studies. Prospective drug trials also support use of cannabidiol and Sirolimus. Low-dose aspirin and antiepileptic drugs administered before any symptoms arise, have delayed the onset of seizure in few patients according to some trials. Studies show that 77% of patients had ocular conditions which comprised commonly glaucoma (46%), strabismus (23%) and choroidal angioma (23%).[11] 100% of the cases reported with epilepsy, with most common partial seizures (simple or complex) (62%). Seizure control was highly variable; 31% of the patient were treated with more than 3 drugs, 15%cases required 3 drugs, and 31% needed 2 drugs, while 23% had good seizure control with monotherapy. One patient was seizure-free after left hemispherectomy. EEG (electroencephalograms)mostly presented spikes, polyspikes, and wave spikes in the brain lobes affected by leptomeningeal angiomatosis (46%). Other neurological symptoms included psychomotor retardation (46%), and mental retardation (46%), hemiparesis (39%), recurrent headaches (39%), stroke-like episodes (23%). 85% of patient had leptomeningeal calcifications in their MRIs in which about 70% had increased calcification. 54% of cases were treated with aspirin showed good outcome.[12] Astoundingly only 3% in patients with a facial port-wine stain had Sturge-Weber Syndrome. Laser treatment have poor results in facial dermatomal port-wine-stained patients. According to nerve dermatomes, port-wine stains at the V3 level had better outcome whereas V2 worst to laser. [13] This syndrome can also manifest neuromuscular, ocular, dermatologic as well as oral features.[14]

CONCLUSION

Sturge Weber Syndrome (SWS) is a rare congenital neurocutaneous disorder due to somatic mutation of GNAQ gene on chromosome 9, with classical symptomatology and neurological imaging findings like “Tram-track” appearance on MRI or gyriform calcification. Neurocutaneous marker like port wine stain (facial nevus flammeus), ocular findings like congenital glaucoma, neurologically seizure being the most common. Pathogenesis includes incomplete formation of underlying anomalous leptomeningeal venous plexus with lack of normal cortical venous drainage ipsilaterally.

Combined team of Neurosurgeon, Neurophysician, Ophthalmologist, Cosmetologist, Physiotherapist, and radiologist can provide the symptom-based treatment.

Treatment options include pharmacological-low dose aspirin, antiepileptics; surgery like Hemispherectomy for seizures, Glaucoma surgeries and laser therapy for port wine stains.

Early investigation and detection of this syndrome has good outcome whereas more studies are required for the definitive treatment of SWS.

Declaration by Authors

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