Scholars Journal of Medical Case Reports **ISSN 2347-6559 (Online)**
Sch J Med Case Rep 2014; 2(9):605-608 **ISSN 2347-9507 (Print)**

(An International Publisher for Academic and Scientific Resources)
www.saspublishers.com DOI: 10.36347/sjmcr.2014.v02i09.009

A Three Year Old Female Child with Type -1 Sturge-Weber Syndrome: A Case Report

J. Narsing Rao[1], Swathi Chacham*[2], U.Narayan.Reddy[3], Naila mazher[4], Afreen[5], Jakkampudi NagaSravani[6]
[1]Professor, [2]Assistant Professor, [3]Head of the Department and Professor, [4, 5,6]Junior Resident, Department of Paediatrics, Deccan College of Medical Sciences, Princess Esra Hospital, Hyderabad-500 002, India

***Corresponding Author:**
Name: Dr. Swathi Chacham
Email: swathi.m.lahiri@gmail.com

Abstract: *Sturge-Weber Syndrome (SWS)* is a neuro-cutaneous disorder characterized by angiomas of face and the central nervous system. These angiomas, are the hallmark of *SWS*. While other phacomatoses are hereditary in nature, *SWS* occurs sporadically and with a frequency of 1 in 50,000. Mutations in GNAQ gene result in altered structure and innervation of cerebral vasculature. A three year old female child presented with recurrent seizures from infancy. General physical examination revealed several hyper-pigmented macules over the trunk and both lower limbs. A purple coloured port wine stain was found on the right side of the face. Imaging (Contrast enhanced CT) of the brain showed extensive pial enhancement in the right parieto-occipital lobe. MRI brain depicted ipsilateral enlargement of choroid plexus, characteristic finding of *Sturge weber syndrome*. We report a rare case of type 1 *SWS* with ipsilateral choroid plexus enlargement and typical pial enhancement.
Keywords: *Sturge-Weber Syndrome*, Port wine stain, Neuro-Cutaneous Disorder, Encephalo-Trigeminal Angiomatosis, Fibronectin

INTRODUCTION

Sturge-Weber Syndrome (SWS) is a rare neurocutaneous syndrome characterized by angiomas involving the face and central nervous system (CNS) [1]. The distinguishing features of *SWS* include seizures, hemianopia, hemiplegia, headache and developmental delay [2, 3]. The reported incidence is 1 in 50,000 [1, 3]. *SWS* have no gender predilection. These changes result from a somatic mutation, in GNAQ gene [4]. This leads to alterations in structure and innervation of blood vessels.

CASE REPORT

A three year old female child admitted with status epilepticus to the emergency department. The child manifested with tonic clonic movement of the left upper and lower limb with deviation of eyes to the left side. But, bowel and bladder incontinence was absent. The seizures required three antiepileptic drugs. The child had postictal drowsiness up to three hours. History of consanguinity and similar complaints were absent in the family. Antenatal and delivery events remained uncontributory. First episode of seizures manifested on fifth day of life, requiring hospitalization and antiepileptic therapy. Despite being on medications, the child suffered from recurrent seizure episodes. The duration and severity of the seizures increased with time. These events co-existed with an underlying developmental delay (developmental quotient-50). Examination showed purple colored port wine stain on the right side of face, involving the forehead, upper eyelid and cheek (as shown in the Fig. 1A). Hyperpigmented macules over the back and abdomen accompanied hemangioma over the feet (as shown in Fig. 1B). Fundus examination showed a normal study. CSF analysis remained normal. These clinical findings suggested *Sturge Weber syndrome* and the child underwent further investigations.

Plain CT had unremarkable findings, with no evidence of volume loss or calcification. Contrast enhanced CT brain (CECT) disclosed extensive pial enhancement in the right parieto-occipital lobe (as shown with arrow in Fig. 2A) and ipsilateral choroid plexus enlargement (as shown with a star in Fig. 2B). MRI (Magnetic resonance imaging) of the brain revealed subtle subcortical white matter hyper intensities (as shown in Fig. 3A, 3B) with ipsilaterally prominent choroid plexus (as shown in Fig. 3B, 3C). SWI (susceptibility weighted imaging) axial image depicted increased subpial vascularity in the right parieto-occipital lobe (as shown in Fig. 3D). These typical findings confirmed *SWS*. The child received supportive and symptomatic care.

Ethical Approval was taken from institute ethics committee.

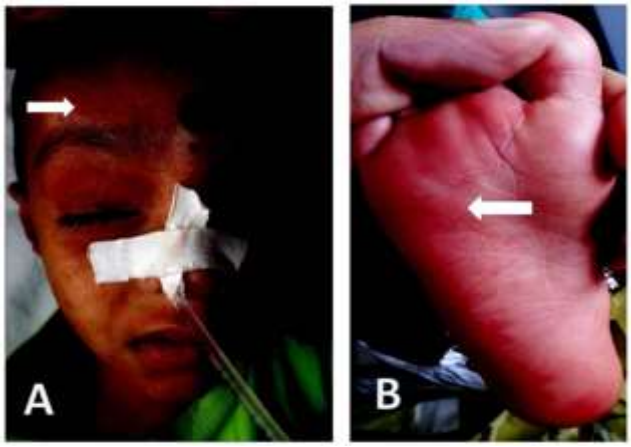

Fig. 1A: Clinical photograph showing purple colored port wine stain on the right side of face, involving the forehead, upper eyelid and cheek (as shown with arrows). **Fig. 1B:** Clinical photograph revealing hemangioma over the feet (as shown with arrows)

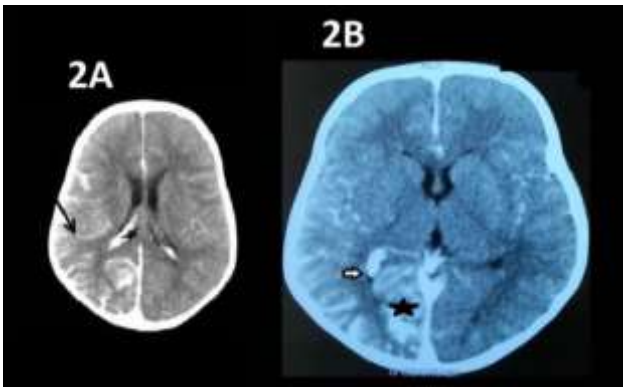

Fig. 2A: Contrast enhanced CT scan shows extensive pial enhancement in right parieto-occipital lobe (as shown with arrow) and ipsilateral enlargement of choroid plexus (as shown with star). **Fig. 2B:** Contrast enhanced CT scan shows ipsilateral enlargement of choroid plexus (as shown with star)

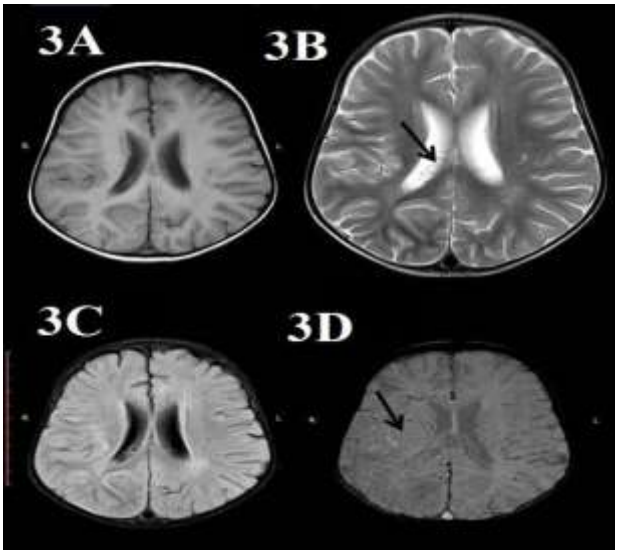

Fig. 3A: MRI brain, T1W axial image demonstrates few punctate hypo intensities in white matter. **Fig. 3B:** MRI brain, T2 weighted axial image shows subtle subcortical white matter hyper intensities with ipsilateral prominent vessels in choroid plexus (as shown with arrow). **Fig. 3C:** MRI brain, FLAIR axial image shows subtle subcortical white matter

hyper intensities with ipsilateral prominent vessels in choroid plexus. **Fig. 3D:** MRI brain SWI axial image reveals increased sub pial vessel density in right parieto- occipital lobe (as shown with arrow)

DISCUSSION

Sturge-Weber Syndrome (SWS), also known as encephalo-trigeminal angiomatosis, is a rare sporadic neuro-cutaneous syndrome [1, 3]. Various studies state an incidence of 1 in 50,000 [1] for SWS. It is characterized by congenital angiomas of the face (nevus flammeus/port wine stain).These are distributed along the ophthalmic or mandibular division of the trigeminal nerve [1, 2]. This port wine stain, a congenital vascular malformation of the dermis, is an initial presentation of SWS [5]. The most common manifestations of this syndrome include seizures (93%) followed by developmental delay and mental retardation (50%). Abnormal development of the vasculature of the eye and the central nervous system accounts to these observations. Other associated symptoms and signs comprise of ocular abnormalities (44%) and hemiparesis (33%). Seizures predominantly occur on the contralateral side of the lesions.

This constellation of findings is characteristic of *Sturge-Weber syndrome* [5]. Likewise, the index case presented with contralateral seizures, developmental delay and port wine stain (PWS). The prevalence of port-wine stains (PWS) is 3 per 1000 live births [3]. Typically, these PWS are faint, pink macules, which progressively darken with age. They may have a well-delineated border or may be diffuse in nature. The standard pattern of distribution is unilateral. However, bilateral involvement or additional port wine lesions in other parts of the body can occur, infrequently [1, 3].

The Roach scale describes three types of *SWS* [6], which are illustrated below:

Type I-Both facial and leptomeningeal angiomas, with / without glaucoma,
Type II-Facial angioma alone (no CNS involvement) with /without glaucoma,
Type III-Isolated leptomeningeal angiomas, usually no glaucoma.

As the index case presented with facial and leptomeningeal angiomas, it denotes type I *SWS.* This syndrome probably occurs from malformation of cutaneous, ocular and cerebral vasculature during early embryonic phase [5]. Intricate molecular interactions lead to altered growth of blood vessels in SWS. This, in turn results in distorted cerebral blood flow, which results in neurologic manifestations [5]. Fibronectin is a high molecular weight glycoprotein which binds to integrins, spanning the cell membrane. This enhances angiogenesis, vascular remodelling, and innervation density [7]. Studies have quoted a differential expression of fibronectin in various blood vessels of the brain in SWS, which might have lead to altered blood flow. In this syndrome, meningeal vasculature had reduced fibronectin expression, in contrast to parenchymal vessels [7].

The prime imaging modalities to diagnose *SWS* include Skull radiograph, Computerized tomography (CT), magnetic resonance imaging (MRI) and functional imaging (PET scan) [8]. Plain skull radiograph shows calcifications, which can also be seen in plain CT brain. Contrast enhanced CT reveals enhancement of pial vasculature along with enlarged choroid plexus. MRI discloses cortical and pial calcifications along with vascular angiomas and cerebral hemiatrophy. Underlying complications like venous thrombosis are better detected with MRI brain and MR angiography. However, Newer imaging modalities like perfusion MRI, single photon emission CT and positron emission tomography (PET) imaging studies aid in the early diagnosis of *Sturge-Weber syndrome* and its complications such as impaired cerebral circulation, more so during prolonged seizures [9]. The index child had these typical radiological features of increased leptomeningeal enhancement with enlarged choroid plexus along with characteristic clinical features of *SWS.*

The possible differential diagnosis of *SWS* include Rendu-Oslar-Weber syndrome (characterized by abnormal dilatation of terminal vessels of skin, mucosa), Maffuci syndromes (multiple angiomas of skin and chondromas of bone) and Von Hippel Lindau disease (a familial syndrome involving hemangioblastoma in the retina and cerebellum, and renal cysts). However the characteristic facial port wine stain along with CECT and MRI brain findings in the index child confirmed *SWS.*

The major reported complications of SWS are cognitive [9] impairments ranging from mild learning disabilities to severe deficits. Impaired cerebral blood flow can result in deep venous thrombosis [10]. Although *SWS* predominantly manifests in infancy and childhood, it can even present in neonatal period [11].

SWS is a multisystem disorder with associated endocrine, psychiatric and ophthalmologic complications. Neurological progression occurs due to impaired blood flow and prolonged seizures. The prime stay of management is early detection and symptomatic and supportive treatment. The role of low dose aspirin to prevent venous thrombosis is still controversial [3]. Intensive measures to prevent seizures and strokes, in young children play a key role. In the current child every effort has been made to diagnose the CNS lesions and control the seizures. The success in the management of seizures with anti epileptic drugs varies, and at times may even require neurosurgical procedures

such as focal cortical resection, hemispherectomy, corpus callosotomy and vagal nerve stimulation (VNS), when the seizures are refractory. Cutaneous PWS is treated with dye laser photocoagulation, which helps in reducing the cosmetic blemish from the cutaneous vascular dilatation. However, the laser photo-coagualtion might increase the risk of glaucoma later on [12].

We emphasize that any cutaneous marker in a child with seizure should alert the physician for the possibility of a neuro-cutaneous syndrome. Also parents of the child should be counselled regarding the refractory nature of the seizures associated and the challenges in the management faced by the physician. Even though the treatment is supportive, early diagnosis and management may help to make the child's life comparatively better.

CONCLUSION

A rare case of *SWS* in three year old female child with characteristic port wine stain, refractory seizures, developmental delay and typical radiological findings.

ACKNOWLEDGEMENTS

Thanks to Dr.Taha, Dr.Aslam, Mr.Mujahid and the nursing staff for helping with the investigations.

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