Romanian Neurosurgery | Volume XXXI | Number 1 | 2017 | January - March

Article

# Sturge Weber Syndrome: review of literature with case illustration

Guru Dutta Satyarthee, Manik Prabhu, Luis Rafael Moscote-Salazar
INDIA, COLOMBIA

DE GRUYTER OPEN

DOI: 10.1515/romneu-2017-0019

*DOI: 10.1515/romneu-2017-0019*

# Sturge Weber Syndrome: review of literature with case illustration

**Guru Dutta Satyarthee[1], Manik Prabhu[1], Luis Rafael Moscote-Salazar[2]**

[1]Department of Neurosurgery, AIIMS New Delhi, INDIA
[2]Neurosurgeon-Critical Care, RED LATINO. Latin American Trauma & Intensive Neuro-Care Organization, Bogota, COLOMBIA

**Abstract**: Sturge-Weber syndrome (SWS) also called as encephalotrigeminal angiomatosis, is a sporadically occurring rare neuro-cutaneous syndrome, characterized by vascular malformation with capillary venous angiomas involving face, choroidal layer of eye globe and leptomeninges responsible for ophthamological as well as neurological signs and symptoms. Authors report an interesting case, a six year old girl, who presented with seizures, facial port wine stain and normal psychomotor development. CT scan showed left cerebral hemiatrophy, left frontal and parieto occipital calcification with cortical calcification in left high frontal convexity. Cranial MRI scan also confirmed finding of left cerebral hemiatrophy and also revealed presence of gyriform cortical calcification, prominent flow voids seen in left basal ganglia. Her seizure is well controlled with antiepileptic medication. The pertinent literature is reviewed and management of such cases is discussed briefly.

**Key words**: Sturge-Weber syndrome, seizure, port wine stain, development

## Introduction

Sturge-Weber syndrome is a rare disorder that occurs with a frequency of 1: 50,000 [1]. SWS is sporadic neuro- cutaneous disease, being characterized by presence of facial port-wine stain, ocular glaucoma and choroidal haemangioma in the eye, leptomeningeal angioma often involving occipital and posterior parietal lobes. This syndrome comprises of constellation of symptoms and signs i.e. including facial nevus, seizures, intracranial hemiparesis, calcification and association of mental retardation [2-8].

## Case illustration

A 6-year-old girl presented to our emergency services with history of left focal motor seizure with secondary generalization lasted about five minutes associated with loss of unconsciousness , frothing of mouth and urinary incontinence. She had six episodes of similar seizures over last two years, which were not associated without any post-ictal neurological deficits. For which, she was kept on phenylhydantoin but not controlled, however, with addition of phenobarbitone, seizures are well controlled and she had no

fresh seizure for last one year.

Parents noticed facial port wine discoloration at birth involving right upper part of face; baby was first issue of non-consanguineous marriage, delivered normally at full term. Perinatal history was unremarkable. She was vaccinated at our institute under supervision of paediatrician, as part of universal immunization programme against diphtheria, pertusis, tetanus and B.C.G. No history of trauma, drug allergy or exanthematous rashes. She had normal psychomotor development. At present patient has no neurological deficit. Patient has venous angiomas involving ophthalmic segment of right trigeminal nerve.

On physical examination, the pulse rate was 110 per minute, respiratory rate 24 per minutes and blood pressure 96/72 mm Hg. There was port-wine stain involving upper half of right upper face. She was incoherent to speech with Glasgow-coma scale of 14/15, with presence of left facial port wine venous angiona (Figure 1 A) both pupils were briskly reacting but uncooperative for fundus evaluation and rest of neurological examination was unremarkable. A peripheral venous access was made by putting intravenous canula, immediately after her arrival in emergency room. Blood sample were taken for blood sugar serum phenytoin level and blood chemistry. Seizures were controlled with intravenous midazolam. Hematological and biochemical profile carried out in emergency care unit was within normal range. Computerized tomography (CT) scan of the head revealed left cerebral hemi atrophy involving left frontal and parieto-occipital lobe with cortical-subcortical calcification (Figure 1 B, C, D) along with enlargement of left lateral ventricle choroid plexus.

Progress was satisfactory and her residual neurological speech deficit resolved within after two hours. Child was discharged on phenytoin and phenobarbitone. MRI scan of brain was carried out during follow-up period revealed, left cerebral hemiatrophy with gyriform cortical calcification, (Figure 2) prominent flow voids seen in left basal ganglia. (Figur 3)

Choroid plexus enlargement seen in left lateral ventricle. (Figure 4) No diffusion restriction on DWI images. Cerebellum and brain stem are normal. Hypertrophy of calvarium on left side also noted. (Figure 5) Ophthalmological evaluation was done, showed no evidence of ocular involvement. She is under our regular follow-up.

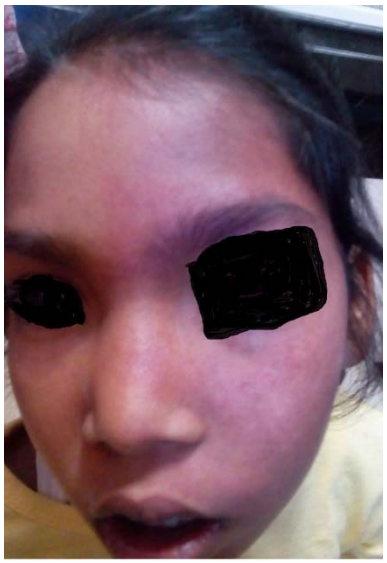
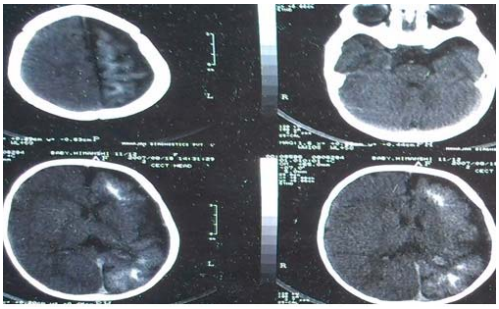
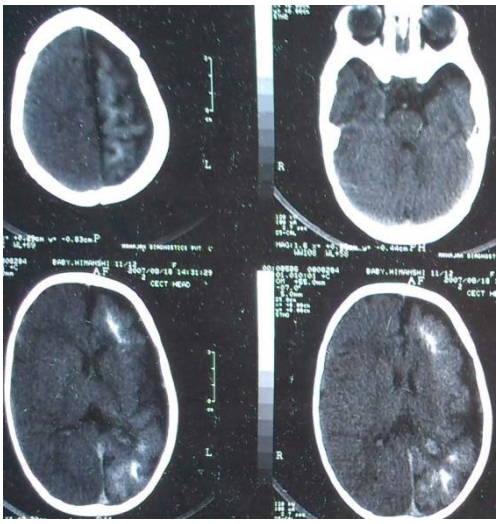


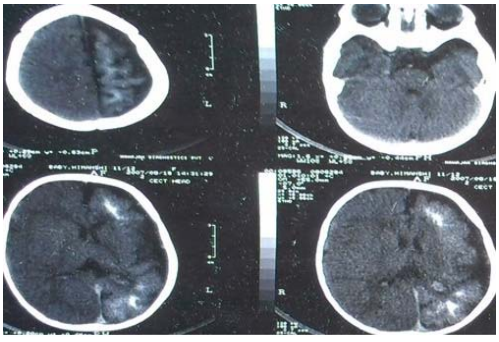
**Figure 1** A - Clinical photograph showing portwine venoios angioma over left side of face



B



C



D

**Figure 1** B, C, D - Contrast enhanced computed tomography head showing atrophy of left cerebral hemisphere with calvarial thickening in a six-year girl with Sturge Weber Syndrome with calcification

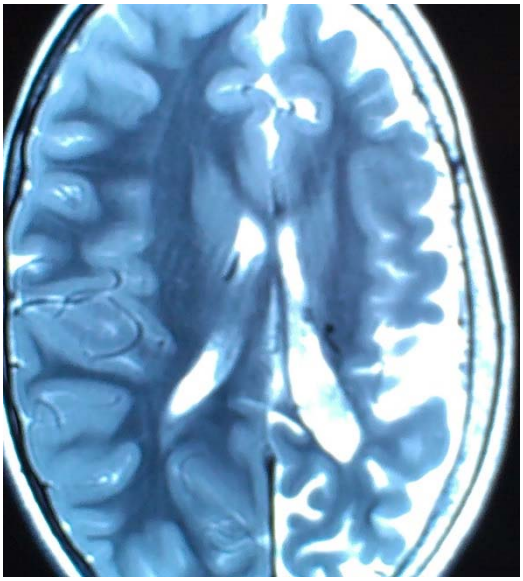


**Figure 2** - MRI brain axial T2 weighted image showing atrophy of left cerebral hemisphere

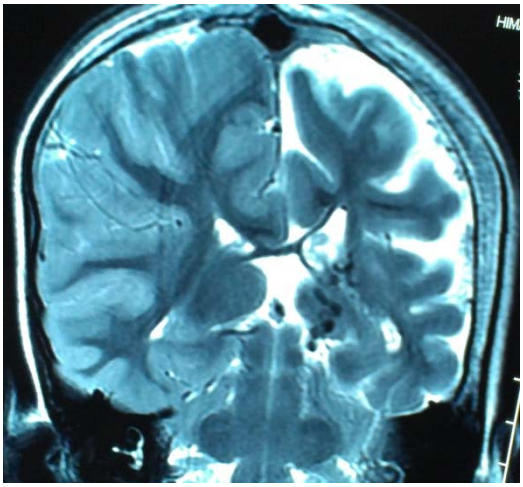


**Figure 3** - MRI brain of six -year- old female with Sturge Weber Syndrome coronal section, T2 weighted image showing atrophy of left cerebral hemisphere

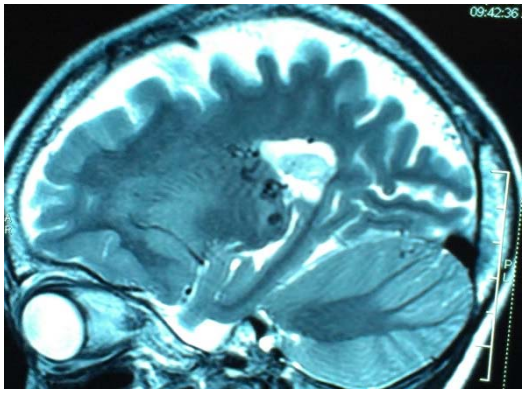


**Figure 4** - MRI brain of a six years girl, T2 weighted image showing presence of left cerebral hemispheric atrophy

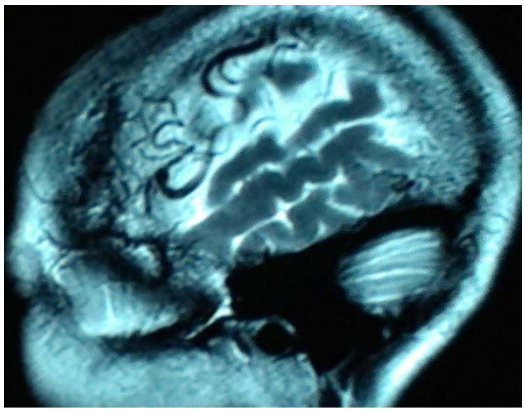


**Figure 5** - MRI brain parasagittal section, T2 weighted image T2 weighted image showing atrophy of left cerebral hemisphere with prominent veins in a case of Sturge Weber Syndrome

## Discussion

Sturge Weber syndrome (SWS) belongs phakomatosis disorders. SWS has constellation of congenital malformation including hamartomatous malformations and lesions involving eye, skin, and central nervous system may be specially affected. In1860, it was first described by Schirmer, further detailed specific description was provided by Sturge in the year1879. Weber, Dimitri and Wissing described the typical gyriform calcifications observed on plain X-ray skull. Krabbe is credited for correlating intracortical calcifications with gyriform calcifications on simple X-ray skull and hence also popularly called as Krabbe syndrome, Sturge Weber Dimitri syndrome, encephalotrigeminal angiomatosis and encephalofacial angiomatosis [3].

Sturge- Weber syndrome is a rare neuro-cutaneous syndrome with estimated incidence of 1 in 20000 to 1 in 50000 live births [9-16]. The characteristic manifestations of SWS are leptomeningeal angiomas, facial port wine venous angioma, and glaucoma. Leptomeningeal angiomas typically tends to involve the occipital and posterior parietal lobe, also considered as the hallmark; however, degree of involvement is highly variable and can be either very extensive to involve ipsilateral lobes of ipsilateral or even extending to involve contralateral cerebral hemispheres. Cutaneous vascular malformation usually occurs in the face, which is usually in the distribution of ophthalmic division of trigeminal nerve [4, 5].

Sturge Weber Syndrome is classified into three types. The type 1 includes angiomas involving face and leptomeningeal as well as the possibility of presence of glaucoma or choroidal lesions. Normally, only one side of the brain is affected. This type represents the commonest type. Type 2 involvement includes a facial angioma (port wine stain) with a possibility of glaucoma developing. However, it is not associated with any

evidence of brain involvement. The final type-3 typically associated with leptomeningeal angioma involvement but facial angioma is absent and glaucoma tends to occur rarely. This type is only diagnosed via brain scan. [14]

SWS may be complete type characterized by presence of skin and central nervous system manifestation, but. Incomplete types are also reported without associated common facial nevus [6]; current case had complete manifestations with presence of port wine stain over upper half on right side of face as well as central nervous system involvement.

Age of being clinically getting symptomatic for Sturge Weber Syndrome is highly variable, spanning from immediate after birth in the neonatal period to the adolescence, although delayed diagnosed as late as sixth decade is also reported. It is usually present at birth with a port-wine stain on the forehead and upper eyelid involving only one side of the face, or rarely the whole face. The birthmark colour can vary from light pink to deep purple depending on complexion of rthe cases, caused due to an overabundance of capillaries around the ophthalmic division of the fifth cranial nerve. Sturge Weber Syndrome is also associated with blood vessels malformation of the pia mater on the same side of the head as the facial angioma. This causes calcification of tissue and degeneration loss of neurons in the cerebral cortex.

Neurological symptoms include seizures, which usually begin in infancy and may worsen with advancing age. There may also be motor deficit or developmental delays and cognitive delays; about 50% cases may suffer with glaucoma.

Muniz reported a two days old baby with acute life threatening event ,who presented to an emergency department to seek management[7, 8]. Focal motor seizures are the most common neurological manifestation in SWS patients, usually present by the age of three years. Many patients have intractable seizures, which may eventually lead to motor deficits and mental retardation [9]. Our patient had normal motor and scholastic performance and had first seizure at age of four year.

Computed tomography of head typically shows curvilinear and gyriform calcifications presence in atrophic areas of brain and associated thickening of calvarial and contrast study shows leptomeningeal enhancement [10, 11]. Characteristic findings observed on cranial MRI include cerebral parenchymal atrophy, enlargement of medullary, periventricular veins and the enlargement of choroid plexus. And may have associated leptomeningeal, cortical gyriform abnormal signal or enhancement. Current case had, left cerebral atrophy with enlargement of periventricular veins with left lateral ventricle choroid plexus demonstrated on MRI brain, while cranial CT scan showed gyriform calcification with left cerebral atrophy and left sided calvarial thickening However other causes of seizure needs exclusion prior to labellling SWS as principal cause of seizure disorder. [12-14]

Important differential for intracranial calcifications at multiple site, cerebral hemiatrophy and includes

Cerebral arteriovenous malformation, infective pathology i.e. TORCH infection, neurocysticercosis, healed cortical infarct, post-radiotherapy induced changes [14-18]

Gupta et al reported a case of hypothalamic glioma occurring in a 7-year-old child, who presented with seizures and diabetes insipidus, underwent near total decompression of the hypothalamic glioma successfully using subfrontal approach. He had a stormy postoperative course due to status epileptics. He remained symptom and seizure free on antiepileptics at last follow-up after three years following surgery. [14]

Treatment revolves primarily around seizure control, with surgical resection only indicated rarely in refractory cases. Ophthalmological examination is also essential to identify and treat ocular involvement [18, 19]

## Conclusion

Prompt and aggressive seizure control with medication is usually sufficient to control seizure; however, in cases of medically refractory seizure surgery may be required. Further, seizure control is essential for psychological and cognitive well being, long term neurological outcome and quality of life of cases suffering with Sturge-Weber syndrome.

***Correspondence***
*Guru Dutta Satyarthee*
*E-mail: drguruduttaaiims@gmail.com*

## References

1. Thomas-Sohl KA, Vaslow DF, Maria BL. Sturge-Weber syndrome: a review. Pediatr Neurol 2004; 30: 303-10
2. Baselga E. Sturge-Weber syndrome. Semin Cutan Med Surg 2004; 23:87-98.
3. Jeevankumar B, Devinder Mohan Thapa, Sturge Weber syndrome: A report of three cases. Indian J Dermatol 2001; 46(3):185-87.
4. Zaki SA, Lad V. Sturge-Weber syndrome with bilateral facial nevus and early cerebral calcification. J Pediatr Neurosci. 2011 Jul; 6(2):114-5.
5. Comi AM. Pathophysiology of Sturge-Weber syndrome. J Child Neurol 2003; 18:509e16
6. Moe P, Seay AR. Neurologic and muscular disorders. In: Hay WW,Hayward AR, Levin MJ, Sondheimer JM, editors. Current Pediatric Diagnosis and Treatment, 16th edn. Singapore: McGraw Hill; 2003. p. 717-92.
7. Muniz AE. Sturge-Weber syndrome presenting as an acute life threatening event. Pediatr Emerg Care 2004; 20:610-2
8. Ferrari L, Coppi E, Caso F, Santangelo R, Politi LS, Martinelli V, Comi G, Magnani G. Sturge-Weber syndrome with an unusual onset in the sixth decade: a case report. Neurol Sci. 2012 Aug;33(4):949-50
9. Sydney Ch'ng, Swee T. Tan. Facial port-wine stains e clinical stratification and risks of neuro-ocular involvement. J Plast Reconstr Aes 2008; 61, 889-93.
10. Hu et al .MR Susceptibility Weighted Imaging (SWI) Complements Conventional Contrast Enhanced T1 Weighted MRI in Characterizing Brain Abnormalities of Sturge-Weber Syndrome. J Magn Reson Imaging. 2008; 28(2): 300–307.
11. Akinpar E. The tram track calcifications. Radiol 2004; 231:515-6.
12. Satyarthee GD, Mahapatra AK. Seizure as presentation of pituitary adenomas; series of four cases. Neurosciences Today 2002; 6: 167-169.
13. Rishabh Pasricha, Satyarthee GD. Seizure occurrence in spinal dysraphism operated cases: series of four cases with Literature Review American J Clinic Neurol Neurosurg 2015; 2, : 41-44.
14. Gupta DK , Satyarthee GD, Sharma MC, Mahapatra AK . Hypothalamic glioma presenting with seizures. a case report and review of the literature. Pediatr Neurosurg. 2006;42(4):249-53.

15. Moore GJ, Slovis TL, Chugani HT. Proton magnetic resonance spectroscopy in children with Sturge-Weber syndrome. J. Child Neurol. 1998; 13 (7): 332-5
16. Pascual-castroviejo I, Viaño J, Moreno F et-al. Hemangiomas of the head, neck, and chest with associated vascular and brain anomalies: a complex neurocutaneous syndrome. AJNR Am J Neuroradiol. 1996; 17 (3): 461-71
17. Griffiths PD. Sturge-Weber syndrome revisited: the role of neuroradiology. Neuropediatrics. 1996;27 (06): 284-94.
18. Comi AM. Update on Sturge-Weber syndrome: diagnosis, treatment, quantitative measures, and controversies. Lymphat Res Biol. 2007;5 (4): 257-64.
19. Shirley MD, Tang H, Gallione CJ et-al. Sturge-Weber syndrome and port-wine stains caused by somatic mutation in GNAQ. N. Engl. J. Med. 2013;368 (21): 1971-9.