

Case Report

Sturge Weber Syndrome (SWS): A case report in an infantErum Afzal¹, Asia Noureen², Muhammad Khalid Iqbal³, Kausar Aftab⁴, Asad Rehman⁵, Rubeena Muien⁶¹ Assistant Professor, Department of Developmental Paediatrics, The Children's Hospital & ICH, Multan.³ Senior Registrar, Department of Developmental Paediatrics, The Children's Hospital & ICH, Multan.^{2,4,5} Post-graduate Resident, Department of Developmental Paediatrics, The Children's Hospital & ICH, Multan⁶ Women Medical Officer, Department of Developmental Paediatrics, The Children's Hospital & ICH, Multan.**Author's Contribution**^{1,2,3,4} Conception of study^{2,5} Experimentation/Study conduction^{1,2,3,4,5} Analysis/Interpretation/Discussion^{2,5} Manuscript Writing¹ Critical Review^{1,3,4} Facilitation and Material analysis**Corresponding Author**

Dr. Asia Noureen,

Post-graduate Resident,

Department of Developmental Paediatrics,

The Children's Hospital & ICH,

Multan

Email: asianoureen900@gmail.com

Article Processing

Received: 31/12/2020

Accepted: 23/09/2021

Cite this Article: Afzal, E., Noureen, A., Iqbal, M.K., Aftab, K., Rehman, A., Muien, R. Sturge Weber Syndrome (SWS): A case report in an infant. *Journal of Rawalpindi Medical College*. 30 Sep. 2021; 25(3): 434-438.

DOI: <https://doi.org/10.37939/jrmmc.v25i3.1548>

Conflict of Interest: Nil**Funding Source:** Nil**Access Online:****Abstract**

Introduction: Sturge Weber Syndrome (SWS), known as encephalotrigeminal angiomatosis, is a rare neurocutaneous disorder and is having a prevalence of 1/20-50,000, live births. The syndrome consists of leptomeningeal angiomas and the angiomas of the face, characteristically in the ophthalmic and maxillary divisions of the trigeminal nerve. The neurological presentations include fits, deficits in cognition, glaucoma, and visual field abnormalities. The overgrowth of the soft tissues and extracranial angiomas can also occur in SWS.

Case presentation: We are presenting a rare case of Sturge Weber Syndrome in an infant who presented at an early stage of this disorder. The index case presented to us with a large port-wine stain and right-sided focal fits. The CT scan brain showed subtle gyriform tram-track calcification of the left cerebral cortex and the left-sided cerebral atrophy. Ophthalmological examination showed glaucoma of the left eye. The clinical and radiological criteria were used to make the final diagnosis of SWS.

Conclusion: The case is being reported in order to increase awareness among medical professionals.

Keywords: Sturge Weber Syndrome (SWS), Port Wine Stain (PWS), Nevi flammeus, Neurocutaneous Syndromes, Phakomatosis, Angiomatosis.

Introduction

Sturge-Weber Syndrome is recognized as encephalotrigeminal angiomatosis which includes hemangiomas of the leptomeninges, facial nevus flammeus (port-wine stain), and glaucoma. SWS can also cause severe complications of an eye by involving conjunctiva and eyelids. The incidence of SWS is 1 in 20-50,000 live births, but it often remains underreported.¹

Sturge-Weber Syndrome is classified into three main types:

Type I has angiomas of the face and leptomeninges (may have glaucoma).

Type II consists of facial angiomas without any neurological manifestations (with possible glaucoma).

Type III involves only leptomenial angiomas (without glaucoma).²

Sturge Weber Syndrome is a rare congenital but is not a hereditary condition. It is usually presented with hamartomas formations, which are the result of the failure of normal development of fetal veins of the involved organs. This causes venous hypertension and subsequently insufficient perfusion of the underlying brain parenchyma resulting in chronic cerebral ischemia of the brain resulting in cerebral atrophy and neurological manifestations.³

The etiology of SWS resides in somatic mosaicism. The molecular and genetic studies showed that somatic mutation involving *GNAQ*, on chromosome 9, was responsible for malformations in the brain and skin of patients with SWS. The laboratory studies confirmed the presence of *GNAQ* mutation in endothelial cells of involved skin capillary abnormalities.⁴

The presence of two out of the three diagnostic criteria is required for making a diagnosis of SWS; those include a facial port-wine stain, raised intra-ocular pressure, and angiomatosis of the leptomeninges. The neurological features of SWS are seizures, weakness, stroke-like episodes, deficits in a visual field, headaches, and cognitive deficits. Patients with the diagnosis of SWS can experience endocrinological abnormalities, impairments incognition, learning problems, behavioral and psychological difficulties, and other medical manifestations.⁵

Case Report

The index case is a nine months old girl presented with 7 days history of right-sided focal fits. She was

delivered at home. She is the second among the two sibs. Pregnancy and the perinatal period were uneventful except that the family noticed a large pinkish-colored spot extending from the left side of the scalp and face and involving the right side of the face sparing the forehead and periorbital area. There were multiple areas of pinkish discoloration on the neck, back, right arm, and left leg. Her elder sib is healthy and there is no family history of such lesions. The clinical and radiological findings were used to make the diagnosis of SWS which included that:

Dermatological findings:

A port-wine stain was presently extending from the left side of the scalp to the face and the neck, involving the lower part of the right side of the face sparing the forehead and periorbital area. (Figure 1) Extensive cutaneous angiomas were present on the back, right arm, and left leg.

Eye Findings:

Table I: eye examination

| Parameters | Right eye | Left eye |
|----------------------|-----------|-------------|
| CD ratio | 0.3 - .35 | 0.45 – 0.50 |
| IOP(mmHg) | 12 | 18 |
| Corneal diameter(mm) | 11 | 12 |

The ophthalmological assessment showed raised intraocular pressure of the left eye and hemangiomas choroid and sclera of both the eyes. (Figure 2)

Systemic Examination:

All other systems were normal on examination.



Figure 1: Port-wine stain involving the left side of the face and right arm



Figure 2: Scleral hemangiomas of both eyes

Developmental assessment:

1. Motor 4 months
2. Cognition 7 months
3. Self-help age-appropriate
4. Socialization 6 months
5. Language poor both receptive and expressive language skills
6. Speech vocalization stage

Remarks: GDD

Radiological Findings:

CT scan brain showed gyriform subtle tram-track calcifications noted in the left cerebral hemisphere with a prominence of extra-axial CSF spaces representing left-sided cerebral atrophy.

MRI brain showed cerebral atrophy and calvarial thickening on T2 weighted images and sub-cortical calcifications on T1 weighted images. (Figure 3)

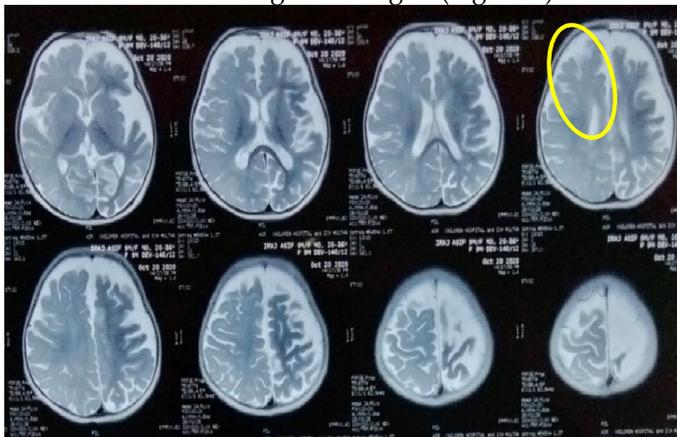


Figure 3: T1 weighted images showing left-sided sub-cortical calcifications

By keeping in view the clinical findings and imaging results, the provisional diagnosis of Sturge-Weber syndrome was concluded. Parents were counselled

about the nature of the disease, its manifestations, complication, and their management. The patient was given anti-epileptics to control fits and pharmacotherapy was started to control the raised intraocular pressure. Parents were also counselled for a follow-up visit in order to monitor complications.

Discussion

Sturge-Weber Syndrome is a neurocutaneous disorder, one of the most common presenting disorders, and it affects approximately 1/20-50,000 live births. It is not an inherited condition, rather occurs sporadically, affecting both genders, and is seen in all races and ethnicities. The features of Sturge-Weber syndrome present at birth in the form of a flammeus nevus on the face (port-wine stain) and later on abnormal vasculatures of the eye and the brain result in a range of complications.⁶

The primary etiology of SWS is considered to be somatic mosaicism. A study involving 4 patients with SWS showed that somatic mosaicism was the main etiology of SWS. An activating somatic mutation in the GNAQ gene, on chromosome 9, was responsible for alterations in the structure and function of blood vessels. The involved mutation was also affecting the expression of extracellular matrix and vasoactive molecules.⁷

Sturge was the first who had given the earliest descriptions of SWS, and Weber, later on, demonstrated the characteristic feature of Sturge-Weber syndrome i.e. cerebral gyriform calcifications. Sturge-Weber syndrome is a heterogeneous disorder with wide variability in its features and natural history. Roach had proposed a schematic classification of the broad spectrum of Sturge-Weber syndrome presentations. These are the following:

Type I has both facial and leptomeningeal angiomas. (May have glaucoma)

Type II has only angiomas of the face and there is no neurological involvement. (May have glaucoma)

Type III has only leptomeningeal angiomas but without a facial flammeus nevus. (No glaucoma)⁸

Cutaneous manifestations:

Unilateral facial nevus flammeus since birth, affecting the first sensory distribution of the trigeminal nerve, is the most prominent clinical finding of SWS. Stains involving the second and third sensory distribution of the trigeminal nerve are also observed, and they can be diffuse and bilateral. Sometimes, small angiomatic nodules may appear within a typical port-wine stain, and in rare instances, a "cobblestone"

pattern of red-purple nodules entirely covers the cutaneous lesion.

Neurological symptoms and signs:

Seizure caused by hypoxia and microcirculatory is another common manifestation of SWS. Partial seizures, which may occur in 70%-90% of those with the disorder by 3 years of age, typically occur contralateral to the neurocutaneous abnormality and worsen with age. Infantile spasms and generalized seizures are also observed.

Other neurological complications:

Secondary to leptomeningeal angiomas included vascular headache, stroke-like episodes, contralateral hemiparesis, hemiatrophy, and hemianopia. About 50%-60% of those with SWS presented mental retardation. Even in those without frank cognitive impairment, intellectual skills are often lower than expected. Chapieski and colleagues noted a mean intelligence quotient (IQ) of approximately 75 and a range from 42 to 127 in 32 individuals for whom IQ scores were available.

Ocular manifestations:

Glaucoma is considered one of the most common presenting ocular manifestations of SWS and is seen in 30-70% of those with SWS, it causes buphthalmos, visual field deficits, and loss of vision; 60% of these patients develop glaucoma during the first year of life due to anterior chamber abnormalities and rest of the 40% develop in childhood or adolescent years as a result of raised episcleral venous pressure. SWS can present with other ocular complications such as vascular abnormalities of the conjunctiva, choroid, episclera, and retina. SWS may also present with oral manifestations. These include oral mucosal vascular hyperplasia, gingival changes of variable severity from minimal vascular hyperplasia to giant masses, which may interfere with the function of the mouth.⁹

Differential Diagnosis

The differential diagnosis of Sturge-Weber Syndrome includes:

- 1: The Klippel-Trénaunay-Weber's syndrome presents with facial nevus flammeus, hypertrophy of soft and bony tissues, and arteriovenous malformations (AVM).
- 2: The Rendu-Osler-Weber's Syndrome or hereditary hemorrhage telangiectasia –that has abnormal vasodilatation of the terminal vessels of the skin, mucosal surfaces, and visceral vessels.
- 3: The Maffucci's syndrome, which presents with cutaneous vascular abnormalities and mucosal abnormalities of lips and palate, dyschondroplasia, and pathological deformities of a long bone. It can cause chondrosarcomas too.

4: The Beckwith-Wiedmann's Syndrome has capillary abnormalities of the central forehead or upper eyelids and with a skin lesion that is very much similar to the Port wine stain.

Other features include macroglossia and mild risk visceral overgrowth and associated neoplasms.¹⁰

Workup for SWS

Radiology:

Computed Tomography (CT) scan of the brain is the diagnostic modality of choice because it can better delineate calcifications, seen as tram-track calcification, but, may be absent in the initial stage of the disease.

MRI brain provides the exact location and extent of leptomeningeal angiomatosis (LAM) and delineates parenchymal abnormalities due to gliosis.

EEG shows background suppression of activity near the angiomas and epileptiform discharges can rise from the involved hemisphere. The PHR most commonly shows epileptiform discharges from the occipital lobe followed by temporal, parietal, and frontal lobes in descending order of occurrence. Some patients have Frontal spikes on PHR can be explained by suppression of the background activity.

Histopathology of the involved tissue in SWS shows vascular abnormalities with capillary malformation and extensive calcification which are in favor of the diagnosis of SWS.³

Soon after the diagnosis of Sturge-Weber syndrome (SWS) is made, a complete ophthalmologic assessment is mandatory to rule out and diagnose glaucoma, because the increased intraocular pressure (IOP) can damage the eye quickly.

In younger children with SWS, examination under general anesthesia

(9GA) or sedation is essential to confirm the diagnosis of glaucoma. Assessment is done by measurement of IOP, corneal diameter, cycloplegic refraction, axial length, and cup-disc ratio. Furthermore, a gonioscopic examination is essential to document angle abnormalities.⁷

Management

For treatment of PWS, a pulsed-dye laser (PDL) can lighten the birthmark. This laser treatment can further decrease the long-term risk of hypertrophy of soft and bony tissues. As a result, functional impairments due to swallowing, speaking, breathing, vision, and hearing can be minimized.

For glaucoma, pharmacotherapy is the first line of management, but it is often not enough tools for the management of aggressive glaucoma. The initial pharmacotherapy to be used is topical beta-blocker, followed by carbonic anhydrase inhibitor. The best-studied topical medication is latanoprost,

progesterone, with effective control of intraocular pressure (IOP) in up to half of the patients. Even with the best possible management, patients with florid glaucoma may be resistant to respond to treatment, and buphthalmos (eye enlargement) and amblyopia may eventually result.

Regarding seizures, the principal aim of pharmacologic therapy is to minimize and eliminate seizures at all. A wide variety of anticonvulsant medications is used to control seizures. We start with an anticonvulsant after the first seizure is documented and the first line anti-seizure drug used in SWS is oxcarbazepine. Other potentially used anticonvulsants are valproic acid, carbamazepine, zonisamide, lamotrigine, and phenobarbital.⁵

Additionally, physiotherapies are essential to control the motor difficulties resulting from neurological insult, and education of caregiver is the priority to increase the intellectual level of the affected child.

Monitoring:

Annual monitoring is recommended for the surveillance of glaucoma for all patients. If a child is found to have raised IOP, it deserves an aggressive treatment that is mandatory to prevent the complications of glaucoma.¹⁰

Conclusion

Sturge Weber Syndrome is one of the most commonly occurring but under-reported syndromes. The diagnosis is based upon the presence of facial port-wine stain, ipsilateral glaucoma, and contralateral seizure. The diagnosis is made by clinical features and is confirmed by radiological modalities such as CT or MRI brain and EEG. Knowledge about this syndrome is important because early diagnosis and aggressive management of glaucoma are necessary to prevent long-term eye complications.

References

1. Javaid U, Ali MH, Jamal S, Butt NH. Pathophysiology, diagnosis, and management of glaucoma associated with Sturge-Weber syndrome. *International ophthalmology*. 2018 Feb 1;38(1):409-16.(NEW R1)
2. Jagtap SA, Srinivas G, Radhakrishnan A, Harsha KJ. A clinician's dilemma: Sturge-Weber syndrome 'without facial nevus'?. *Annals of Indian Academy of Neurology*. 2013 Jan;16(1):118.(R6)
3. Ansari S, Ozair E. Sturge-Weber Syndrome: Rare Neurocutaneous Disorder. Volume 5, Issue 2. April-June, 2015 Page | 40 <https://www.jpmonline.com/>(R2)
4. Huang L, Couto JA, Pinto A, Alexandrescu S, Madsen JR, Greene AK, Sahin M, Bischoff J. Somatic GNAQ mutation is

enriched in brain endothelial cells in Sturge-Weber syndrome. *Pediatric neurology*. 2017 Feb 1;67:59-63(R7).

5. Bachur CD, Comi AM. Sturge-weber syndrome. *Current treatment options in neurology*. 2013 Oct 1;15(5):607-17. DOI: 10.1007/s11940-013-0253-6(R8)

6. Comi AM. Sturge-Weber syndrome. In *Handbook of clinical neurology* 2015 Jan 1 (Vol. 132, pp. 157-168). Elsevier. <https://doi.org/10.1016/B978-0-444-62702-5.00011-1>(R1)

7. <https://emedicine.medscape.com/article/1177523-overview#a1>

8. Bhagat S, Gupta G, Singh S. Unusual presentation of sturge weber syndrome. *Journal of case reports*. 2015 Apr 14;5(1):111-5. [http://dx.doi.org/10.17659/01.2015.0028\(R4\)](http://dx.doi.org/10.17659/01.2015.0028(R4))

9. Jing ZH, Li NY, Zhou XJ, Wang JD, Zhang RS. Sturge-Weber syndrome: a case report and review of literatures. *Chinese Medical Journal*. 2010 Jan 1;123(1):117-21.

10. DOI: 10.3760/cma.j.issn.0366-6999.2010.01.023(R5)

11. Neto FX, Junior MA, Ximenes LS, de Souza Jacob CC, Junior AG, Palheta AC. Clinical features of Sturge-Weber syndrome. *Intl Arch Otorhinolaryngol*. 2008;12(4):565-70. http://www.arquivosdeorl.org.br/conteudo/acervo_eng_print.asp?id=573