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Case Report

# Sturge-Weber Syndrome: A Case Report of a 12-Year-Old with Facial Angioma and Seizure Disorder

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## Abstract

### Background

Sturge-Weber Syndrome (SWS) is a rare congenital disorder characterized by facial port-wine stain, leptomeningeal angiomas, and ocular abnormalities such as glaucoma. It results from a somatic mutation in the GNAQ gene, leading to abnormal vascular development. Clinical features vary widely, with many children initially appearing healthy before developing neurological, ophthalmic, and developmental issues. Early recognition and multidisciplinary management are crucial for improving outcomes, especially in resource-limited settings.

### Case presentation

A 12-year-old girl presented with recurrent seizures, progressive left eye vision loss, and a longstanding port-wine stain over her left periorbital region. Examination revealed optic atrophy, elevated intraocular pressure, and neuroimaging showed cerebral atrophy with characteristic gyriform calcifications. She was diagnosed with Type I SWS and managed with antiepileptics, aspirin, and glaucoma treatment. Her developmental delay and cognitive difficulties underscored the need for comprehensive care and support.

### Conclusion

This case highlights the typical features of SWS, emphasizing the importance of early clinical suspicion when facial port-wine stain is associated with neurological and ocular signs. Prompt diagnosis allows for timely management to control seizures, preserve vision, and support developmental needs. In resource-limited environments, a high index of suspicion and multidisciplinary collaboration are vital for optimizing patient outcomes.

**Keywords:** sturge-weber syndrome; facial angioma; port-wine stain; seizure disorder

## Introduction

Sturge-Weber Syndrome (SWS) is a rare congenital condition that leaves a visible and often life-changing mark on those affected. It is best known for the characteristic facial port-wine stain, a birthmark that may seem harmless at first but often signals deeper neurological and ocular involvement [1]. The condition results from abnormal blood vessel development involving the skin, brain, and sometimes the eyes [2].

Children with SWS may begin life appearing healthy, only to develop seizures, weakness on one side of the body, or vision problems as they grow [3]. These symptoms can place a heavy emotional and social burden not only on the child but also on their family, especially when access to specialized care is limited [4].

The syndrome occurs sporadically, usually due to a somatic mutation in the GNAQ gene, and its estimated frequency is between 1 in 20,000 to 50,000 live births [5,6]. The severity and combination of symptoms vary widely; some patients experience only minor skin lesions, while others develop significant neurological impairment [7].

Early recognition and multidisciplinary management are key to improving outcomes and supporting both the physical and emotional wellbeing of affected children [8]. Here, we present the case of a 12-year-old child with facial angioma and recurrent seizures, illustrating the clinical features, diagnostic process, and challenges encountered in managing Sturge-Weber Syndrome in a resource-limited setting.

## Case Presentation

A 12-year-old girl was brought to the outpatient department by her father after experiencing an episode of generalized tonic-clonic seizure one week prior to presentation. The seizure lasted approximately 10 seconds, with no frothing from the mouth and no loss of bladder or bowel control. She regained full consciousness shortly after the episode.

According to her father, this was not the first time she had convulsions. The first episode occurred when she was about nine months old, followed by several recurrent seizures during early childhood. Over the years, her condition had been fairly well controlled with carbamazepine 200 mg twice daily, and she had remained seizure-free for the past eight months. There was no history of stroke-like episodes, speech problems, or weakness of the limbs.

In addition to the seizures, her father reported a gradual decline in vision over the years, which had progressed to total loss of sight in the left eye during the last two months. He also mentioned a purplish birthmark around her left eye, which had been present since birth and remained unchanged in size. The family had grown accustomed to it, thinking it was a harmless mark (Figure 1). More recently, her teachers had raised concerns about her poor school performance and difficulty in concentration, prompting further evaluation.

***Developmental Milestones:*** Her developmental history revealed that she had delayed milestones, sitting independently at around 18 months and walking at 3 years of age. The father could not recall the timing of other milestones.

**Family and social history:** There was no family history of seizures, developmental delay, or similar skin lesions.

**On examination,** the girl was alert, calm, and cooperative. A well-demarcated purplish discoloration (port-wine stain) was seen over the left periorbital and malar region, corresponding to the ophthalmic (V1) and maxillary (V2) branches of the trigeminal nerve (Figure 1). There were no similar lesions elsewhere.

**Neurological examination** was largely normal, with no facial asymmetry or motor weakness. However, she had absent light perception in the left eye, while the right eye's vision was normal. Deep tendon reflexes and muscle tone were symmetrical, and there were no sensory deficits.

**Ophthalmologic evaluation:** revealed optic atrophy and raised intraocular pressure in the left eye, consistent with secondary glaucoma. Fundoscopy of the right eye was normal.

**Musculoskeletal system:** normal gait, no deformities on the hands, normal joints of upper and lower limbs with normal range of motions no tenderness notes, normal back no tenderness no deformities noted.

**Cardiovascular examination:** normal radial pulses, good volume and regular, non-collapsing, no jugular venous distention, normal precordium, apex beat at 5th intercostal space no heaves normal heart sounds.

**Respiratory examination:** normal chest shape no any deformity, bilateral air entry with vesicular sounds

**Abdominal examination:** normal abdominal shape, no distention no scars, flat umbilicus, no tenderness on palpation, no organomegaly normal bowel sounds and tympanic note on percussion.

A head CT scan demonstrated left cerebral cortical atrophy with tram-track gyriform calcifications, particularly in the parietal and occipital lobes, findings characteristic of Sturge-Weber Syndrome (Figure 2 and figure 3).

Based on the clinical picture and imaging, a diagnosis of Sturge-Weber Syndrome (Type I) was made. The patient was maintained on carbamazepine 200 mg twice daily for seizure control, aspirin 75 mg once daily to improve cerebral perfusion and reduce vascular complications, and timolol eye drops for glaucoma management.

She and her family were counseled on the chronic nature of the condition and the importance of adherence to medication, regular ophthalmologic and neurologic follow-up, and educational support to address her learning difficulties. The father was encouraged to continue providing emotional and social support, as these play an essential role in her overall quality of life.

**Figure 1:** Facial port-wine stain involving the left periorbital and malar region, corresponding to the ophthalmic (V1) and maxillary (V2) branches of the trigeminal nerve.

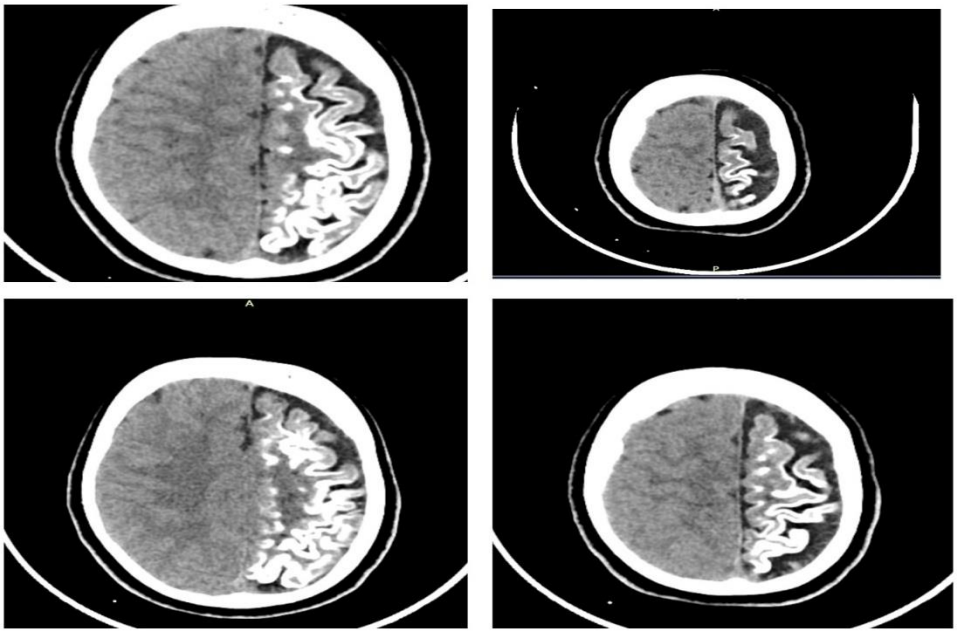

**Figure 2:** non-contrast CT scan showing left cerebral cortical atrophy with tram-track gyriform calcifications, predominantly in the parietal and occipital lobes.

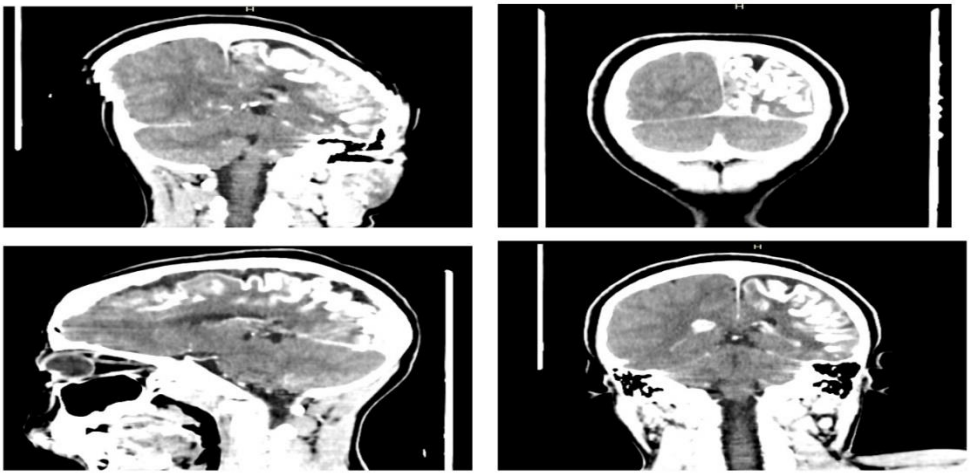

**Figure 3:** non-contrast CT scan showing left cerebral cortical atrophy with tram-track gyriform calcifications, predominantly in the parietal and occipital lobes.

## Discussion

Sturge-Weber Syndrome (SWS) is a rare, congenital neurocutaneous disorder caused by a somatic activating mutation in the GNAQ gene, leading to abnormal vascular development in the skin, brain, and eyes [1,2]. The classic triad includes facial port-wine stain (nevus flammeus), leptomeningeal angiomas, and glaucoma.

In this case, the child presented with a port-wine stain over the left periorbital region, seizure disorder, and progressive unilateral visual loss, representing the typical features of Type I SWS. The tram-track gyriform calcifications and cortical atrophy observed on CT scan reflect chronic venous stasis and cortical ischemia caused by leptomeningeal angiomas [3,4].

Seizures are the most common neurological manifestation of SWS, often starting within the first year of life [5]. Our patient's seizure onset at nine months aligns with this typical pattern. The long-term seizure control with carbamazepine is consistent with best-practice management. Low-dose aspirin therapy may reduce microthrombotic complications and improve cerebral perfusion [6].

Ocular involvement, particularly glaucoma, occurs in up to 70% of patients with SWS and may lead to irreversible vision loss if untreated [7]. In this patient, glaucoma and optic atrophy explained her progressive visual decline. Timolol eye drops were used to control intraocular pressure.

Developmental delay and cognitive impairment are common in SWS, especially when the parietal and occipital lobes are affected, as in this case [8]. Educational support and cognitive interventions are therefore essential.

Management of SWS is symptomatic and multidisciplinary, requiring coordination between neurologists, ophthalmologists, dermatologists, and psychologists. This case underscores the challenges faced in resource-

limited settings, where access to advanced imaging and specialized care may be limited, highlighting the importance of clinical vigilance, timely diagnosis, and compassionate care.

## Conclusion

This case highlights the classical features of Sturge-Weber Syndrome and underscores the importance of recognizing the condition early, particularly when a facial port-wine stain is accompanied by neurological or ocular symptoms. Early diagnosis allows timely seizure control, prevention of visual loss, and educational support for affected children.

In resource-limited settings, where access to advanced imaging and specialized care may be restricted, a high index of clinical suspicion and multidisciplinary collaboration remain crucial. Beyond medical management, providing psychological and social support to the child and family is essential to improving overall quality of life.

**Abbreviations:** SWS: Sturge-Weber Syndrome CT: Computed Tomography

## Acknowledgment

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### Patient Consent

The patient's guardian signed an informed consent form and agreed the case to be published in a scientific journal

## Conflict of Interest

The authors declare no conflicts of interest related to this case report.

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### Authors contributions

The authors contributed significantly to the case report

## References

1. Shirley MD, Tang H, Gallione CJ, Baugher JD, Frelin LP, Cohen B, et al. (2013). Sturge–Weber Syndrome and Port-Wine Stains Caused by Somatic Mutation in GNAQ. N Engl J Med. 368(21):1971–1979.
2. Sturge–Weber syndrome. In: Handbook of Clinical Neurology [Internet]. Elsevier; 2015 [cited 2025 Nov 10]. p. 157–68. Available from: https://linkinghub.elsevier.com/retrieve/pii/B9780444627025000111
3. Pascual-Castroviejo I, Pascual-Pascual SI, Velazquez-Fragua R, Viaño J. (2008). Sturge-Weber Syndrome. Study of 55 Patients. Can J Neurol Sci J Can Sci Neurol. 35(3):301–307.
4. Di Rocco C, Tamburrini G. Sturge–Weber syndrome. Childs Nerv Syst. 2006 Aug;22(8):909–921.
5. Sujansky E, Conradi S. (1995). Outcome of Sturge-Weber syndrome in 52 adults. Am J Med Genet. 57(1):35–45.
6. Lance EI, Sreenivasan AK, Zabel TA, Kossoff EH, Comi AM. (2013). Aspirin Use in Sturge-Weber Syndrome: Side Effects and Clinical Outcomes. J Child Neurol. 28(2):213–218.
7. Thomas-Sohl KA, Vaslow DF, Maria BL. (2004). Sturge-Weber syndrome: A review. Pediatr Neurol. 30(5):303–310.
8. Powell S, Fosi T, Sloneem J, Hawkins C, Richardson H, Aylett S. (2021). Neurological presentations and cognitive outcome in Sturge-Weber syndrome. *Eur J Paediatr Neurol.* 34:21–32.

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