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CASE REPORT

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Sturge weber syndrome: A case report

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Key Clinical Message

This report presents a 14-year-old male with seizures and facial port-wine stains, who upon further evaluation was found to have SWS. Early diagnosis and consistent treatment of Sturge–Weber syndrome in children are essential to prevent seizures and improve quality of life. Anti-seizure medications play a crucial role in preventing and controlling seizures.

KEYWORDS

case report, neurocutaneous syndromes, port-wine stain, seizure, Sturge–Weber syndrome, tram track sign brain

1 | INTRODUCTION

Sturge Weber Syndrome (SWS) is a rare sporadic genetic neurocutaneous syndrome with a reported incidence of 1:50,000.[1] It was first described by Schirmer in 1860 and later by Sturge in 1879.[2]

SWS is caused by somatic mosaic mutations in the GNAQ gene,[3] which is situated on the long arm of chromosome 9.[4] GNAQ encodes Gαq, a G-protein subunit crucial for cell signaling. The normal function of GNAQ is to regulate cellular activities via signaling pathways. During the sixth week of intrauterine development, an embryonal vascular system emerges around the cephalic region of the neural tube, which usually regresses by the ninth week. Activating mutation of GNAQ in SWS causes dysregulated signaling, leading to the persistence of these structures leading to angiomas of the leptomeninges, face, and eyes.[5,6] A leptomeningeal angioma affecting the occipital and posterior parietal lobes, eye abnormalities (choroidal hemangioma and glaucoma), and facial port-wine stains, typically in ophthalmic and maxillary distributions of the trigeminal nerve, are the hallmarks of SWS.[7]

Complete SWS refers to both CNS and facial angiomas, while incomplete SWS refers to only one affected area without the other. Classification is done using the Roach scale.[8]

1. Type I: Leptomeningeal and facial angiomas; possibly with glaucoma.
2. Type II: Facial angiomas only, possibly with glaucoma.
3. Type III: Isolated leptomeningeal angiomas, typically without glaucoma.

Seizures are the most common symptom. Diagnosis is clinico-radiological and supported by genetics. Symptomatic management, particularly seizure control, is crucial.

All the authors have read and approved the final manuscript.

2 | CASE HISTORY AND EXAMINATION

A 14-year-old boy presented to the emergency department with the complaint of one episode of abnormal body movement. The episode lasted for 4 min, involved all four limbs, and was associated with frothing from the mouth and tongue bite. The episode occurred while the child was awake at school. After the episode, he regained consciousness but remained confused for a few minutes. On further questioning with the parents, it was found that he had similar episodes four years back for which he was taking levetiracetam prescribed by the local pharmacy. The parents believed that the disease was cured and thus the medication was stopped since the past six months. No history of recurring headaches, stroke-like events, head trauma or similar family history was reported.

On examination, vital signs were within normal limits. A two-centimeter laceration was present on the lateral aspect of the tongue. Characteristic reddish discoloration was noted on the right side of the face, involving parts of the forehead and scalp, eyebrow, and upper eyelid. These lesions, identified as port wine stains, had been present since birth (Figure 1). Neurological examination, including assessment of cognition, was within normal limits. An ophthalmologic examination revealed no signs of glaucoma.

2.1 | Investigations

Routine laboratory investigations were within normal limits. CT scan of the head showed tram track ribbon-like cortical calcification involving the right temporal and occipital lobes with no evidence of volume loss. (Figure 2).

Magnetic resonance imaging (MRI) brain disclosed curvilinear cortical and subcortical calcifications in the right posterior temporal lobe, accompanied by notable leptomeningeal enhancement and prominent ipsilateral choroid plexus (Figure 3).

Based on the totality of history, clinical examination, and findings on neuroimaging, a diagnosis of Sturge–Weber syndrome was made.

3 | RESULTS AND TREATMENT

Our patient was managed with anti-seizure medications and was discharged on levetiracetam and low-dose aspirin. Counseling regarding the disease and the need for medication adherence was also given. On a six-month follow-up, the patient was seizure-free and was doing well.

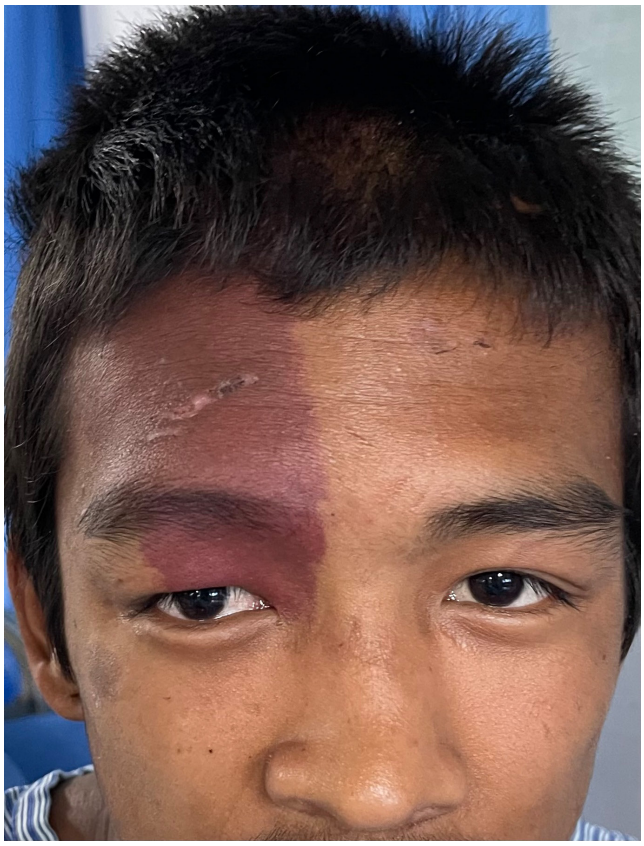

FIGURE 1 Port Wine stain on the right side of the face, along cranial nerve V1 distribution.

4 | DISCUSSION

SWS is a rare neurocutaneous condition characterized by facial, ocular, and brain (leptomeningeal) angiomas, often identified by port-wine stains.

Port wine stains are congenital (0.3%–0.5% incidence in neonates) vascular malformations characterized by ectasia of capillary and postcapillary venules in the skin and mucosa,[9] named after the deep red hue that they leave on the skin or mucosa.[10] They mostly occur unilaterally, however bilateral involvement and extension to the neck, limbs, and other parts of the body have been reported. These lesions were thought to appear along the dermatomes supplied by the ophthalmic and maxillary divisions of the trigeminal nerve and the involvement of the ophthalmic division area is considered typical in SWS.[5,11] These lesions become darker, more nodular, and hypertrophic over time[9] and tend to bleed profusely when traumatized.[2]

Early diagnosis and treatment are essential, especially when the upper face is affected, as there is a 15%–50% risk of brain and/or eye involvement.[12]

In patients with SWS, leptomeningeal angiomas frequently develop unilaterally in the parietal and occipital areas. The presence of an angioma alters blood flow

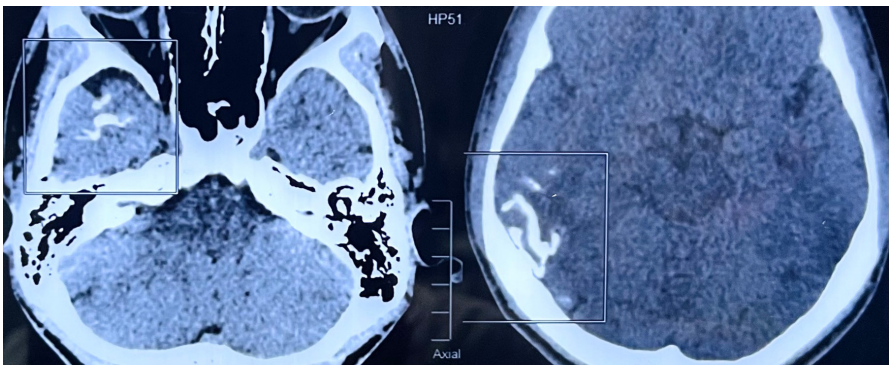

FIGURE 2 CT scan head showing tram track calcifications in right temporal and occipital lobes.

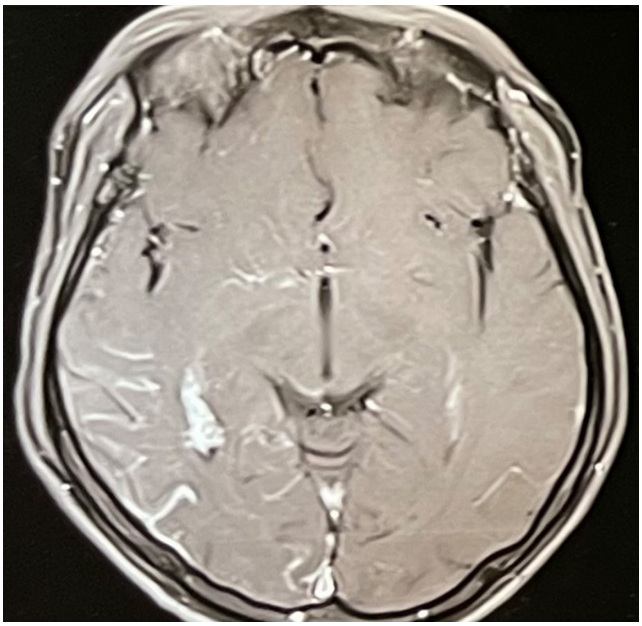

FIGURE 3 Postcontrast brain MRI showing prominent leptomeningeal enhancement along the right LCVM with prominent ipsilateral choroid plexus.

dynamics, leading to calcium deposits in the cerebral cortex. This can result in debilitating neurological conditions like seizures, mental disability, autistic behavior, hemiplegia, hemiparesis, recurring headaches, stroke-like events, psychomotor retardation, and mental retardation.[6,13,14] Seizures could be atonic, tonic, or myoclonic and are the most frequent neurologic manifestations of SWS that appear from infancy to adulthood.[5]

Epilepsy affects around 75%–90% of patients with SWS brain involvement, with the majority developing it within the first year of life. Early onset of seizures has been linked to decreased cognitive performance, affecting the neuro quality of life. Preventing epilepsy is crucial for people with SWS as it raises the risk of stroke and brain damage.[15]

Antiseizure medications are commonly prescribed as the initial treatment for individuals with epilepsy associated with SWS. Medication alone has been found to achieve adequate seizure control in about 40% of cases of SWS, commonly used ones being carbamazepine, oxcarbazepine and levetiracetam.[16] However, the optimal regimen for treating seizures in SWS is still unknown, and individual responses to medications may vary. Clinical decisions should be based on a thorough assessment of each patient's specific circumstances, taking into account factors such as medical history, comorbidities, and potential side effects of the medications.

Ocular involvement can cause various eye conditions such as hemianopia, glaucoma, buphthalmos, and choroidal hemangioma. Intraoral angiomatosis can affect the lips, palate, gingiva, floor of the mouth, and buccal mucosa.[2]

Differential diagnoses include Maffuci's syndrome, Von Hippel Lindau disease, Rendu-Oslar-Weber syndrome, angio-osteodystrophy syndrome and Klippel Trenaumy-Weber syndrome.[17]

The diagnosis is based on radiological imaging, which shows leptomeningeal vascular malformation on contrast-enhanced T1-weighted MRI and cortical and subcortical calcification on head computed tomography (CT).[18] EEG, magnetic resonance spectroscopy, and fluorodeoxyglucose-positron emission tomography (FDG-PET) may also aid in patient evaluation but are not commonly utilized and are not recommended. In individuals above the age of one, MRI is the most often used diagnostic modality. A CT scan identifies calcification, the most prevalent of which is a gyriform calcification, which is referred to as a "tram-track sign."[19,20] The use of ionizing radiation in CT scans restricts its application.

There is no particular therapy for SWS. The type and intensity of clinical characteristics influence treatment and prognosis. Almost all individuals experience altered personality development when port wine stain is present, and it can cause severe psychological distress.[17] Dermabrasion, tattooing, and flash lamp pulsed dye lasers can help to improve port wine stains.[2] The use of an AED and aspirin is done for symptomatic treatment.[18] Anti-seizure medications for seizure control,

preventative and headache therapy, and glaucoma treatment including lowering intraocular pressure are all part of the medical care provided to individuals with SWS. Patients with glaucoma, refractory seizures, or some illnesses connected with SWS, such as scoliosis, are better off undergoing surgery. Vagal nerve stimulation, corpus callosotomy, hemispherectomy, focal cortical resection, and surgical intervention for diffuse choroidal hemangiomas with retinal detachment and glaucoma are among the surgical procedures for SWS.[5]

In our case, the patient experienced the first attack of seizure at the age of 10 years and the reddish port-wine stain was seen in the child from the birth itself which gradually darkened with age. Typical features of SWS revealed in CT scan head and MRI brain proved invaluable in establishing the diagnosis. Our patient was also counseled about the nature of the disease, the need of medication adherence and the consequences of not taking them. Laser photocoagulation for port wine stain was refused owing to financial constraints. Possible treatment modalities were also discussed.

5 | CONCLUSION

Seizures are a predominant concern in patients with SWS. Seizure control through strict adherence to anti-seizure medications can significantly improve the quality of life.

AUTHOR CONTRIBUTIONS

Mitesh Karn: Conceptualization; data curation; formal analysis; investigation; methodology; writing – original draft; writing – review and editing. **Aachal Barma:** Conceptualization; data curation; formal analysis; writing – original draft; writing – review and editing. **Liladhar Ojha:** Conceptualization; data curation; formal analysis; methodology; supervision; writing – original draft. **Suman Bhatta:** Formal analysis; methodology; resources; supervision; validation; writing – review and editing. **Puja Neupane:** Data curation; formal analysis; investigation; methodology; software; supervision; validation. **Kusum Dhital:** Data curation; formal analysis; investigation; project administration; supervision; validation. **Anjan Sharma:** Data curation; formal analysis; software; supervision; writing – original draft. **Suraj Acharya:** Data curation; investigation; methodology; writing – original draft; writing – review and editing. **K. C. Unika:** Methodology; resources; software; writing – original draft; writing – review and editing. **Asmita Barma:** Formal analysis; software; supervision; validation; writing – original draft; writing – review and editing. **Gunjana Rawal:** Formal analysis; investigation; methodology; resources; supervision; writing – original draft; writing – review and editing. **Devraj Mahato:** Conceptualization; data curation; validation; writing – original draft.

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CONFLICT OF INTEREST STATEMENT

The authors report no conflicts of interest.

DATA AVAILABILITY STATEMENT

Data sharing is not applicable to this article as no new data were created or analyzed in this study.

ETHICS STATEMENT

No ethical approval is necessary.

CONSENT

Written informed consent was obtained from the patient's guardian to publish this case report and accompanying images.

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