MEDISAINS
Jurnal Ilmiah Ilmu - Ilmu Kesehatan

Case Report

Pediatric sturge–weber syndrome diagnosed by computed tomography imaging in a resource-limited setting: a case report

Komang Okky Maharani Ciptana Putri [1]✉, Mario B. Realino Nara [2], Defranky Theodorus [2]

[1] *Medical Doctor, Assistant of Pediatric Department, dr. T.C. Hillers Regional General Hospital, Sikka, East Nusa Tenggara, Indonesia*
[2] *Pediatrician, Department of Pediatric, dr. T.C. Hillers Regional General Hospital, Sikka, East Nusa Tenggara, Indonesia*

ARTICLE INFORMATION

Received: October 07, 2025
Revised: October 29, 2025
Accepted: November 12, 2025

KEYWORDS

Sturge-Weber Syndrome; Port-Wine Stain; Neurocutaneous Syndromes; Capillary Malformations, Congenital, 1; Tomography, X-Ray Computed

CORRESPONDENCE

Phone: 082237782598
E-mail: raniciptanaputri@yahoo.com

A B S T R A C T

Background: Sturge–weber syndrome (SWS) is a rare neurocutaneous disorder marked by facial capillary malformations, leptomeningeal vascular anomalies, and ocular complications such as glaucoma. Diagnostic evaluation and management can be challenging in resource-limited settings where magnetic resonance imaging (MRI) and genetic testing are unavailable.

Case presentation: An 8-year-old girl presented with recurrent generalized tonic–clonic seizures. Examination revealed a unilateral port-wine stain involving the right trigeminal distribution and gingiva, along with decreased visual acuity and elevated intraocular pressure consistent with secondary glaucoma. Non-contrast head computed tomography (CT) showed gyriform cortical–subcortical calcifications with a tram-track appearance and right hemispheric atrophy, supporting the diagnosis of Sturge–Weber Syndrome. The patient was treated with valproic acid and topical timolol, resulting in good seizure control and improvement in ocular symptoms. She remains under regular pediatric and ophthalmologic follow-up.

Conclusions: This case demonstrates that clinical recognition combined with characteristic CT findings allows reliable diagnosis of SWS in the absence of MRI or genetic testing. Medical management provided favorable short-term outcomes, underscoring the importance of early detection, multidisciplinary care, and structured monitoring in resource-limited environments.

INTRODUCTION

Sturge–weber syndrome (SWS), or encephalofacial angiomatosis, is a rare congenital neurocutaneous disorder with an estimated incidence of 1 in 50,000 live births.[1] It arises from a sporadic somatic mutation in the GNAQ gene during embryogenesis rather than hereditary transmission.[1,2] The syndrome is classically characterized by a facial capillary malformation (port-wine stain) within the trigeminal nerve distribution, accompanied by leptomeningeal vascular malformations that may affect the brain and eyes.[3] Clinically, patients commonly present with seizures, hemiparesis, developmental delay, signs of elevated intracranial pressure, and ocular manifestations such as glaucoma.[3,4]

Most documented SWS cases originate from tertiary hospitals equipped with advanced diagnostic tools, including contrast-enhanced brain MRI and genetic confirmation of GNAQ mutations.[5–7] Several studies also underscore the role of surgical interventions—such as hemispherectomy, lobectomy, and glaucoma surgery—particularly for refractory epilepsy or severe ocular complications.[6,7] However, literature describing the diagnostic and management challenges in resource-limited settings remains scarce. In such environments, head CT often becomes the only accessible neuroimaging modality, making classical radiological signs, such as the tram-track gyral calcification, essential for early recognition and timely intervention.[8]

This case report is important because it highlights the diagnostic pathway and conservative management of SWS in a facility with limited imaging and no genetic testing capability—a context rarely represented in published literature. Unlike reports that emphasize surgical management in high-resource centers, this study presents a child with SWS who achieved clinical stability and a favorable outcome through medical therapy alone. The purpose of this report is to describe the clinical presentation, neuroimaging characteristics, and successful non-surgical management of pediatric Sturge–Weber Syndrome in a resource-limited setting.[8,9]

https://doi.org/10.30595/medisains.v23i3.28415

CASE PRESENTATION

An 8-year-old girl was brought to the emergency department after experiencing two episodes of generalized tonic–clonic seizures, each lasting approximately five minutes prior to arrival. The events were characterized by rhythmic jerking of all extremities and upward eye deviation, followed by full recovery of consciousness. The seizures were accompanied by mild fever and headache. Pregnancy, delivery, and neonatal histories were unremarkable, with no reports of infection, bleeding, or asphyxia. Since birth, the patient had a persistent reddish lesion on the right half of her face, which the family regarded as a birthmark.

Her medical history revealed a similar seizure episode at eight months of age, for which she received antiepileptic therapy for several months. The medication was discontinued due to long-term seizure-free status. The patient reported blurred distance vision and occasional right-eye pain. Growth and developmental milestones were age-appropriate, and there was no history of limb weakness, behavioural disturbances, learning difficulties, or endocrine symptoms. There was no family history of seizures, cutaneous vascular lesions, or ophthalmologic abnormalities.

On examination, her vital signs were within normal range. A sharply demarcated erythematous capillary malformation involving the right forehead, cheek, upper lip, and gingiva was noted, consistent with a port-wine stain localized to trigeminal nerve branches V1–V3 (Figures 1 and 2). No additional vascular lesions or hemangiomas were present. Neurological examination showed intact consciousness, normal muscle tone and strength, symmetric physiological reflexes, negative bilateral Babinski signs, and no focal deficits or hemiparesis. Cardiovascular, respiratory, and abdominal examinations were normal. Ophthalmologic examination revealed decreased visual acuity in the right eye (>3/60), tortuous conjunctival arteries, increased cup-to-disc ratio, and elevated intraocular pressure (N+1/P), consistent with secondary glaucoma.

A non-contrast head CT demonstrated cortical and subcortical calcifications (347 HU) with a classic tram-track appearance in the right parietal, temporal, and occipital lobes. Asymmetric cerebral size was observed, with right hemispheric atrophy (Figure 3). Sleep EEG indicated epileptogenic discharges with mild cerebral dysfunction. Advanced imaging, including contrast-enhanced brain MRI, was unavailable due to resource limitations. Genetic testing for GNAQ mutations and retinal angiography could not be performed for similar reasons. Routine laboratory investigations, including complete blood count, were normal.

Based on clinical findings and available investigations, the patient was diagnosed with classic Sturge–Weber Syndrome, accompanied by secondary glaucoma and epilepsy. The treatment plan consisted of valproic acid for seizure control and topical timolol to reduce intraocular pressure, as advised by the ophthalmology team. The patient was scheduled for regular follow-up at pediatric and ophthalmology clinics to monitor seizure frequency, visual status, and intraocular pressure.

The expected outcomes were satisfactory seizure control without recurrence, stabilization of intraocular pressure, and prevention of progressive optic nerve damage. During three days of inpatient care, no further seizures occurred, and intraocular pressure improved compared with initial measurements. The patient was discharged in stable condition with maintenance therapy and planned outpatient monitoring.

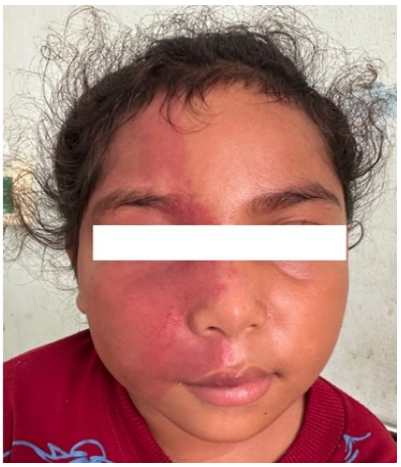

Figure 1. Port-wine stain on the right face within the V1–V2 distribution of the trigeminal nerve.

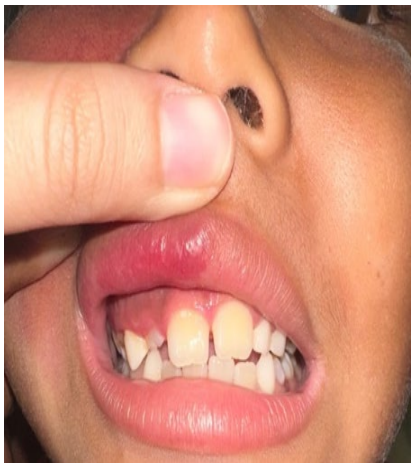

Figure 2. Gingival involvement with thickening of the upper right gingiva.

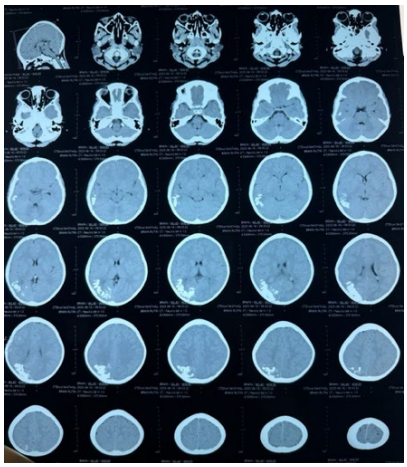

Figure 3. Non-contrast head CT showing cortical–subcortical calcifications with a tram-track sign in the right parietal, temporal, and occipital lobes, alongside right hemispheric atrophy.

DISCUSSION

This case demonstrates the classical clinical and radiological spectrum of SWS in a child managed successfully in a resource-limited setting. The presence of a facial capillary malformation within the trigeminal nerve distribution, early-onset seizures, ocular involvement, and unilateral cortical calcifications represents a pattern frequently observed in SWS with cerebral involvement. Cutaneous and mucosal changes, including gingival thickening, typically reflect progressive vascular ectasia, consistent with previous studies reporting that more than 80% of patients with brain involvement also exhibit orofacial manifestations.[10,11] Neurologically, seizure onset within the first year of life is strongly associated with underlying leptomeningeal angiomas, chronic hypoperfusion, and gliosis.[12,13] This patient experienced recurrent generalized seizures, aligning with data from previouse study who reported that 60–70% of children with SWS develop epilepsy, often requiring long-term therapy.[14]

In the present case, valproic acid was selected as the initial antiepileptic agent due to its broad-spectrum efficacy, suitability for both focal and generalized seizure types, and relatively favorable neurodevelopmental safety profile in pediatric populations.[15,16] Evidence from Wang et al. also supports comparable seizure control between valproic acid and levetiracetam, with fewer behavioral adverse effects in those receiving valproic acid.[17] These considerations, along with limited pharmacologic availability at the treating facility, justify the choice of valproic acid as the first-line therapy. The patient's seizure cessation during hospitalization further reflects an appropriate therapeutic response.

Ocular involvement in this case, characterized by elevated intraocular pressure and optic nerve cupping, is among the most common complications of SWS. Secondary glaucoma arises due to elevated episcleral venous pressure or structural anomalies of the anterior chamber angle.[21] Timolol was selected as initial therapy, consistent with current pediatric evidence showing comparable intraocular pressure reduction to prostaglandin analogs but with fewer side effects.[18-20] In settings where advanced ophthalmologic interventions are limited, early initiation of topical β-blockers may help delay disease progression. Surgical intervention remains the standard of care for refractory cases; however, this patient demonstrated symptomatic improvement with medical therapy alone.

Imaging findings played a central role in establishing the diagnosis. Despite the known superiority of contrast-enhanced MRI for detecting leptomeningeal vascular malformations,[24] non-contrast CT remains a valuable diagnostic modality in resource-limited settings. The tram-track calcifications observed in this patient represent a well-recognized hallmark of SWS and often correlate with chronic hypoxic injury and gliosis.[22,23] EEG findings of epileptogenic activity further supported the clinical diagnosis and guided antiepileptic therapy selection. The combination of characteristic cutaneous findings and supportive CT and EEG results allowed for a confident diagnosis despite the unavailability of MRI and genetic testing.[25]

Throughout observation, the patient demonstrated good clinical improvement, including cessation of seizures and reduction of intraocular pressure. These outcomes are consistent with reports suggesting that medically managed SWS can achieve favorable functional results when treatment adherence and multidisciplinary follow-up are maintained.[26] Regular monitoring remains essential, particularly for seizure recurrence, visual function, and neurodevelopmental progression.

Overall, the clinical trajectory of this patient reinforces the importance of early recognition of hallmark SWS features and illustrates how timely, evidence-based medical management can achieve favorable outcomes even when advanced imaging and genetic testing are unavailable. This case highlights that, in resource-constrained environments, careful clinical evaluation supported by essential imaging modalities such as CT can effectively guide diagnosis and management while preventing long-term neurological and ophthalmologic complications.

CONCLUSIONS AND RECOMMENDATION

This case highlights that Sturge–Weber Syndrome can be reliably identified through its distinctive clinical features and characteristic CT findings, even in the absence of MRI or genetic testing. Conservative treatment combining antiepileptic therapy and topical management for glaucoma produced a favorable short-term outcome, demonstrating that multidisciplinary care remains effective in resource-limited environments. Early recognition, treatment adherence, and structured follow-up are crucial to prevent neurological and ophthalmological deterioration. Strengthening access to essential diagnostic tools and long-term monitoring is needed to improve outcomes for children with SWS in low-resource settings. Further research is warranted to evaluate long-term prognosis and the applicability of current management strategies in Indonesian pediatric populations.

REFERENCES

1. Sánchez-Espino LF, Ivars M, Antoñanzas J, Baselga E. Sturge-Weber syndrome: a review of pathophysiology, genetics, clinical features, and current management approaches. *Appl Clin Genet*. 2023;16:63-81. https://doi.org/10.2147/TACG.S363685
2. 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;368:1971-1979. https://doi.org/10.1056/NEJMoa1213507
3. Valery CB, Comi AM. Sturge-Weber syndrome: updates in pathogenesis, diagnosis, and treatment. *Ann Child Neurol Soc*. 2023;1:186-201. https://doi.org/10.1002/cns3.20031
4. Ketaren RJ, Cenadi JC. Sturge-Weber syndrome: a

glimpse into a rare clinical diagnosis. *Magna Neurol*. 2025;3:128-131. https://doi.org/10.20961/magnaneurologica.v3i2.1823
5. Solomon C, Comi A. Sturge-Weber syndrome: updates in translational neurology. *Front Neurol*. 2024;15:1493873. https://doi.org/10.3389/fneur.2024.1493873
6. Joslyn KE, Truver NF, Comi AM. A review of Sturge-Weber syndrome brain involvement, cannabidiol treatment, and molecular pathways. *Molecules*. 2024;29(22):5279. https://doi.org/10.3390/molecules29225279
7. El Hachem M, Diociaiuti A, Galeotti A, et al. Multidisciplinary, multicenter consensus for the care of patients affected with Sturge-Weber syndrome. *Orphanet J Rare Dis*. 2025;20:3527. https://doi.org/10.1186/s13023-024-03527-w
8. Raval DM, Rathod VM, Patel AB, Sharma B, Lukhi PD. Sturge-Weber syndrome: a rare case report. *Cureus*. 2022;14:e28786. https://doi.org/10.7759/cureus.28786
9. Karn M, Barma A, Ojha L, et al. Sturge-Weber syndrome: a case report. *Clin Case Rep*. 2024;12:e9452. https://doi.org/10.1002/ccr3.9452
10. Yeom SE, Comi AM. Updates on Sturge-Weber syndrome. *Stroke*. 2022;53:3769-3779. https://doi.org/10.1161/STROKEAHA.122.038585
11. Disse S, Ramantani G, Küpper H, et al. Sturge-Weber syndrome in a multinational pediatric cohort: a systematic analysis of different types. *Orphanet J Rare Dis*. 2025;20:3769. https://doi.org/10.1186/s13023-025-03769-2
12. Sabeti S, Ball KL, Bhattacharya SK, et al. Consensus statement for the management and treatment of Sturge-Weber syndrome: neurology, neuroimaging, and ophthalmology recommendations. *Pediatr Neurol*. 2021;121:59-66. https://doi.org/10.1016/j.pediatrneurol.2021.04.013
13. Yeom SE, Cohen B, Weiss CR, et al. Genetic testing in the evaluation of individuals with clinical diagnosis of atypical Sturge-Weber syndrome. *Am J Med Genet A*. 2023;191:983-994. https://doi.org/10.1002/ajmg.a.63106
14. Mozaffari K, Krishnakumar A, Chen JS, et al. Seizure outcomes in children with Sturge-Weber syndrome undergoing epilepsy surgery: an individual participant data meta-analysis. *Seizure*. 2023;107:43-51. https://doi.org/10.1016/j.seizure.2023.03.008
15. Senthil S, Durgam SS, Ali H, Pratinya KG, Krishnamurthy R, Mandal AK. Treatment outcomes of primary combined trabeculotomy with trabeculectomy in early-onset glaucoma with Sturge-Weber syndrome. *J Glaucoma*. 2024;33:340-346. https://doi.org/10.1097/IJG.0000000000002355
16. Formisano M, di Pippo MC, Scuderi L, Abdolrahimzadeh S. Current concepts on diffuse choroidal hemangioma in Sturge-Weber syndrome. *Ophthalmic Genet*. 2021;42:375-382. https://doi.org/10.1080/13816810.2021.1910963
17. Wang S, Liu QZ, Zhao R, et al. Seizure, motor, and cognitive outcomes after epilepsy surgery for patients with Sturge-Weber syndrome: results from a multicenter study. *Neurology*. 2024;103. https://doi.org/10.1212/WNL.0000000000209525
18. Wen T, Wang L, Luo H, Tang L. Sturge-Weber syndrome secondary glaucoma: from pathogenesis to treatment. *Eye Vis (Lond)*. 2025;12:16. https://doi.org/10.1186/s40662-025-00432-6
19. Shah AD, Alexieff P, Tatachar P. Sturge-Weber syndrome: a narrative review of clinical presentation and updates on management. *J Clin Med*. 2025;14:2182. https://doi.org/10.3390/jcm14072182
20. Kaplan EH, Kossoff EH, Bachur CD, et al. Anticonvulsant efficacy in Sturge-Weber syndrome. *Pediatr Neurol*. 2016;58:31-36. https://doi.org/10.1016/j.pediatrneurol.2015.10.015
21. Altavilla E, De Giacomo A, Greco AM, et al. New trigger for stroke-like episode in Sturge-Weber syndrome: a case report. *Children (Basel)*. 2025;12:589. https://doi.org/10.3390/children12050589
22. Karagianni MD, Brotis AG, Tasiou A, et al. Hemispherotomy revised: a complication overview and a systematic review meta-analysis. *Brain Spine*. 2023;3:101766. https://doi.org/10.1016/j.bas.2023.101766
23. Smegal LF, Vedmurthy P, Ryan M, et al. Cannabidiol treatment for neurological, cognitive, and psychiatric symptoms in Sturge-Weber syndrome. *Pediatr Neurol*. 2023;139:24-34. https://doi.org/10.1016/j.pediatrneurol.2022.10.014
24. Lo W, Marchuk DA, Ball KL, et al. Updates and future horizons on the understanding, diagnosis, and treatment of Sturge-Weber syndrome brain involvement. *Dev Med Child Neurol*. 2012;54:214-223. https://doi.org/10.1111/j.1469-8749.2011.04169.x
25. Gupta SS, Joslyn KE, McKenney KD, Comi AM. Biomarker development in Sturge-Weber syndrome. *J Neurodev Disord*. 2025;17:9640. https://doi.org/10.1186/s11689-025-09640-6
26. Smegal LF, Sebold AJ, Hammill AM, et al. Multicenter research data of epilepsy management in patients with Sturge-Weber syndrome. *Pediatr Neurol*. 2021;119:3-10. https://doi.org/10.1016/j.pediatrneurol.2021.02.006