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**CASE REPORT** **OPEN ACCESS**

# STURGE - WEBER SYNDROME: A GLIMPSE INTO A RARE CLINICAL DIAGNOSIS

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**ABSTRACT**

**Background:** Sturge-Weber syndrome is a spectrum of neurocutaneous disorders characterized by angiomas involving the face, choroid, and leptomeninges. Patients present with the typical "port wine stain", acute seizure onset, and increasing hemiparesis.
**Case:** A 1-year-old female presented to the ED with a seizure that coincided with a fever. The patient had two generalized tonic-clonic seizures in a day, which lasted less than 5 minutes. Before this, the patient had a history of seizures that started at 6 months old. The patient had an erythematous patch covering the right side of her face, along with left-sided weakness and positive plantar reflex on the left foot. Head non-contrast MRI scan showed a hypointense lesion/blooming artifact in the right frontal lobe on T2FFE and a prominent vascular structure on the right frontal lobe, accompanied by the widening of multiple veins and sinuses.
**Discussion:** Sturge-Weber syndrome is complete when both facial angioma and leptomeningeal involvement are present. Diagnosis is confirmed through clinical features, with seizures and hemiparesis correlating to imaging findings. In our patient's case, a type I Sturge-Weber syndrome diagnosis was made. The patient was started on oral anticonvulsants and future controls.
**Conclusion:** Sturge-Weber syndrome is a clinical syndrome with many neurological complications. The lack of specific treatment urges an early diagnosis, as it can affect the patient's outcome.

**Keywords:** leptomeningeal angioma, port-wine stain, Sturge-Weber syndrome



## Introduction

Sturge-Weber syndrome, also known as encephalotrigeminal angiomatosis, is a spectrum of neurocutaneous disorders characterized by angiomas involving the face, choroid, and leptomeninges. Patients typically present with seizure as the first neurological manifestation, with the typical "port wine stain" or "nevus flammeus", the facial capillary vascular malformation, which is typical in cases of Sturge-Weber syndrome. Increasing hemiparesis will follow along with the seizures. Sturge-Weber syndrome diagnoses are based on the typical clinical symptoms, facial appearance, and MRI findings. Since there is no specific treatment for Sturge-Weber syndrome, clinicians should aim to minimize seizure activity using anticonvulsants. Surgical treatment is an option if the patient has refractory seizures following medication.[1]

This case report describes a 1-year-old female who presented with two episodes of generalized tonic-clonic seizures, accompanied by characteristic facial features of Sturge-Weber syndrome. A definitive diagnosis of Sturge-Weber syndrome was confirmed following a thorough physical examination and necessary diagnostic tests. The patient's clinical presentation aligned with the known manifestations of this rare neurocutaneous disorder, reinforcing the diagnosis.

## Case Report

This case was reported from Rumah Sakit Siloam Hospitals Lippo Village in March 2024. We report the case of a 1-year-old female who presented to the emergency department with complaints of a seizure that coincided with a fever. The patient also presented with vomiting and diarrhea. Further history was taken, and it was noted that the patient had two generalized tonic-clonic seizures in a day, which lasted below 5 minutes and coincided with a fever. The seizure subsided after giving the patient a dose of suppository diazepam. The patient has had a history of seizures; the first onset started at 6 months old; the patient has never had any medications for the seizures, antenatal history proved unremarkable, and they had no trouble at delivery. The patient weighed 11.1 kg, had no observable growth delays, and was fully vaccinated for up to 1 year. Further physical examination revealed that the patient had an erythematous patch covering the right side of the patient's face, causing concerns about a port wine stain. A neurological examination revealed left-sided weakness. The plantar reflex was found to be positive on the left foot. The patient was then diagnosed with acute symptomatic seizure with suspected Sturge Weber syndrome, accompanied by acute gastroenteritis, and subsequently sent for a head MRI scan and electroencephalogram testing (Figure 1).

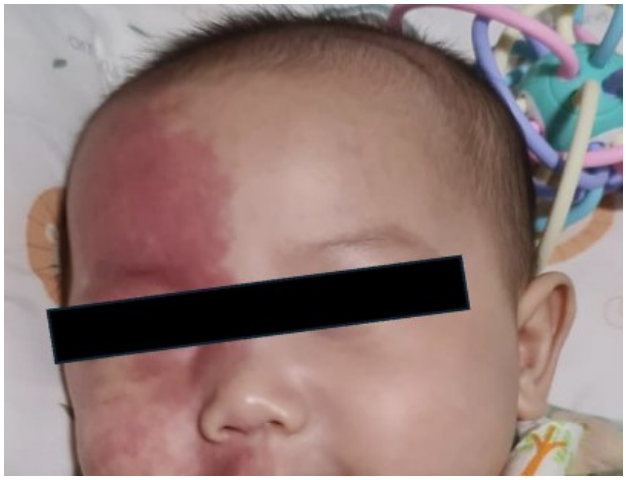

**Figure 1.** Our patient's facial angioma represents the typical "port-wine stain" found in Sturge-Weber Syndrome

Electroencephalogram testing showed that the patient had 3rd-degree abnormal EEG readings that showed slow and asymmetrical background, vertex, and spindle waves over the right hemisphere. Head non-contrast MRI scan showed a hypointense lesion/blooming artifact in the right frontal lobe on T2FFE, suggestive of intracranial calcifications, with correlation to a non-contrast CT scan and a prominent vascular structure on the right frontal lobe, widening of the superior sagittal sinus, rectus sinus, inferior sagittal sinus, vein of Galen, and the internal cerebral vein, and cortical atrophy in the right frontal and parietal lobe (Figure 2). The findings are suggestive of Sturge-Weber syndrome, and a diagnosis of Type I Sturge-Weber syndrome was made. The patient was started on IV phenytoin medications for her acute gastroenteritis. During the inpatient stay, no seizures were observed, and the patient was discharged after symptoms of acute gastroenteritis subsided and prescribed oxcarbazepine 50 mg bidaily, as well as levetiracetam 200 mg bidaily for oral anticonvulsants.

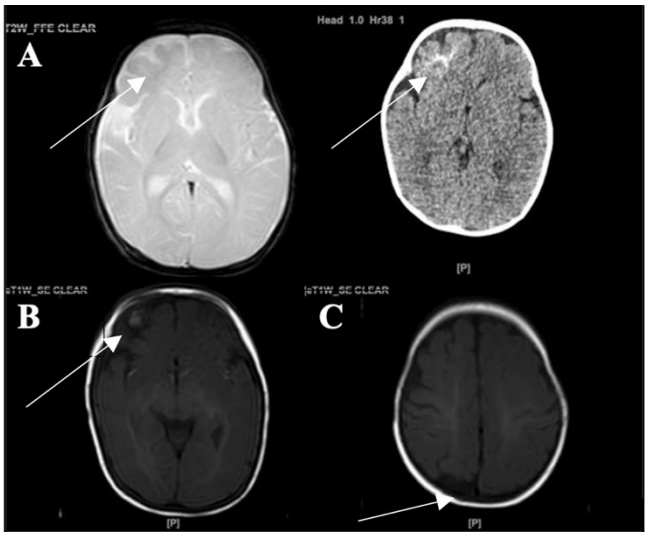


**Figure 2.** (A) Non-contrast head MRI correlates with a non-contrast head CT scan, showing calcification and atrophy of the right frontal lobe and widening of the vein of Galen. (B) A non-contrast head MRI showed a prominent vascular structure in the right frontal lobe. (C) Noncontrast head MRI showing widening of the superior sagittal sinus

## Discussion

Sturge-Weber syndrome is the third most common neurocutaneous disorder, following neurofibromatosis and tuberous sclerosis. It is classically associated with the facial "port mnbv0063 wine stain" in the 1st region of the trigeminal nerve, glaucoma, and ipsilateral occipital leptomeningeal angiomata.[1] Sturge-Weber syndrome impacts approximately 1 in 20.000 live births and is not inherited. Instead, they occur sporadically, with no specific demographics in particular, with it occurring in both males and females with a sex ratio of 1:1. The overall risk of Sturge-Weber syndrome associated with any facial cutaneous vascular malformation is approximately 8%, and rarely does Sturge-Weber syndrome with leptomeningeal angiomata present without any facial involvement.[2,3,4] Activating R183Q GNAQ somatic mutation remains the most common somatic mutation in the pathophysiology of Sturge-Weber Syndrome; recent studies show that GNA11 and GNB2 somatic mutations are related to Sturge-Weber Syndrome.[5,6]

Sturge-Weber syndrome is referred to as complete when both facial angioma and leptomeningeal involvement are present. The Roach Scale has been used for the classification of Sturge-Weber syndrome, which is as follows:

1. Type I: Both facial and leptomeningeal angiomas; may have glaucoma (classic Sturge-Weber syndrome)
2. Type II: Facial angioma, alone with no CNS involvement, may have glaucoma
3. Type III: Isolated leptomeningeal-brain angioma, usually no glaucoma[2,7,8]

According to the above classification, our patient has met the type I Sturge-Weber syndrome requirements. However, Bhagat et al. have reported a case of Sturge-Weber Syndrome where angiomas appear on the extremities.[9] Intracranial angiomatosis is usually unilateral and almost always correlates with the unilateral distribution of the facial angiomatosis, as seen with our patient. Bilateral intracranial involvement, which is reported in approximately 15% of patients, is associated with a higher incidence of seizures and overall poor outcome, as reported by Sugano et al.[11] The mean age of onset of seizures was 24 months in patients with unihemispheric involvement and 6 months with bihemispheric involvement. Our patient's first onset was at 6 months old, making it possible for early diagnosis despite the mean age of seizure onset. Despite the mean age of onset, Saad et al. have reported a case of adult-onset and re-emergence of seizures in a patient with Sturge-Weber syndrome after being seizure-free for almost 20 years.[12]

The golden standard for diagnosis involves a CT scan or MRI. The vascular malformation of the brain is associated with enlarged and tortuous leptomeningeal vessels. In correlation to our patient's imaging, we found a prominent vascular structure in the right frontal lobe. Gyriform calcifications can be seen on neuroimaging and are classically described as the "tram track sign," a telltale sign for Sturge-Weber syndrome.[13,14] This sign is best seen on CT scan imaging. Still, since CT uses ionizing radiation, MRI of the brain with contrast is recommended. The most common locations of these calcifications appear in the occipital or posterior parietal/temporal lobes; however, in our patient's case, the calcifications appear in the right frontal lobe.[15] The MRI findings depend on the stage of the disease, where in the early phase, findings of transient hyperperfusion are seen in leptomeningeal enhancements, such as serpiginous enhancements along the sulci. This is seen with contrast MRI of the head, which was not done in our patient. In the late phase, cortical atrophy is seen alongside a lack of superficial cortical veins with prominent deep medullary or subependymal veins and an enlarged choroid plexus.[16,17] In correlation with our patient's findings, there is a widening or prominence of the superior sagittal sinus, rectus sinus, inferior sagittal sinus, vein of Galen, and the internal cerebral vein, alongside the presence of cortical atrophy, suggesting that our patient has entered the late stage of Sturge-Weber syndrome. Early detection of patients with asymptomatic port-wine stains is possible within 3 months, allowing for earlier intervention.

To date, there is no specific treatment for Sturge-Weber syndrome. The primary aim of treatment in patients is to minimize the occurrence of seizures using anticonvulsive medications. Most patients are treated with either oxcarbazepine, levetiracetam, or a combination of both. Oxcarbazepine has been observed to have better seizure control than levetiracetam and is usually combined with low-dose aspirin.[2,18] Surgery may be considered in patients with pharmacoresistant refractory seizures, which includes the consideration of hemispherectomy or a focal resection of the seizure focus, with studies showing promising results.[19] Topical eye drops that reduce intraocular pressure can be started in patients with glaucoma.[8] As for our patient, we started IV phenytoin for inpatient seizure prophylaxis, continued by bidaily oxcarbazepine 50 mg and bidaily levetiracetam 200 mg, with plans and future controls being made. The prognosis for Sturge-Weber Syndrome is influenced by the extent of hemispherical involvement and the age at which seizures begin. A study by Harmon et al. demonstrated that patients with greater hemispheric involvement and earlier onset of seizures often exhibit lower cognitive function. The study found a notable association between male gender and reduced cognitive performance, suggesting males may be more adversely affected than females. However, the authors noted that these sex differences require further research to clarify their implications, highlighting the complexity of Sturge-Weber Syndrome and the need for ongoing studies to understand the relationships between these factors and cognitive outcomes.[20]

## Conclusion

Sturge-Weber syndrome is a clinical syndrome with a vast spectrum of clinical manifestations. Often, in our practice, patients come in during the late stage of the disease with multiple neurological complications, as is the case with our patients. The lack of specific treatments urges an early diagnosis, as controlling future neurological complications can affect the patient's outcome.

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## References


1. Shirley MD; Tang H; Gallione CJ; Baugher JD; Frelin LP; Cohen B; North PE; Marchuk DA; Comi AM; Pevsner J; Sturge-Weber Syndrome and port-wine stains caused by somatic mutation in Gnaq. The New England journal of medicine. U.S. National Library of Medicine; 2015. DOI: 10.1056/NEJMoa1213507
2. Valery CB, Comi AM. Sturge–Weber syndrome: Updates in pathogenesis, diagnosis, and treatment. Annals of the Child Neurology Society; 2023. 1(3):186-201. 10.1002/cns3.20031
3. Higueros E, Roe E, Granell E, Baselga E. Sturge-Weber syndrome: A review. Actas Dermosifiliográficas. 2017. 108(5):407-417. DOI: 10.1016/j.ad.2016.09.022
4. Yeom S, Comi AM. Updates on Sturge-Weber syndrome. Stroke. 2022. 53(12):3769-79. DOI: 10.1161/STROKEAHA.122.038585
5. Hildebrand MS, Harvey AS, Malone S, Damiano JA, Do H, Ye Z, McQuillan L, Maixner W, Kalnins R, Nolan B, Wood M. Somatic GNAQ mutation in the forme fruste of Sturge-Weber syndrome. Neurology: Genetics; 2018. 4(3):e236. DOI: 10.1212/NXG.0000000000000236
6. Sánchez-Espino LF, Ivars M, Antoñanzas J, Baselga E. Sturge-Weber syndrome: a review of pathophysiology, genetics, clinical features, and current management approaches. The Application of Clinical Genetics; 2023. 63-81. DOI: 10.2147/TACG.S363685
7. Comi AM. Sturge–Weber syndrome. Handbook of clinical neurology; 2015 Jan. 132:157-68. Retrieved on August 15, 2024. Available from: https://www.sciencedirect.com/science/article/abs/pii/B9780444627025000111
8. Wang Y, Geng G, Hu W, Zhang H, Liu Y, Gao Z, Zhang H, Shi J. Epileptic seizures as an initial symptom for Sturge-Weber syndrome type III: A report of two cases. Experimental and Therapeutic Medicine; 2024. 28(1):1-8. DOI: 10.3892/etm.2024.11856
9. Bhagat S, Gupta G, Singh S. Unusual presentation of Sturge-Weber syndrome. Journal of case reports; 2015. 5(1):111-5. DOI: 10.17659/01.2015.0028
10. Sherwani OA, Patra PC, Ahmad SA, Hasan S. Sturge-Weber syndrome: A rare case report. Cureus; 2023. 15(12). DOI: 10.7759/cureus.33056
11. Sugano H, Iimura Y, Igarashi A, Nakazawa M, Suzuki H, Mitsuhashi T, Nakajima M, Higo T, Ueda T, Nakanishi H, Niijima S. Extent of leptomeningeal capillary malformation is associated with severity of epilepsy in Sturge-Weber syndrome. Pediatric Neurology; 2021. 117:64-71. DOI: 10.1016/j.pediatrneurol.2020.11.004
12. Saad AH, Omar SM, Elgilli AA, Omer IA, Jalaleldeen MH. An Atypical Seizure Onset and Re-Emergence in a Refugee with an Undiagnosed Sturge-Weber Syndrome: A Case Report from a Limited Setting. International Medical Case Reports Journal; 2024. 31:615-20. DOI: 10.2147/IMCRJ.S380907
13. Pinto AL, Chen L, Friedman R, Grant PE, Poduri A, Takeoka M, Prabhu SP, Sahin M. Sturge-Weber syndrome: brain magnetic resonance imaging and neuropathology findings. Pediatric Neurology; 2016. 58:25-30. DOI: 10.1016/j.pediatrneurol.2016.01.004
14. Sudarsanam A, Ardern-Holmes SL. Sturge–Weber syndrome: from the past to the present. European Journal of Paediatric Neurology; 2015. 18(3):257-66. DOI: 10.1016/j.ejpn.2014.12.004
15. Bar C, Pedespan JM, Boccara O, et al. Early magnetic resonance imaging to detect presymptomatic leptomeningeal angioma in children with suspected Sturge–Weber syndrome. Developmental Medicine & Child Neurology; 2020. 62(2):227-33. DOI: 10.1111/dmcn.14247
16. Neerupakam M, Reddy PS, Babu BA, Krishna GV. Sturge Weber syndrome: A case study. Journal of Clinical and Diagnostic Research: JCDR; 2017. 11(5):ZD12. DOI: 10.7860/JCDR/2017/25593.9891
17. Sinawat S, Auvichayapat N, Auvichayapat P, Yospaiboon Y, Sinawat S. 12-year retrospective study of Sturge-Weber syndrome and literature review. J Med Assoc Thai; 2015 Jul. 97(7):742-50. Retrieved on August 15, 2024. Available from: https://pubmed.ncbi.nlm.nih.gov/25265774/
18. Kaplan EH, Kossoff EH, Bachur CD, et al. Anticonvulsant efficacy in Sturge-Weber syndrome. Pediatric neurology; 2016. 58:31-6. DOI: 10.1016/j.pediatrneurol.2016.01.005
19. Wang S, Pan J, Zhao M, Wang X, Zhang C, Li T, Wang M, Wang J, Zhou J, Liu C, Sun Y. Characteristics, surgical outcomes, and influential factors of epilepsy in Sturge-Weber syndrome. Brain; 2022. 145(10):3431-43. DOI: 10.1093/brain/awac227
20. Harmon KA, Day AM, Hammill AM, Pinto AL, McCulloch CE, Comi AM, Ball KL, Fisher BJ, Juhász C, Kim H, Koenig J. Quality of life in children with Sturge-Weber syndrome. Pediatric neurology; 2019. 101:26-32. DOI: 10.1016/j.pediatrneurol.2019.07.004