

## Case Report

# “Sturge – Weber Syndrome: A Case Report and Review of Literature”

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### Article History

Received: 04.04.2022

Accepted: 09.05.2022

Published: 12.05.2022

### Journal homepage:

<https://www.easpublisher.com>

### Quick Response Code



**Abstract:** Sturge-Weber syndrome is a rare, sporadically occurring neurocutaneous syndrome characterized by port wine stain (facial nevus flammeus), congenital glaucoma, and anomalous leptomeningeal angiomatous malformation. Port wine stain is usually the first component of the syndrome. Seizures of the side contralateral to the port wine stain occur early in infancy and worsen with age. Radiological investigations, like computed tomography and magnetic resonance imaging are most useful, playing a pivotal role in demonstrating the cerebral changes. We report the case of a three year old child presenting with port wine stain over left half of the body, weakness of right upper and lower limb with an episode of seizure. The child was known to have seizures from 6 months of age and was on irregular anticonvulsant treatment. This case highlights the various neurological manifestations of Sturge-Weber syndrome and how imaging helps to characterize each.

**Keywords:** SWS, port wine stain, seizures.

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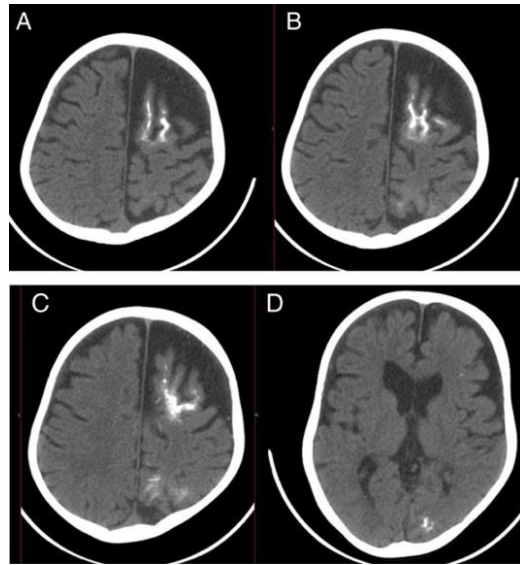
## INTRODUCTION

Among the neurocutaneous syndromes, Sturge-Weber syndrome (SWS) is rare and unique as it is one of the very few phakomatoses that are sporadic and not inherited. It is also one of the most disfiguring syndromes, as a prominent nevus flammeus is seen in majority cases. The reported prevalence of SWS is 1 in 50,000 live births. It is characterized by facial capillary malformations (classically referred to as angiomas even though they are not tumors), accompanied by variable degrees of ocular and neurological anomalies. The exact etiology is unknown but the primary defect may be a developmental insult affecting precursors of tissues that originate in the promesencephalic and mesencephalic neural crest, which later give rise to vascular and other tissue malformations in the meninges, eye, and the dermis. Radiological investigations are most useful, with computed tomography and magnetic resonance imaging playing a pivotal role in demonstrating the cerebral changes.

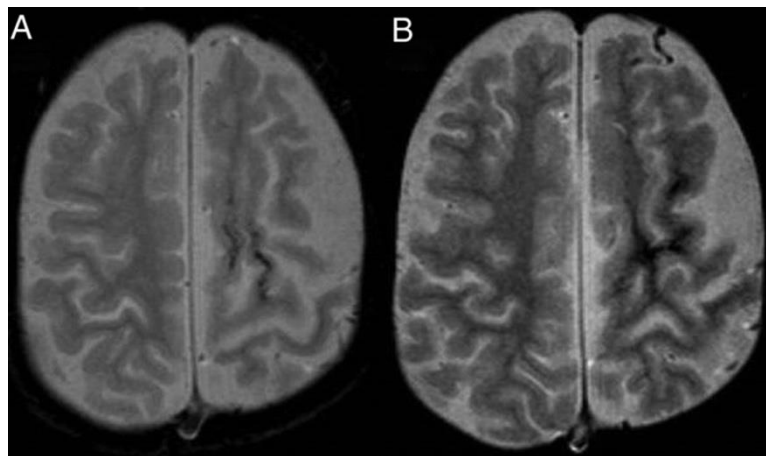
## CASE REPORT

A case of a 3-year-old child with developmental delay presented to the paediatric department with a 3-day history of high-grade fever with chills and rigours. There was associated weakness of the right upper and lower limbs with an episode of seizure that lasted for 5 mins. The child was known to have seizures from 6 months of age and was on irregular anticonvulsant treatment, which was stopped 2 months earlier.

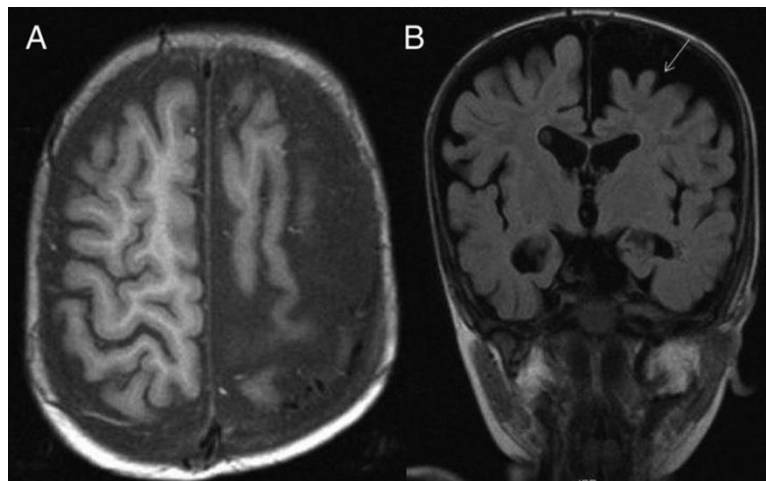
Clinical examination revealed macrocephaly, port-wine stains on the left half of the body and reduced tone in right upper and lower limbs. CT and MRI of the brain revealed subcortical calcifications depicting ‘tram-track’-like appearance in the left frontal, parietal and occipital lobes (figures 1 and 2). Hemiatrophy of the left frontal lobe (figure 3) with prominence of ventricles, basal cisterns, Sylvian fissures, left choroid plexus (figures 4 and 5) and hyperostosis of the calvarium was seen.



**Fig-1**



**Fig-2**



**Fig-3**

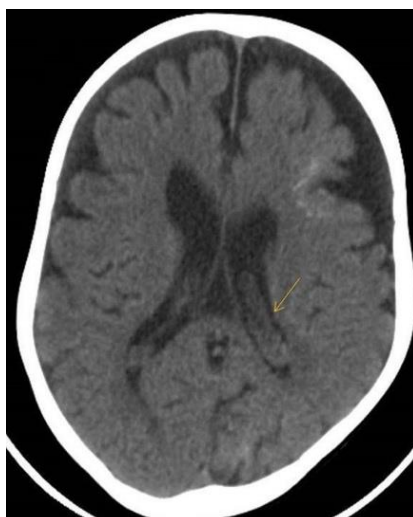


Fig-4

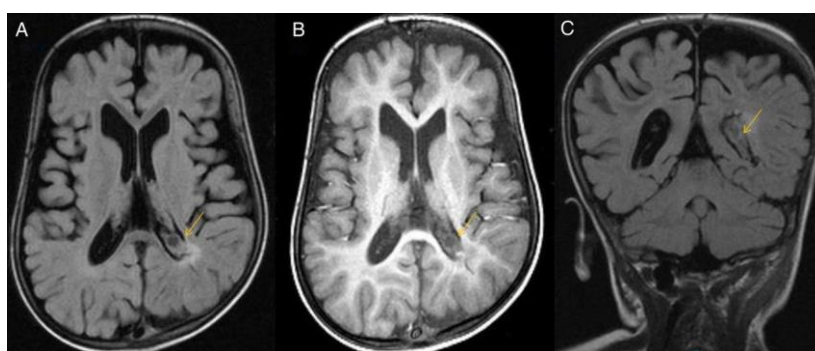


Fig-5

## DISCUSSION

Sturge-Weber syndrome, also known as encephalotrigeminal angiomas, is a rare neurocutaneous syndrome with estimated prevalence of 1 in 50,000 live births. Its hallmarks are variable combinations of Capillary malformation of skin (port wine stain) in the distribution of trigeminal nerve, Retinal choroidal angioma (with or without glaucoma) and cerebral capillary-venous leptomeningeal angioma. Thus SWS is a vascular phakomatosis, which means that unlike the commoner phakomatoses like neurofibromatosis, tuberous sclerosis, or Von Hippel Lindau disease, it manifests without associated neoplasms but with predominant vascular manifestations. The imaging studies are useful for confirming the diagnosis of Sturge-Weber syndrome and evaluating the extent of intracranial involvement and may be important when clinical stigmata are atypical or not yet developed. Both CT and MR imaging are sensitive in providing anatomic information and diagnosis. The clinical finding of port wine stain and imaging findings like cortical subcortical calcifications, the absence of white matter abnormalities like gliosis, and absence of changes in the skull vault make helps in the diagnosis of SWS. Treatment of SWS consists of control of seizures. Hemispherectomy has been recommended early in life in case of refractory seizures, first for better seizure control and also to

promote intellectual development. The prognosis of this case is poor due to early onset of seizures and extensive leptomeningeal angiomas, which will lead to early developmental delay and special educational requirements.

## CONCLUSION

SWS is a rare, sporadic condition due to primary venous dysplasia causing impaired venous outflow and subsequent cerebral ischemia. Certain types can progress to intractable epilepsy and may necessitate radical surgical intervention. Radiologic imaging plays several key roles in the management of SWS patients. MRI confirms the diagnosis of intracranial involvement and helps document the extent of involvement. CT is more sensitive than MRI for detecting cortical calcifications. Imaging plays a pivotal role in diagnosis of this condition.

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**Cite This Article:** Aravind S, Shree varshini.T, Anbumani, Prabakaran M (2022). "Sturge – Weber Syndrome: A Case Report and Review of Literature". *EAS J Radiol Imaging Technol*, 4(3), 26-29.