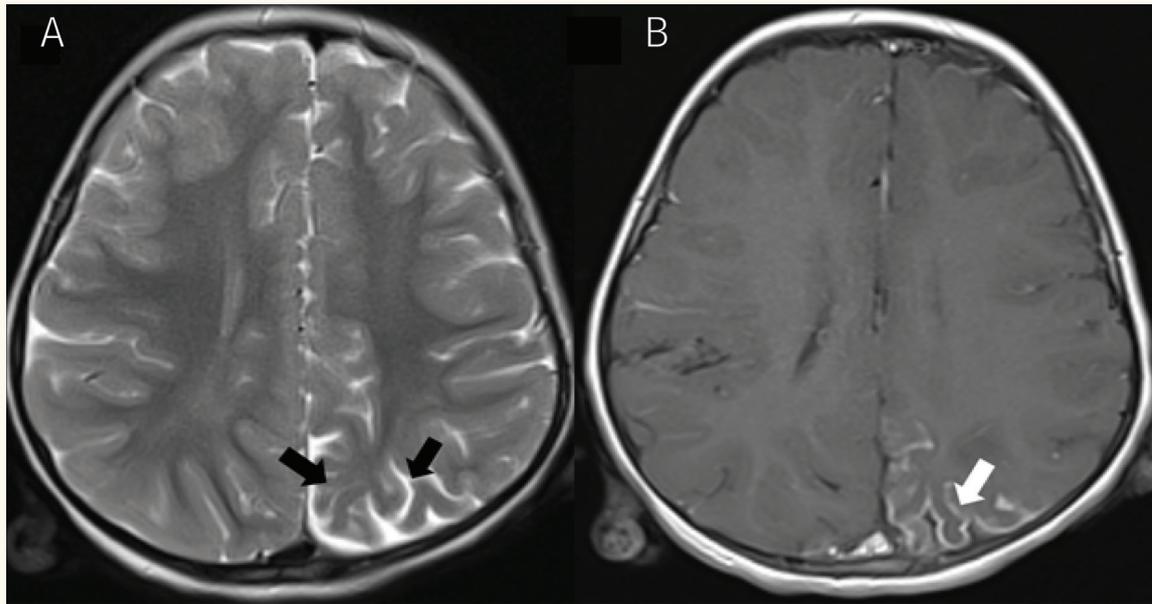


## Three-year-old Girl with Sturge-Weber Syndrome without Facial Nevus

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طفلة عمرها ثلاثة أعوام مصابة بمتلازمة استريج - فيبر من دون وحمه الوجه

روشان كول، رينيث ماني و رانا شعيب حامد

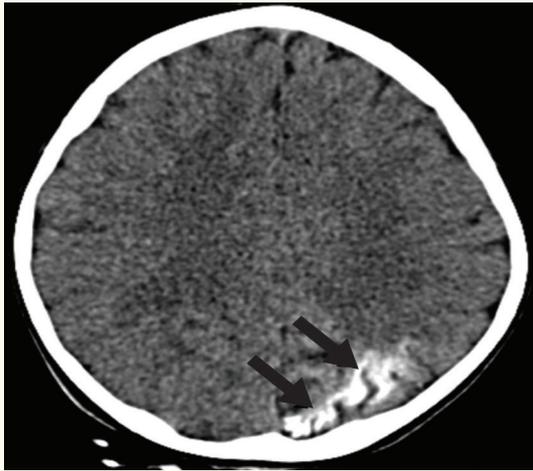


**Figure 1 A & B:** Axial images of (A) T2-weighted magnetic resonance imaging (MRI) brain scan showing focal left occipitoparietal brain atrophy (black arrows) and (B) T1-weighted post-contrast MRI showing cortical enhancement in the gyri (white arrow) consistent with leptomeningeal angiomatosis.

**S**TURGE-WEBER SYNDROME (SWS) IS A neurocutaneous disorder. Classic SWS is characterised by the presence of *nevus flammeus* (port-wine stain) involving the first sensory branch of the trigeminal nerve, ipsilateral leptomeningeal angiomatosis, choroidal angioma and glaucoma.<sup>1</sup> The condition is autosomal recessive in inheritance and reported from all over the world. The presence of a cranial component without facial *nevus* and eye involvement is labelled as type three and is uncommon. We report a girl with SWS syndrome and partial epilepsy who had only the cranial component of the syndrome.

A three-year-old girl was referred to Sultan Qaboos University Hospital (SQUH), Oman, for evaluation and management of her uncontrolled partial motor seizures. She had been born to consanguineous

parents with an uneventful birth. There was nothing relevant in the perinatal period or the family history. The recurrent episodes of seizures had started at the age of three months. The seizures took the form of left-sided gaze deviation with tonic-clonic movements of the left half of the body lasting for five minutes. There were no precipitating factors such as fever or infections. During these episodes, no loss of consciousness or secondary generalisation was noted. The episodes were not followed by post-ictal weakness. She was diagnosed with epilepsy and prescribed carbamazepine in a peripheral hospital. Another anti-epileptic drug was added as the seizures were not controlled. She continued to have one to two episodes per week even with two anticonvulsants. At this time, a third anti-epileptic medication, sodium valproate, was started and the



**Figure 2:** Axial image of the plain computed tomography brain scan showing gyriform hyperdensity in the left occipitoparietal area (arrows) consistent with cortical calcification.

previous drugs were discontinued. With the sodium valproate her seizures were better controlled (one episode every 2–3 months) and she was referred to SQUH for further management. Her development was normal for her age. The clinical examination on presentation showed growth parameters (head circumference, height, weight) in the 25<sup>th</sup> centile with no dysmorphic features and no neurocutaneous markers. She did not have any focal neurological deficit and there was no organomegaly. There was no visual field defect. The seizure history was reviewed and a diagnosis of left simple partial motor seizures was made and the cause investigated. Levetiracetam was added to the treatment for better control of her seizures. This drug is specifically designed for partial seizures and has few side-effects. The baseline blood investigations were normal. The electroencephalogram showed intermittent high-amplitude slowing in the left posterior region with no epileptiform discharges. Spikes and spike-waves are the diagnostic hallmark for epilepsy, but in a child with a structural lesion these abnormalities may not be present. The magnetic

resonance imaging (MRI) scan of the brain revealed a focal left occipitoparietal brain atrophy with enhancing *gyri* [Figure 1]. The brain features shown on the MRI scan were consistent with leptomeningeal angiomatosis. A computed tomography (CT) scan of the brain revealed calcification in the angiomatosis area [Figure 2] thus confirming the diagnosis of SWS. The ophthalmic examination was normal. Since there were no skin lesions, a diagnosis of SWS type 3 was made.

## Comment

SWS is classified into three types, depending upon the extent of the components involved. Type 1 involves the skin, eye and brain. In type 2, there is no brain involvement and in type 3 only the brain is affected. This is due to the variable expressivity seen in inherited diseases. The incidence of classical SWS is 1/50,000 live births, but the incidence of SWS without facial nevus is not known.<sup>2</sup> In one series of 28 patients, three cases were seen with an isolated cranial component, comprising 10.7%.<sup>2</sup> The diagnosis of SWS type 3 is made by CT and MRI scanning and histopathology.<sup>2</sup> SWS patients need treatment for the associated seizures and glaucoma. Surgical therapy should be considered in patients with drug-resistant epilepsy.<sup>1,3</sup>

## References

1. Lo W, Marchuk DA, Ball KL, Juhász C, Jordan LC, Ewen JB, et al. Updates and future horizons on the understanding, diagnosis, and treatment of Sturge-Weber syndrome brain involvement. *Dev Med Child Neurol* 2012; 54:214–23. doi: 10.1111/j.1469-8749.2011.04169.x.
2. Jagtap SA, Srinivas G, Radhakrishnan A, Harsha KJ. A clinician's dilemma: Sturge-Weber syndrome 'without facial nevus'! *Ann Indian Acad Neurol* 2013; 16:118–20. doi: 10.4103/0972-2327.107725.
3. Siri L, Giordano L, Accorsi P, Cossu M, Pinelli L, Tassi L, et al. Clinical features of Sturge-Weber syndrome without facial nevus: Five novel cases. *Eur J Paediatr Neurol* 2013; 17:91–6. doi: 10.1016/j.ejpn.2012.06.011.