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Sturge–Weber Syndrome: About a Case

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Abstract **Case Report**

Sturge–Weber syndrome (SWS) is a rare, congenital neurocutaneous disorder characterized by facial capillary malformations (port-wine stains), leptomeningeal angiomas, and neurological manifestations such as seizures and developmental delay. We report the case of a 16-year-old male with a history of drug-resistant epilepsy and cerebral palsy, who presented with altered consciousness. Neuroimaging revealed gyriform cortical calcifications, ipsilateral cerebral hemiatrophy, and a temporal arachnoid cyst. This case highlights the role of CT and MRI in diagnosing SWS and identifying associated anomalies.

Keywords: Sturge–Weber syndrome, cortical calcifications, cerebral hemiatrophy, arachnoid cyst, epilepsy.

INTRODUCTION

Sturge–Weber syndrome (SWS) is a rare sporadic phakomatosis, with an estimated incidence of 1 in 20,000–50,000 live births, caused by a somatic mosaic activating mutation in the GNAQ gene [1,2]. It is defined by the classic triad of facial capillary malformation (port-wine stain), leptomeningeal angiomatosis, and ocular abnormalities, most notably glaucoma [3]. Neurological manifestations, including seizures, hemiparesis, and cognitive impairment, often appear in infancy or early childhood [4]. Neuroimaging is essential for diagnosis, demonstrating pathognomonic gyriform ("tram-track") calcifications and ipsilateral cortical atrophy. This report presents a case of SWS in an adolescent, with emphasis on the imaging findings and differential diagnosis.

OBSERVATION

We report the case of a 16-year-old male, followed since early childhood for drug-resistant epilepsy and cerebral palsy, presented with loss of consciousness for three days. On examination, he was comatose with a flat port-wine stain over the right fronto-orbital region. There were no signs of acute meningeal irritation.

On clinical examination, we have objectified a flat cutaneous angioma at the level right fronto-orbital. A cerebral CT scan was performed (Figure 1) showing Gyriform cortical calcifications in the right fronto-temporo-parietal region, ipsilateral cortico-subcortical atrophy, and an Incidental right temporal arachnoid cyst. An MRI is also performed (Figure 2) out highlighting Right cerebral hemiatrophy with associated ex vacuo dilatation of the lateral ventricle, ipsilateral gyriform calcifications (hypointense on susceptibility-weighted imaging), and a right temporal cystic lesion consistent with an arachnoid cyst.

associated with voids of gyriform signals on the T2* and SWI sequences. Thus, the diagnosis of Sturge Weber syndrome was discussed, and the patient was referred for further care.

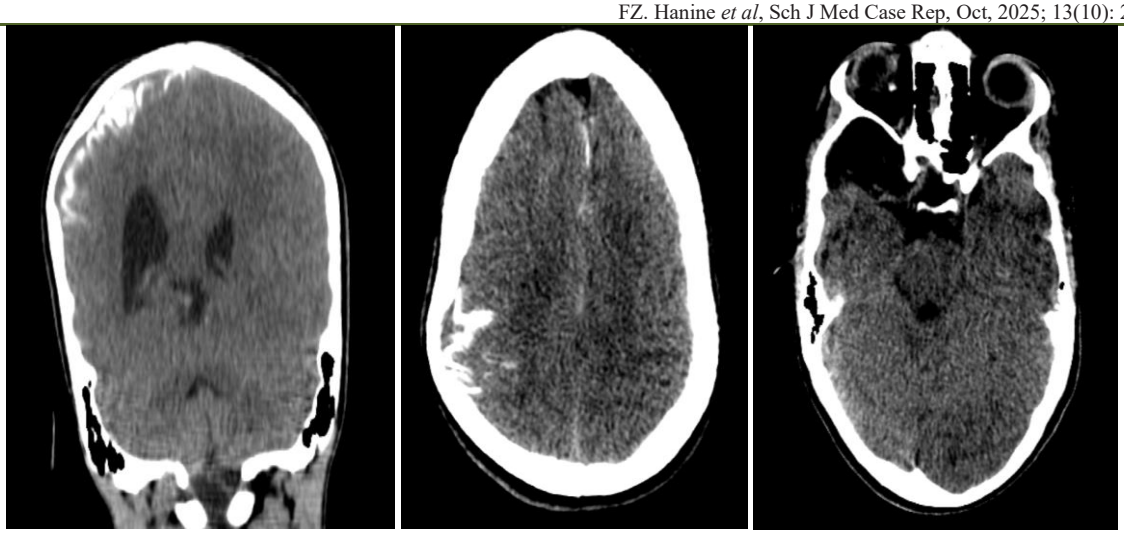

Figure 1: Non-contrast CT scan showing gyriform cortical calcifications in the right fronto-temporo-parietal region with ipsilateral cortical atrophy and a right temporal arachnoid cyst

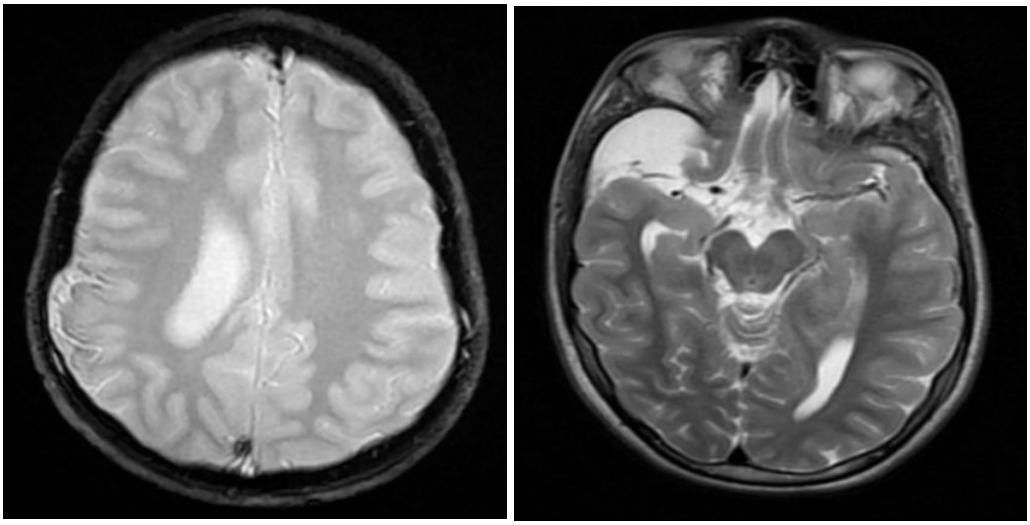

Figure 2: Axial T2 and T2 * weighted MRI showing voids of gyriform signals with right cerebral hemiatrophy and a right temporal arachnoid cyst

DISCUSSION

SWS results from a somatic activating mutation in GNAQ, leading to abnormal capillary–venous development in skin, leptomeninges, and ocular structures [2,5]. Neurological manifestations are mainly due to chronic venous stasis, hypoxia, and secondary cortical injury [6]. Seizures occur in up to 80% of patients, often refractory to treatment [7].

Neuroimaging features:

- CT: Characteristic gyriform cortical calcifications, typically in the occipital or posterior parietal lobes, ipsilateral to the facial angioma.
- MRI: Demonstrates leptomeningeal enhancement, cortical and white matter atrophy, and venous anomalies. SWI and T2* sequences are useful for detecting calcifications.
- Perfusion MRI: May reveal chronic hypoperfusion in affected regions.

Our patient also presented with a temporal arachnoid cyst, which, although likely incidental, can contribute to seizure activity. Differential diagnoses include post-infectious calcifications (e.g., CMV, toxoplasmosis), hypoxic–ischemic encephalopathy, and other phakomatoses such as tuberous sclerosis.

Early diagnosis is crucial, as aggressive seizure control and management of vascular anomalies may improve long-term neurological outcomes [8].

CONCLUSION

This case illustrates the typical presentation of SWS with classic clinico-radiological features. CT and MRI remain essential for diagnosis and follow-up. Recognition of associated anomalies, such as arachnoid cysts, is important for comprehensive management.

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