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CASE REPORT PEER REVIEWED | OPEN ACCESS

Sturge–Weber syndrome: About a case

Sara Habib Chorfa, Kenza Sidki, Soumya El Graini, Amal Lahfidi, Meryem Fikri, Firdaous Touarssa, Mohamed Jiddane

ABSTRACT

Facial port-wine stains are capillary malformations, which can reveal, very rarely, Sturge–Weber syndrome (SWS). The SWS is a severe neurocutaneous syndrome, which involves a facial port-wine stain, ophthalmologic abnormalities, and neurologic signs.

Neuroimaging (computed tomography [CT]-scan/ angio-magnetic resonance imaging [MRI]) provides the diagnosis of SWS and the best age to perform the exam is not established. When a newborn has a facial port-wine stain reaching V1, ophthalmologic examination must be performed in the first months of life, as well as neuroimaging, a treatment of the port-wine stain must be considered. We report the case of a child in whom SWS was suspected based on facial angioma and pharmaco-resistant epilepsy.

Keywords: Facial angioma, Leptomeningeal angiomas, MRI

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INTRODUCTION

Sturge–Weber syndrome, or encephalotrigeminal angiomatosis, is phakomatosis characterized by facial port wine stains and pial angiomas. It is part of a wide spectrum of possible phenotypes included in the cerebrofacial arteriovenous metameric syndrome (CAMS).

Sturge–Weber syndrome is a rare syndrome, with an incidence estimated at 1 in 20,000–50,000 [1]. The diagnosis is usually obvious on account of a congenital facial cutaneous capillary malformation and if there are neurological symptoms or findings, magnetic resonance imaging (MRI) of the brain is undertaken with gadolinium contrast to detect leptomeningeal capillary-venous malformations [2].

CASE REPORT

We report the case of a 7-year-old child admitted for status epilepticus with no notion of awareness between seizures, all evolving in a context of apyrexia, which motivated the family to consult urgently in our service. On examination, the girl was conscious, with no sensory-motor deficit; somatic examination revealed a flat cutaneous angioma on the face occupying the territory of the trigeminal nerve. Brain MRI showed the presence of serpiginous vascular structures of venous origin in the left frontotemporal, right frontoparietal, and peri-mesencephalic regions, calcifications in the right parieto-occipital region, and enlarged cortical gyri with marked contrast enhancement, with partial extension to angiomatous malformation (Figures 1–3).

Electroencephalogram (EEG) revealed a focus of right parieto-occipital region and the ophthalmological examination revealed no abnormality. Therefore, the diagnosis of Sturge–Weber syndrome was retained, and the child was put on medical treatment based on anti-epileptics with good clinical improvement (regression of the seizures).

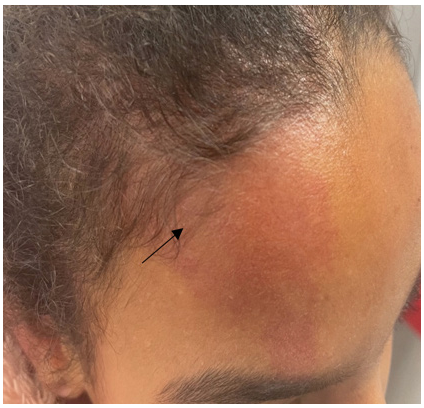

Figure 1: Photograph of the patient's face, showing the facial angioma, involving the V1 territory of the trigeminal nerve.

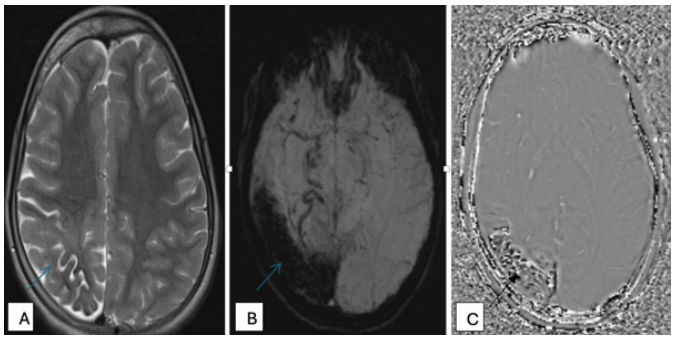

Figure 2 (A–C): Brain MRI showing a venous anomaly in the right parieto occipital cortical and extensive area with hyposignal in axial T2-weighted and SWI (blue arrow in A and B), and corresponding signal dropout in C (black arrow), related to calcifications.

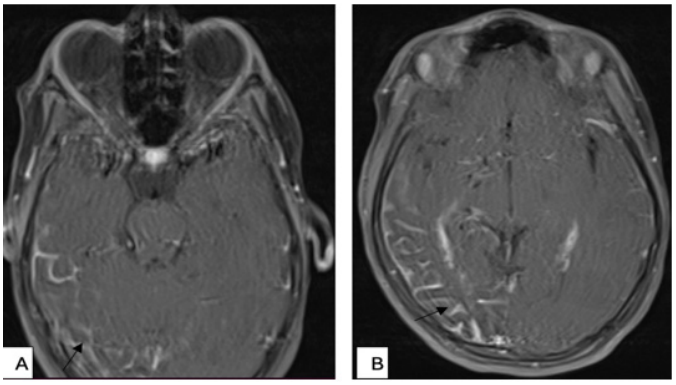

Figure 3(A and B): Brain MRI with injection gadolinium showing a leptomeningeal angiomas (black arrow).

DISCUSSION

Sturge–Weber–Krabbe syndrome is a very rare neurocutaneous and ocular phacomatosis with malformative vascular substratum, where the cutaneous involvement is often unilateral reaching the territory of the trigeminal nerve. Its presence is very suggestive of the diagnosis but its absence does not exclude it. It is characterized by a port-wine stain birthmark on the forehead and upper eyelid on one side of the face. The birthmark can vary in color from light pink to deep purple and is caused by an overabundance of capillaries (small blood vessels) around the trigeminal nerve just beneath the surface of the face [3].

The ocular abnormalities are dominated by glaucoma (30–70%), a choroidal angioma (found in 40–50% of the cases), and a retinal detachment. Neurological signs are dominated by epilepsy (75–90% of cases), often early and severe, partial motor seizures of the contralateral hemibody in 70% of cases. Motor deficit and mental retardation are found in 50% of cases [4]. Psychiatric disorders, which have been described, remain rare. A classification has been proposed by Roch and Coll which describes 3 forms of Sturge–Weber syndrome. Type 1 (classic) includes intracranial and facial manifestations; Type 2 includes facial involvement only without central changes; and Type 3 includes intracranial manifestations only. Our case belongs to Sturge–Weber syndrome Type 1 according to Roch and Coll [5].

Cross-sectional imaging plays a key role in the diagnosis of Sturge–Weber syndrome. Magnetic resonance imaging and cerebral CT-scan will look for a cerebral atrophy, could be focal or hemispherical, but often ipsilateral to the angioma; intracranial calcifications in the shape of an "S," gyriform or train rail, subcortical at the level of the meningeal arteries and the cortical veins; enlargement and calcifications of the choroid plexus ipsilateral to the angioma; cortical and gyriform enhancement. In most cases (82%), angiography is abnormal and demonstrates absent superficial cortical veins with abnormal and enlarged deep venous drainage [6].

The treatment can vary, preventive, and curative, based on anti-epileptics. As for the surgical treatment of the pial angioma, it consists of a hemispherectomy and must be retained for unilateral forms of severe and serious evolution of epilepsy or intellectual regression.

CONCLUSION

Sturge–Weber syndrome is a congenital vascular disease characterized by a wine-colored facial angioma, a leptomeningeal angioma and neurological complications (epilepsy, focal neurological deficits, and intellectual disability). The diagnosis is clinical and brain imaging looks for the presence of a leptomeningeal angioma and cerebral calcifications. Treatment is symptomatic.

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Sara Habib Chorfa – Conception of the work, Acquisition of data, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Kenza Sidki – Design of the work, Acquisition of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Soumya El Graini – Design of the work, Drafting the work, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Amal Lahfidi – Interpretation of data, Drafting the work, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Meryem Fikri – Interpretation of data, Drafting the work, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

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Mohamed Jiddane – Acquisition of data, Interpretation of data, Drafting the work, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

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Authors declare no conflict of interest.

Data Availability

All relevant data are within the paper and its Supporting Information files.

ABOUT THE AUTHORS

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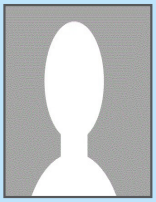

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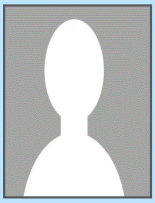

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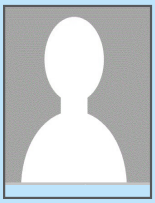

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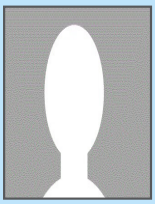

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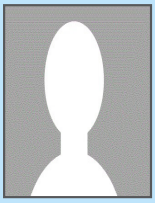

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