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Sturge-Weber Syndrome A Case Report

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Abstract **Case Report**

Sturge-Weber syndrome (SWS) or encephalo-trijeminous angiomatosis combines congenital facial angioma, leptomeningeal angioma and choroidal angioma. Diagnosis is made with MRI, ideally before ocular complications arise. In Sturge-Weber syndrome, neurodevelopmental outcome depends on recognition of the signs of severity and appropriate therapeutic management of the epilepsy. We report the case of a 9-year-old boy in whom Sturge-Weber syndrome was suspected on the basis of a facial angioma and drug-resistant epilepsy.

Keywords: Sturge-Weber, MRI, angioma.

INTRODUCTION

Sturge-Weber syndrome (SSW) is a neurocutaneous angiomatosis combining congenital facial angioma, leptomeningeal angioma (LMA) and choroidal angioma. Affected patients present with epilepsy in 75-90% of cases. It is a rare disease with an unpredictable course [3].

MRI is the main imaging modality recommended in clinical practice. Other imaging modalities (CT, PET) have a limited role and may be used for other clinical investigations or specific research [1].

CASE REPORT

We report the case of a 9-year-old child referred to our Radiology Department for drug-resistant epileptic seizures. No epilepsy was noted in the family history, and there was no evidence of consanguinity. Le showed good psychomotor development. Clinical examination revealed a facial angioma.

The EEG revealed a focus of right fronto-parietal distress, and the ophthalmological examination was normal.

A cerebral MRI scan (Figure 1) showed atrophy of the right cerebral hemisphere, leading to a decrease in volume, with significant homolateral parieto-occipital leptomeningeal enhancement, the site of calcifications in the T2* signal void. Hypertrophy of the choroid plexuses of the right VL was also noted, as was dilatation of the right fronto-parietal transcerebral veins.

In view of the clinical and radiological findings, the diagnosis of Sturge Weber syndrome type 1 was accepted.

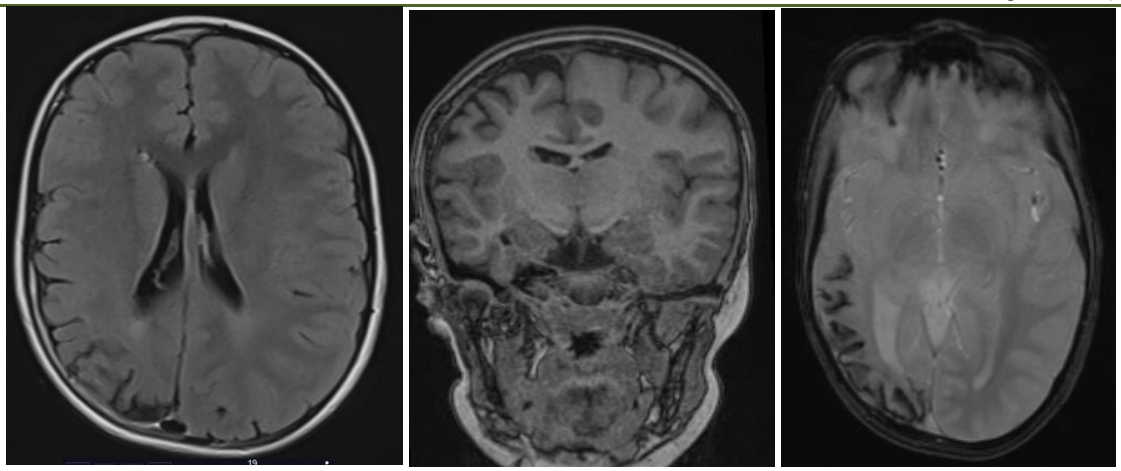

Figure 1: Right parieto-occipital atrophy associated with white matter-gray matter dedifferentiation with calcifications in signal gaps

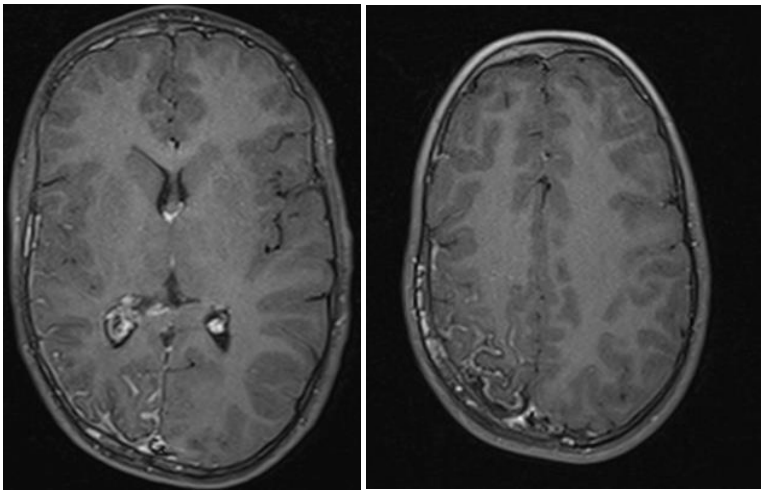

Figure 2: T1 sequence after Gadolinium injection Significant right parieto-occipital leptomeningeal enhancement and right VL choroid plexus hypertrophy

DISCUSSION

Sturge-Weber syndrome is a very rare neurocutaneous and ocular phacomatosis with a vascular malformative substrate.

The incidence of Sturge-Weber syndrome is not perfectly known, but is estimated at 1 per 20,000 to 50,000 live births. In France, it is estimated at 1 case per 100,000 births. In Morocco, there is as yet no exhaustive register of these patients. The disease is not familial but sporadic, affecting both sexes equally, and has been described in all ethnic groups [10, 15, 16, 18].

Neurological signs are dominated by epileptic seizures (75-90% of cases), often early and severe, bilateral in over 90% of cases. These seizures become progressively refractory to medication and are often poorly managed by the patient and family. Motor deficit and mental retardation are present in 50% of cases [6, 15-17].

The facial nevus is often unilateral, affecting the territory of the trigeminal nerve. Its presence is highly suggestive of the diagnosis, but its absence does not rule it out. Ocular involvement is dominated by glaucoma (30-70%), with choroidal angioma found in 40-50% of cases. The fundus should therefore be examined for reddish retinal elevation [6, 10, 14].

A classification has been proposed by Roch *et al*., who describe 3 forms of Sturge Weber:
Type 1 (classic): intracranial and facial manifestations;
Type 2: Facial involvement only, without central changes;
Type 3: Intracranial manifestations only.

Our case belongs to Sturge Weber syndrome type 1 according to Roch et Coll. Our patient's main symptom was convulsive seizures, with no motor deficit or mental retardation. The facial cutaneous angioma was a major factor in the suspicion of the diagnosis, even before imaging.

Cross-sectional imaging plays a vital role in the diagnosis of Sturge Weber syndrome. MRI is the examination of choice for early detection and follow-up

of this pathology [6, 13]. A cerebral CT scan will reveal: focal or hemispheric cerebral atrophy, often homolateral to the angioma; intra-cranial "S"-shaped, gyriform or train-rail calcifications, subcortical to the meningeal arteries and cortical veins; hypertrophy and calcifications of the choroid plexus, homolateral to the angioma; cortical, gyriform contrast.

Cerebral MRI, which is more sensitive than CT [13], reveals the following:
In the early phase, leptomeningeal enhancement (in the form of serpiginous enhancement along the sulci), with diffusion restriction in the case of associated ischemia.

In the late phase, there is a T2 hypersignal in the region of gliosis, with a decrease in pial enhancement associated with cortical atrophy. Gyriform calcifications are best seen on T2* or SW1 sequences (susceptibility-weighted images) and appear as areas where there is no increase in diffusion [15, 16].

Angiography is no longer performed except in cases of severe epileptic disease, when palliative hemispherectomy is proposed; its value lies in better assessing the extension of the angioma [5].

Functional brain imaging is not commonly used. It has specific indications, often enabling early diagnosis. It studies cerebral glucose metabolism using positron emission tomography (PET), and regional cerebral blood flow using functional imaging such as single photon emission tomography (SPECT) [4, 12]. These examinations look for: in the early stages of the disease: transient regional hypermetabolism of the cortex at the level of the pial angioma; in the advanced stages: hypometabolism in PET and hypoperfusion in SPECT at the level of calcified areas.

The EEG is often abnormal, showing a slowing of background activity in one or both hemispheres in relation to cerebral distress [2, 6], as confirmed in our case.

Treatment is multifaceted, preventive and curative, based on antiepileptic drugs. Surgery may be considered in patients with refractory seizures. For glaucoma, medical treatment is often preferred, given the risk of significant intra- and post-operative complications, but is often ineffective [5, 8, 11]. In our patient's case, surgical treatment is planned following a multidisciplinary decision.

CONCLUSION

Sturge-Weber syndrome (SWS) is a rare mesoblastic phacomatosis classically characterized by facial and leptomeningeal angiomatosis, often responsible for epilepsy. Other disorders, such as glaucoma, are also possible. Diagnosis is made by magnetic resonance imaging (MRI) and computed tomography (CT). Treatment is based on the symptoms, and is best undertaken on a multidisciplinary basis.

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