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Research Article

STURGE WEBER SYNDROME: AN EXCEPTIONAL CAUSE OF EPILEPSY IN CHILDREN (A CASE REPORT)

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A R T I C L E I N F O

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A B S T R A C T

Sturge-Weber syndrome (SWS) or encephalo-facial angiomatosis, is a rare congenital neuro-cutaneous and ocular syndrome. It comprises two types of malformations: congenital facial capillary angioma and leptomeningeal capillaro-venous angioma, most often homolateral parieto-occipital. The most frequent clinical neurological presentation is epileptic seizures. Magnetic resonance Imaging (MRI), plays an important role in establishing the diagnosis. We report a case of SSW grouping the different elements of the original triad: planar angioma of the hemiface, leptomeningeal angiomatosis and choroidal angioma with glaucoma.

INTRODUCTION

Sturge-Weber-Krabbe syndrome (SWK) is a neurocutaneous angiomatosis combining congenital facial angioma, leptomeningeal angioma (LMA) and choroidal angioma. It is usually a non-familial, sporadic condition affecting all races and also both sexes with discrete male predominance, clinically affected patients present with epilepsy in 75-90% of cases. Tomodensitometry and magnetic resonance imaging are considered the key to diagnosis of this syndrome.

Patient and observation

We report the case of an 11 year old female child, with a history of intellectual retardation, admitted for first episode of seizure. There was no personal or familial epilepsy and no consanguinity. The beginning of the symptomatology was brutal by the installation the same day of tonic-clonic seizures left hemi-body, having lasted 30 mn and yielding under Diazepam, the whole evolves in a context of fever quantified 38°C. On admission, the patient was febrile at 38°C, somnolent, without sensory-motor deficits and the rest of the examination showed a cutaneous angioma on the face occupying the territory of the right V (figure 1). A cerebral CT scan was performed and showed calcifications and a right temporo-occipital leptomeningeal hemangioma. (Figure 2). The EEG showed a focus of temporo-occipital suffering on the right side. The diagnosis of sturge-weber syndrome revealed by a convulsive seizure was retained; an ophthalmological examination was completed and glaucoma was found.

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The patient was put under medical treatment based on antiepileptic drugs and local treatment of her glaucoma with good clinical evolution (recovery of consciousness and cessation of seizures).

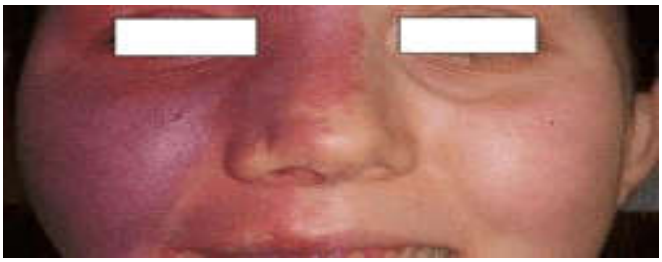

Figure 1 cutaneous planar angioma occupying the territory of the right V

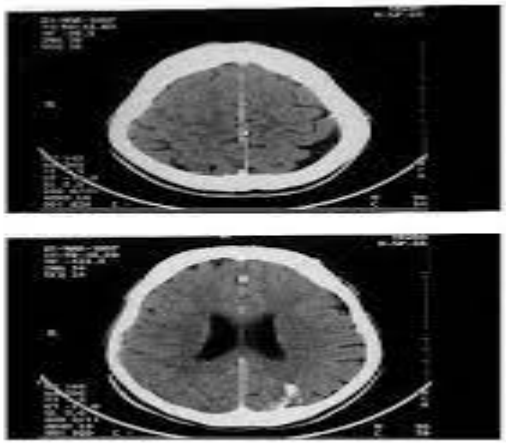

Figure 2 CT: Calcifications and leptomeningeal temporo-occipital hemangioma

DISCUSSION

Sturge-Weber syndrome (SWS), a rare congenital neurocutaneous syndrome, was first described in 1879 by Sturge in a 6 ½-year-old patient with facial angioma, glaucoma, and convulsions. Weber reported in 1922 the existence of cerebral calcifications in a patient with the same symptomatology and specified the clinical features of the syndrome. The term "sturge-weber syndrome" was adopted in 1935 by Professor Hilding Bergstrand in recognition of the two clinicians [1].

The current incidence of SSW is between 1/20 000 and 1/50 000 live births [2]. The disease affects both sexes equally and is described in all ethnic groups [3]. This condition is not familial but sporadic ; however, rare familial cases have been reported, including one case of identical twins [4,5]. In our patient, no familial cases of SWS were found.

The current definition of SSW includes a cutaneous plane angioma of the face reaching the V1 territory, neurological malformations (which may be responsible for convulsions, mental retardation or neurological deficits) and ophthalmological abnormalities (choroidal angioma, congenital glaucoma), which are inconsistently present.

The facial angioma that characterizes SWS consists of vascular telangiectasias, transforming into fibrosis giving a thick spongy skin tissue. Its wine color is due to the presence of deoxygenated blood in the vascular spaces. Skin involvement is not constant : this angioma is seen in 90% of cases. It is usually unilateral but in 16 to 37% it is bilateral and then it is a factor of bad prognosis. It conforms roughly to the trigeminal territory and the risk of associated cerebral involvement is estimated at 30% when the V territory is involved [6]. The cutaneous plane angioma was present in our patient; unilateral occupying the right V territory.

Epilepsy is present in 75 to 90% of cases in the literature. The epilepsy is most often early and severe, present from the first years of life. In most cases, it manifests itself before the age of one year and even before the age of six months, and in this case a convulsive state may be revealing. Comitial seizures take several forms: partial motor seizures in the hemisphere contralateral to the facial angioma (69%), tonic or clonic seizures, generalized seizures (25%) immediately or mostly secondary, more rarely myoclonus or spasms (2%). Fever and infections often precipitate the onset of epilepsy [6]. This is the case of our patient, the mode of revelation was partial left hemicorporeal convulsive seizures contralateral to the facial angioma.

Motor déficit is observed in about 50% of cases [7]. Hemiparesis or hemiplegia are frequently diagnosed in the first months of life. These neurological deficits may be acquired gradually over time or may be the result of a stroke like associated with seizures and/or migraine [8]. This deficit predominates in the upper limb and in particular in the hand, most often respecting the face. It is a deficit that is initially flaccid and then spastic. Most often walking is possible by mowing, while the use of the hand is sometimes impossible. The affected limbs are often hypotrophic. Aphasia is rare. Our patient has no motor deficit.

It seems that 50-60% of the patients are affected by mental retardation [9], according to several authors, there seems to be a close relationship between epilepsy and mental deterioration. Intellectual development appears to be normal until the onset of epilepsy. Early and poorly controlled epilepsy can rapidly lead to mental deterioration. Our case does not agree with the data in the literature, our patient presents with intellectual delay while presenting the first episode of convulsive seizure.

While epilepsy and mental retardation are the most common symptoms in SWS, migraine like headache has been recognized as the most important feature of SWS, 28% of patients followed for SWS may have headache with the clinical features of migraine [10]

In SSW, ocular involvement is inconstant [11] and is observed in one third of cases [10]. Glaucoma is the major ocular involvement (71%), but other lesions can be observed, such as buphthalmia or retinal detachment [12,13]. In our patient, ocular involvement was present, and the ophthalmological examination showed glaucoma.

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 [14]. Our case belongs to the Sturge Weber syndrome type 1 according to Roch *et al*. SWS with intracranial involvement should be suspected in every patient with facial skin involvement suggesting the syndrome. Cross-sectional imaging plays an important role in the diagnosis of Sturge Weber syndrome. Cerebral CT will look for: cerebral atrophy, focal or hemispheric, often homolateral to the angioma; intracranial calcifications in the shape of an "S", gyriform or train track of seat under cortical, at the level of the meningeal arteries and the cortical veins; hypertrophy and calcifications of the choroid plexus, homolateral to the angioma; cortical contrast, gyriform [14]. Cerebral MRI, which is more sensitive than CT [15], makes it possible to objectify : early signs even before the clinic; pie-merid angioma: interest of MRA sequences++; choroid plexus angioma; venous development abnormalities; cerebral atrophy; cerebral calcifications in hypo signal on all sequences: T2-weighted +++ gradient echo (GE) sequences; polymicrogyria, lissencephaly or localized pachygyria. In our patient, brain imaging such as cerebral CT was performed showing a temporo-occipital leptomeningeal hemangioma homolateral to the location of the cutaneous angioma.

Functional brain imaging is not common practice, but it has specific indications, often allowing an early diagnosis, by studying cerebral glucose metabolism by positron emission tomography (PET) and regional cerebral blood flow by functional imaging such as single photon emission tomography (SPECT) [16,17]: at an early stage of the disease: transient regional hypermetabolism of the cortex at the level of the pial angioma; at an advanced stage: hypo-metabolism in PET and hypo-perfusion in SPECT at the level of calcified areas. The EEG is often abnormal showing a slowing down of the background activity in one or both hemispheres in relation to cerebral suffering [18,19]. This is the case of our patient, the EEG showed a focus of right temporo-occipital pain.

Treatment is multifaceted [6]. Antiepileptic treatment has two components: curative and preventive. Curative treatment is often dictated by the type of epilepsy and must be rigorous because of the major risk of motor deficit. Preventive treatment is recommended in newborns [20]. The surgical treatment of pial angioma is controversial. It consists of a

hemispherectomy and should be considered for unilateral forms of severe evolution either by the seriousness of the epilepsy or by the importance of the intellectual regression [7]. For glaucoma, medical treatment is often preferred because of the significant risks of intra- and post-operative complications [21]. Plane angioma can be treated without particular danger by pulsed dye laser. Studies on the pathophysiology of SSW have clearly underlined the contribution of venous congestion, stasis and thrombosis in the progression of the symptoms of this syndrome. It has been suggested that ischemic phenomena are more involved than epileptic phenomena in the neurological deterioration of patients with SSW; thus, paroxysmal episodes may respond better to platelet anti-aggregants than to anticonvulsants [22]. In this case the role of aspirin seems very plausible [23]. Studies have reported a 65% decrease in the frequency of strokes with antiplatelet drugs without improvement in the frequency of seizures [24]. Our patient benefited only from antiepileptic drugs, with good clinical improvement, and did not need surgical treatment.

The prognosis of SSW is serious, the elements of poor prognosis are: the bilaterality of the facial angioma, the existence of a rebellious or precocious epilepsy before the age of one year and the appearance before 2 years of a motor deficit [6]. These elements are absent in our patient, so it is a sturge weber syndrome with a good prognosis.

CONCLUSION

The diagnosis of Sturge-Weber syndrome should be suspected in patients with planar facial angioma occupying the trigeminal territory. In the face of this suspicion, brain imaging and an ophthalmologic examination should be performed to allow early diagnosis, initiate treatment as early as possible and therefore improve the mental and motor future of the affected child.

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