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Case Report

Sturge-Weber Syndrome: Presentation of a Clinical Case through a Literature Review

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Abstract

Sturge-Weber syndrome (SSW) is a congenital and sporadic neurocutaneous disorder of a non-hereditary nature, although some familial cases have been described, it has currently been associated with a somatic activating mutation in the GNAQ gene (Protein GQ subunit Alpha G) on chromosome 9q21. Due to its low prevalence, it belongs to the group of so-called rare diseases. It is characterized by the presence of a characteristic skin lesion called port wine stain or nevus flammeus, very common in childhood (1/300 newborns) and persists throughout life, associated with leptomeningeal angiomatosis or angiomatosis in the choroid of the eye, epileptic seizures as the most frequent neurological manifestation that can be associated with significant cognitive impairment in these patients.

Case report: A 27-year-old male patient presented with a history of "port wine stain" since birth, with epileptic seizures that were difficult to control at the beginning. Diagnosis was made by clinical presentation, computed tomography, electroencephalogram, and CSF characteristics.

Keywords: Sturge; Weber; Port Wine Stain; Leptomeningeal Angiomatosis; Epileptic Seizures

Introduction

Sturge-Weber syndrome (SSW), also called encephalotrigeminal angiomatosis, is a neurocutaneous syndrome with an incidence of 1/50000-1/230000 live births without distinction of race or sex [1]. It was first described in 1860 by Schirmer (a German ophthalmologist), in 1879 Sturge described it in full, and by 1922, Weber completed the table with a description of radiological alterations [2]. Its origin is considered during the first trimester of gestation and around 2013 Shirley., *et al*. detected a mutation in the GNAQ gene, which is composed of 7 exons that cover a region of 310,993 nucleotides and are involved in cell growth through signals through cell membrane receptors via MAP kinases. Therefore, an activating mutation would increase it signaling leading to the capillary malformations observed in this syndrome. The earlier the mutation in embryonic development, the greater the impact on the affected structures of the skin, eye, and brain [3]. For better management, there is a classification of Sturge-Weber syndrome according to the Roach scale into 3 types

- **Type 1:** It is the most frequent. Facial and cerebral involvement. You may develop glaucoma.

- **Type 2:** It has facial involvement, not brain involvement, and may develop glaucoma.
- **Type 3:** It is the least common of all and the most difficult to diagnose. He has brain involvement only without the presence of a port wine stain or eye involvement [4].

The hallmark of this entity is a facial cutaneous venous dilation known as nevus flammeus or birthmark in port wine. It is clinically diagnosed based on the typical cutaneous, central nervous system, and ocular abnormalities associated with it. It also involves other locations, such as the oral cavity or the airway [5]. The new findings on this syndrome open the door to new research on etiology, the establishment of new treatments and report a variety of symptoms. For this reason, we present a clinical case following a review of the literature.

Case Report

We present a 27-year-old male patient, university student, with a history of bilateral facial angioma since birth, diagnosed with epilepsy since childhood, with frequent admissions due to status epilepticus and follow-up by ophthalmology due to the presence of bilateral choroidal hemangiomas.

- **Physical Examination:** Patient with good general condition who cooperates with the interrogation, conscious, oriented in time, space and person, with cognitive functions without alterations. The patient presents with bilateral facial hemangioma along the entire length of the trigeminal nerve and its branches, as well as bilateral choroidal hemangiomas and soft tissue hypertrophy (upper lip). They do not have angiomas anywhere else on the body.
- **Complementary**: Chemistry: Within normal limits. Lumbar puncture (cerebrospinal fluid): moderate increase in protein. EEG: unilateral reduction of base amplitude in the right hemisphere with wave spikes and polymorphous delta activity with secondary generalization.

Levetiracetam has been used as a treatment in this patient for generalized tonic-clonic epileptic seizures from adolescence to the present, with adequate control of these, and regular follow-up by the specialty of Neurology and Ophthalmology respectively.

CT Scan

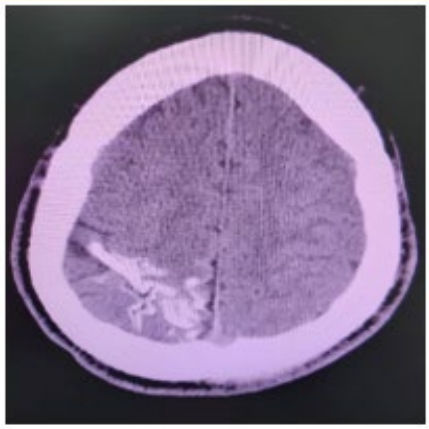

Figure 1: Presence of calcifications at the right parietal level, slight signs of cortical atrophy and abnormal venous drainage.

Discussion

Many authors have referred to the symptomatology of Sturge-Weber syndrome and its typical patient with facial angioma present from birth [6]. This was the case in this patient, but not all patients with facial angiomas have this syndrome. Among the cutaneous manifestations, capillary or venular malformation at the facial level is its hallmark, with variable size, lateralized and of color ranging from pale pink to purpuric, unilateral, although bilateral cases have been reported with distribution in the area of sensory innervation of the three branches of the trigeminal nerve: the frontal branch (V1), the maxilla (V2), and the mandibular (V3) [7,8] and may be associated with lesions of the choroidal vessels of the eyes or leptomeninges of the brain and may be located in other areas of the body [9]. In a study by Tallman., *et al*. in 310 patients with facial angioma, 85% were unilateral and 15% bilateral [7,10]. The incidence of SSW with bilateral facial angioma has been reported in 32% and is associated with a higher prevalence of intracranial abnormalities [11].

It has been suggested that initially patients may appear neurologically normal, but there are factors that suggest a progressive course of cortical injury in this syndrome. The most notable manifestation is epileptic seizures (75%-90%), appearing at any age with a predominance in childhood (during the first year of life) [1,7]. they are typically focal onset and usually evolve into generalized tonic-clonic seizures; Infantile spasms, myoclonic or

atonic crises are not exclusive [12]. Studies identified by the Sturge-Weber Foundation found seizures in 83% of patients. Headache is the second most common neurological symptom and affects 30-45% of patients in the form of migraines with aura and hemiplegia (28%), tension headaches and glaucoma headache [13].

Ocular manifestations describe glaucoma (60%) and diffuse choroidal hemangioma (40-50%), almost always unilateral and ipsilateral, but bilateral cases associated with bilateral nevus flammeus have been described. Other symptoms include: buphthalmos, heterochromia of the iris (10%); and homonymous hemianopsia [14,15].

Other manifestations include stroke-like events [16]; macrocephaly, developmental delay; intellectual disability (60%); learning disabilities; autism spectrum disorders (24%); ADHD (40%) [14,17], emotional symptoms in 50% (depression, anxiety and low self-esteem) and sleep disturbances or in the form of sleep-due insomnia (26-57%). Hydrocephalus and intracranial hemorrhage have rarely been reported, but it is presumed that they may be secondary to impairment of cerebrospinal venous drainage [16].

In our case, the port-wine stain was bilateral, affecting the three branches of the trigeminal nerve, associated with bilateral choroidal hemangiomas and unilateral leptomeningeal involvement, without involvement of other areas.

Although in most cases, facial angioma is evident from birth in SWS, cases have been reported in which it may be absent [5]. Epileptic seizures as the only neurological manifestation so far began in childhood with focal status epilepticus, the reason for several consecutive admissions with variation in adolescence to generalized tonic-clonic seizures that were better controlled with oral anticonvulsant treatment. In view of the absence of a history of developmental delay in childhood, motor deficit and adequate intellect with active work status, another symptomatology in this patient of those mentioned above is denied.

Laboratory tests in this entity are usually normal and proteins are elevated in the CSF, presumably due to microhemorrhage, which are common in SSW [1]. In our case there was no bleeding**.** With respect to neurophysiological studies, electroencephalography helped in the diagnosis of these patients.

According to references in a Canadian study, EEG was normal in only 4%, with background suppression in 74% (unilateral in 64% and bilateral in 10%) and epileptiform discharges in 22 cases [18]. In the patient presented, there was evidence of unilateral reduction of the base amplitude in the right hemisphere with wave spikes and polymorphous delta activity with secondary generalization.

Among the neuroimaging studies that confirm CNS involvement, it has been reported that magnetic resonance imaging is superior to tomography in this syndrome. However, the diagnosis is usually evident on plain skull radiography. Bernal and Altman reported abnormal activation patterns in occipital areas on functional magnetic resonance imaging (fMRI) in patients with SWS [18,19]. In this case, the diagnosis was obtained by a computed tomography of the skull showing the presence of calcifications at the right parietal level, discrete signs of cortical atrophy, and abnormal venous drainage.

The differential diagnosis included: Klippel-Trenaunay-syndrome de Weber; Beckwith – Wiedemann syndrome, Dyke-Davidoff-Masson syndrome, Siderosis, Calcification secondary to intrathecal methotrexate therapy and meningitis, Rendu-Osler-Weber syndrome – Hereditary Hemorrhagic telangiectasias; Von Hippel- Lindau; Wyburn-Mason syndrome – Retinal arteriovenous angioma; Shapiro- Shulman; Divry- Van Bogaert; Bannayan-Zonana; Cobb syndrome and others [1,12,13,20].

In Sturge-Weber syndrome, the specific treatment is not delimited, but generally includes prevention and prophylaxis of the associated symptoms, in the case of convulsive seizures, use of anticonvulsants and surgical options in refractory cases [22]. Erba and Cavazzuti estimated that 40% of patients with SWS could become candidates for epilepsy surgery, excluding those with good control such as our patient who is compensated. [21]. For port wine stain, it is treated with laser photocoagulation of dye, which has been useful in reducing cosmetic imperfections caused by cutaneous vascular dilation which can have some psychological impact [24]. In cases associated with glaucoma, the goal is to control intraocular pressure to prevent optic nerve injury, and ophthalmologists consider surgical therapy as the mainstay of treatment [23].

Conclusion

In general, Sturge-Weber syndrome is a challenge for any health professional, due to the complexity of its varied symptoms and the entities to which it can be associated, for this reason it is necessary to carry out an exhaustive study of the patient to establish a timely diagnosis and try to achieve a better development to achieve ophthalmological improvements neurological and cutaneous with current therapeutics.

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