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Sturge - Weber Syndrome: Literature Review and Report of a Clinical Case

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Abstract

In this study, a clinical case of 8-year-old male, with a history of seizures from 7 months of age associated with Sturge Weber syndrome, under treatment with oxcarbazepine and phenobarbital is presented. At 3 years of age, during his hospitalization he presented several tonic-clonic seizures, cardiorespiratory arrest, for which he began advanced CPR for two minutes. An angioresonance of the brain was performed, which reported angiomatosis in the left sigmoid sinuses and brain magnetic resonance imaging reported left temporal parietal occipital cortical atrophy associated with linear and nodular millimeter calcifications. Thanks to current research, it is known that the syndrome is caused by a mutation in the GNAQ gene, however, its etiology is not yet proven. The development of this clinical case provides important information on the onset and clinical evolution of WSS manifested with facial angioma, glaucoma and seizures at an early age.

Keywords: *Sturge Weber Syndrome, Facial Hemangioma, Mutation, Epilepsy, Angiomatosis, Glaucoma.*

1. Introduction

Sturge-Weber syndrome is a congenital neurocutaneous disorder, with an incidence of 1 in 20,000 - 50,000 births, characterized by a port wine-colored facial and scalp spot, leptomeningeal angiomatosis and glaucoma, caused by a change in the GNAQ gene on chromosome 9 (9q21.2). It does not have a clear genetic pattern so there is no direct evidence of hereditary predisposition. It is necessary to make an early diagnosis since there may be complications such as glaucoma, vascular stenosis, epilepsy,

neurological and neurocognitive affectations. This article aims to describe the case of a rare syndrome, its clinical manifestations and treatment.

2. Materials And Methods

A descriptive study was conducted, with the purpose of describing a clinical case of Sturge Weber, based on the review of the physical medical history of the clinical case of the Metropolitan Hospital of Quito, after the informed consent of the parents. On the other hand, a bibliographic review was carried out where the analysis of original articles and systematic reviews was executed in a general way in the systematization of the background of the research topic. The PubMed and Scielo databases were consulted; in the Google Scholar search engine, keywords such as: Sturge Weber syndrome, epilepsy, facial hemangioma, port wine spot, angiomatosis, were used as a complement to the review.

3. Results and Discussion

Sturge – Weber Syndrome (SSW) also called encephalotrigeminal angiomatosis, is an alteration in neuroectodermal development, which generates neurocutaneous disorders with facial angiomas and in most cases leptomeningeal involvement ipsilateral to the facial nevus, the skin of the face, scalp, in ophthalmic and maxillary regions, occurs in 1 in 50,000 people (Suárez et al., 2020). This angioma is known as Port Wine Spot, it can occur in extracranial regions and soft tissues (Pila et al., 2010). Its etiology is unknown, although it is suspected of a deficient development of embryological vascularization, this error affects a specific area of the neural crest area, responsible for the origin of the connective tissue of the facial dermis, the ocular choroid and the pia mater (Prato et al., 2006; Sarti et al., 2019).

Epidemiology

It occurs in 10% of neonates with port wine stain on the distribution of cranial nerve V (trigeminal nerve); it affects both sexes. There is one case for every 2 500 dermatological patients, 1 for every 25 000 paediatric patients and 1 in 50 000 births (Marcher, 2014). IN ECUADOR THERE ARE NO REPORTS OF THE DISEASE

Physiopathology

It is due to a change or mutation in the GNAQ gene on chromosome 9 (9q21.2), this is responsible for encoding the G-alphaq protein, which is important in several growth factors with effect on the dilation of blood vessels and for neurotransmitters; this mutation during the fetal stage causes a network of blood vessels not to regenerate normally, After week 5 to 9 of the embrazo, remaining as a membrane of the blood vessel, corresponding to the area of distribution of the trigeminal nerve, it is worth mentioning that the eyes may be affected, due to an increase in fluid pressure in the eye, which is known as glaucoma (Uvebrant, 2016). It does not present a clear genetic pattern or direct evidence of hereditary predisposition, the syndrome occurs in all races and with equal frequency in both sexes, but arises as a new mutation that is not transmitted to the next generation (Prato rt al., 2006; Uvebrant, 2014).

Clinical Manifestations

It is classified into three types according to the organs that are involved: Type I, involves hemangiomas on the face and surface of the brain, along with epilepsy and glaucoma. Type II, hemangioma present on the face, glaucoma may occur. Type III, unusual shape and involves angiomas on the surface of the brain (Prato et al., 2006).

About 75% to 90% of patients have seizures, which usually begin at one year of age. Seizures are usually focal, but they can be generalized. In 25-50% of cases, hemiparesis of the side contralateral to the nevus is observed in port wine. Sometimes hemiparesis gets worse, especially in patients whose attacks cannot be controlled; Between 50% of patients have intellectual disability, and a greater number show some kind of learning difficulty, so there may be developmental delay. On the other hand, glaucoma usually occurs at birth or appears later. The eyeball may increase in size and protrude out of the orbit (buftalmos).

Treatment

This focuses on symptoms, so anticonvulsants for status epilepticus and drugs to treat glaucoma. A hemispherectomy is sometimes performed if patients have intractable seizures. In some cases, low-dose aspirin is given, starting at the time of diagnosis, to help prevent strokes or decrease progressive hemispheric atrophy presumably by preventing thickening in abnormal capillaries. Selective photothermolysis or pulsed dye laser can lighten nevus in port wine, however, treatment trials with topical beta-blockers are underway (Suarez et al., 2010).

Clinical Case

An 8-year-old male patient with a history of seizures since 7 months of age associated with Sturge Weber syndrome. In treatment with oxcarbazepine and phenobarbital for crises difficult to control. At 3 years of age, he was admitted due to fever, weakness, tonic seizures of the right upper limb and right hemiparesis. *Filiation data:* Patient born and resident in Guaranda, Ecuador; ORH+ blood group *Personal data:* product of second pregnancy, normal prenatal controls (echoes), born at 42 weeks by cephalovaginal delivery without pathologies during childbirth In his personal history he presents facial hemangioma since birth with Sturge Weber syndrome, at 7 months he debuted with tonic-clonic seizures so he required hospitalizations until he was 1 year 9 months, in which he presented a crisis difficult to control. No important neurological family pathological history is collected, asthmatic father. The positive data collected on physical examination were: in the neurological system, the patient asleep, reactive to management without signs of neurological focality, isochoric pupils normorreactive to light, hypotonic, otoscopy: bilateral erythematous ear canals; digestive: abdomen, apparently painful on palpation in deep hypogastrium, diminished hydro-air sounds; infectious: febrile During the third day of hospitalization was performed under sedation, angioresonance. After the procedure, in the recovery room he presented a seizure administering the usual medication, he remained stable for 1 hour. After this time, she presented a new seizure described by the mother as tonic-clonic movements of the right upper limb, midazolam was administered without response and presented cardiorespiratory arrest that required advanced CPR for 2 minutes; admitted to the PICU with minimal response to intense painful stimulus, shallow breathing with poor air intake on the left side, chest X-ray detected left atelectasis. Inside the patient's clinic I present: Neurological evolution, brain CT showed cortical cerebral atrophy and left parietoccipital intraparenchymal calcifications.

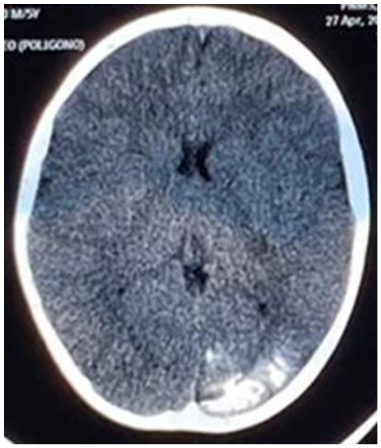


Figure (1). CT scan of the brain.

Source: Clinical history of the Metropolitan Hospital

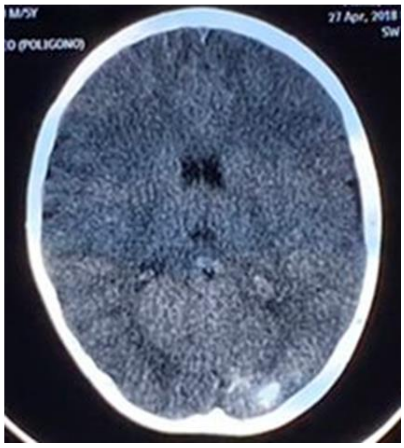


Figure (2). CT scan of the brain.

Source: Clinical history of the Metropolitan Hospital

After the administration of contrast, in Cerebral CT Angiography there were no stenosis or vascular dilations, small vascular uptakes in the left parietal region, thickening of intraocular soft densities at the level of the posterior vitreous chamber.

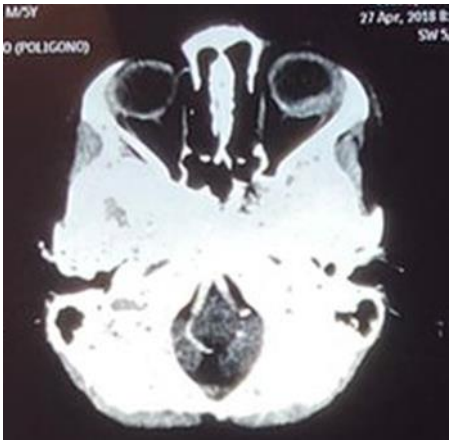
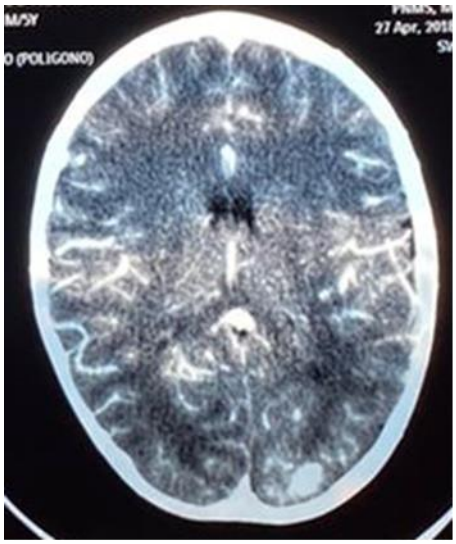


Figure (3). Cerebral CT angio.

Source: Clinical history of the Metropolitan Hospital



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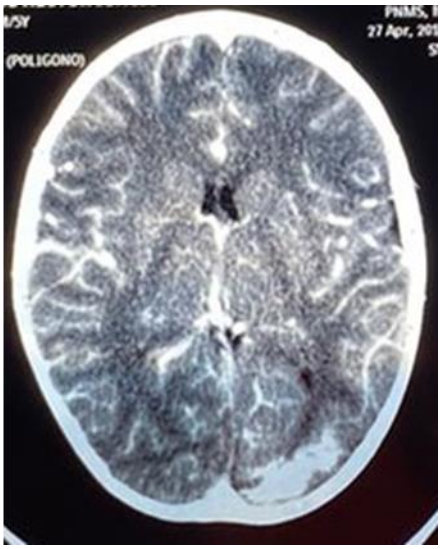

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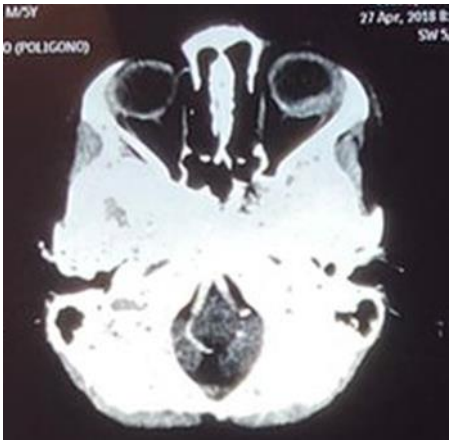

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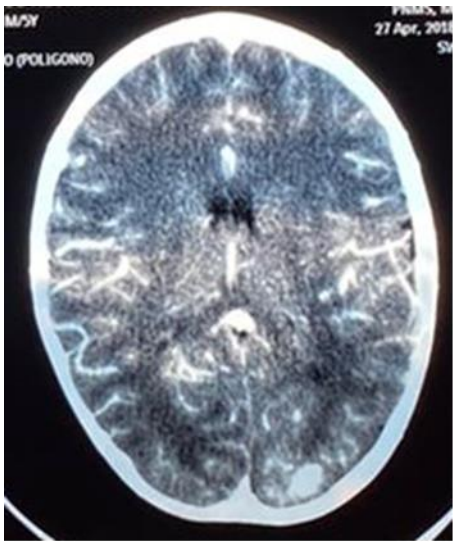

Figure (4). Cerebral CT angio.

Source: Clinical history of the Metropolitan Hospital

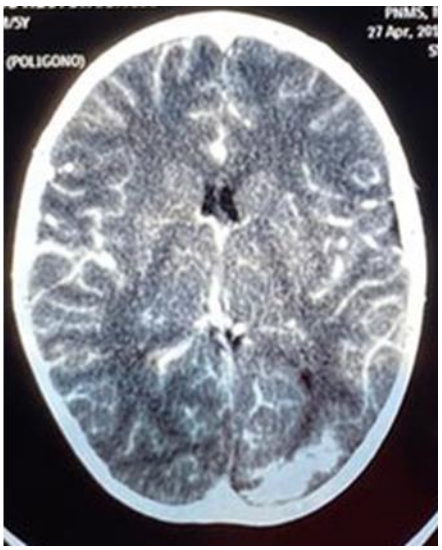

Figure (5)Cerebral CT angio.

Source: Clinical history of the Metropolitan Hospital

On brain MRI: left temporoparietooccipital cortical atrophy associated with linear and nodular millimeter calcifications; After gadolinium contrast, left orbital angiomatosis of external posterior retinal predominance and cortical with ecstatic cortical veins draining in the left transverse and sigmoid sinus was observed; Hypertrophy of the left choroid plexus with increased vascularization at the parieto-occipital level.

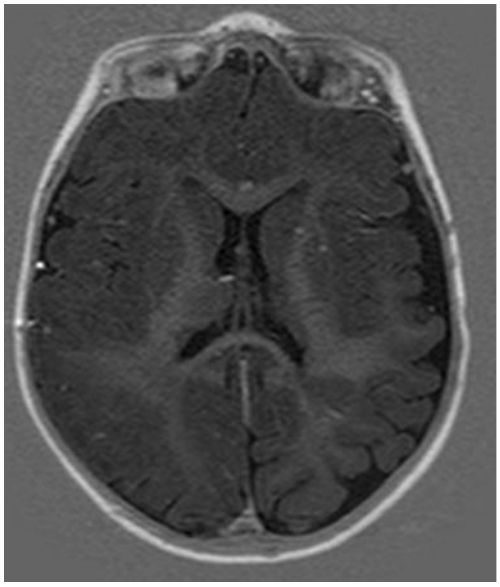

Figure (6).Brain MRI
Source: Clinical history of the Metropolitan Hospital

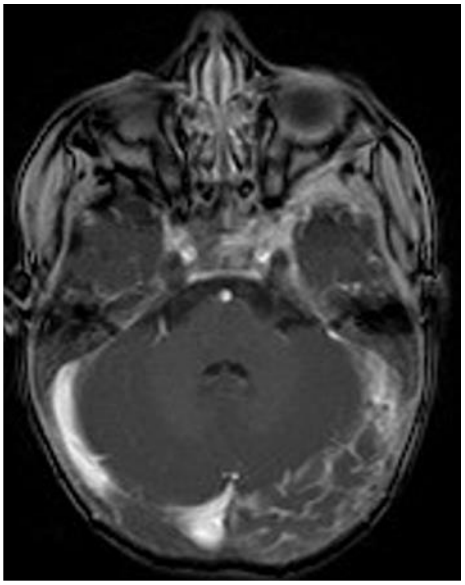

Figure (7). Brain MRI
Source: Clinical history of the Metropolitan Hospital

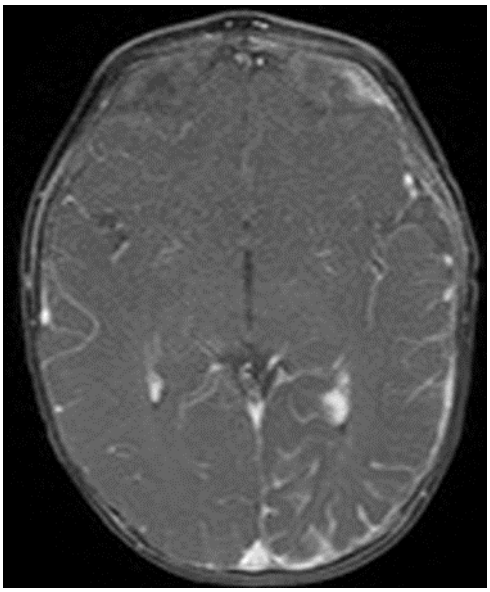

Figure (8). Brain MRI
Source: Clinical history of the Metropolitan Hospital

The encephalogram reported slow tracing of the right quadrant, with no record of epileptiform activity

The ophthalmological study found:

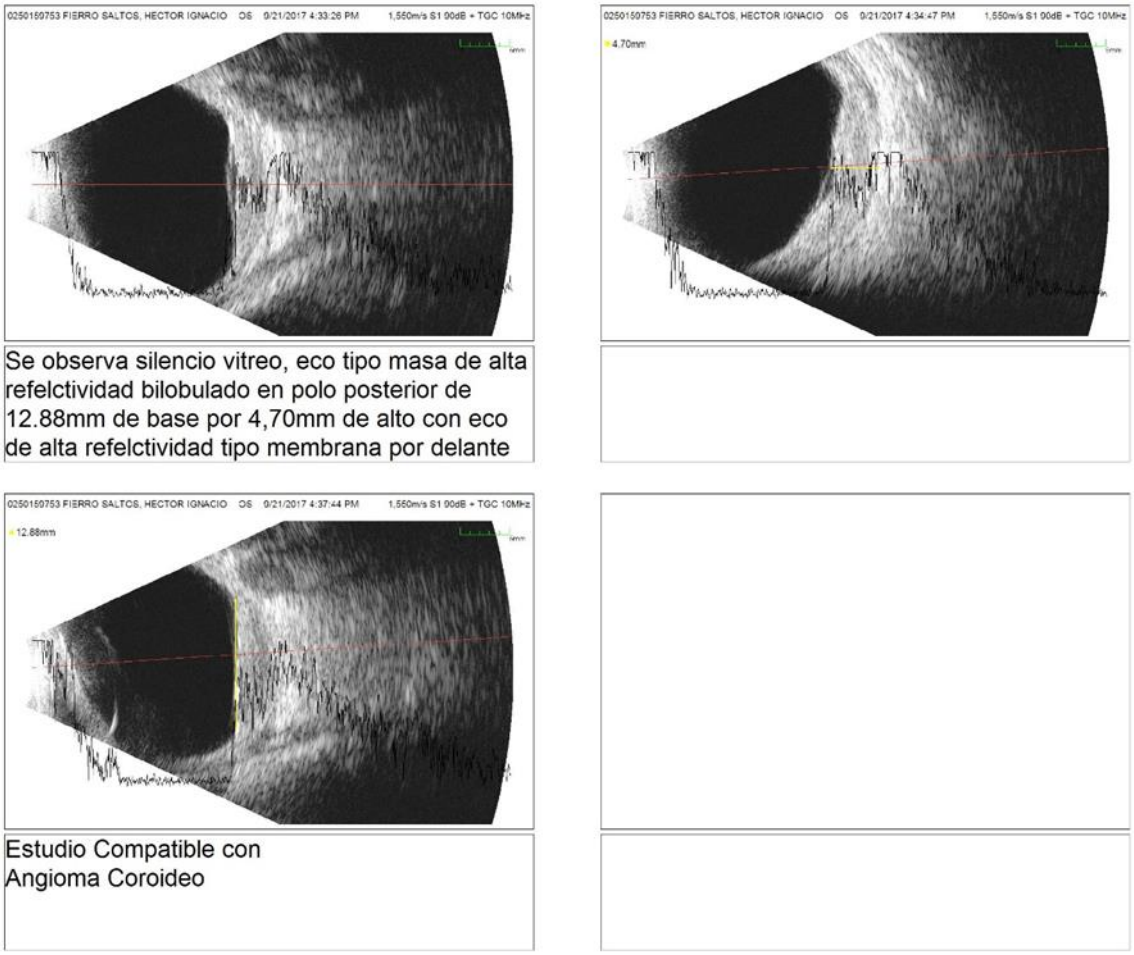

Figure (9). Ocular and orbital ultrasound
Source: Clinical history of the Metropolitan Hospital

Cardiovascular evolution on admission to PICU presented hypotension for 2 occasions so norepinephrine was administered. Respiratory evolution was performed a culture of tracheal secretion, evidenced Streptococcus Viridans and Neiseria spp. At 8 days he was discharged with hemodynamic stability without presenting seizures, with final diagnosis Sturge Weber syndrome / Peripheral Angiopathy in diseases classified elsewhere. He is currently followed up in consultations and is treated with oxacarbaxepin and phenobarbital anticonvulsants at pediatric doses.

Sturge Weber syndrome is a rare disorder of neuroectodermal development, there is the presence of facial angioma of progressive growth, from a pink spot to a dark and nodular spot, this can come with accompanying lesions in the choroid vessels of the eyes or the leptomeninges of the brain, often ipsilateral to the nevus, On the other hand it predisposes to calcifications, cerebral atrophy and convulsions (*Sturge, 2019)*. In the disease there are no reports of the existence of racial difference so that anyone and regardless of sex can be affected, in the case presented of the patient angioma was evidenced in the hemiface and hemicranium, accompanied by glaucoma, left leptomeningeal involvement, calcifications and cerebral atrophy, diagnosis of SSW.

Currently there are few studies that assertively explain the clinical evolution of the disease (Marileydis et al., 2021; Neto et al., 2008). Epilepsy occurs in 70-90% of patients due to the high sensitivity of the cerebral cortex to impaired blood circulation, seizures usually begin in one part of the brain and produce spasms on the opposite side of the body, these appear at any time, however the earlier they appear the greater the risk of hemiplegia, (Wilson et al., 2019) therefore the patient has a high risk of focality due to early onset of epilepsy (7 months) and presented right-sided hemiparesis

on two occasions after the seizure status According to recent reports, the disease can be suspected in the prenatal period by ultrasound or MRI that reveal the presence of unilateral hemispheric calcifications, focal hemispherical atrophy and changes in the white matter,4 in this case the mother underwent 4 ultrasounds which all showed normal images and it was not until after birth that they detected the syndrome (Santos & Cavalheiro, 2010). It is worth mentioning that the European Organization for Rare Diseases (EURODIS) reports that there are more than 6,000 rare diseases worldwide of which 80% are of genetic origin and are often chronic and life-threatening. In Ecuador, the Ministry of Public Health (MSP) defined only 106 orphan pathologies. Tuberous sclerosis and Sturge Weber syndrome are not considered (*Telegraph, 2014;* Kwok et al., 2019*).*

4. Conclusion

This article described the case of a rare syndrome such as Sturge Weber Syndrome and explained the clinical manifestations and existing treatments for this disease

Thus, thanks to current research it is known that the syndrome is caused by a mutation in the GNAQ gene, however, its etiology is not yet proven. The development of this clinical case provides important information on the beginnings and clinical evolution of SSW manifested with facial angioma, glaucoma and seizures at an early age.

Conflict of interest:

The authors declare no conflict of interest.

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