Original Article

Sturge-Weber Syndrome: Literature Review And Case Report

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Abstract

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 patch, leptomeningeal angiomatosis and glaucoma, caused by a change in the GNAQ gene on chromosome 9 (9q21.2). It has no clear genetic pattern so there is no direct evidence of hereditary predisposition. Early diagnosis is necessary as there may be complications such as glaucoma, vascular stenosis, epilepsy, neurological and neurocognitive impairment. This article aims to describe the case of a rare syndrome, its clinical manifestations and treatment. Case report: 8-year-old male patient with a history of seizures since 7 months of age associated with Sturge Weber syndrome, treated with oxcarbazepine and phenobarbital. At the age of 3 years, during his hospitalization, he presented several tonic-clonic seizures, cardiorespiratory arrest and was started with advanced CPR for two minutes. Brain angioresonance was performed, which reported angiomatosis in left sigmoid sinuses and encephalic magnetic resonance imaging, which reported left temporal parietal occipital cortical atrophy associated with linear and nodular millimetric calcifications. Thanks to current research it is known that the syndrome is produced 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 course of WSS manifested by facial angioma, glaucoma and seizures at an early age.

Keywords: Sturge Weber syndrome, facial hemangioma, mutation, epilepsy, angiomatosis, glaucoma.

INTRODUCTION

Sturge – Weber Syndrome (SSW) also called encephalotrigemonal angiomatosis, is an alteration in neuroectodermal development, which generates neurocutaneous disorders with facial angiomas and in most cases ipsilateral leptomeningeal involvement to the facial nevus, the skin of the face, scalp, ophthalmic and maxillary regions, occurs in 1 in 50,000 people (Suarez-Amor et al, 2010). This angioma is known as Port wine colored 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 & Peraza, 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. One case is observed for every 2,500 dermatological patients, 1 for every 25,000 pediatric patients and 1 in 50,000 births (Higueros et al, 2017). 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 pregnancy, remaining as a membrane of the blood vessel, corresponding to the distribution area 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 & Peraza, 2006; Uvebrant, 2016).

Clinical Manifestations

It is classified into three types according to the organs involved:

1. Type I, it involves hemangiomas on the face and surface of the brain, along with epilepsy and glaucoma.

2. Type II, hemangioma present on the face, glaucoma may occur.

3. Type III, unusual shape and involves angiomas on the surface of the brain. (Prato & Peraza, 2006)

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

Treatment

This focuses on the symptoms, so anticonvulsants for status epileptics 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 stroke 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. (Suárez-Amor et al, 2010)

Clinical Case

An 8-year-old male patient with a history of seizures from 7 months of age associated with Sturge Weber syndrome. In treatment with oxcarbazepine and phenobarbital for crises of difficult control. At the age of 3 he was admitted due to fever, decay, tonic seizures of the right upper limb and right hemiparesis.

Parentage data: Patient born and resident in Guaranda, Ecuador; ORH+ blood type

Personal data: product of second gestation, normal prenatal controls (echoes), born 42 weeks per delivery, cephalovaginal 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 of difficult control.

No important neurological family pathological history is collected, asthmatic parent.

The positive data collected on physical examination were: in the neurological system, the sleeping patient, reactive to management without signs of neurological focality, normoreactive isochoric pupils to light, hypotonic, otoscopy: bilateral erythematous ear canals; digestive: abdomen, apparently painful to palpation in deep hypogastrium, decreased hydro-aerial noises; 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, 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 cardiorespiratory arrest that required advanced CPR for 2 minutes; was 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.

Within the patient's clinic I present:

Neurological evolution, brain CT showed cortical cerebral atrophy and left parietoccipital intraparenchymal calcifications. The samples were taken from the medical history of the Metropolitan Hospital, Quito-Ecuador.

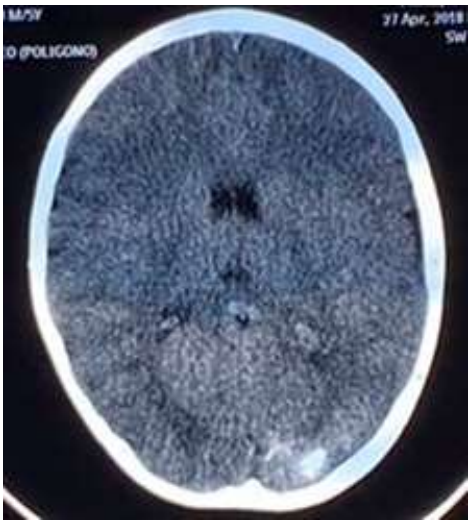

Figure 1: CT scan of the brain.

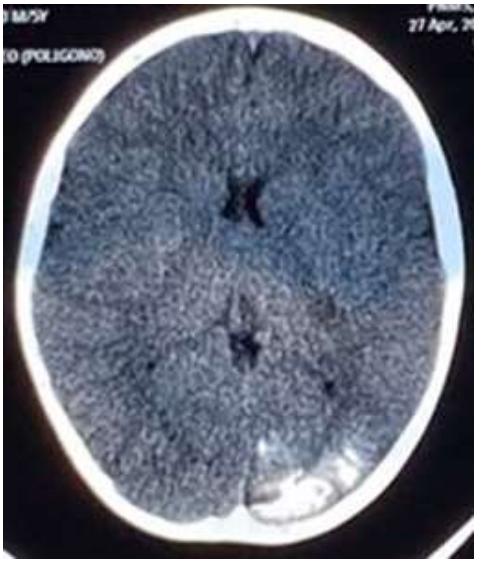

Figure 2: CT scan of the brain.

After the administration of contrast, in Cerebral CT there was no evidence of 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.

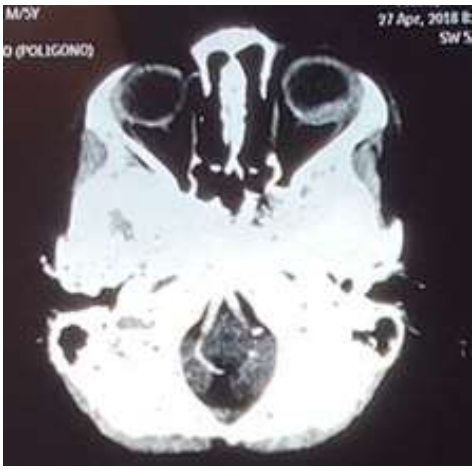

Figure 3: Angio TC Cerebral.

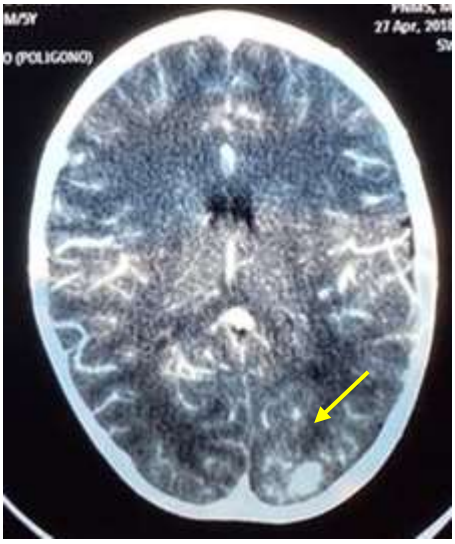

Figure 4: Angio TC Cerebral.

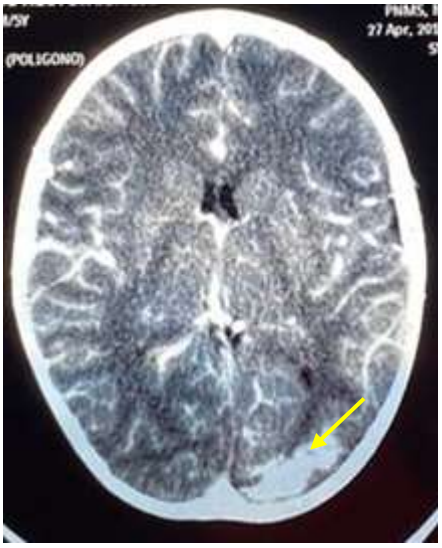

Figure 5: Angio TC Cerebral.

On brain magnetic resonance imaging: left temporoparietooccipital cortical atrophy associated with linear and nodular millimeter calcifications; Gadolinium postcontrast left orbital angiomatosis of external posterior retinal predominance and cortical with ecstatic cortical veins of drainage in left transverse and sigmoid sinus was observed; Hypertrophy of the left choroid plexus with increased vascularization at the parieto-occipital level

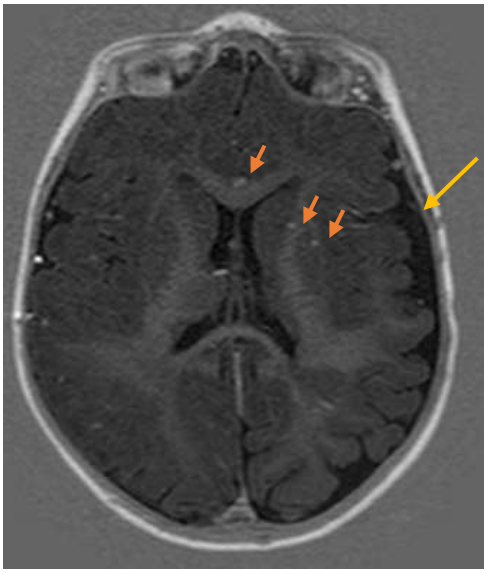

Figure 6: Brain MRI

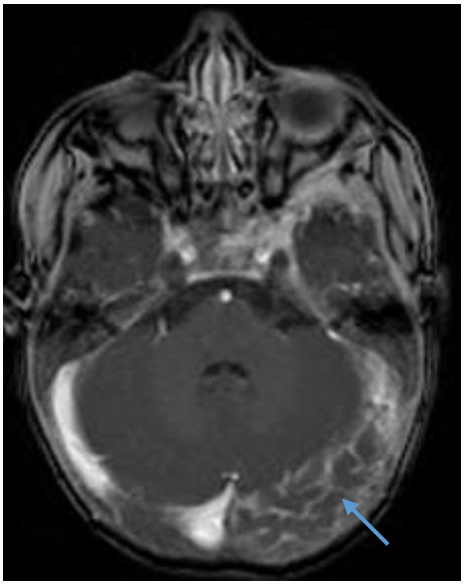

Figure 7: Magnetic resonance imaging

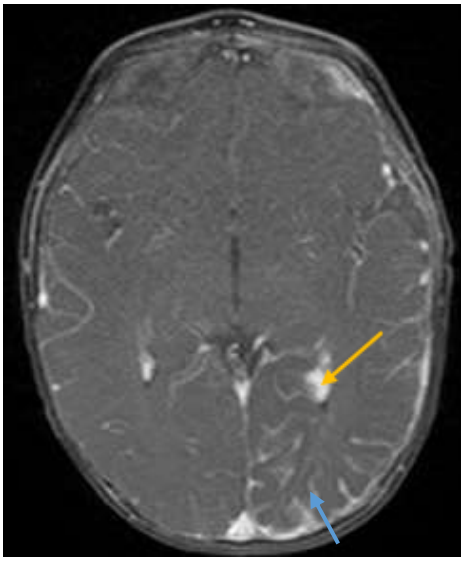

Figure 8: Magnetic resonance imaging

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

In the ophthalmological study it was found:

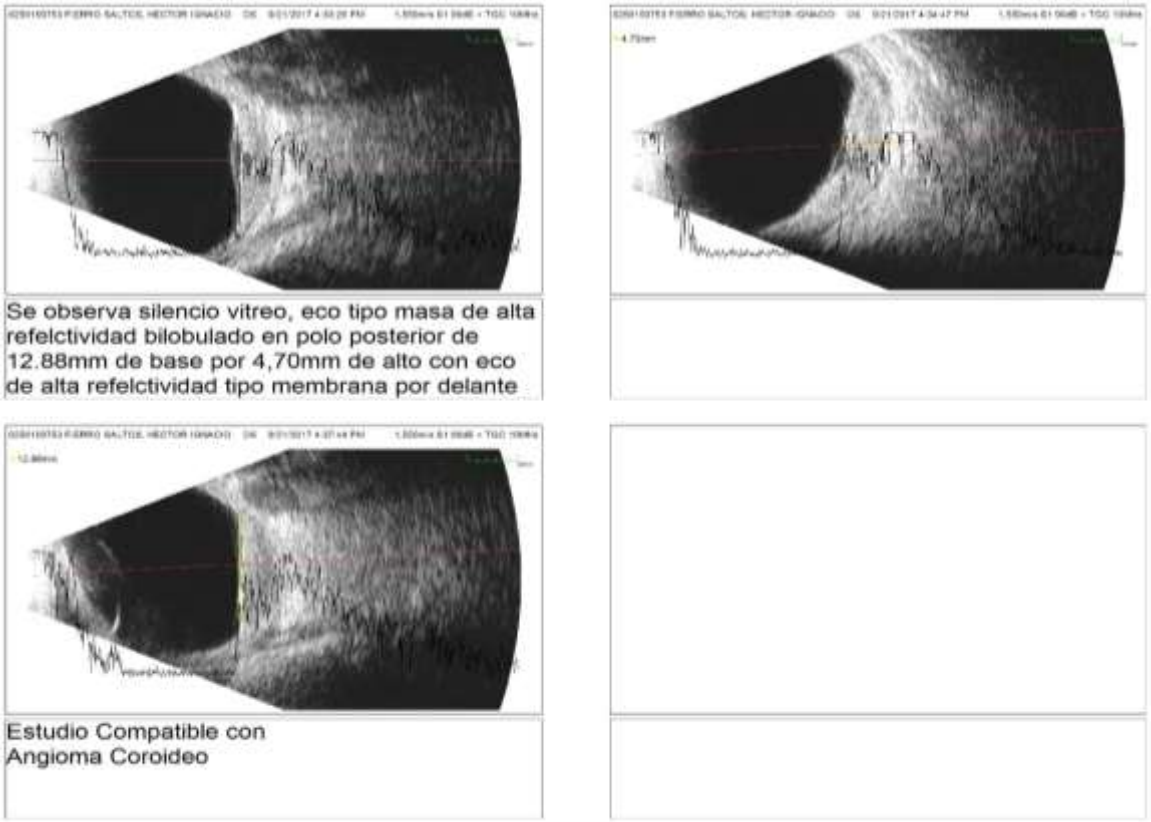

Figure 9: Ocular and orbital ultrasound.

Cardiovascular evolution upon admission to PICU presented hypotension for 2 occasions so norepinephrine was administered.

Respiratory evolution was performed a culture of tracheal secretion, Streptococcus Viridans and Neiseria spp. were evidenced.

At 8 days he was discharged with hemodynamic stability without presenting seizures, with final diagnosis of Sturge Weber syndrome / Peripheral Angiopathy in diseases classified elsewhere.

He is currently being followed up in consultations and is being treated with oxacarbaxepin and phenobarbital anticonvulsants at pediatric doses.

METHODS

A descriptive study was conducted with the purpose of describing a clinical case of Sturge Weber, based on the review of the physical clinical 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. We consulted the database of PubMed and Scielo; in the Google Scholar search engine, keywords such as: Sturge Weber syndrome, facial hemangioma epilepsy, Port wine spot, angiomatosis, were used as a complement to the review.

DISCUSSION

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 choroidal vessels of the eyes or the leptomeninges of the brain, often ipsilateral to the nevus, on the other hand, it predisposes to calcifications, brain atrophy and seizures (Velásquez-Gallego et al, 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 hemincium, 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.

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, (Londoño et al, 2018) therefore the patient has a high risk of focality due to early onset of epilepsy (7 months) and presented hemiparesis of the right side for two occasions after the convulsive 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 hemispheric atrophy and changes in the white matter, (Higueros et al, 2017) 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.

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 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. (Posada et al, 2008)

CONCLUSIONS

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 initiation and clinical evolution of VSS manifested with facial angioma, glaucoma and seizures at an early age.

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