

Clinical Features of Sturge-Weber Syndrome

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SUMMARY

- Introduction:** The Syndrome of Sturge-Weber is a rare condition of congenital development, and is characterized by a neurocutaneous disorder with angiomas wrapping the leptomeninges and the face skin, mainly in the course of ophthalmic (V1) and maxillary (V2) branches of the trigeminal nerve.
- Objective:** To review the literature about the Sturge-Weber Syndrome with emphasis on the current aspects.
- Method:** The following databases were searched: EMedicine, Encyclopedia of Medicine, FindArticles, LILACS, MEDLINE, Merck Manuals On-Line Medical Library and Scielo, and the searches applied the terms: Sturge-Weber Syndrome, neurocutaneous syndromes, encephalo-trigeminal angiomatosis, nevus flammeus, in articles published between 1991 and 2007.
- Literature's Review:** The most characteristic clinical statement is the presence, since the birth, of nevus flammeus, that generally reaches one half of the face and may stretch out up to the neck; in addition, other clinical manifestations may be present, like the corticocerebral angiomatosis, cerebral calcifications, epilepsy, ocular and buccal affections and mental retardation. The diagnosis is established by means of the inquiry of neurological and ophthalmic alterations in patients with a characteristic nevus flammeus, allied to the clinical data of complementary exams such as Computerized Tomography. The treatment consists basically of controlling the already confirmed clinical manifestations and preventing from the appearing of other alterations, mainly buccal and ocular.
- Conclusion:** This syndrome is not much frequent, but it needs to be early diagnosed, since it brings a series of complications to its carriers when not treated, specially because of reaching the Nervous Central System. The health professionals have to be suitably able to recognize its characteristic signs and symptoms, and so improve the quality of life of these patients.
- Keywords:** Sturge-Weber syndrome, congenital, angiomatosis, otorhinolaryngology.

INTRODUCTION

The Sturge-Weber Syndrome (SWS) or encephalotrigeminal angiomatosis, or even craniofacial angiomatosis, is specifically congenital, non-hereditary condition of rare development (1,2), although the literature presents case reports inherited in an autosomal recessive and dominant manner (3), namely of unknown etiology, marked by angiomas involving the leptomeninges and the facial skin, preferably in the first and second trigeminal division course: the ophthalmic and maxillary branches, respectively (4), and therefore an association in which the cerebral, cutaneous and ocular hemangiomas prevail (3).

In spite of being uncommon, this is the most frequent disease among the neurocutaneous syndromes - specially with vascular predominance -, and its proportion is of 1/50.000 births (1,2).

Clinically, the most specific finding of the SWS is the presence, in the birth, of the nevus flammeus, also known as port wine stain (3); in addition to this, other clinical manifestations may be present, mainly those regarding changes to the central nervous system, such as convulsive crises and mental retardation, resulting from the leptomeningeal angioma (1).

The diagnosis is frequently defined by the syndrome characteristic nevus; however, we remark besides such lesion is not an essential condition for the diagnosis, in spite the disease rarely occurs without the port wine stain (5), it's also necessary to make differential diagnosis with other syndromes of similar affection, hence the importance of the complimentary exams performance, such as Magnetic Resonance (MR) and Computerized Tomography (CT) (6).

Due to the wide spectrum of its symptomatology, the SWS treatment primarily depends on its clinical features, but it's basically related to the convulsive crises control, and ranges from a possible medication therapy to a surgical intervention (7).

Before these facts and the condition of being a multifactor character syndrome, and of etiology still strictly known and therefore of difficult prevention, it presents the need to perform a literature review, aiming at clarifying its specific signs and symptoms in order to alert the scientific community as for its research and knowledge, both for its relevance and for the importance of its early diagnosis, by means of all its unique diagnosis.

LITERATURE REVIEW

Etiopathogenesis

The Sturge-Weber Syndrome was initially described by SCHIRMER, in 1860, and was then specified by STURGE (1878), who associated the dermatological and ophthalmic changes to the disease's neurological manifestations. In 1992, WEBER complemented it through the documentation of these patients cerebral radiologic alterations (8).

Although its etiology is unknown, we believe the SWS's clinical manifestations derive from a common embryologic base. Some authors say it's from a congenital malformation resulting from changes in the ectoderm, mesoderm and neural crest derivatives, in which we observe the persistence of a vascular plexus around the cephalic neural tube portion, that characterizes the leptomeningeal and face angiomatosis (9), between the fourth and the eighth intra-uterine life (7,10).

We depart from the understanding that between the fifth and the eighth uterine life, the optic vesicle and the occipital lobe visual cortex are anatomically close to each other, and allow the prevalence of the association of the nevus flammeus, along the ophthalmic branch of pair V of cranial nerves, with the choroid hemangioma and the involvement of the occipital lobe, and a failure in the development of superficial cortical veins would support the occurrence of the event and thus the venous superficial draining loss would deviate the blood to the meninges in development, causing the formation of abnormal vascular channels and originating the leptomeningeal angiomatosis (10).

Recent studies indicate the absence of normal cortical veins cause stasis and thrombosis of the angiomatous bed with hypoxia and chronic ischemia of the cerebral tissue, and a progressive deposition of calcium in the more external cortical layers and in the subintimal space of capillaries and small vessels of the second, third and fourth cortical layers, so that the venous leptomeningeal angioma on the cortical surface interferes with the normal supply of oxygen to the subjacent cortex, which affects the intima-pia. Therefore, the SWS anatomopathological lesion is progressive and prevails in the parietal and parieto-occipital lobes. The ipsilateral brain hemisphere is thus atrophic in a great part of the cases (11, 12).

Some authors also believe there may be a somatic mutation in the ectoderm, mesoderm and neural crest, that is, a dominant autosomal hereditary disorder that may lead to the overproduction of angiogenic factors (5).

Clinical Manifestations

The SWS presents with neurological (brain anomaly), cutaneous, ocular and oral manifestations that may (or not) be associated with one another; however the most evident clinical manifestation is the presence of the nevus flameus or port wine stain on the face, which normally follows the course of branches V1 and V2 of the trigeminal nerve (11).

Some authors classify the SWS in three types: Type I, relative to the appearing of cerebral, leptomeningeal angiomas and glaucoma, forming the disease's classical form; Type II, regarding the appearing of facial angioma and glaucoma, without evidence of intracranial disease; and Type III, relative to the presence of isolated leptomeningeal angioma and absence of glaucoma, considered to be a rare condition (13).

Despite it's not an essential condition of the syndrome, the Port wine stain is the most frequent finding of the disease, generally present since the birth, and affects half of the face, and may expand to the neck with a characteristic to darken with age, ranging from reddish in the beginning to purple in time. It's worth remarking that only carriers of the nevus flameus, along the ophthalmic branch, develop the syndrome in its classical form (7).

The ipsilateral leptomeningeal angiomatosis also represents one of the main signs of the syndrome, may present with a progressive nature and commonly leads to cerebral calcifications, epileptic convulsive crises, contralateral hypertrophy and hemiparesis and mental retardation (10).

The epilepsy is the most common neurologic manifestation that occurs as a result of the angiomatosis, and affects from 75% to 90% of the cases. In the patients with unilateral involvement, the convulsive attacks correspond to 72% of the neurological disorders, while in patients with bilateral neurologic affection, this number increases to 93%. The generalized tonicoclonic motor focal convulsions occur more frequently in the initial phase of life, in 45% of these patients; the crises begin before the first year of life, with a peak of incidence between 4 and 6 months of age; however, in older children and adults, we may also observe a complex partial convulsion (13,14).

Hemiplegia and other neurological disorders affect the opposite side of the nevus flameus. Mental retardation corresponds to 50% and hemiparesis to 30% of the neurologic disorders (6).

The ocular angioma, also present in the syndrome, appears in 30% of the cases, affects the choroids and the ocular sclerotic, and is ipsilateral to the cutaneous angioma

(14). Vascular malformation problems in conjunctive, episcleral vein, choroid and retina, in addition to glaucoma - present in about 30% to 70% of the ophthalmic affections - represent the SWS syndrome ocular clinical manifestations (7).

Such ocular manifestations result from the pressure increase, lead to glaucoma and buftalmia, and the latter is more common than glaucoma, due to prenatal intraocular hypertension. The homonymous hemianopia, another neurological complication that affects the visual field, occurs invariably when the occipital lobe is affected. Other congenital ocular anomalies are present, such as coloboma of the iris and deformities in the lens of the eye. Clinically we have retroorbital pains and visual deterioration (5).

It's recommended that children with SWS are reevaluated constantly, due to the strong relationship between the upper eyelid and glaucoma, even in the normal intraocular pressure measurements (6).

Oral manifestations are also present in approximately 38% of the cases, and they may involve hemangiomatic lesion in the lip, oral mucosa, gum, tongue and palatine region (10).

The oral manifestations are generally unilateral and finish abruptly in the middle line. We observe ipsilateral gingival hyperplasia, characterized by an increase in the vascular component and gingival hemorrhage at minimal traumatism. The accumulation of food and the presence of bacterial plates may intensify inflammation and gingival hyperplasia (15).

The macroglossia and the maxillary bone hypertrophy found in some patients may cause a bad occlusion and facial asymmetry. Other factors, such as dilantinic therapy, used by the patients in the treatment of convulsive crises, may exacerbate the gingival increase picture (16).

Diagnosis and Complimentary Exams

Although it's not present in all cases and isn't pathognomonic in the syndrome, the diagnosis is based on the presence of the nevus flameus in the face, and, according to some authors, the association between convulsion and the nevus, irrespectively of the neurological loss, is sufficient for the syndrome's clinical diagnosis (5), although others defend the idea that, besides both these findings, it's also necessary one of the following manifestations to be present: Contralateral hemiparesis and hemiatrophy, mental retardation and glaucoma or buftalmia findings. However, taking into account the other

existing systemic manifestations, there's the need for the use of other diagnostic methods (1).

As for the cerebral disturbances, for example, the Computerized Tomography documents the intracranial calcifications and unilateral cerebral atrophy; while the cerebral vascular malformations are more easily viewed through Magnetic Resonance (1). A few cases of syndrome without the nevus flammeus were diagnosed by Magnetic Resonance, Computerized Tomography and angiography (5).

The MR exam is a better characterizer of the leptomeningeal angiomas, as well as the cerebral parenchyma and parenchymal venous abnormalities. While the CT technique represents the best diagnostic means compared to the MR in the cortical calcifications detection (17), since the studies demonstrate the cranial radiographies may reveal giriform calcifications in "rail line" in the affected side, and remark the location of the leptomeningeal angiomas above the cerebral cortex ipsilateral to the nevus flammeus (7). However, it's worth remarking that some traces indicate such giriform calcifications are invisible to the conventional radiographic exam, even after two years of age (6).

The ophthalmologic changes may be observed through the retinal examination (14) and also ocular ultrasonography, which showed to be an important method for diagnostic complementation, and contributed for valuable data concerning the diagnosis and the location of the choroid hemangiomas (16).

Important findings were also described of the ultrasonic biomicroscopy (UBM) in patients with the Sturge-Weber Syndrome associated or not with glaucoma. Such study comments the UBM not only as a diagnostic method, but also as a promissory method in the study of the syndrome ocular lesions (2).

The oral manifestations are observed in radiographies through the identification of horizontal and vertical bone re-absorptions with loss of the hard lamina, which commit the teeth of the side involved by the lesion and obviously by the clinical observation of other changes, such as gingival hyperplasia (5).

Differential Diagnosis

The Sturge-Weber Syndrome differential diagnosis includes the angio-osseous-hypertrophic Syndrome, also known as Klippel-Trénaunay-Weber's syndrome that presents with Port wine stains in the face ends, soft and osseous tissues hypertrophy, in addition to other features of the SWS (10).

We still have the Rendu-Osler-Weber's Syndrome or hereditary hemorrhage telangiectasis - an uncommon, dominant autosomal condition - whose features are abnormal vascular dilatation of the skin terminal vessels, mucosae and viscera. The disease involves the lips, buccal mucosa, pharynx, conjunctives and face, in addition to neurofibromatosis, elliptocytosis and arteriovenous aneurysm (6).

We also consider the Maffucci's syndrome, which presents with vascular malformations in the skin and mucosae (lips and palate), dyschondroplasia, and may also lead to long bones pathological deformities or fractures and cause chondrosarcomas (6).

Another situation is the occurrence of occipital calcification in the absence of the nevus flammeus; such fact requires the research of celiac disease associated with epilepsy and brain calcifications to differ from SWS (17).

The Beckwith-Wiedmann's Syndrome requires differential diagnosis, because such capillary malformation in the central region of the forehead or upper eyelids presents with a lesion similar to the Port wine stain, associated with macroglossia and some risk associated to visceral neoplasms due to the visceral overgrowth (18).

Treatment

The treatment of the Surge-Weber's Syndrome is variable, and depends on the nature or intensity of its possible clinical features.

Usually, the Port wine nevus in the face may be improved by the use of new techniques of laser therapy or through the use of cosmetics (7,18).

However, we remark the first direction for treatment must focus the control of the convulsive crises, with administration of suitable medication. In case the convulsive crises become uncontrollable, several neurosurgical procedures have been used - for instance lobectomy or hemispherectomy - aiming at diminishing such crises and make the patients more sensitive to the medication treatment. Nevertheless, the great limitation to the surgical intervention is its high morbimortality (7).

In addition, physiotherapies to mitigate the motor difficulties resulting from hemiparesis and reeducation to increase the intellectual level are also recommended (7, 18).

The annual monitoring as for glaucoma is recommended for all patients, specially children and, if applicable, it deserves an aggressive treatment (19).

The patients with oral changes must be submitted to oral cavity exams periodically and perform a detailed control of the bacterial plate, since it may aggravate the vascular picture and extreme cares are also crucial with surgical procedures in the oral cavity, due to its trend to flow with intense hemorrhage (1).

FINAL COMMENTS

The large spectrum of clinical manifestations of the Sturge-Weber Syndrome shows its multifactor nature and therefore its difficult early diagnosis. Even the classical manifestations, such as facial nevus flammeus, convulsive crises, glaucoma and gingival changes, contribute for the late diagnosis before their differential diagnoses, which also form uncommon and not much known syndromes (1).

As the SWS's etiopathogenesis is not strictly known, its prevention is then hardened and its early diagnosis is critical, since it allows the control of future complications, mainly those relating to the Central Nervous System, considering the inexistence of specific treatments for such pathology (18).

The Sturge's disease requires major cares in the performance of invasive procedures, which improves the surgical planning and the therapeutic proposal to be instituted, and prevents the significant interurrences arising out of hemorrhages that elevate so much this disease's morbimortality (20).

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