Sturge-Weber syndrome: a report of 14 cases

Lucia Parisi,1 Teresa Di Filippo,1 Sabina La Grutta,1 Rosa Lo Baido,2 Maria Stella Epifanio,1 Maria Esposito,3 Marco Carotenuto,3 Michele Roccella1
1Department of Psychology, University of Palermo; 2Psychiatric Clinic, University of Palermo; 3Clinic of Child and Adolescent Neuropsychiatry, Second University of Naples, Italy

Abstract

Sturge-Weber-Krabe syndrome (SWS), also known as encephalotrigeminalangiomatosis and named the forth facom atosis, recall the names of the authors who first described it in its basic clinical, radiological and anatomo-pathological aspects. We report here 14 cases of Sturge-Weber disease. In 6 of these, despite what had been previously described in literature, an extension of the angiom a has been noted in other parts of the body. The study of these subjects stresses not only the need for a pharmacological/neuropsychomotor intervention, but also the need of a psychotherapeutic approach, for the emotional and affective implications that could derive from this syndrome. The reported cases are similar to those presented in literature for their main features. In particular, two elements are interesting: i) the exceptional diffusion of the red nevus onto the whole hem isphere; and ii) the evaluation of the way the patients live the disease, which has not been previously considered in literature. We can conclude that SWS is a multisystem disorder that requires the neurologist to be aware of the possible endocrine, psychiatric, ophthalmological, and other medical issues that can arise and impact on the neurological status of the patients.

Case Report

Our study reports 14 cases of Sturge-Weber disease, 6 male and 8 female patients, studied in follow up. We observed 10 cases since they presented seizures, and 4 cases for the presence of a severe picture of psychomotoric retardation. In 5 of 10 cases, the seizures started within the first year of life. Some cases (n=3) present an extension of the angiom a in other body parts, and, particularly in one case, the nevus extends itself on the branch of trigem inus, exceptionally to the whole face, corresponding to the first and the second frontal area and also on the cheek in a distribution involving the trigem inal nerve distribution area and vascular malformation(s) of the brain (leptomeningealangiom a) with or without glau-coma.4-10 Although SWS is a congenital disorder, which usually presents in infancy, occasionally neurological symptoms first present in adulthood. Most affected individuals do survive into adulthood with varying degrees of neurological impairment, including epilepsy, hemiparesis, visual field deficits, and cognitive impairments ranging from mild learning disabilities to severe deficits.5-7 SWS is a multisystem disorder that requires the neurologist to be aware of the possible endocrine, psychiatric, ophthalmological, and other medical issues that can arise and impact the neurological status of these patients. Seizures, stroke-like episodes, glaucoma, headache, and developmental delay are frequent associated features.11

Some of these clinical features have only recently been described.4-10

This study describes 14 subjects with SWS who express various characteristics of the clinical spectrum of the syndrome.

Discussion

Sturge Weber Syndrome (SWS), also called encephalotrigeminalangiomatosis, is a sporadically occurring neurocutaneous syndrome, characterized by vascular malformation with capillary venous angiom a that involve face, choroid of eye and leptomeninges with resulting neurological and orbital manifestations.8 Genetic studies have stressed only some rare case of familial connection. As far as the nature of angiopathy is concerned, the vascular alterations is not an angioblastoma.5-12 The prevalent orientation is to consider the alteration as a malformation that starts prematurely during intrauterine life.6 The subarachnoid and choroid angiodysplasia, like the cutaneous one, is made by mature vessels, without any note of endothelial proliferation; that explains why it is so resistant to the ionogetic radiation treatments with antiblastic chemotherapies, withglycoactive steroids.10-12 The nevus of the face, evident already at the birth, has the color of port-wine, it is located in the orbital area, extending itself generally in the frontal area and also on the cheek in a distribution corresponding to the first and the second branch of trigeminus, exceptionally to the whole hem isphere. It can also be located in the oral
cavum, in the pharynx, in the throat, in the tongue, inside the nose, on the eyelid conjunctiva, in the episclera, in the retrobulbar orbit causing proptosis with hipomobility of the bulb and through the orbital fissures; and it can have endocranial development in the extrudal site.\textsuperscript{5,14-16} It is usually unilateral and rarely bilateral. At first the lesion is flat, just palpable and it decolorizes on vitropression. Over the years, a progressive development of the anomalous vascularity has been noted and the nevus becomes prominent, thicker, darker, mameloned, and sometimes hypertrophic.\textsuperscript{12-18}

The leptomeningealangiomatosis is located mainly in the occipital areas and in occipital parietal areas of a cerebral hemisphere.\textsuperscript{5,12,16-24} Its structural and developing characteristics recall those of the red nevus (it is also a progressive developing ectasiant capillary venous angiodyplasia).\textsuperscript{5,12,21} Under the vascular mass the cerebral tissue meets a gradual process of atrophy with a dilatation of the homolateral ventricle and increase insubarachnoid spaces. The subintimal calcification of meningeal arteries is associated to the cerebral atrophy; it extends gradually at the intima and at the media giving to the vessels the appearance of petrified tubes. The calcification process takes place mainly in the intravascular parenchyma with accumulation, in the external layers of the cortex, of pulverized granules of calcium salts that form heaps which progressively increase and replace the cerebral tissue.\textsuperscript{4,5,12,17,19}

The calcium accumulations, absent at birth and exceptional in the newborn, are formed later. They translate themselves into a peculiar radiological picture of the syndrome: the ring-shape calcifications, appearing with parallel spires, are 2-3 mm apart.\textsuperscript{12,23}

Severe calcification in the affected hemisphere is related to severely decreased perfusion in underlying white matter and is associated with more severe epilepsy in SWS patients.\textsuperscript{19}

The presence of cerebral calcifications, evident at CAT and at MR has a negative prognostic meaning since it stresses the unavoidable and irreversible deterioration of the child’s condition.\textsuperscript{5,10,20} The direct visualization of the leptomeningealangiomia is very constant with tomodosiometry or with arteriography; the encephalon NMR reveals an atrophy and with tom odensitometry or with arteriography; the involvement of the white mass and of the deep venous dilatations on sequences in T2. Recently sequences have also been used in T1 after an injection of gadolinium in order to visualize the leptomeningeal angiomia.\textsuperscript{5,11,16,19}

A congenital and hypsilateralangiomia of the chorioid can be present: this angiomia can determine a light blue staining of the iris (even in the case of the dark color of the other eye). It is usually located at the posterior pole in the inter papillary macular area where a diskshapepigmented white-gray stain turns to red.\textsuperscript{5,17,21} The structural and evolutive characteristics of this vascular anomaly recall those of the leptomigestic nevus the area of which is related to it.\textsuperscript{5,21}

The glaucoma is the secondary effect often evident directly afterbirth. In its pathogenesis, the dilatation and congestion of episcleral vessels seem to have a relevant role. The hypertension, acting in a phase of the development in which the structures of containment of the globe (sclera and cornea) are more pliable, causes exhaustion and then dilatation.\textsuperscript{5,8,19,22} The glaucoma can be present at birth with megophthalmia, megalocornea and other anomalies such as coloboma of the iris, deformity of the lens, or it can be revealed later; there is relatively early evidence of the compromised visual acuity.\textsuperscript{5,9,10,23}

Ninety percent of cases present focal or generalized convulsions that often represent the onset of neurological symptoms and quite often these start during the first year of life.\textsuperscript{17,22} The seizures, which at first can be of the Jacksonian type, controlateral and later generalized, later become more and more frequent, and show little sensitivity to pharmacological treatment.\textsuperscript{19-25}

The seizures are associated to controlateral hemiparesis. The first attacks are temporary; later become persistent with spastic alterations, disturbances of sensibility, deficit of limb growth of the hemiparetic side. Moreover, they are associated to homonymous hemianopsia, a consequence of the angiomatosis location of the occipital lobe.\textsuperscript{21,25}

The outcome of hemispherectomy in pediatr ic patients is good for those with refractory epilepsies, such as West syndrome, Lennox-Gastaut syndrome, epileptic encephalopathy with continuous spikes and waves during slow sleep, and startle epilepsy arising from a hemispheric lesion associated with hemiplegia.\textsuperscript{24-27}

The aphasia is frequent and follows the seizur al symptomatology revealing itself just the first one of them.\textsuperscript{16-22}

There are varying degrees of mental retardation and this is often serious. It has been noted in 80% of cases, sometimes in early infancy, other times after normal psychological development after the early infancy; for this reason it has been called nevus amentia.\textsuperscript{18,25}

The deterioration generally takes place simultaneously to the beginning and the repetition of seizures.

A significant quadratic relationship was found between IQ and extent of severe (but not total) hypometabolism.\textsuperscript{25} Seizure variables also contributed significant variance to cognitive functions. Results suggest that intermedi ate size of severe hemispheric hypometabolism is associated with the worst cognitive outcomes, and small or absent lesions, with the best cognitive outcomes; children in whom a very large extent of the hemisphere is severely affected are likely to have relatively preserved cognitive function.\textsuperscript{23,25}

White matter (WM) loss is associated with cognitive impairment in SWS.\textsuperscript{25} Nevertheless cognitive and fine motor functions are related to diffusion abnormalities in specific ipsilateral, mostly frontal, WM regions.\textsuperscript{25}

Quaintness in behavior, hyperactivity, irritability, muddle, suicide and homicidal trends are frequent.\textsuperscript{4,17,24}

The most frequent diagnoses were mood disorder (31%), disruptive behavior disorder (25%), and adjustment disorder (25%). A substance-related disorder was the most frequent in adults (67%).\textsuperscript{29} A significant association was found between disruptive behavior disor der not otherwise specified and more left frontal and left parietal involvement.\textsuperscript{26} A trend toward significant association of having a seizure in the past three months with disruptive behavior disorder not otherwise specified

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Conclusions

Sturge-Weber-Krabbe syndrome is a multi-system disorder that requires the neurologist to be aware of the possible endocrine, psychiatric, ophthalmological, and other medical issues that can arise and impact the neurological status of these patients.5,13

The characteristics of the reported cases are similar to those presented in literature. Two elements are interesting: the exceptional diffusion to the whole hemicorpo of the red nevous and the evaluation of the living patterns related to the disease that is not consid-
ered in literature.

On the contrary, this is an important factor since new pharmacological therapies can obtaina good control of the epilepsy and conse-
quently an extension of life, and less deteriora-
tion of the patient’s condition.

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