

CONSIDERATIONS ON A STURGE-WEBER-KRABBE CASE

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Abstract

The Sturge-Weber-Krabbe syndrome is a complex form of neuroangiomas, having as diagnostic elements cutaneous, cerebral and ocular angioma. The authors present a 9 years old male patient diagnosed with extensive cutaneous angiomas. The child requires follow-up, as the evolution and prognosis of the disease can be unfavourable, by neurologic disorders and mental retardation aggravation.

Key words: Sturge Weber, diagnosis, child.

Introduction

Sturge-Weber-Krabbe angiomas (SWK) is a complex form of neuroangiomas, characterised by ocular, cutaneous and cerebral angiomas. The disease is sporadic; there is no direct evidence of a hereditary transmission. Like other neurocutaneous syndromes, SWK syndrome has an incomplete penetrance, with a great variability of clinic manifestations. The disease is rare, the frequency is 1/10.000 live births, without sex predilection.

Case presentation

The authors present an 8 years old male patient diagnosed with extensive cutaneous angiomas, accompanied by ocular and neuropsychiatric disorders.

The parents are young and healthy, and have other three healthy children. Both maternal and paternal relatives have no pathology associated to the case.

Personal history: first born child, from a normal pregnancy, delivered normally at full term, weight at birth 3000g, with no neonatal suffering, was natural alimentation up to 6 months, current vaccinations done adequately.

Psychomotor development milestones were delayed; the child presents a moderate mental retardation accompanied by behavioral disorders.

Clinical examination:

- adequate staturponderal development: weight 24kg, height 130cm;
- extended deep red, flat, non-prominent angioma on head, neck, right hemithorax, upper limbs and right lower limb;
- recurrent right anterior epistaxis;
- congenital hyperpigmentation of the iris – iris heterochromia;
- moderate mental retardation, normal neurological findings, and intermittent episodes of seizures;
- the rest of clinical examination found no other abnormalities



Fig. 1. Non-prominent angioma on head, neck, right hemithorax.



Fig. 2 extended deep red, flat, non-prominent angioma on head, neck, , right hemithorax, upper limbs and right lower limb

Paraclinic investigations

Biological tests: hemogram and complete blood count, acute phase reactants, seric proteins electrophoresis, common urine tests, seric levels of calcium, phosphate and magnesium, alkaline phosphatase, urea, creatinine, aminotransferases – all found in normal limits.

Ophthalmologic examination: anterior segment of the right eye – flat well-circumscribed angioma surrounding

the sclerocorneal limbus, violaceous sclera on a 4-5mm perilimbic-ring. Bilateral – congenital iris hyperpigmentation, heterochromia of iris. Visual acuity: right eye- light perception, left eye -1.

Fundus examination: normal discs, congenitally pigmented retina, and normal retinal blood vessels.

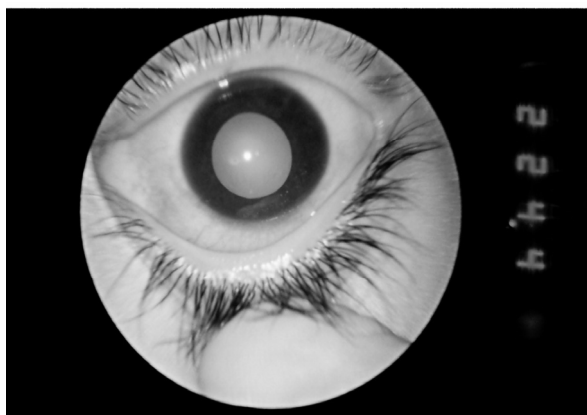


Fig. 3: Right eye, violaceous sclera on a 4-5mm perilimbic-ring.

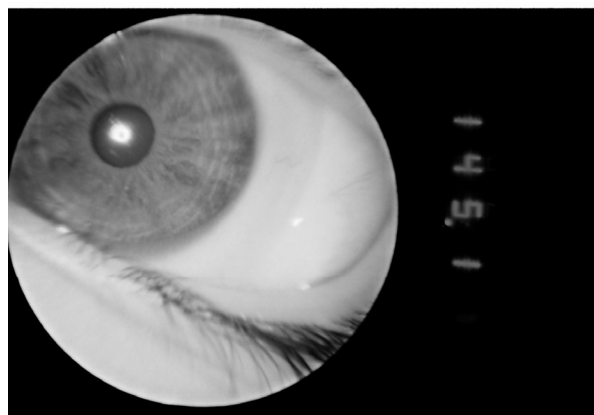


Fig. 4: Left eye.

Electroencephalogram: dominant alpha slight high voltage activity with irregular aspect, slow polymorph delta

and gamma waves; isolated, sharp high voltage synchronic waves – all in central derivations.

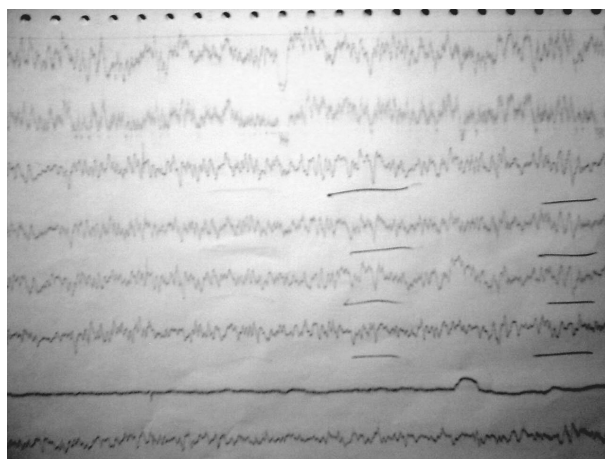
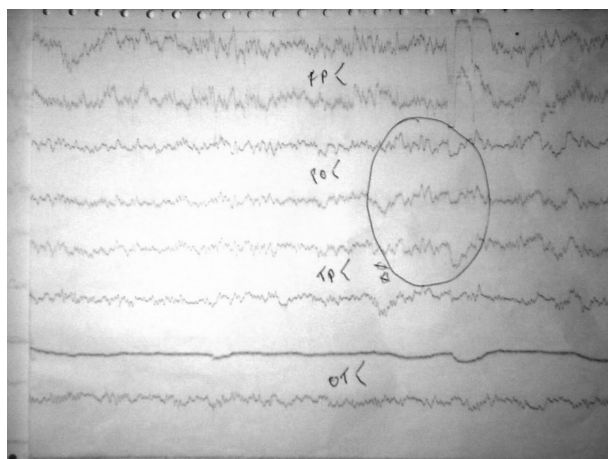


Fig. 5: E.E.G.

Cranial, thoracic and abdominal CT-scan – showed no evidence of angioma.

Otorhinolaryngologic exam found angioma placed on the nasal mucosa.

Discussions

The diagnosis was made on the presence of cutaneous lesions: multiple, extensive flat angiomas, predominant on right hemibody, repeated convulsions,

accompanied by moderate delayed cognitive skills and on anamnestic dates – absence of related pathology in family members.

We consider that the expression of cerebral angiomas are EEG alterations, seizures and the mental retardation and behavioral disorders, although cranial CT-scan found no signs of cerebral angioma.

Ocular manifestations include: flat angiomas – right eye, violaceous sclera, congenital hyperpigmented iris,

iris heterochromia, and congenital pigmented retina – bilateral. Right eye decreased visual acuity is probably explained by a tumoral compressive mass of the optic pathway which couldn't be found at CT-scan.

No manifestations of congenital glaucoma were found.

The SWK Syndrome was first described in 1879, and then Weber in 1922 and Krabe in 1934 made a complete description. Association of cutaneous and cerebral angiomas together with ocular manifestations characterizes the complete encephalotrigeminal angiomatosis.

Cutaneous lesions can lead to making the diagnosis from the first presentation. Classical angiomas are found on lower face, in the trigeminal nerve territory. Often angiomas extend also to the neck, trunk and abdomen as in our case. Cutaneous angiomas are presented in 90% of cases.

Cerebral angiomatosis is the second characteristic of the disease. Clinical manifestations are: seizures, migraine episodes in later childhood, hemiparesis, hemiplegia, constant and progressive mental retardation.

The most frequent ocular manifestations are: choroidal haemangioma and glaucoma. Classic forms of the disease include glaucoma- if this misses, in the presence of cutaneous and cerebral angiomas, the association is called Jahnke syndrome.

Other crude paucisymptomatic forms were included into distinct syndromes:

- Schirme syndrome – with precocious glaucoma and buphthalmos;

- Lawford syndrome – late glaucoma with chronic evolution, without buphthalmos;

- Moles syndrome – choroidal angioma, without enlargement of the eye;

- Brushfield-Wyatt – association of face angioma and calcified angioma in the contralateral cerebral hemisphere.

These entities are no longer considered as apart syndromes, but as forms of SWK syndrome.

The Roach Scale is used for classification:

- type I - includes facial and leptomeningeal angiomas; may have glaucoma;

- type II – facial angioma alone, no cerebral involvement; may have glaucoma;

- type III –isolated leptomeningeal angioma, or another cutaneous localization; usually no glaucoma.

Conclusions

The particularity of the case is the extension of cutaneous lesions accompanied by neuropsychic manifestations (seizures, mental retardation) in a child whose cerebral lesions and glaucoma were not found during investigations.

The child requires follow-up, as the evolution and prognosis may become dramatic, by aggravation of the neurological and mental disorders.

References

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