Sturge-Weber syndrome

Encephalofacial cavernous haemangiomatosis; Encephalotrigeminal haemangiomatosis

DESCRIPTION

Sturge-Weber syndrome (SWS) is one of the phakomatoses ('birthmarks'), the congenital or heritable conditions characterised by haemartomas and/or neoplasms throughout the body. The other phakomatoses include neurofibromatosis, tuberous sclerosis, ataxia-telangiectasia, Von Hippel Lindau syndrome and Wyburn-Mason syndrome.

The hallmark of SWS is the generally unilateral haemangiomas of the ipsilateral face, eye, meninges and brain. Haemangiomas are vascular haemartomas, being a benign congenital tissue malformation made up of cells in their normal location but malformed or misarranged. SWS is associated with an increased risk of glaucoma; an elevated intraocular pressure (IOP) secondary to either structural changes in the angle or an increased episcleral venous pressure. Progressive neurological changes in SWS can lead to epilepsy, haemiparesis and mental retardation.

SWS does not show a definitive inheritance pattern. There appears to be no predilection for either race or gender. The condition is usually diagnosed at birth because of the facial appearance.

SYMPTOMS

In cases where glaucoma occurs very early in life, tearing and photophobia are likely symptoms.

SIGNS

- Congenital facial haemangioma, also known as *nevus flammeus* or a 'port-wine' stain. The haemangioma of the face typically corresponds to the territory of the first and second divisions of the trigeminal nerve. Facial hypertrophy of the area may occur (Figure 1). If the haemangioma involves the upper eyelid, glaucoma is more likely
- Choroidal haemangioma is seen in about half of SWS cases (see also Choroidal haemangioma). Diagnosis of the diffuse orange-red choroidal haemangioma may be assisted by a comparison of fundus background colour with the fellow eye, ultrasound examination and fluorescein angiography
- Glaucoma may develop in approximately one-third of cases, and in about two-thirds of these, onset is in the first two years of life. An early rise in IOP can lead to buphthalmos, with an enlarged eye and cloudy cornea (Figure 1). Infantile glaucoma may lead to strabismus, amblyopia and anisometropia
- Other potential ocular signs include conjunctival and episcleral vascular abnormalities and telangiectasia (Figure 2), iris heterochromia and secondary exudative retinal detachment.

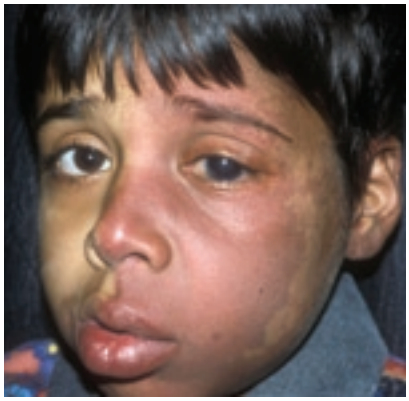

Figure 1 Sturge-Weber syndrome. The left eye is enlarged and cornea cloudy due to glaucoma. The facial haemangioma occupies the distribution of all three divisions of the trigeminal and some degree of tissue hypertrophy is present. (Courtesy of LV Prasad Eye Institute, Hyderabad, India)

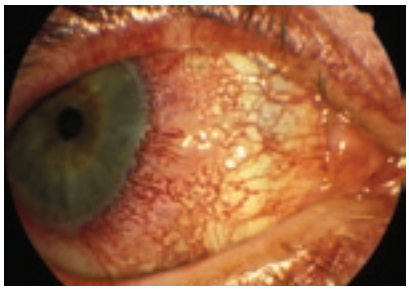

Figure 2 Conjunctival and episcleral vascular abnormalities and telangiectasia

PREVALENCE

SWS is rare, its actual prevalence being unknown.

SIGNIFICANCE

There may a reduced life expectancy in those patients who exhibit the full spectrum of the disease, in particular severe epilepsy and mental retardation. Glaucoma in SWS can prove refractory to treatment with a poor prognosis.

DIFFERENTIAL DIAGNOSIS

Other even less common conditions may show a facial haemangioma, for example, Klippel-Trenaunay-Weber syndrome. Choroidal haemangioma should be differentiated from choroidal melanoma and a serous detachment of the retinal pigment epithelium.

SEE ALSO

Congenital glaucoma, other phakomatoses listed above.

MANAGEMENT

Topical medication

An attempt to manage glaucoma, if present, may be initially made with aqueous suppressants. Alternatively, these medications can be used as an adjunct therapy following surgical intervention.

Laser and incisional surgery

Cases where the angle structures are anomalous may benefit from goniotomy. Filtration surgery may also be considered. Laser photocoagulation, radiation therapy and photodynamic therapy have been used to treat retinal detachment and enhance the resolution of sub-retinal fluid.

Refractive correction or therapy

Treatment of any strabismus and amblyopia should be initiated.

Review

Careful review is indicated for the development of glaucoma and retinal detachment. Neurological and dermatological manifestations may also require review.

The full series of these articles is available in the book *Posterior Eye Disease and Glaucoma A-Z* by Bruce AS, O'Day J, McKay D and Swann P. £39.99. For further information click on the Bookstore at **opticianonline.net**

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