Leber's congenital amaurosis

DESCRIPTION

The word 'amaurosis' refers to partial or complete vision impairment, particularly when there is no obvious cause. Leber's congenital amaurosis is a retinal dystrophic disease that is inherited as an autosomal recessive trait.

It is characterised by a bilateral severe vision loss, if not blindness, at birth or shortly thereafter, and should be suspected if an infant presents with vision loss, nystagmus and poor pupil reflexes in the presence of normal or near normal fundi. The few cases that retain reasonable vision in their early years tend to decline with time so that in adulthood they are legally blind. A variety of fundus changes may lead to a difficulty in diagnosis (see below).

Most patients are otherwise normal although certain potential systemic associations may be seen. These include mental retardation, deafness and renal, skeletal and cardiac abnormalities.

SYMPTOMS

There are few symptoms other than poor vision in those old enough to appreciate it. The parents may have noted roving eye movements and strabismus, as well as a poking or rubbing of the eyes which is known as the oculo-digital sign.

SIGNS

External signs include nystagmus and strabismus. Keratoconus is quite common and has been related to the propensity for eye rubbing. Continuous eye rubbing (oculo-digital syndrome) may cause a loss of orbital fat, leading to enophthalmos. High hypermetropic and high myopic refractive errors may be seen, with the former being more common. Pupil reflexes to light are sluggish and may be absent altogether. Cataracts frequently develop in the second decade.

As noted above, fundus changes can be very variable. The fundi may appear normal at birth and in infancy, with changes developing in the first few years of life. These include a peripheral pigmentary degeneration, the appearance of white flecks and a salt-and-pepper mottling, macular dysplasia and raised optic discs (pseudopapilloedema). With time optic disc pallor and attenuated arterioles develop. Fluorescein angiography is rarely helpful to the diagnosis and both photopic and scotopic components of the electroretinogram are severely abnormal or even absent.

Infant child with oculodigital syndrome in Leber's congenital amaurosis. Figure courtesy of Kanski J J *Clinical Ophthalmology* 4th Edition, Butterworth-Heinemann

PREVALENCE

It has been estimated that Leber's congenital amaurosis has a prevalence of about 3 per 100,000 live births.

DIFFERENTIAL DIAGNOSIS

If peripheral pigmentary changes are present there may some confusion with retinitis pigmentosa. Similarly, macular dysplasia can resemble macular scarring from toxoplasmosis. Optic atrophy as an early finding in some Leber's patients may be confused with autosomal recessive optic atrophy. The disease also needs to be distinguished from congenital stationary night blindness and achromatopsia.

MANAGEMENT

Advice

There is no treatment for the ocular complications of this disease.

Genetics

Paediatric and genetic consultations should be obtained.

Refractive correction or LVAs

If necessary, spectacles should be prescribed following refraction under cycloplegia to enhance any remaining vision.

The full series of these articles will be 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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