Retinoschisis - Juvenile X-linked

DESCRIPTION

Retinoschisis is a splitting within the sensory retina, and may occur in age-related degenerative and X-linked recessive forms. The age-related degenerative form is described in the previous condition (*Optician*, June 20).

X-linked recessive retinoschisis is a splitting of the inner retina at the level of the nerve fibre layer. It occurs almost always in males and is congenital and bilateral, with the affected gene being localised to the short arm of the X chromosome. The carrier females do not appear to be detectable with the ophthalmoscope. X-linked retinoschisis appears to affect all races, but with many cases being reported from Finland, it may be the most common X-linked disease found in that country. There are no known associations between this condition and systemic diseases.

SYMPTOMS

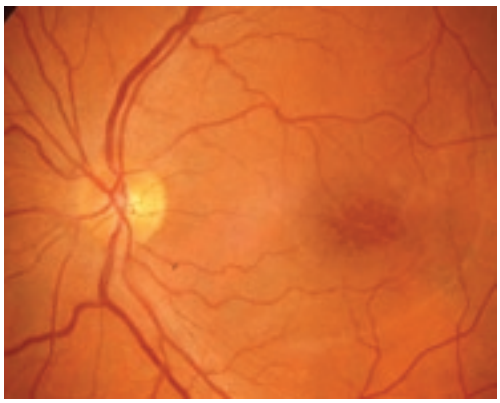
Affected males typically present with an unexplained loss of vision in both eyes during the first decade of life. At this age, vision is usually at the level of 6/9 or 6/12. There is a slow inexorable decline over the ensuing decades with vision being around 6/18 to 6/24 at age 20 and falling to 6/60 by 60 years of age. The advent of vitreous haemorrhage may cause a more acute vision loss.

SIGNS

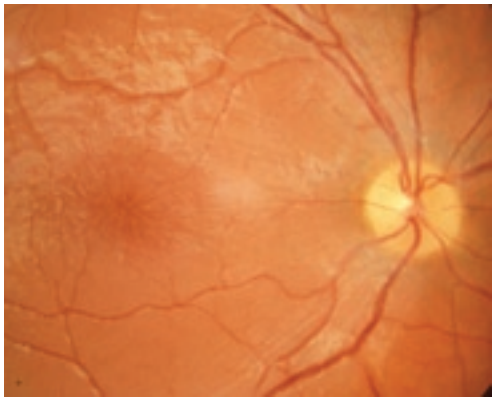
The classical finding of a cystic schisis at the maculae is present in almost all cases from an early age. They radiate in a stellate or cartwheel configuration often accompanied by retinal folds. Unlike similarly appearing changes in cystoid macular oedema, there is no leakage on fluorescein angiography. With age the characteristic appearance changes to one of pigmented macular atrophy.

In half of all cases the macular changes are accompanied by a peripheral degenerative retinoschisis, most commonly found in the inferior temporal quadrant of the fundus.

Vitreous haemorrhage may arise from a superficial neovascular tuft or bleeding with a peripheral cyst. Vitreous veils and retinal detachment can ensue. A star-shaped posterior subcapsular cataract may develop in some patients in adulthood.



Juvenile X-linked retinoschisis in a 36-year-old male patient. Vision was 6/36 in each eye



Juvenile X-linked retinoschisis at the right macula of an 11-year-old boy. Vision was 6/12 in each eye. His younger brother and two male first cousins also showed the dystrophy

PREVALENCE

Rare.

DIFFERENTIAL DIAGNOSIS

Age-related peripheral retinoschisis; Rhegmatogenous retinal detachment; Cystoid macular oedema; Goldmann-Favre disease.

MANAGEMENT

Additional investigations

Visual field testing will usually reveal relative central scotomas and absolute scotomas consistent with areas of peripheral retinoschisis such as constrictions of the superior nasal field. Colour vision testing is not helpful in the diagnosis.

The b-wave of the electroretinogram is typically abnormal in all cases. The electro-oculogram is generally normal until late in the disease process. Dark adaptation is also usually normal.

Refractive correction or LVAs

There is no treatment available for the retinoschisis at the macula. Low vision services and genetic counselling should be considered.

Incisional surgery

While a complicating vitreous haemorrhage usually spontaneously resolves, a vitrectomy may be contemplated in cases where the haemorrhage is persistent. If there is a risk of retinal detachment or if a retinal detachment has occurred, appropriate surgery should be entertained.

Review

In view of the potential for loss of visual acuity and possible complications, annual review is indicated.

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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