Stargardt's disease

Fundus flavimaculatus

DESCRIPTION

Stargardt's disease is an autosomal recessively inherited dystrophy of the macula. Rarely, autosomal dominant forms have been described. In the recessive form of Stargardt's disease, the ABCR gene, felt by some to be mapped to the short arm of chromosome 1, has been identified as the causative agent. The autosomal dominant form of the condition has been mapped to chromosome 6. The term *fundus flavimaculatus* refers to the yellow fleck-like deposits which are typically seen in this condition, and at one time, Stargardt's disease and *fundus flavimaculatus* were thought to be separate entities.

SYMPTOMS

Typically, a child or young adult will present with a progressive, bilateral, symmetrical loss of vision. The rate of vision loss is usually fairly slow to about 6/12, and thereafter can be more rapid to 6/60.

SIGNS

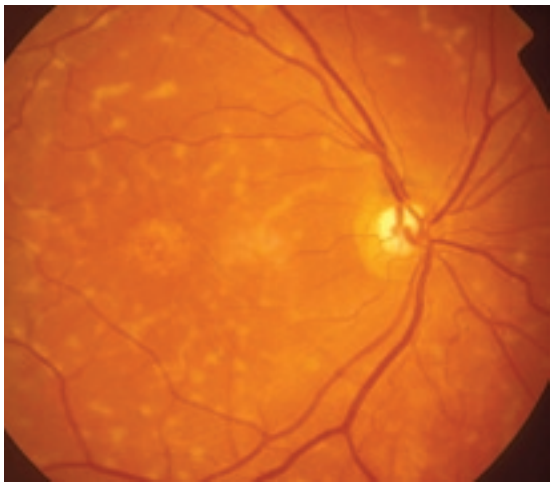
In the early stages of the disease, there may be little to see with the ophthalmoscope, despite significant visual symptoms. Eventually, the macula assumes an atrophic beaten-metal appearance due to atrophy of the RPE with surrounding soft yellow flecks that are often characteristically fish-tailed or pisciform. These deposits at the level of the retinal pigment epithelium (RPE), which appear to be lipofuscin material, may involve a wide area of the central retina. In some cases, macular atrophy predominates and may have a bull's-eye appearance, whereas in others the yellow flecks are the most obvious feature.

PREVALENCE

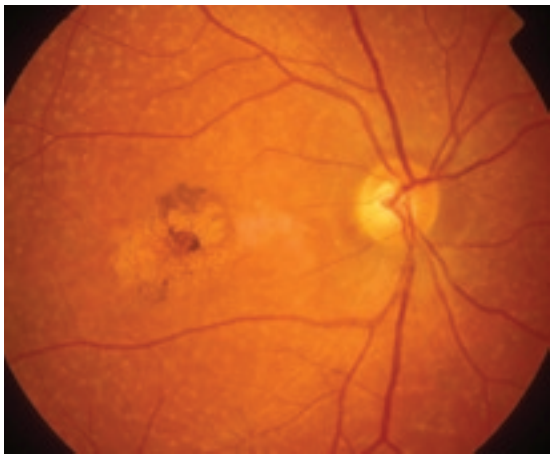
Stargardt's disease has a prevalence of about 1 in 20,000. It is the pre-eminent cause of degeneration of the macula affecting the RPE in the first two decades of life, comprising about 7 per cent of all retinal dystrophies.

DIFFERENTIAL DIAGNOSIS

Other conditions that may produce deposits or atrophy in the macular region include: Drusen, toxic maculopathies – from, for example, chloroquine, tamoxifen and talc, Cone dystrophy, *Fundus albipunctatus* and *Retinitis punctata albecens*.



Stargardt's disease of the right fundus of a 35-year-old male, showing early macular atrophy and flavimaculatus fleck changes at the retinal pigment epithelial level



Stargardt's disease of the right fundus of an 18-year-old male low vision patient. Advanced macular atrophy and yellow flavimaculatus flecks affected both fundi

SEE ALSO

Non-physiological visual loss.

MANAGEMENT

Additional investigations

Testing with an Amsler grid usually reveals central abnormalities and typically macular photostress recovery times are prolonged. Visual field testing may reveal a loss of central sensitivity with normal peripheral fields. Macular hyperfluoresence and a silent choroid may be seen with fluorescein angiography. Electrophysiological testing will usually reveal a normal ERG in the early stages of the disease, becoming more affected as the disease progresses. The EOG is also usually normal.

Advice

Unfortunately, early in the disease process, labels such as malingering or psychogenic amblyopia are sometimes used. People who malinger are often aggressive and are trying to avoid some activity, such as going to school or military service. Psychogenic amblyopes, unlike malingerers, are usually mild mannered and actually experience their symptoms. Signs may include a variable visual acuity, visual field and colour vision defects of a non-organic nature, normal findings when examining the anterior eye, pupil responses and fundus, and convergence and accommodation insufficiency. Such labels only serve to delay the correct diagnosis.

Refractive correction or LVAs

There is no treatment for this condition. Low vision and other rehabilitative services may be beneficial and genetic counselling should be considered.

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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