Pattern dystrophies of the retinal pigment epithelium

Butterfly dystrophy, Sjögren's recticular dystrophy, Fundus pulverulentus, Adult vitelliform dystrophy or degeneration

DESCRIPTION

The name pattern dystrophy of the retinal pigment epithelium (RPE) groups together a series of macular conditions previously named with a variety of descriptive terms and eponyms. Some examples include butterfly dystrophy, Sjögren's reticular dystrophy, fundus pulverulentus and adult vitelliform dystrophy or degeneration (adult Best's disease).

These conditions tend to share the following features:

- An autosomal dominant inheritance pattern
- A minimal effect on visual function, certainly in the early stages
- A good prognosis for vision.

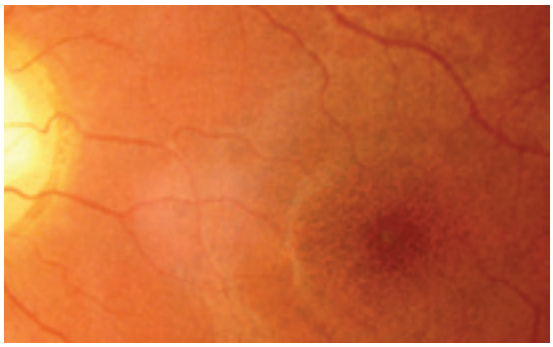
The fact that different types of pattern dystrophy can be seen within the same family lends support to the notion of a single disease process. Also the pattern of the macular pigmentation may change with the passage of time. The Sjögren's reticular type can be inherited as an autosomal recessive trait. In the autosomal dominant form of the disease, the peripherin/RDS gene, which is located on chromosome 6, is thought to be defective.

Reports of associations with systemic diseases have been few and include *pseudoxanthoma elasticum*, myotonic dystrophy and maternally inherited diabetes and deafness.

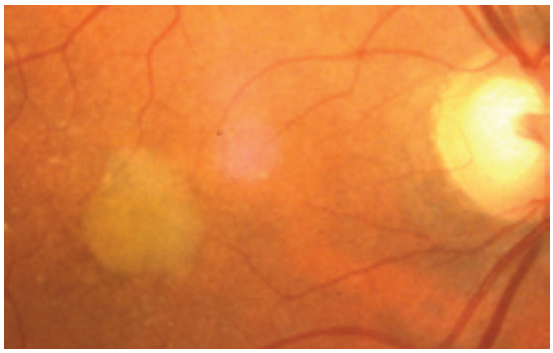
The yellow deposits in pattern dystrophies are due to the accumulation of lipofuscin within enlarged RPE cells. Lipofuscin accumulates with age and is present in abnormal amounts in many degenerations and can overlay choroidal naevi and melanomas.

SYMPTOMS

The pattern dystrophies may well be discovered during a routine eye examination. Visual acuity usually remains stable until older age, when some decline in vision may be noted. Usually there is little effect on colour vision, dark adaptation and the visual field, although isolated examples of paracentral visual field defects and a tritan colour vision deficiency have been described.



Pattern dystrophy (fundus pulverulentus) of the macula in a 12-year-old boy, with pepper and salt-like array of yellow pigment deposits at the macula. The boy's mother and his two brothers had the dystrophy as well as three first cousins, his mother's sister and brother and his maternal grandmother



Adult vitelliform (Best's) disease in a 60-year-old female. Vision was normal

SIGNS

Possible patterns include:

- A circumscribed, raised yellow lesion (adult vitelliform dystrophy)
- Butterfly and reticular arrangements of pigment
- Multiple yellow deposits which may simulate fundus flavimaculatus and punctate pigment mottling (fundus pulverulentus).

The lesions are typically bilateral and symmetrical. Electrophysiological testing may reveal an abnormal electro-oculogram, and a mild reduction in the electroretinogram in older patients. In old age, the disease can mimic dry or atrophic age-related macular degeneration in appearance.

PREVALENCE

Rare.

DIFFERENTIAL DIAGNOSIS

Other conditions with macular pigmentary and yellow deposits: Stargardt's disease; Best's vitelliform dystrophy; Cone dystrophies, Drusen; Age-related macular degeneration.

MANAGEMENT

Additional investigations

With fluorescein angiography the RPE pigment deposits in the macular area will typically hypofluoresce as they block the underlying choroid. Any adjacent areas of RPE atrophy will hyperfluoresce. Ocular coherence tomography may assist with differential diagnosis. Photostress test (glare recovery) and Amsler grid are also useful tests in macular evaluation.

Advice

There is no treatment for the pattern dystrophies and patients should be reassured that the condition should not significantly affect their vision, at least until old age. Even then, the degree of vision loss may be quite mild.

Review

Regular review should be undertaken to monitor any change. The examination of other family members to identify further cases is always useful.

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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