Cone dystrophies

Progressive cone degeneration
Cone-rod dystrophy
Rod-cone dystrophy

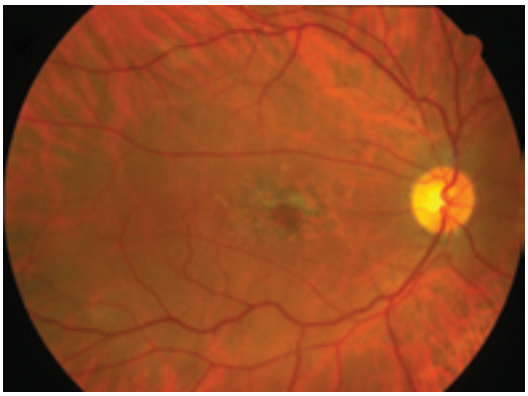

FIGURE 1. Granular pattern of pigmentary change at the macula

DESCRIPTION

These are a heterogenous group of conditions usually characterised by an early, bilateral and slowly progressive loss of central vision. Although frequently sporadic in nature, when inherited, it is usually as an autosomal dominant trait. Autosomal recessive and X-linked recessive forms have been described. Autosomal dominant cone dystrophy has been mapped to chromosome 17. In some cases of cone dystrophy, rod dysfunction will also develop over time (cone-rod degeneration), whereas in others, rod dysfunction may precede that of the cones (rod-cone degeneration).

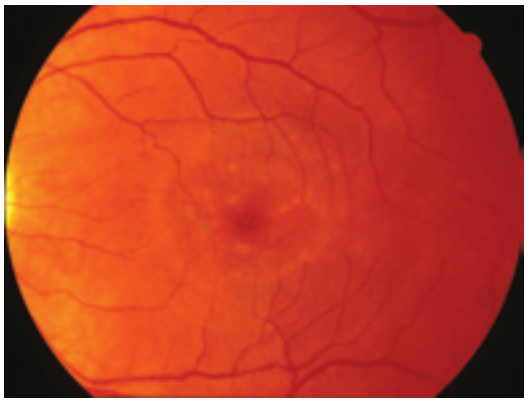

FIGURE 2. Bull's eye pattern of retinal pigment epithelial atrophy

SYMPTOMS

The two most common initial symptoms in cases of cone dystrophy are blurred vision and photophobia. Once vision has declined to a level of 6/12 to 6/18, colour vision may be severely affected. Red-green colour vision defects are most commonly seen. Vision loss may well commence in the first decade of life, declining slowly to around 6/60 by the third or fourth decade. Some patients are quite sensitive to glare and will complain of difficulty adjusting to changing light levels. Some difficulty with night vision is usually present in cases of rod-cone degeneration, with this sub-type carrying the worse overall prognosis for vision.

SIGNS

Fundus changes in these conditions are very variable. The fundus may appear normal in the early stages of the disease, even in the presence of blurred visual acuity. Later in the disease process, a bull's eye or central pattern of RPE atrophy may be observed. Other potential fundus signs include pigment clumping in the macular region, a tapetal sheen at the maculae, peripheral pigmentary changes that may lead to confusion with retinitis pigmentosa and temporal pallor of the optic disc. Nystagmus is commonly seen.

Examination of the visual fields typically shows central, paracentral and pericentral ring scotomas. Central scotomas are likely to deepen and enlarge with time. Peripheral visual fields are usually normal to begin with, but in patients where rod involvement ensues, peripheral sensitivity becomes depressed.

PREVALENCE

Rare.

DIFFERENTIAL DIAGNOSIS

Other conditions that may lead to bull's-eye or geographic RPE changes at the maculae, and pigmentary changes in the fundus such as: Chloroquine maculopathy; Stargardt's disease; Age-related macular degeneration; Central areolar choroidal dystrophy; Retinitis pigmentosa; Optic atrophy.

MANAGEMENT

Additional investigations

Electrophysiological studies are important in establishing a definitive diagnosis. Typically with the ERG in cone dystrophy, the photopic 'B' wave is depressed and the 30Hz flicker shows a reduced amplitude. Rod involvement, either early or late, leads to a reduction in rod-mediated ERG responses and reduced dark adaptation. Electro-oculography is unlikely to provide further information. Fluorescein angiography may reveal macular RPE window defects at an early stage in cases of cone dystrophy, and may also aid in the more accurate delineation of bull's eye macular changes early in development.

Refractive correction and low vision assistance

There is no treatment for cone dystrophy. Low vision services are appropriate. Tinted spectacles or contact lenses have been suggested to reduce photophobia and thereby improve vision.

Topical medication

Weak miotics may be used during the day such as 0.5 per cent pilocarpine.

Genetic counselling

Patients and their relatives may be offered genetic counselling.

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 www.opticianonline.net.

◆ **Adrian Bruce** is a Chief Optometrist at the Victorian College of Optometry and a Senior Fellow, Department of Optometry and Vision Sciences, The University of Melbourne.
◆ **Justin O'Day** is an Associate Professor in the Department of Ophthalmology, The University of Melbourne and Head Of Neuro-Ophthalmology Clinic, Royal Victorian Eye and Ear Hospital.
◆ **Daniel McKay** is a Medical Officer at the Royal Victorian Eye & Ear Hospital.
◆ **Peter Swann** is Associate Professor in the School of Optometry, Queensland University of Technology.