Choroideraemia

DESCRIPTION

Choroideraemia is an X-linked recessive condition with progressive, bilateral atrophy of the choroid and overlying retinal pigment epithelium. Affected patients develop night blindness in childhood, progressive constriction of the visual fields in adulthood, and profound visual impairment later in life. No specific treatment is available. The inheritance pattern implies that only males are affected, while females are unaffected carriers (although rare cases of carriers with severe disease have been reported).

SYMPTOMS

Night blindness (nyctalopia) is typically noted in boys in the first decade, but may be delayed up to age 30. Loss of peripheral vision often becomes evident in the teenage years. Central vision is usually severely impaired by middle age.

SIGNS

The first signs to appear are areas of pallor or irregular pigmentation in the peripheral fundus, representing patches of choroidal and retinal pigment epithelium atrophy. With disease progression, more extensive atrophy allows the underlying choroidal vessels to become more prominent. By this time, constriction of the peripheral visual fields is evident. Later in life, further choroidal atrophy may expose the underlying, pale sclera. Visual acuity is usually reduced to 6/60 or worse by age 50. The optic disc, retinal vessels and colour vision remain relatively normal throughout the course of the disease. Routine examination of other family members is essential. Female carriers often develop fine, mottled areas of brown pigment in the peripheral fundus.

PREVALENCE

Rare (approx one in 100,000). All daughters of affected males are carriers. No male children will be affected if the mother is not a carrier. Half of the male children of female carriers will develop choroideraemia, and half of the female children will be carriers.

SIGNIFICANCE

Most patients are legally blind by age 50 to 60 years. No specific treatment is available. Genetic counselling is critical.

DIFFERENTIAL DIAGNOSIS

Retinitis Pigmentosa, Phenothiazine Retinopathy, Gyrate Atrophy.

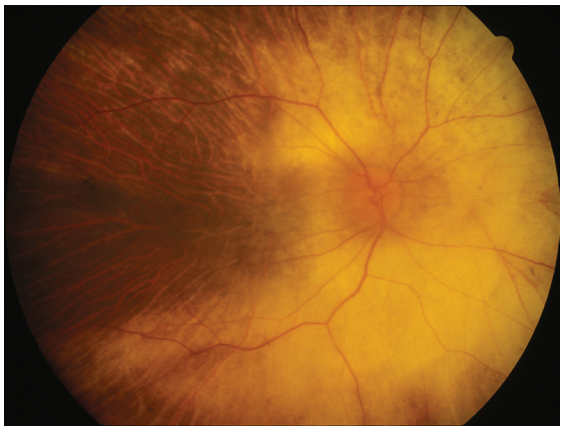

FIGURE 1. Extensive RPE and choroidal atrophy, exposing underling choroidal vessels and sclera

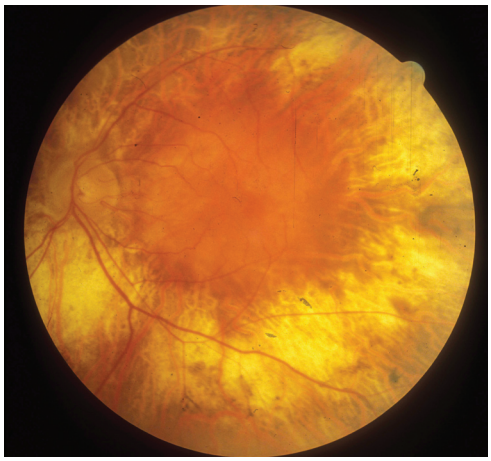

FIGURE 2. Choroideremia in an adult male patient. Vascular attenuation is present

SEE ALSO

Congenital stationary night blindness.

MANAGEMENT

Ocular tests, imaging investigations

In the early stages of choroideraemia, dark adaptation thresholds are increased, and the electroretinogram (ERG) is reduced. The ERG subsequently becomes unresponsive. Fluorescein angiography reveals normal retinal vessels and hyperfluorescent patches corresponding to areas of chorioretinal atrophy.

Advice

No effective treatment is available for choroideraemia. Yearly review, patient education, low vision aids and correction of refractive errors will allow patients to obtain maximum benefit from their remaining vision.

Genetics

Genetic counselling provides families with an understanding of the implications of the disease, and may affect family planning. Genetic tests have been developed to identify affected patients and female carriers; prenatal diagnosis via choroinic villus sampling is also available.

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.