



Retinitis pigmentosa

DESCRIPTION

Retinitis pigmentosa (RP) is a bilateral and usually symmetrical pigmentary dystrophy that at first predominantly affects the retinal rods, with the retinal cones more involved as the condition progresses. RP encompasses a group of conditions, some purely ocular in nature and others that have significant systemic components. Examples of the latter include Bassen-Kornzweig syndrome, Refsum's disease, Usher's syndrome, Laurence-Moon and Bardet-Biedl syndromes, and Kearns-Sayer syndrome. RP may be inherited as an autosomal dominant, autosomal recessive or an X-linked recessive trait. An autosomal recessive pattern is the more common, followed by the dominant type. The X-linked type is the least common. The recessive forms of the disease tend to occur earlier in life and are more visually destructive.

SYMPTOMS

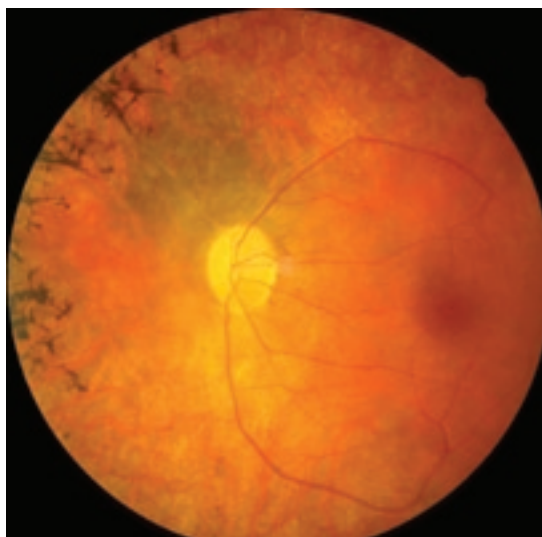
The pre-eminent symptom is poor night vision (nyctalopia, night blindness). Good central vision is usually retained until late in the disease but can be affected by associated ocular conditions. Progressive visual field loss leads to a disabling tunnel vision. These patients are often quite sensitive to glare.

SIGNS

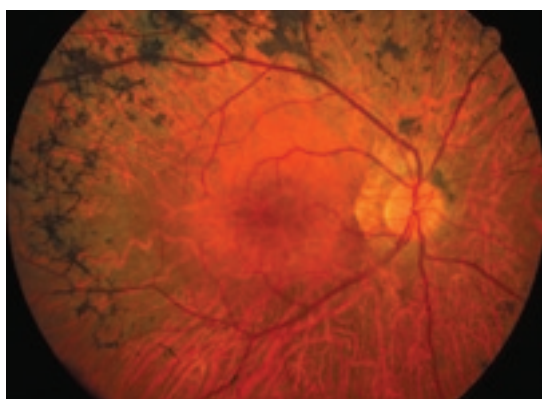
The earliest signs of retinal pigmentary epithelial hypertrophy and atrophy afflict the equatorial and mid-peripheral fundus, and if detected at this stage, the term retinitis pigmentosa *sine pigmento* is sometimes applied. These are followed by the classically described areas of spidery, bone-spicule pigmentary clumps which typically have a perivascular disposition. These pigmentary changes progress both peripherally and centrally, and correspond to the annular scotoma in the visual field.

In time, optic atrophy supervenes with the optic disc, often described as having a waxy pallor and the retinal arterioles become attenuated. The macula may show cystoid oedema, and epiretinal membranes may develop.

Optic nerve head drusen, open-angle glaucoma, keratoconus and myopia are more common in RP patients. Vitreous opacities are often noted and posterior subcapsular cataract is a frequent complication.



The left fundus of a 25-year-old white male with retinitis pigmentosa. Optic atrophy and vascular attenuation accompanied bone-spicule pigmentation. There is a coincidental choroidal naevus above the optic disc



Bone spicule pigmentation in retinitis pigmentosa

PREVALENCE

Uncommon to rare (approximately 1 in 5,000); the most common hereditary retinal degeneration.

DIFFERENTIAL DIAGNOSIS

Poor night vision: congenital stationary night blindness, choroideremia, gyrate atrophy of the choroid and vitamin A deficiency. Similar retinal appearance: drug toxicities eg thioridazine and chloroquine, trauma, old retinal detachment and inflammatory disorders such as syphilis and congenital rubella.

MANAGEMENT

Additional investigations

Visual field assessment will show mid-peripheral losses. A severe attenuation of electrophysiological responses may be seen early in the disease process. Dark

adaptation will be adversely affected. If systemic disease association is suspected, neurologic or blood workup may be beneficial.

Genetic

There is no treatment for RP. It is important that patients and their relatives are offered genetic counselling and they often benefit from their interaction with RP support groups.

Refractive correction or low vision aids

Low vision and other rehabilitative services must be considered as the disease progresses.

Incisional surgery

Patients who have significant symptoms from posterior subcapsular cataract should be considered for cataract surgery.

Oral medication

Cases complicated by cystoid macular oedema may benefit from oral carbonic anhydrase inhibitors. Some believe that vitamin A supplementation may be helpful in some cases. One report indicated that the progression of electrophysiological deficits may be retarded by vitamin A. This approach, however, is somewhat controversial.

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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