



Gyrate atrophy

DESCRIPTION

Gyrate atrophy of the choroid and retina is a rare metabolic chorioretinal dystrophy that is inherited as an autosomal recessive trait. It is bilateral and slowly progressive, commencing as a mid-peripheral atrophy of choroidal vessels, the RPE and photoreceptors. Even though the central retinal areas appear normal in the earlier stages of the disease, RPE and photoreceptor damage has been demonstrated. Gyrate atrophy is associated with a many-fold increase in the levels of the amino acid ornithine in body fluids, due to a deficiency of the enzyme ornithine ketoacid aminotransferase (OAT). The gene for OAT has been mapped to chromosome 10. Because OAT is a vitamin B6-dependent enzyme, dietary supplementation with vitamin B6 has been utilised as a treatment regimen to reduce plasma ornithine levels. However, only a minority of patients respond to this mode of therapy. Few systemic associations with gyrate atrophy have been reported. Electromyographic testing may reveal abnormalities in most patients, although few attest to overt muscle weakness.

SYMPTOMS

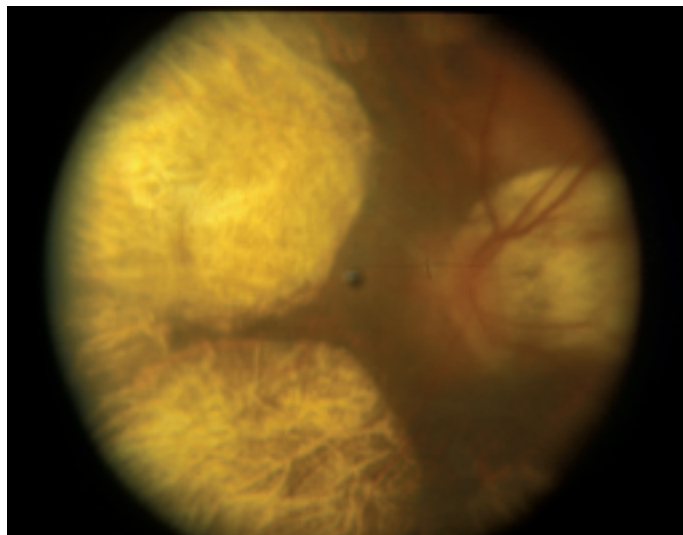
Typically, patients with gyrate atrophy develop night vision difficulties in their teens and 20s, and sometimes in the first decade.

In the early stages, central vision is good. As the condition advances, central vision deteriorates either from involvement of the maculae in the disease process or from potential complicating factors such as cystoid macular oedema or posterior subcapsular cataract.

SIGNS

The classical signs are mid-peripheral areas of chorioretinal atrophy that have scalloped edges and are separated from each other by thin bands of pigmentation.

These expand both towards the central areas of the fundi and peripherally. The maculae are usually spared until late in the course of the disease. Optic atrophy and thinning of the retinal arterioles are noted later on. Other intraocular changes that have been described include posterior subcapsular cataract, posterior vitreous detachment, epiretinal membranes and cystoid macular oedema. These patients may also have high degrees of myopia and astigmatism.



Essential gyrate atrophy of the choroid
(Courtesy of LV Prasad Eye Institute)

Defects in the visual field match the atrophic areas in the fundi, initially being mid-peripheral scotomas that may coalesce to form an annular scotoma similar to that seen in retinitis pigmentosa. Progression of these defects leaves patients with markedly reduced visual fields usually by their fifth decade.

Electrophysiological studies, including the electroretinogram, electro-oculogram and dark adaptation, typically give markedly abnormal findings. Carriers of gyrate atrophy usually have normal fundi.

PREVALENCE

The prevalence of gyrate atrophy is unknown. There have been reports suggesting a higher number of cases in Finland, and a prevalence of 1 in 50,000 has been postulated in that country. The disease is rare.

DIFFERENTIAL DIAGNOSIS

Choroideraemia; Atypical retinitis pigmentosa; Pavingstone degeneration; Chorioretinal atrophy from high myopia; Pigmentary retinopathy from thioridazine, Congenital stationary night blindness.

MANAGEMENT

Genetics

As mentioned, gyrate atrophy is inherited as an autosomal recessive trait that has been mapped to chromosome 10.

Oral medication and diet

Plasma ornithine levels may be reduced by the administration of vitamin B6 (pyridoxine hydrochloride). Only a small proportion of patients prove to respond to this treatment and there is little evidence that it alters the progression of the disease. Another approach is to reduce the dietary intake of protein, an arginine-restricted diet, and there is some evidence that this, especially if started at an early age, may slow the disease process.

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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