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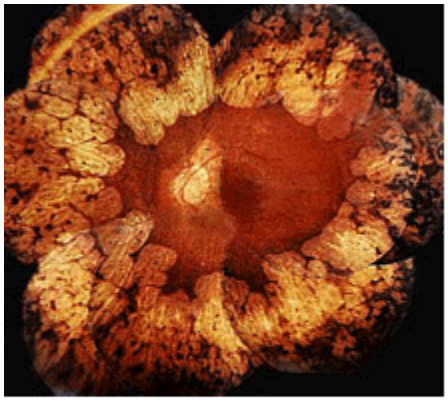
**NLM Citation:** National Center for Biotechnology Information (US). Genes and Disease [Internet]. Bethesda (MD): National Center for Biotechnology Information (US); 1998-. Gyrate atrophy of the choroid and retina.
**Bookshelf URL:** https://www.ncbi.nlm.nih.gov/books/


# Gyrate atrophy of the choroid and retina



The retina of a patient with gyrate atrophy of the choroidand retina of the eye caused by ornithine aminotransferase (OAT) deficiency. [Image credit: Muriel Kaiser-Kupfer, NEI, NIH, Bethesda, MD, USA and David Valle, Johns Hopkins University, Baltimore, MD, USA.]

People suffering from gyrate atrophy of the choroid (the thin coating of the eye) and retina face a progressive loss of vision, with total blindness usually occurring between the ages of 40 and 60. The disease is an inborn error of metabolism.

The gene whose mutation causes gyrate atrophy is found on chromosome 10, and encodes an enzyme called ornithine ketoacid aminotransferase (OAT). Different inherited mutations in OAT cause differences in the severity of symptoms of the disease. OAT converts the amino acid ornithine from the urea cycle ultimately into glutamate. In gyrate atrophy, where OAT function is affected, there is an increase in plasma levels of ornithine.

It is already known that reduction of the amino acid arginine in the diet has a salutary effect on most patients. Current lines of research into the disease include: (1) investigating how variant mutations of the alleles (versions of the gene inherited) interact in order to cause the differing symptoms of the disease and (2) work on mouse models of the disease is furthering our understanding, which is hoped will lead to a true cure.

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