Ophthalmologic abnormalities in long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency: presentation of a long-term survivor

Veit Sturm

PURPOSE
Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency is one of the recently discovered defects of mitochondrial fatty acid beta-oxidation surprisingly associated with ophthalmologic abnormalities. The presentation of a long-term survivor may enlarge the clinical spectrum associated with this disorder.

METHODS
A 12-year retrospective review of the clinical course of a 19-year-old long-term survivor was performed. The author concentrated on characteristic ophthalmologic measures: visual acuity, refraction, ophthalmoscopy, visual fields, and electroretinography.

RESULTS
The author found a milder course than described in the literature, although very few case reports of long-term survivors have been published. The patient developed slower circumscribed atrophy of the choroid, retinal pigment epithelium, and retina.

CONCLUSIONS
Because of therapeutic and prenatal diagnostic opportunities in LCHAD deficiency, it is important to recognize this severe disorder early in its course. This may lead to a milder course and better prognosis due to early dietary therapy.