Report of a Case. A 24-year-old woman had been diagnosed with abetalipoproteinemia at age 2 years. The diagnosis was based on peripheral blood acanthocytosis, low serum cholesterol levels, absence of serum lipoprotein, and characteristic lipid deposits in mucosal cells on small bowel biopsy. She was treated with vitamin E injection and oral vitamins A, D, and K. Results of initial ocular examinations were within normal limits.

At age 18 years, the patient voluntarily discontinued treatment with vitamin supplementation; afterward she reported worsening of night vision and progressive field changes. She was evaluated at the Ocular Genetics Clinic of The Hospital for Sick Children, Toronto, Ontario, at age 24 years. Best-corrected vision was 20/20 OU. Refraction was −0.75+0.50×165° OD and −1.00+1.00×175° OS. Ophthalmoscopy showed bilateral and symmetric helicoid peripapillary changes (**Figure 1**), equatorial retinal pigment epithelium (RPE) mottling, and attenuation of blood vessels but absence of true angioid streaks. There was no sign of inflammation. Fluorescein angiography showed a large peripapillary defect with no evidence of leakage or angioid streaks (**Figure 2**). A Goldmann visual field examination revealed bilateral constriction of the peripheral field and enlargement of the blind spot. Electroretinogram recordings were severely attenuated to all stimuli.

The serum level of vitamin A was 0.20 µmol/L (normal range, 1.05-3.14 µmol/L), whereas for vitamin E, it was 7.0 µmol/L (normal range, 12.0-46.0 µmol/L). Vitamin therapy was restarted, and 6 months into treatment there was no progression of symptoms.

Comment. Helicoid peripapillary chorioretinal atrophy is a rare disorder characterized by winglike chorioretinal atrophy emanating from the optic disc. A dominant form was recently mapped to chromosome 11p15.[4] The differential diagnosis of HPCD includes serpiginous choroiditis, angioid streaks, malignant myopia, paravenous retinochoroidal atrophy, and radial lattice reti-

Helicoid Peripapillary Chorioretinal Degeneration in Abetalipoproteinemia

Abetalipoproteinemia is a rare autosomal recessive disorder characterized by the absence of apolipoprotein B.[1] It is caused by mutations of the microsomal triglyceride-transfer protein gene.[2] Ocular manifestations include retinitis pigmentosa–like changes, nystagmus, ophthalmoplegia, ptosis, cataracts, anisocoria, and angioid streaks.[3] Helicoid peripapillary chorioretinal degeneration (HPCD) is characterized by chorioretinal atrophy that radiates from the optic disc as winglike extensions.[4] No systemic disease has been associated with HPCD. We report a case of HPCD associated with abetalipoproteinemia.

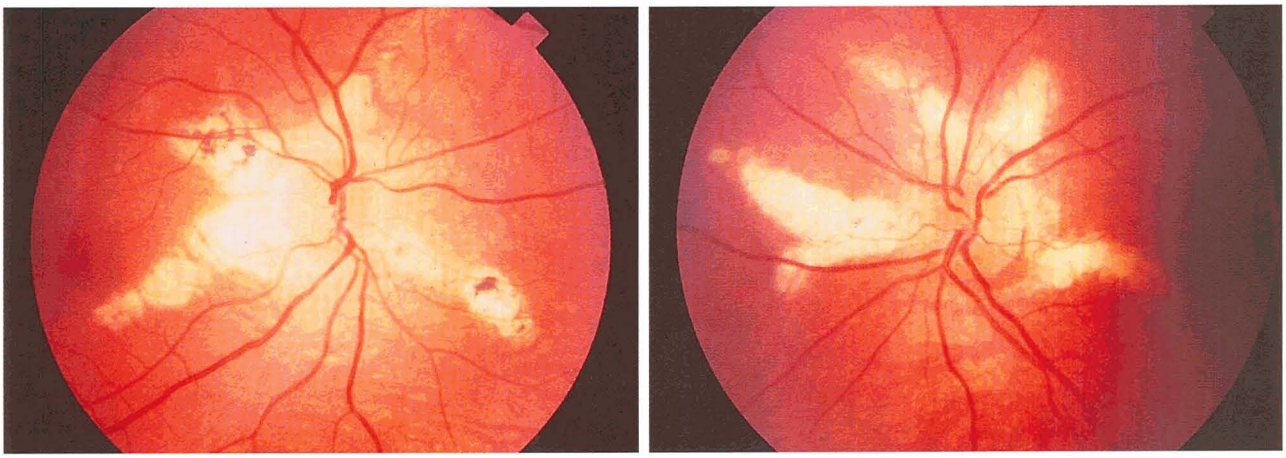

Figure 1. *Fundus photographs showing bilateral symmetrical wing-shaped atrophy radiating from the optic discs, compatible with helicoid peripapillary chorioretinal degeneration in the right (left) and left (right) eyes.*

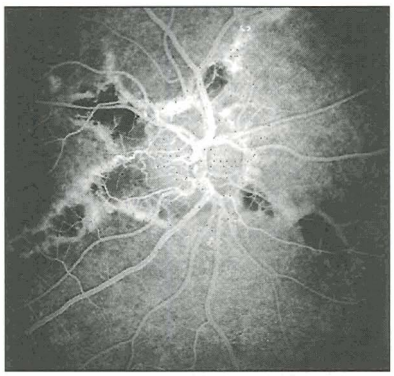

Figure 2. *Corresponding late-phase fluorescein angiography of the patient's right eye showing the absence of dye leakage from the peripapillary lesions.*

nal degeneration.[5] The absence of inflammatory signs, the symmetry of the fundus lesions, the characteristic wing-shaped atrophy, and the absence of leakage on fluorescein angiography in our patient support the diagnosis of HPCD.[5]

Brazitikos and Safran[6] suggested that HPCD is caused by dysplastic abnormalities of the peripapillary RPE, which predispose it to damages from mechanical stretching of the globe during growth, progressive tearing of the RPE, and subsequent chorioretinal atrophy. The pathogenesis of HPCD, and its possible relationship to angioid streaks, remains unclear.

Vitamin E deficiency has been implicated as a cause of retinal changes in abetalipoproteinemia.[7] Vitamin E acts as a free radical scavenger and prevents oxidative injury to membrane lipids.[1] Deficiency of vitamin E may decrease its protective effect on the RPE, predisposing the RPE to the tearing mechanism proposed by Brazitikos and Safran.[6]

To our knowledge, this case represents the first association of HPCD with abetalipoproteinemia. Whether HPCD represents a true distinct manifestation of abetalipoproteinemia or a variant of angioid streaks remains to be clarified.

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