Image in cardiology

Extremely low high-density lipoproteins and corneal opacities

Lipoproteínas de alta densidad extremadamente bajas y opacidades corneales

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Received 18 October 2022; Accepted 21 November 2022

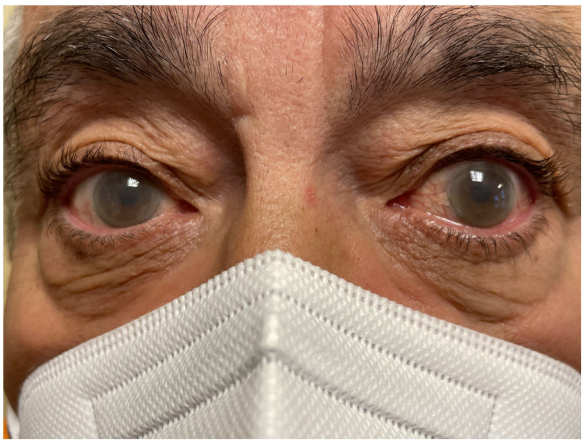

Figure 1.

Primary hypoalphalipoproteinemia is an uncommon disorder characterized by the abnormal production of apolipoprotein (apo) A-I, resulting in extremely low levels of high-density lipoprotein cholesterol. The image shown is of a 60-year-old man, with consanguineous parents, with high-density lipoprotein cholesterol levels of 7 mg/dL (40-60), triglycerides of 135 mg/dL, total cholesterol of 119 mg/dL, low-density lipoprotein cholesterol of 83 mg/dL, lipoprotein A of 15.8 mg/dL (< 30), and apoB of 99 mg/dL (55-140). He was on treatment with simvastatin 10 mg daily.

Examination findings of note included xanthelasma and complete corneal arcus with bilateral corneal opacities that did not affect visual acuity (figure 1). The patient gave written consent for his photographs to be taken and for the case to be published. The initial diagnostic suspicion was a lecithin-cholesterol acyltransferase deficiency, as corneal opacities are classically associated with this condition, the partial form of which is known as *fish-eye disease*. A genetic study was performed, which found a homozygous mutation in the apoA-1 gene associated with primary hypoalphalipoproteinemia (NM_000039.3(APOA1):c.67C >T (p.Gln23*)). For an accurate diagnosis, genetic testing is essential. Currently, management consists of controlling low-density lipoprotein cholesterol levels, and the prognosis is determined by premature onset of atherosclerotic disease.

FUNDING

No funding was received for the writing and publication of this work.

AUTHORS' CONTRIBUTIONS

J. Costas Eimil: writing and design. P. Sánchez-Sobrino: review and approval.

CONFLICTS OF INTEREST

The authors declare no conflicts of interest.

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Available online 18 January 2023

https://doi.org/10.1016/j.rec.2022.11.012