Multimodal imaging evaluation in Lipemia Retinalis: A Case Report

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Introduction

Severe hypertriglyceridemia is defined as elevated levels of lipids in the blood (>/2000mg/dL) and is a result of primary (genetic) causes or a combination of secondary conditions, such as diabetes mellitus. (1). Ocular findings of hypertriglyceridemia include xanthelasma, iris xanthomas and lipemia retinalis. (1). The last one is characterized by creamy-white retinal vessels, salmon colored retina, and lipid infiltration and, in most cases, visual acuity is not affected. (2,3). Lipid-lowering therapy leads to normalization of fundus appearance and restoration of visual acuity (1).

Case Report

An 18-year-old woman followed at the endocrinology department due to hypertriglyceridemia since she was 3 months old, was referred for ophthalmological examination. Visual acuity was 20/20 in both eyes (OU), biomicroscopic examination of the anterior segment of OU was normal, whereas fundus examination revealed bilateral creamy-white retinal vessels and a salmon-pink retina in OU. Multimodal evaluation was performed and revealed, in optical coherence tomography (OCT), the presence of highly reflective and engorged retinal superficial capillaries and hyperreflective dots in the inner retinal layers; Fundus Autofluorescence, OCT Angiography (OCT-A) and Fluorescein Angiography were normal in OU. Laboratory exams were performed and revealed a triglyceride level of > 1505 mg/dL (normal <90mg/dL), with lipemic venous sample. The patient was referred for genetic testing, which detected a homozygous mutation in the LPL gene, confirming a lipoprotein lipase deficiency. She continues to be monitored by the endocrinology department for management of serum triglycerides.

Discussion

Lipemia retinalis is asymptomatic, with preserved visual acuity in most cases, and occurs when the plasma triglycerides levels are near to 2000 to 2500 mg/dL or more. (3). The classic appearance of cream-colored retinal vessels occurs due to scattering of light by the triglyceride-laden chylomicrons and this resolves when serum lipid levels are reduced. (2). Deficiency of lipoprotein lipase is one of the primary causes of hypertriglyceridemia and is an autosomal recessive disease with an incidence of less than 1:1.000.000. (3). The consequent increase in systemic triglycerides have been shown to contribute to an increased risk for future myocardial infarction, acute coronary syndrome, and strokes events, and systemic treatment is recommended. (1). This includes restriction of fat in the diet and elevated intake of protein and carbohydrate. Usually, no treatment is required for the lipemia retinalis itself. (4).

A complete retinal evaluation of these patients includes performing multimodal imaging, in which OCT can highlight different findings, such as engorged and highly reflective superficial retinal vessels shadowing the underlying structures (because of the higher density of contents flowing through them) (5) and hyperreflective dots, that seems to represent extravasated lipids. (2). Furthermore, changes in the electroretinogram of these patients have already been described, with decreased rod and cone amplitudes associated with lipemia retinalis, with resolution after systemic treatment. (6). Most recently, Alsarhani et al. described OCT-A findings in patients with lipemia retinalis, which demonstrated intact superficial and deep retinal capillary plexus. (7).

These patients must be monitored in a multidisciplinary manner for better systemic management.

Figures

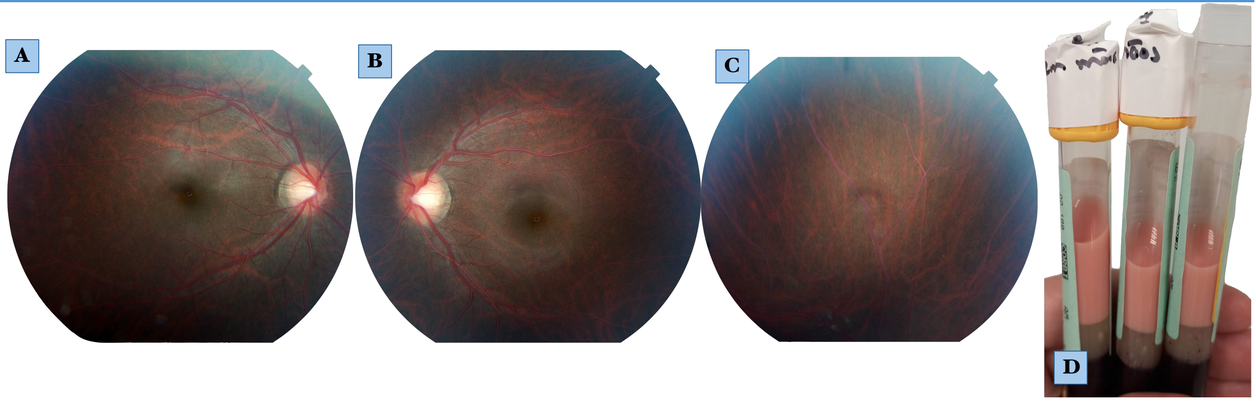

Figure 1 – A – C: Color fundus photograph of OU revealing bilateral creamy-white retinal vessels.
D: Lipemic venous sample.

Figures

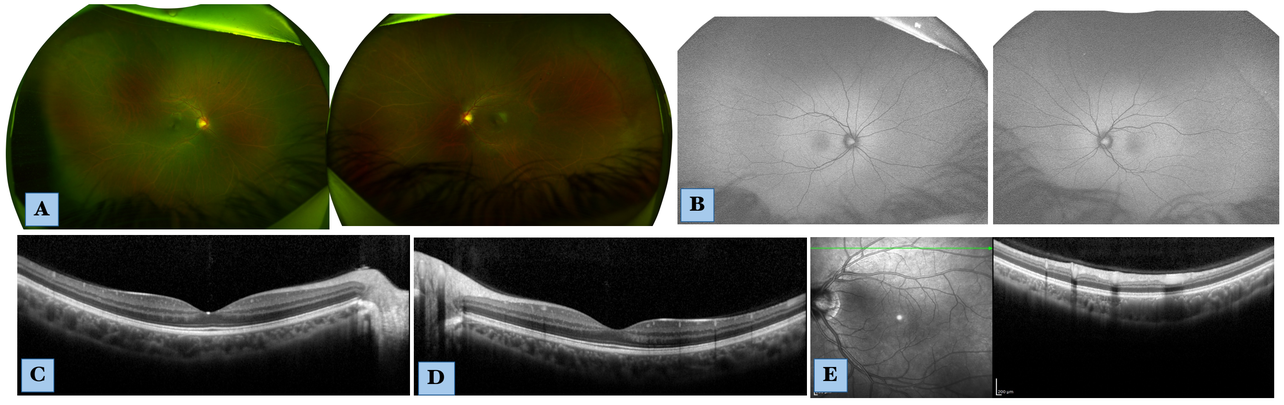

Figure 2 – A: Color fundus widefield imaging of OU revealing bilateral creamy-white retinal vessels.
B: Fundus autofluorescence widefield without alterations.
C , D: Macular **OCT** of OU showing hyperreflective dots in inner retina.
E: OCT in superior temporal arcade revealing highly reflective and engorged retinal vessels.

Figure 3: Fluorescein angiography of OD (A;C) and OS (B;D) ; without alterations in early (A; B) and late phases. (C; D)

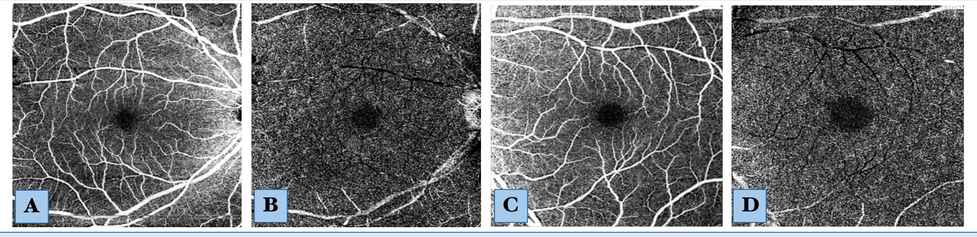

Figure 4 – A –J: OCT-A of OD (A;B) and OS (C;D) without alterations in Superficial capillary plexus (A;C) and Deep Capillary Plexus (B;D)

References

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