Lipaemia retinalis – A case report on a life-saving timely ocular diagnosis

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Case report

Introduction

Lipemia retinalis (LR) is a clinical diagnosis where patient presents with milky white retinal vessels. It results from hypertriglyceridaemia of more than 1000mg/dl. Though asymptomatic initially, it can be associated with life threatening complications if undiagnosed. This case report describes incidental diagnosis of a LR patient who subsequently recovered following severe metabolic disorders.

Case report

A 36 year old Caucasian male was referred to the Emergency Department by the optician who noticed white retinal vessels as an incidental finding at a routine sight test. Patient had no visual symptoms. He declined significant past medical or family history of vascular diseases. There was no history of ocular trauma or surgery. His visual acuities were of 6/5 in each eye. The intraocular pressures were normal. His colour vision tested on the Ishihara chart was normal. Dilated fundoscopy revealed bilateral milky white retinal vessels with no evidence of haemorrhages or exudates. There was no optic disc swelling. His systemic examination was unremarkable.

![Fundal photos of both eyes showing characteristic whitish retinal vessels.](image1)

![OCT Macula scan showing engorged retinal vessels and intraluminal hyper-reflective materials.](image2)

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With a clinical diagnosis of lipemia retinalis, urgent hematological investigations (full blood count (FBC), erythrocyte sedimentation rate (ESR), C-reactive protein (CRP) level, lipid profile, random blood sugar level (RBS), liver and renal function test) were done.

While awaiting the test results, the next day the patient developed symptoms and signs of an acute abdomen and was immediately admitted to the medical unit. His hematological results confirmed markedly elevated triglyceride levels of 4425mg/dL with evidence of septicemia (CRP - 140mg/l) and hyperglycaemia (RBS - 413mg/dl). His FBC reported as lipaemic sample and required a repeat assessment. Ultrasound scan confirmed acute pancreatitis with ascites.

Patient was started on intravenous antibiotics, insulin and oral fenofibrate. He recovered from the episode of acute abdomen following 5 days of inward treatment. Ophthalmological review was planned for 3 months after first presentation, with repeat dilated fundoscopy and OCT Macula which were normal. Genetic analysis revealed mutation of lipoprotein lipase gene coding, which was the cause for his LR.

**Discussion and conclusion**

LR is a rare oculo-metabolic condition which is associated with homozygous lipoprotein lipase gene mutation. Historically a case of lipemia retinalis has been described by Heyl et al, in 1880.

The triglycerides in the food, when absorbed through the intestines, are commonly transported from intestine to the systemic circulation as chylomicrons where they are rapidly removed from the plasma under normal conditions. Chylomicrons are the largest lipoprotein macromolecules when compared with other types of lipoproteins such as high density lipoproteins (HDL), low density lipoproteins (LDL) and very low density lipoproteins(VLDL). Hypertriglyceridaemia occurs when there is genetic defect in the metabolism of chylomicrons, resulting in the abnormal accumulation of triglycerides.

The chylomicronaemia syndrome occurs when the serum triglyceride levels exceed 1000 mg/dl, and it is characterized by eruptive xanthoma, lipemia retinalis, and abdominal pain and/or pancreatitis.

Above mentioned lipid metabolism is governed by genetically inherited enzymes and hence the enzymatic assay of suspected patients can reveal the underlying genetic defect as revealed by this case.

The literature on symptom analysis of LR cases, highlights that except for few published cases, most of the patients have no visual symptoms and lipemia retinalis is commonly detected incidentally, during a routine ophthalmic workup. Hence visual acuity remains normal in such patients, but the associated severe but easily treatable metabolic status and ocular complications needs early attention. In our case too, the patient didn’t have any visual impairment, however some published cases of LR have presented with reduced visual acuity due to inner retinal ischemia or the development of retina vein occlusion.

Ocular examination revealing the creamy-white appearance of retinal vessels is due to scattering of light by the triglyceride-laden chylomicrons in plasma. At early stages it starts in the peripheral vessels and when the disease severity increases it involves central retinal vessels. Characteristic salmon color of the retina is seen in severe cases.

Systemic examination of LR patients should include examination of the cardiovascular system for murmurs, carotid or renal bruits, and peripheral pulses. Xanthomas can be seen on buttocks and elbows which suggest deposition of excessive chylomicrons. Hepatosplenomegally should be examined to rule out possible pancreatic disease.

When investigated with optical coherence tomography (OCT) scan through the affected vessels in the retina, it highlights the engorged retinal vessels, as well as back shadowing due to the presence of intraluminal hyper-reflective materials in the vessels of the inner nuclear and ganglion cell layer. This was similarly seen in the OCT scan done on the case described.

Haematological assessment with fasting lipid profile is needed to diagnose hypertriglyceridaemia. The blood specimen will appear lipaemic, with a creamy white lipid layer in a standing tube of blood, which was similarly seen in our patient.

Although different treatment options are published in the literature, a prompt reversal of hyperlipidemia is the key to a better prognosis. A low-fat diet without breast milk supplementation is said to be effective in treating infants with severe LR. Correction of the lipid levels may reverse the abnormal findings within one week. Lipid-lowering drugs (Fibrates) are the key medical treatment to reduce triglycerides. In addition as glycemic control is affected in LR patients, it should be controlled with insulin accordingly. Although surgical options are rarely needed Capitena et al revealed a case of rapid resolution of infantile LR following exchange transfusion.
The differential diagnosis for lipemia retinalis includes leukemia, advanced diabetic eye disease with sclerotic vessels, intravascular calcification of retinal vessels secondary to renal failure and hyperparathyroidism, retinal branch retinal artery or vein occlusion and hypertensive retinopathy. In leukemia, the veins appear reddish pale, while the arteries are pale yellow. In lipemia, arteries and veins are of the same color and can be differentiated only by caliber as the veins are of a larger caliber. Renal failure and hyperparathyroidism, can be diagnosed by the relevant hematological tests. Branch vessel occlusion and hypertensive retinopathy show other associated ocular features.

As this is an inherited condition, patients present in their early years of life with systemic pathology, such as hepatosplenomegaly, acute pancreatitis, acute myocardial infarction and stroke13. As mentioned above, both life threatening (stroke/ischaemic heart diseases) and vision threatening (retinal vein occlusions) complications are associated with LR, early diagnosis and treatment are essential to prevent such complications. Hence this patient had an early clinical diagnosis and urgent treatment was started to prevent the life and vision threatening complications.

References