

## Lipemia retinalis in a child: an overview of disease and a case report

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**Abstract:** Lipemia retinalis is a rarely described condition which occurs in certain types of both primary and secondary hyperlipidemia. We report a case of six month old female child of lipemia retinalis with ocular manifestations. In this presentation we have included an overview of clinical manifestation and treatment plan.

**Keywords:** Lipemia retinalis, Hyperlipidemia, Chylomicrons.

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### INTRODUCTION

Lipemia retinalis was first described by Hexyl in 1880 [1]. It is a rare condition seen with primary as well as secondary hyperlipidemia [2]. The principal fundal abnormality in lipemia retinalis is a milky discolouration of retinal vessels and retinal vessels give fluorescent hue on fundoscopic examination. These findings can vary in degree from barely detectable peripheral vessel changes to cream colouration of all retinal vessels with a 'salmon' -coloured fundus. We present a case of lipaemia retinalis of a child and its management.

### CASE HISTORY

Six month old female baby weighing 4.6 kg was admitted in Paediatric ward with complaints of not gaining weight, fever and vomiting for past 3 days. Child was the first baby of the couple with no history of miscarriage or foetal death of sibling death, borne at term by normal vaginal delivery at a nursing home with birth weight of 3.2 kg with uneventful antenatal natal and postnatal periods. Mother did not have any specific illness throughout the pregnancy. The child was on exclusively breast feeding since birth. There was no significant family history of any chronic illness, recurrent pain abdomen or unexplained death.

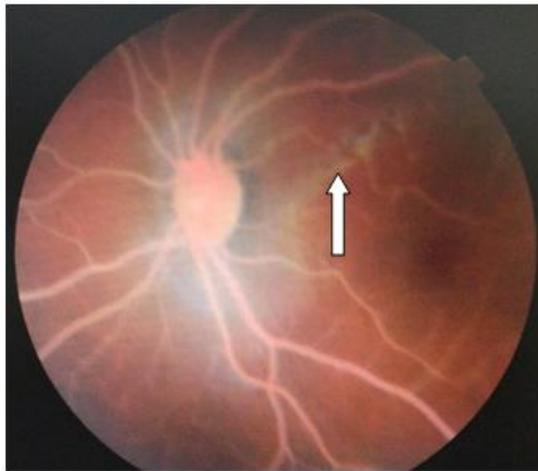
On general physical examination the child was active and had hepato-splenomegaly without dysmorphic features. The child has normal liver and kidney function test with sterile urine and blood cultures. Ultrasonography showed hepato-splenomegaly with normal liver texture. During sampling, her blood was found to be highly viscous pink in colour and milky. The child was referred to Eye OPD for fundus examination. On Ophthalmic examination Child was following light, Pupillary reaction was normal, pupils were isochoric, ocular motility was full, corneas were transparent, anterior segment examination was normal.

Fundus examination revealed that vessels in the posterior pole and peripheral area of each eye had a creamy fluorescent appearance. Both arterioles and veins were flat and ribbon like and it was difficult to differentiate retinal arterioles from veins. Optic disc and macula were normal. Lipid exudates were also seen. This was typical fundal picture of lipaemia retinalis. Both mother and father had serum triglyceride levels mildly deranged (210 mg/dl and 175 mg/dl respectively), with all other parameters within normal range.

Treatment of child was initiated with MCT oil and special fat free diet was advised to restrict fat intake to less than 15-20% of daily diet with avoidance of saturated fats. Lipid lowering agent fenofibrate was started in the dose of 3 mg/kg/day. Child was reviewed after 2 wks for repeating lipid profile. Results of lipid profile and fundoscopic findings have shown marginal improvement. Child is now on regular follow-up.



**Fig-1: White arrow showing whitish colour retinal vessels**



**Fig-2: Arrow showing Lipid infiltration in Retina**

#### DISCUSSION:

The incidence of lipaemia retinalis correlates approximately with the level of plasma triglycerides. However, the relationship is indirect because the key factor is the level of chylomicrons (the large lipoproteins) which serve to transport triglycerides. To a lesser extent slightly smaller VLDL (very low density lipoprotein) macromolecules are also involved. The fundal appearance is believed to be caused by the visualisation of increased levels of chylomicrons in the circulating retinal blood. However, not all patients with even grossly elevated chylomicrons or triglyceride levels show evidence of lipaemia retinalis, demonstrating that other factors, such as haematocrit and difference in translucency of the retinal and choroidal vessels, are important.

The variation of the aspect in the fundus has been graded:

- Grade I (early) - white and creamy aspect of the peripheral retina vessels;
- Grade II (moderate) - the creamy colour of the vessels extends towards the optic disc;
- Grade III (marked) - the retina appears with a salmon colour and all vessels arteries and veins present a milky aspect [2, 3].

The retina's white milky aspect occurs when the plasma levels are near to 2000 to 2500 mg/dL or more [1, 2]. Vinger *et al.*; [4] believed that the changes in the fundus happen in consequence of the elevated levels of circulating chylomicrons at the retinal vessels however, Winder AF [5] suggested that these high levels are not sufficient to cause the lipaemic creamy serum. Some authors [6] justified that in some cases with elevated levels of chylomicrons and triglycerides lipaemia retinalis was not present suggesting that changes in haematocrit and in the translucence of the Retinal and choroid vessels should be considered.

We started fat free diet because it has been observed that once the hyperlipidemia has been

detected and the fat in the diet has been restricted, the clinical findings can be reversed to normal. The deficiency of lipoprotein lipase is rare in hyper-lipo proteinemia, and is characterized by the high levels of chylomicrons in serum. It is an autosomal recessive disease with an incidence of less than 1:1.000.000. Lipoprotein lipase is present in the vascular endothelium and breaks the chylomicrons into glycerol and free fatty acids; the deficiency of lipoprotein lipase causes the accumulation of chylomicrons [7].

The predetermined treatments for these cases are restriction of fat in the diet (less than 10 g/day) and elevated intake of protein and carbohydrate.

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