



## CREAMY WHITE RETINAL VESSELS IN AN INFANT: LIPEMIA RETINALIS AS A CLUE TO UNDERLYING METABOLIC DISORDER

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### ABSTRACT

Lipemia retinalis is a rare ophthalmic manifestation characterized by creamy discoloration of retinal vessels, typically seen with extreme hypertriglyceridemia. It may occur in both primary and secondary hyperlipidemias, with familial chylomicronemia and familial combined hyperlipidemia being the most common primary causes. This is a case report of a 4-month-old female infant referred for ophthalmic evaluation after incidental detection of pale, creamy retinal vessels during a routine examination. Fundoscopic evaluation revealed grade III lipemia retinalis, correlating with markedly elevated serum triglyceride (4,200 mg/dL) and total cholesterol (750 mg/dL) levels. This case highlights the importance of recognizing lipemia retinalis as an early indicator of potentially life-threatening lipid disorders in infancy, facilitating timely intervention to prevent complications such as pancreatitis and cardiovascular disease.

**KEYWORDS:** Lipemia Retinalis, Hypertriglyceridemia, Infant, Case Report

Lipemia retinalis is an uncommon but distinctive ocular sign of marked hypertriglyceridemia. First described by Heyl in 1880, it is frequently associated with familial hyperchylomicronemia and may also be seen in familial combined hyperlipidemia, which has a prevalence of 1–2% in the general population. It is characterized by a creamy discoloration of retinal blood vessels, ranging from a subtle salmon-pink to a milky white appearance, correlating with triglyceride concentration. Fundoscopic changes usually become apparent when plasma triglycerides exceed 2,500 mg/dL. The condition itself does not impair vision, but its presence often indicates an underlying severe lipid metabolism disorder requiring urgent intervention. (Shinkre and Usgaonkar, 2019; Jain *et al.*, 2017)

Hyperlipidemias are broadly classified as primary (genetic) or secondary (acquired). Primary hyperlipidemias include disorders such as familial chylomicronemia, familial combined hyperlipidemia, and familial hypercholesterolemia. Secondary causes are related to lifestyle and systemic diseases such as diabetes mellitus, hypothyroidism, liver or kidney disease, and chronic alcohol abuse.

Lipemia retinalis typically begins with involvement of the peripheral retinal vessels and progresses toward the posterior pole. The grading system by Vinger *et al.* classifies the condition as:

- Grade I: Creamy white appearance of peripheral retinal vessels

- Grade II: Creamy discoloration extending toward the optic disc
- Grade III: Diffuse milky retinal vessels with salmon-pink background

Review of published literature indicates that lipemia retinalis in neonates and young infants is rare, with only a handful of cases reported. In some instances, detection has been incidental during retinopathy of prematurity screening. The youngest reported cases include a 3-day-old female with GPIHBP1 mutation and a 27-day-old male with familial chylomicronemia. (Chaudhury *et al.*, 2015; Vinger and Sachs, 1970; Zahavi *et al.*, 2013)

Although lipemia retinalis is visually striking, it often goes unnoticed in routine pediatric assessments unless a fundoscopic examination is performed. In neonates and infants, this finding can be the first clinical clue to a serious metabolic disorder. Failure to detect and treat severe hypertriglyceridemia early can result in pancreatitis, hepatic dysfunction, and premature cardiovascular disease. (Wani *et al.*, 2015; Jain *et al.*, 2017; Hegele *et al.*, 2018)

### Methodology Approach

This case report compiled following:

1. Clinical Evaluation: Routine paediatric examination followed by detailed ophthalmic assessment using indirect ophthalmoscopy.

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2. Laboratory Investigations: lipid profile, liver and renal function tests, and genetic lipid disorder screening.
3. Grading: Fundus changes were graded as per Vinger *et al.*'s classification.
4. Management: Dietary fat restriction and parental counselling.
5. Follow-Up: Lipid levels and fundus appearance monitored at six weeks post-intervention.

## RESULTS

### Findings

- Fundus: Grade III lipemia retinalis in both eyes (figure 1).
- Laboratory: Triglycerides 4,200 mg/dL; Total cholesterol 750 mg/dL; HDL 12 mg/dL.
- Diagnosis: Likely familial combined hyperlipidemia.



**Figure 1: Lipemia retinalis**

### Management Outcome

After six weeks of dietary modification, triglycerides reduced to 600 mg/dL, and retinal changes regressed centrally with improvement in vessel color.

### DISCUSSION

Lipemia retinalis is a pathognomonic sign of marked hypertriglyceridemia, with the degree of vascular discoloration correlating to triglyceride levels. In infants, early diagnosis is critical to preventing systemic complications. This case is notable for the unusually high lipid levels and severe (Grade III) retinal changes at such a young age.

### CONCLUSION

Persistent and uncontrolled hypercholesterolemia and hypertriglyceridemia in infancy can cause life-threatening complications, including premature atherosclerosis, pancreatitis, and hepatic failure. Recognition of lipemia retinalis during fundus examination provides a crucial diagnostic opportunity for early intervention and improved prognosis.

### Future Scope

- Routine fundus screening in neonates with high lipid profiles.
- Genetic testing for early identification of at-risk infants.

- Long-term studies to assess outcomes of early dietary intervention in familial lipid disorders.
- Development of neonatal-specific lipid-lowering therapies with proven safety profiles.

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