Milky Blood in an Infant

Marta P. Soares[1], Diogo Rodrigues[2], Raquel Ferreira[2], Sílvia Sequeira[1]

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A 36-day-old previously healthy boy, born after an uneventful pregnancy and vaginal delivery, was admitted with a history of respiratory distress after a four-day course of diarrhea. The clinical examination showed signs of sepsis (grunting, fever, poor perfusion, tachycardia, tachypnea, subcostal retractions) and hepatosplenomegaly. During sampling, his blood, serum and plasma were found to be milky (Figs. 1 and 2). Laboratory investigation showed 2.26 x 10^{12} red blood cells/L, hematocrit 19.1%, leukocytosis (13.9 x 10^{9} cells/L) and neutrophilia (neutrophils 87%), severe hypertriglyceridemia (24 129 mg/dL) and hypercholesterolemia (1208 mg/dL). Other laboratory tests such as hemoglobin, C-reactive protein, lipase and amylase levels were unobtainable due to lipemic samples.

Initially, sepsis was assumed and confirmed by blood culture which revealed *Citrobacter braaki.*

His management included fasting, fluid therapy, a red blood cells transfusion, a partial exchange transfusion and an antibiotic course of eight days, with clinical and laboratory improvement. On day five the child seemed well, and blood tests revealed both a total cholesterol of 281 mg/dL and triglycerides of 707 mg/dL. Oral feeding was started with a low-fat polymeric diet showing continuous improvement.

Ultrasonography showed hepatomegaly with hepatic and pancreatic steatosis, and ophthalmologic examination revealed *lipemia retinalis* (white colored retinal vessels). Molecular studies revealed a homozygous pathogenic variant in the *LPL* gene confirming the suspicion of a lipoprotein lipase deficiency (OMIM#238600).

The lipoprotein lipase deficiency is an ultra-rare autosomal recessive disorder and is characterized by severe hypertriglyceridemia, recurrent acute pancreatitis, cutaneous xanthomata, hepatosplenomegaly and *lipemia retinalis*.[1-3,5] It can also cause pancreatic abscess and pancreatic necrosis.[1-4]

Severe hypertriglyceridemia manifests itself as milky blood / serum / plasma as described in this patient.[1]

Figure 1. Blood samples on day one.

Figure 2. Blood and serum samples.

Keywords: Infant; Hyperlipoproteinemia Type I; Hypertriglyceridemia/diagnosis; Hypertriglyceridemia/therapy; Lipoprotein Lipase/deficiency; Rare Diseases

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

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1. Metabolic Unit, Hospital de Dona Estefânia, Lisbon, Portugal
2. Pediatric Intensive Care Unit, Hospital de Dona Estefânia, Lisbon, Portugal
Corresponding Author
Sílvia Sequeira
silviasasequiera@gmail.com
Hospital de Dona Estefânia, Rua Jacinta Marto, 1169-045 Lisboa, Portugal
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Consent for publication

Consent for publication was obtained.

Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

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WHAT THIS REPORT ADDS

- Milky blood / plasma is usually a manifestation of chylomicronaemia / severe hypertriglyceridemia and may be associated to a primary monogenic disorder.
- Treatment may be essentially a dietary fat restriction as in our case report.
- We emphasize the importance of a quick diagnosis and management in order to improve the outcome.

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