

## Case Report

# A neonatal hypertriglyceridemia presenting with respiratory distress: a rare case report

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### ABSTRACT

Neonatal hypertriglyceridemia is a very rare condition. Diagnosis in neonatal period is very difficult and is usually diagnosed when acute pancreatitis sets in. Early diagnosis is important as it can prevent the complications associated with the condition that is acute pancreatitis and pancreatic necrosis. Here we present a case of neonatal hypertriglyceridemia who presented to us with respiratory distress but was diagnosed early due to the presence of highly viscous and milky blood. This holds importance as early treatment can reduce the complications and morbidity associated with familial hypertriglyceridemia.

**Keywords:** Hypertriglyceridemia, Milky blood, Neonatal

### INTRODUCTION

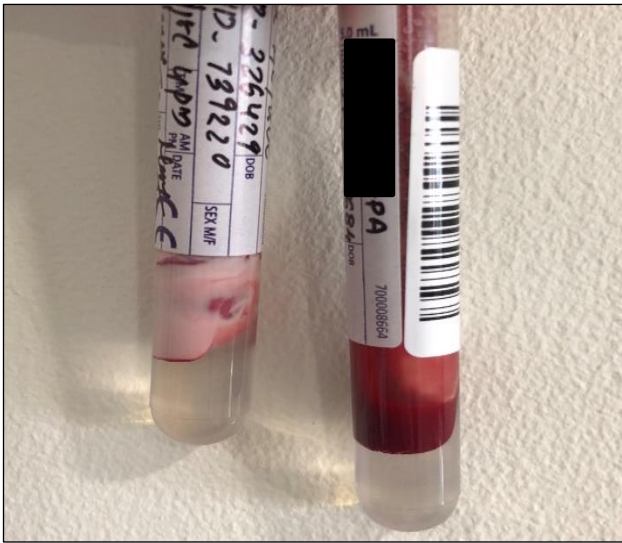
Familial hypertriglyceridemia (FH) is a very rare condition occurring in around 1 % of population.<sup>1</sup> Plasma triglyceride levels are a measure of triglyceride rich lipoproteins and their metabolic remnants which are derived either from diet or produced by liver. Hypertriglyceridemia is defined as plasma triglycerides (Tg) concentration above 150 mg/ dL in a fasting state, while severe hypertriglyceridemia (HTg) is defined as a level of Tg above 885 mg/dL, or according to some authors, above 500 mg/dL.<sup>2,3</sup> According to Fredrickson's classification of hyperlipoproteinemia, the only 2 types that supports the presence of chylomicrons in plasma are type I and V. The differences between type I and V consist in the level of cholesterol and frequency. Type I hypertriglyceridemia is defined by the presence of chylomicrons, normal level of total cholesterol, very high level of Tg, and a relative frequency below 1%, whereas type V hypertriglyceridemia is characterized by the presence of chylomicrons and very low-density

lipoproteins, an increased level of cholesterol with an elevated level of Tg, and relative frequency slightly higher, about 5%.<sup>4</sup> Lipid disorders can occur as a primary event or secondary to the underlying disease. These children usually present with xanthomas, which may cause thickening of the Achilles tendon or extensor tendons of the hands, or cutaneous lesions on the hands, elbows, buttocks, or knees.<sup>5</sup> Here we present a case of neonatal hypertriglyceridemia who presented with pyrexia and respiratory distress.

### CASE REPORT

A 28 days old male baby, delivered as a late preterm to a primigravida mother by LSCS with uneventful antenatal and postnatal events, was admitted to NICU with respiratory distress and suspect sepsis. The child was exclusively breastfed. During sampling the blood was milky and highly viscous (as shown in Figure 1). The child had hepatosplenomegaly. The child had raised hemoglobin with reduced hematocrit. Normal liver and

kidney function test with sterile blood and urine culture were reported. Ophthalmoscopic examination reported lipemia retinalis. Echocardiography was reported normal.



**Figure 1: Sample showing milky blood compared with control.**

Serum analysis was done for LDL, VLDL and HDL by electrophoresis, TG and S. cholesterol by spectrophotometry, Apo A and B by immunoturbidimetric technique. Results were obtained as S. triglyceride 40,000 mg/dl (<150 mg/dl), VLDL 8000 mg/dl (<30 mg/dl), S. cholesterol 70 mg/dl (<170 mg/dl), HDL 8 mg/dl (40-60 mg/dl), LDL 52 mg/dl (<110 mg/dl), other investigations were Apo A (< 40 mg/dl), Apo B (< 40 mg/dl), CRP was highly reactive 120 mg/l (<5 mg/l). Complete blood counts revealed Hb of 30.4 g/dl with PCV of 20.2 %, S. lipase 231 U/L (22-51 U/L), T3 3.53 pg/ml (2.6-4.2 pg/ml), fT4 0.93 ng/ml (0.58-1.64 ng/ml), TSH 2.01 uIU/ml (<10 uIU/ml)

The child was started on lipid lowering agents Gemfibrozil, a fibric acid derivative, fat soluble vitamins, medium chain triglycerides, docosahexaenoic acid.

Blood sampling done after 2 weeks for lipid profile showed blood red in colour and less milky. Lipid profile revealed S. triglyceride 11,439 mg/dl (< 150 mg/dl), VLDL 2400 mg/dl (<30 mg/dl). Blood samples have been sent for clinical exome sequencing to a genetic laboratory. Results are awaited. Both parents were screened for familial hyperlipidemia reported triglyceride levels in higher normal range.

## DISCUSSION

Hypertriglyceridemia is defined as plasma triglyceride above the 95<sup>th</sup> percentile for age and sex.<sup>6</sup> It is a rare disorder in infancy. Serum triglyceride values above 1000 mg/dl occurs in 1 in 5000 persons. The serum is opalescent in these cases due to increase in VLDL. Lipid

disorders occurs either as primary event or secondary to an underlying disease.

Primary hypertriglyceridemia is the result of various genetic defects leading to disordered triglyceride metabolism. The familial disorders of triglyceride-rich lipoproteins include both common and rare variants of the Frederickson classification system.

These include chylomicronemia (type I), familial hypertriglyceridemia (type IV), and the more severe combined hypertriglyceridemia and chylomicronemia (type V). Hepatic lipase deficiency also results in a similar combined hyperlipidemia.<sup>1</sup> Familial Chylomicronemia (Type I Hyperlipidemia) is an autosomal recessive rare single-gene defect due to by mutations affecting clearance of apoB lipoproteins occurring approximately 1 in 1,000,000.

The disease usually presents during childhood with acute pancreatitis. Familial Hypertriglyceridemia (Type IV Hyperlipidemia), an autosomal dominant disorder occurring in approximately 1 in 500 individuals is characterized by elevation of plasma triglycerides >90th percentile (250-1,000 mg/dL range), often accompanied by slight elevation in plasma cholesterol and low HDL. Hypertriglyceridemia Type V, more severe characterized by increased levels of chylomicrons as well as VLDL particles (Frederickson type V) have Triglyceride levels are often >1,000 mg/dL.

These patients often develop eruptive xanthomas in adulthood, whereas type IV hypertriglyceridemia individuals do not. The most effective treatment modality is dietary triglyceride restriction by restriction of fat, sugars and carbohydrates. Niacin in hypertriglyceridemia is not recommended in infants.

According to the National Cholesterol Education Program (NCEP), normal triglyceride level is 150 mg/dl (1.7 mmol/l).<sup>7</sup> Early diagnosis is important to prevent further complications that is. atherosclerosis, pancreatitis and pancreatic necrosis, although pancreatic function deteriorates very slowly.<sup>8,9</sup> A study by Brunzell, reported that triglyceride induced pancreatitis occurred at plasma levels > 2000 mg/dl. Recommended triglyceride levels < 1000 mg/dl can be used as a threshold.<sup>10</sup>

In present study we suspected the case as hypertriglyceridemia type V in view of reported highly raised triglyceride levels and VLDL levels and low HDL levels. Low hematocrit levels were observed with high triglyceride levels and hematocrit levels improved with decreasing triglyceride levels. Lipid lowering drugs and fat-soluble vitamins improve lipid parameters and with a multidisciplinary approach we can prevent neonatal mortality.

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## REFERENCES

1. Boman H, Hazzard WR, Albers JJ, Cooper MN, Motulsky AG. Frequency of monogenic forms of hyperlipidemia in a normal population. *Am J Hum Genet* 1975;27(6):A19.
2. Hegele RA, Ginsberg HN, Chapman MJ, Nordestgaard BG, Kuivenhoven JA, Averna M, et al. The polygenic nature of hypertriglyceridemia: implications for definition, diagnosis, and management. *Lancet Diabetes Endocrinol.* 2014;2:655-66.
3. Pejic RN, Lee DT. Hypertriglyceridemia. *JABFM.* 2006;19:310-6.
4. Fredrickson DS, Lees RS. A system for phenotyping hypolipoproteinemia. *Circ.* 1965;31:321-7.
5. Kliegman R, Stanton B, Geme JW, Schor NF, Behrman RE. *Nelson textbook of pediatrics.* 20<sup>th</sup> Edition. Philadelphia, PA: Elsevier; 2016.
6. Brunzell JD, Miller NE, Alaupovic P. Familial chylomicronemia due to a circulating inhibitor of lipoprotein lipase activity. *J Lipid Res.* 1983;24(1):12-9.
7. Foil-Fonseca JC, Levy E, Godard M, Lambert M. Familial lipoprotein lipase deficiency in infancy: clinical, biochemical, and molecular study. *J Pediatr.* 1998;133(3):417-23.
8. Yuan G, Al-Shali KJ, Hegele RA. Hypertriglyceridemia: its etiology, effects and treatment. *Can Med Assoc J.* 2007;176(8):1113-20.
9. Labossiere R, Goldberg IJ. Management of hypertriglyceridemia. *Therapeutic Lipidol.* Totowa: Humana Press; 2008:201-20.
10. Brunzell JD, Bierman EL. Chylomicronemia syndrome. *Med Clin North Am.* 1982;66:455-63.

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