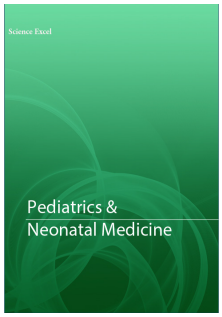
Pediatrics and Neonatal Medicine

Case Report




# A Case of "Milky Blood". Presentation of a Puerto Rican Infant with Severe Hyperlipidemia

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**Abstract**

Familial Chylomicronemia's incidence is one in a million which would explain why it has been rarely described in the literature as initially presenting in infants. In fact, this is one of the few reported cases, and the first reported in a Puerto Rican infant. The Incidental finding of lipemic "milky" blood led to the discovery and consequent manifestations of disease that followed.

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Infants, Familial chylomicronemia syndrome, Hyperlipidemia.



## Introduction

Familial Chylomicronemia syndrome is a rare autosomal recessive disorder with an incidence of one in a million. It leads to increased levels of triglycerides, cholesterol and chylomicrons causing blood to appear lipemic, which is usually an incidental finding in asymptomatic patients. There are only a few cases described in literature as presenting in infants. We report the first Puerto Rican infant with this finding of lipemic blood in whom we suspected the disorder following the symptoms that manifested with this syndrome..

## Case Report

This is the case of a 1-month old female infant who had an incidental finding of Hypertriglyceridemia and Hypercholesterolemia after her primary care physician ordered a laboratory work up due to an upper respiratory tract infection. The blood sample was found to be highly lipemic for which lipid panel was ordered revealing triglyceride levels at 15,125 mg/dL and cholesterol at 929 mg/dL. Patient's previous history includes 39 weeks of gestation without any significant prenatal complications, who soon after birth presented with meconium plug syndrome and neonatal sepsis as well as persistent vomiting after feedings with abdominal distention which required partially hydrolyzed formula. She received TPN (Total Parenteral Nutrition) (Repak 20% Lipids) for four days during the perinatal period with adequate evolution and discharge home.

The patient was subsequently hospitalized due to the hyperlipidemia and admitted to the pediatric intensive care unit where she was evaluated and managed by an endocrinologist who recommended treatment with fibrate; the geneticist suggested a diet with medium chain triglycerides, vitamins A, D, E and K, and carnitine supplementation. Ophthalmological evaluation found Lipemia Retinalis. Cardiologist evaluation was remarkable for Peripheral Pulmonic Stenosis. Patient was transfused with PRBC's due to symptomatic anemia and evaluated with lipids electrophoresis, Mutation Panel for Lipoprotein Lipase and discharged home after four days. Outpatient treatment included: Gemfibrozil, diet with Pregestimil 20, hematinics, folic acid and levocarnitine. At 4 months of age the patient developed skin lesions that resembled xanthomas (Figure 1 and 2), distributed over her arms, legs, face, and abdomen that were complicated with an infection for which she was hospitalized and treated with Ampicillin and Cleocin. At 6 months of age she was admitted to hospital due to acute pancreatitis, lipase was found to be 391 U/L, amylase was 30 U/L, Triglycerides 3,028, and Cholesterol 198 (Table 1).

Her development has been age appropriate. Height, weight, head circumference and BMI have always trended within normal limits. Parents describe her as an active and happy infant with good appetite and sleeping patterns, and who "does not appear to be ill."

In her most recent Well Child appointment she appeared with mild gross motor delay and not yet able to sit on her own or crawl. She was found to have truncal hypertonicity that causes her to lean back when seated. Physiatrist evaluation was recommended.

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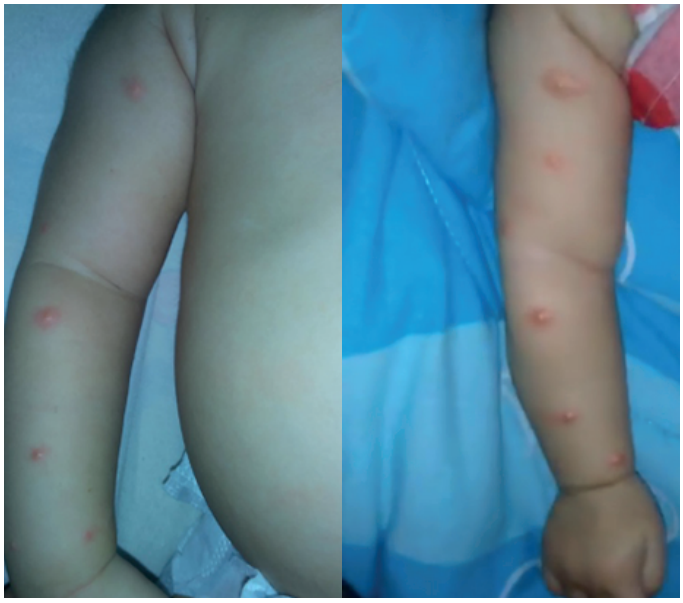

***Figure 1.*** *Progression of skin lesions found on patient's right arm. They first appeared as papules and some later developed into yellowish plaques.*

She has no allergies or sensitivities to medications, food, environmental factors or latex. Her immunizations are up to date.

Father's medical history is remarkable for liver problems, and episodes of pancreatitis. Mother and siblings have no relevant conditions, neither does her extended family. Patient is undergoing genetic counseling in the hopes of getting the appropriate testing to confirm the suspected diagnosis.

Patients current treatment regimen consists of Vitamin D 1,000 units daily, Vitamin B-12 1mg daily, Folic Acid 1mg daily, Gemfibrozil 20mg/kg daily, Atorvastatin 5mg daily, and Levocarnitin 2 ml PO BID (50mg/kg/day). Diet includes a formula rich in medium chain triglycerides (Pregestimil 20) and orange and green vegetables (Gerber 2nd foods).

## Discussion

There are various pathologies that may lead to disorders causing hyperlipidemia. One we would like to highlight in this report is Familial Chylomicronemia. Lipoprotein lipase (LPL) enzyme and cofactor Apolipoprotein C-II gene mutations, as well as lipoprotein lipase inhibitors have been found as causes of Familial chylomicronemia, a rare autosomal recessive disorder [2]. The literature reports cases of infants and children with this disease to have consanguineous parents, for this patient however, her parents have no such relationship.

In cases due specifically to LPL enzyme deficiency or impairment, patients have trouble metabolizing triglycerides leading to increased amounts in the bloodstream; it generally manifests within the first decade of life, 25% occur in infancy [3]. In asymptomatic patients, diagnosis is often an incidental finding of lipemic blood. It is characterized by marked elevation of serum triglycerides, cholesterol and chylomicrons. Consequently, leading to recurrent attacks of pancreatitis,

***Table 1.*** *Laboratory values of pancreatitis hospitalization.*

| Parameter | Result | Reference values |
|---|---|---|
| Sodium | 136 | 130 - 147 mEq/L |
| Potassium | 4.9 | 3.4 – 5.6 mEq/L |
| Chloride | 107 | 95 – 108 mEq/L |
| Carbon dioxide | 14 | 20 – 30 mEq/L |
| Anion gap | 19.2 | 6 – 12 mEq/L |
| BUN | Less than 5 | 2 – 20 mg/dL |
| Creatinine | Less than 0.15 | 0.3 – 0.7mg/dL |
| Glucose | 105 | 60 – 100 mg/dL |
| Calculated osmolality | 260.0 | 266 – 295 mOSm/kg |
| Calcium | 9.2 | 8.8 – 10.8 mg/dL |
| Total Bilirubin | 0.5 | < 2 mg/dL |
| AST | 45 | 9 – 80 units/L |
| ALT | Less than 12 | 12 – 45 units/L |
| Alkaline Phosphatase | 204 | 150 – 420 units/L |
| Total Protein | 6.1 | 6 – 8.3 gm/dL |
| Albumin | 3.6 | 1.9 – 4.9 g/dL |
| Albumin/Globulin Ratio | 1.4 | 3.4 – 5.2 g/dL |
| Triglycerides | 3028 | < 75 mg/dL |
| Cholesterol | 198 | < 170 mg/dL |
| LDL Cholesterol | TNP | < 110 mg/dL |
| VLDL Cholesterol | TNP | < 30 mg/dL |
| HDL Cholesterol | 13 | >45 mg/dL |
| Total Cholesterol/HDL Ratio | 15.00 | < 3.3 mg/dL |
| Amylase | Less than 30 | 30 – 100 units/L |
| Lipase | 391 | 3 – 216 units/L |

TNP: Test not performed

eruptive xanthomas, hepatosplenomegaly and Lipemia Retinalis [3]. This is true in homozygotes who often present in infancy, as opposed to heterozygotes who do not express severe clinical symptoms [1].

Although this patient still has no genetic evidence to confirm clinical diagnosis, we suspect that according to her medical history she is manifesting symptoms of Familial Chylomicronemia due to a deficiency of LPL. The patient has demonstrated all possible symptoms over time.

At present there is no FDA approved treatment for Familial Cylomicronemia. Administration of lipid lowering agents, such as with this patient, has been used in other reported cases. Dietary modification is crucial in managing disease. These patients require a restricted intake of fats. Medium chain triglycerides are preferred because they leave the intestine directly into the portal vein, and do not require transport on chylomicrons [1]. Table 2 shows the most recent laboratory values that demonstrate the effectiveness of both treatment and dietary modifications in this patient.

***Table 2.*** *Recent laboratory values.*

| Parameter | Result | Reference values |
|---|---|---|
| Cholesterol | | < 170 mg/dL |
| HDL Cholesterol | | >45 mg/dL |
| Total Cholesterol/HDL ratio | | < 3.3 mg/dL |
| Non-HDL Cholesterol | | < 120 mg/dL |
| Triglycerides | | < 75 mg/dL |
| Amylase | | 30 – 100 units/L |
| Lipase | | 3 – 216 units/L |
| CPK | | 5 – 200 units/ L |
| AST | | 9 – 80 units/L |
| ALT | | 12 – 45 units/ L |

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