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CASE STUDY

AN INFANT PRESENTING WITH MILKY SERUM: A RARE CASE REPORT

Varsha S, Shubha HV, Vivek TG, Vijaya C

[1]*Assistant Professor, Department of Pathology, Sapthagiri Institute Of Medical Sciences & Research Centre , Bangalore, INDIA -560090*
[2]*Lab Head, SRL Diagnostics, Fortis Hospital, Rajajinagar, Bangalore, INDIA-560086*
[3]*Consultant Pathologist, Apollo Diagnostics, Ramamurthy Nagar, Bangalore, INDIA-560016*
[4]*Professor & HOD, Department of Pathology, Sapthagiri Institute Of Medical Sciences & Research Centre , Bangalore, INDIA -560090*

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ABSTRACT: Lipid disorders can occur either as primary event or secondary to an underlying disease. Primary dyslipidemias are associated with overproduction or impaired removal of lipoprotein. An abnormality in the lipoprotein itself or in the lipoprotein receptor induces impaired removal of lipoprotein. Hypertriglyceridemia is defined as having plasma triglyceride above the 95th percentile for age and sex. It is a rare disorder in childhood. We present a case of six months old male infant who was admitted in our hospital with the history of anemia. Infant was detected to have highly viscous and milky serum during the sampling of blood coincidentally. The serum triglyceride and cholesterol levels were markedly raised. There were no typical signs of hyperlipidemia like growth failure, dysmorphic features, hepatosplenomegaly, eruptive cutaneous xanthomas or lipemia retinalis. There was also no positive family history of hyperlipidemia. A clinical diagnosis of Type I hypertriglyceridemia was made and the infant was put on lipid lowering drugs and iron drops. Infant responded well to the treatment. Hemoglobin levels improved with the improving lipid profile parameters. Early diagnosis and intervention can therefore, improve the prognosis by reducing the complications and preventing the premature death.

KEYWORD: Dyslipidemia, Hypertriglyceridemia, Milky serum, Anemia, Familial Chylomicronemia Syndrome, Hyperlipidemia

INTRODUCTION :

Hypertriglyceridemia is defined as having fasting plasma triglyceride levels above the 95th percentile for age and sex [1,2,3]. It is a rare disorder in childhood. According to the National Cholesterol Education Program (NCEP), normal triglyceride level is <150 mg/dl (<1.7 mmol/l) [4].

Primary hypertriglyceridemia is the result of various genetic defects leading to disordered triglyceride metabolism. Secondary causes are acquired and may include a high-fat diet, obesity, diabetes, hypothyroidism, and certain medications (e.g., estrogen and tamoxifen) [5].

Corresponding Author:
Dr. Shubha. H.V,
Lab Head, SRL Diagnostics, Fortis Hospital, Rajajinagar, Bangalore, INDIA-560086

According to Fredrickson's classification (WHO), Primary hypertriglyceridemia includes type 1, 2b, 3, 4 and 5. This is based on serum lipid concentration, appearance of serum after centrifugation, and lipoprotein electrophoresis. Primary hypertriglyceridemia includes familial chylomicronemia (Type 1), familial combined hyperlipoproteinaemia (Type 2b) familial dysbetalipoproteinaemia (Type 3), familial hypertriglyceridemia (Type 4) and primary mixed hyperlipidemia (Type 5) [6]. In the pediatrics group, high levels of triglyceride, in more than 90%, is secondary to conditions such as obesity and type 2 diabetes mellitus [3]. We describe a case of six months old male infant who was admitted in our hospital with the history of anemia. Infant was coincidentally detected to have highly viscous and milky serum. The infant was evaluated further and a clinical diagnosis of Type I hypertriglyceridemia was made and was treated accordingly.

CASE REPORT:

A six months old male infant was referred to the department of paediatrics of our hospital with the complaints of lethargy, irritability and poor feeding since a week. The infant was born at term by normal vaginal delivery with a birth weight of 2.8 kg and uneventful antenatal and postnatal periods. Exclusive breast feeding was done since birth. Developmental milestones were appropriate for the age. On physical examination, the child had severe pallor. However, there was no growth failure, dysmorphic features or hepatosplenomegaly. During the sampling of blood for various haematological and biochemical investigations, the child's serum was found to be highly viscous and milky **(Figure 1)**. This was purely a coincidental finding in our case. The routine blood counts revealed Hb- 6.8g/dl, RBC- 2.43 millions/cumm, HCT -22.6 %, WBC -11.7 thousand/cumm, platelets- 4.54 lakhs/ cumm. Biochemical investigations showed very high levels of S. cholesterol- 350 mg/dl, S. triglycerides- 950 mg/dl, HDL cholesterol- 9mg/dl, LDL cholesterol- 250 mg/dl. Liver and kidney function tests were normal.

A clinical diagnosis of Type I hypertriglyceridemia was conferred upon. The infant was further evaluated. However, the infant did not show any typical signs of hyperlipidemia like eruptive cutaneous xanthomas, lipemia retinalis or the history of recurrent acute pancreatitis. There was also no family history of hypertriglyceridemia. Both parents and other sibling of the index case had normal lipid profile parameters. The child was put on lipid lowering drugs, iron drops and a low fat formula. Blood transfusion was also done. The child was discharged and the parents were asked to review again for further workup. After 6 weeks, repeat lipid profile and routine blood counts were done. The Hb level had improved significantly to 11.8 g/dl with the lowering of serum cholesterol levels to 160 mg/dl and serum triglycerides to 250 mg/dl. Early diagnosis and medical intervention in our case helped in achieving significantly great results.

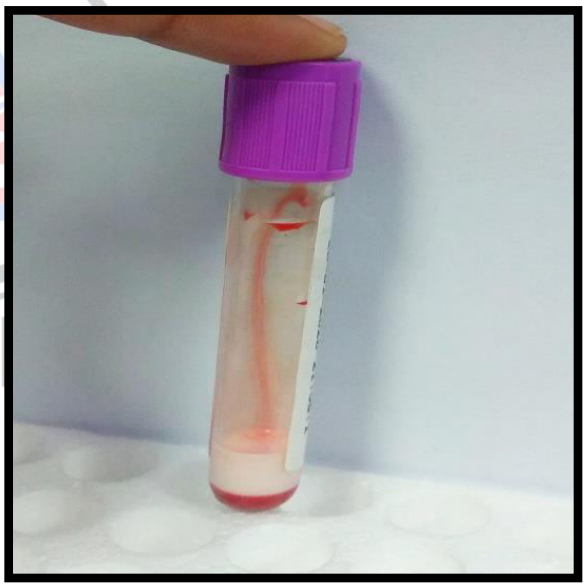

Figure 1: Patient's blood sample showing milky serum

DISCUSSION:

Fasting plasma triglyceride levels that are, typically above the 95th percentile for age and sex is known as hypertriglyceridemia [1,2,3]. According to Fredrickson's classification (WHO), Primary hypertriglyceridemia includes five types- familial chylomicronemia (Type 1), familial combined hyperlipoproteinaemia (Type 2b) familial dysbetalipoproteinaemia (Type 3), familial hypertriglyceridemia (Type 4) and primary mixed hyperlipidemia (Type 5) [7].

Monogenic disorders that cause abnormal levels of plasma cholesterol and triglycerides have received much attention due to their role in metabolic dysfunction and cardiovascular disease. Type I Familial chylomicronemia syndrome (FCS) is characterized by hyperchylomicronaemia and hypertriglyceridemia, confirmed by lipoprotein lipase deficiency [7, 8]. It is very rare syndrome with prevalence of 1 in 1 million for homozygotes and 1 in 500 for heterozygote [9,10].

FCS is characterized by severe hypertriglyceridemia with episodes of abdominal pain, recurrent acute pancreatitis, eruptive cutaneous xanthomata, hepatosplenomegaly, and lipemia retinalis. However, evidence suggests that presentation during infancy can be heterogeneous and may include other signs such as pallor, anemia, jaundice, irritability, and diarrhea. These manifestations are variable in the time and severity of presentation [11]. In the present case, the infant had complaints of lethargy, irritability and poor feeding. On examination the infant had severe pallor. A clinical diagnosis of type 1 hyperlipidemia was given as the serum was lipemic with markedly elevated levels of serum cholesterol and triglycerides.

In a similar study conducted by Chaurasiya OS et al [12], a two month old male baby was found to have milky serum. S. Cholesterol was 2,200 mg/dl and triglycerides 13,292 mg%. There was no dysmorphic feature. Child was anemic and had hepatosplenomegaly and xanthomas. There was positive history of hyperlipidemia. In our case, there was no family history of hyperlipidemia. However, genetic testing could not be done.

Dietary restriction of fats to 20 g/day or less or to 15% or less of the total energy intake is the basic treatment modality in this case [13]. However, initiating a combination of fibrates, niacin and or fish oil to lower triglyceride levels to below 500 mg/dL is the primary goal [6]. Dietary restriction of fats will prevent elevation of serum triglycerides further, thereby preventing complications of hypertriglyceridemia like pancreatitis, cardiovascular disease [14]. But a significantly persistent high triglyceride level necessitates pharmacological intervention. There has been a general reluctance to use drug therapy to treat lipid abnormalities in children; however, increasing evidence suggests effectiveness and short-term safety similar to those in adults [15]. Lipid lowering drug and low fat formula dramatically improve lipid parameter and prevent premature death. Early diagnosis and medical intervention can therefore, prevent complications and helps the infant to lead a near normal lifestyle by reducing the hospital admissions.

CONCLUSION:

Hypertriglyceridemia is a serious yet treatable cause of milky serum. The case highlights the importance of identifying this unusual and rare condition in an infant as it leads to serious consequences if undiagnosed and left untreated. Timely medical intervention by lipid lowering drugs and simple dietary modifications like restriction of fats help in achieving great results. Typical clinical symptoms and signs may not necessarily appear in all the cases. Early diagnosis is of utmost importance to prevent the complications and thereby, improve the prognosis.

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