Case Report

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A Case Report of Familial Chylomicronemia Syndrome

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Received: 7 September 2020 **Revised:** 12 October 2020 **Accepted:** 8 November 2020

ARTICLE INFO

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Keywords:
Chylomicronemia;
Hypertriglyceridemia;
Hemolytic Anemia;
Leukocytosis;
Pancreatitis

ABSTRACT

Background: Diagnosis of neonatal chylomicronemia, as a very rare condition, is very difficult and usually is diagnosed when acute pancreatitis sets in. Early diagnosis can prevent the complications such as acute pancreatitis and pancreatic necrosis which are associated with the condition.

Case Presentation: A 5.5 month- old female breastfed baby presented to us suffering from splenomegaly because of respiratory infection. Anemia and leukocytosis were seen in laboratory data. The result of bone marrow aspiration (BMA) performed to diagnosis was normal. Following the study, the patient had a high triglyceride, which improved with the treatment of symptoms and blood indices.

Conclusion: Our case reports a rare disorder that was initially admitted with suspicion of malignancy, organomegaly, anemia and leukocytosis. In the course of hospitalization, the diagnosis of malignancy was rejected after BMA, and chylomicronemia was diagnosed and the patient's leukocytosis and high uric acid were eliminated by treatment of the disease and the patient's symptoms were improved.

Introduction

Familial chylomicronemia syndrome (FCS) is a rare autosomal recessive disorder caused by mutations in lipoprotein lipase, apolipoprotein C-II deficiency or the presence of inhibitors to lipoprotein lipase, resulting in accumulation of chylomicrons in plasma and hypertriglyceridemia. There are several complications caused by elevated triglycerides in patients which acute pancreatitis is the most serious being episodes.[1-3] Due to its rarity and lack of specificity of signs and symptoms, the

recognition and correct diagnosis of the disease is challenging.[4] FCS is characterized by marked elevation of triglyceride and chylomicron levels, leading to lipemic plasma, recurrent attacks of acute pancreatitis, eruptive xanthoma, hepatosplenomegaly, lipemia retinalis.[2] Its prevalence is approximately 1 in 1 million for homozygotes.[5-7] Twenty- five percent of cases of familial chylomicronemia syndrome manifest during infancy; however, extremely rare cases manifest during the neonatal period. Pink- colored blood, milky white supernatant and falsely elevated pseudohyponatremia can be caused by severe hypertriglyceridemia. Lab studies usually show very high levels of triglycerides.[6] Treatment of patients with familial chylomicronemia requires severe dietary fat restriction to maintain fasting TG levels below 850 mg/dl to reduce the risk of pancreatitis.[8-10]

Case Presentation

The patient was a 5.5 month-old female baby of non-consanguineous parents admitted to Shahid Sadoughi hospital due to respiratory infection who was accidentally considered as suffering from splenomegaly in physical examination. Anemia and leukocytosis were found in laboratory tests. She did not have poor feeding, vomiting, restlessness, fever, any change in urination and defecation, diarrhea, constipation, and melena. The patient was the third child of a G3L3Ab0 mother created by a caesarian. Her parents had no relationship. There was no case history for the mother during pregnancy and she was also not on drugs during pregnancy. The patient's family history, medication-taking history and medical allergy were negative. The patient was breastfed and baby food was not started yet. She was pale, not jaundiced and didn't have a syndromic face. The fontanel was open, but not wide. The nasal bridge was not wide and the ears did not have a low set. The patient didn't have a cleft palate. The chest was not deformed in observation. There was no reduction in lung voice sound during auscultation. There were also no rales and wheezing, and sounds from two sides were normal and symmetrical in cardiac auscultation. There was no heart murmur and extra sound. No apparent lesion and scar was seen in stomach examination. No mass was touched. A splenomegaly about 6-5 cm under the edge of the ribs was touched. Hepatomegaly was not obvious.

In the patient laboratory data: The results of laboratory tests showed high uric acid, WBC, bilirubin and TG, and low hemoglobin (Table 1).

Table 1. laboratory data

Test	Preliminary result	Unit
BS	165	
urea	9	mg/dl
cr	0.4	mg/dl
Bili T	3.1	mg/dl
Bili D	1.1	mg/dl
ALT	30	U/L
AST	160	U/L
LDH	4546	U/L
Retic	1.8	
W.B.C	55700	3/ml^ X10
PLT	275000	
Hb	9.9	mg/dl
CRP	Weakly positive	mg/l
Coombs D & I	Negative	
Uric Acid	19	mg/dl
ESR	4	
CHOL	132	mg/dl
TG	1284	mg/dl
HDL	25	U/L
Na	135	mEq/L
K	3.3	mEq/L
Ferritin	112	ng/ml
Lipase	218	U/L
Amylase	26	U / ml

Peripheral blood smear: The patient's REC morphology

- Severe anisocytosis, hypochromia, dacryocytosis & schistocytosis
- 47NRBC /100WBC

The patients second PBC Sample

- RBC Morphology: Aniso + poikilo+ Hypo +Target+sphrocytosis +
- 3NRBC / 100WBC

Primarily the patient hospitalized at an

emergency department, considering the breast-fed signs the necessary tests requested, ultrasound was performed and peripheral blood smear (PBS) was requested. Numerous nucleated red blood cells (NRBCs) were observed in patient's PBS that patients' leukocytosis was justifiable based on. Because of the milky blood, cholesterol and triglyceride were checked. In the reported samples, patient had high TG and normal cholesterol. Considering endocrine consultative symptoms, medical history and tests related to the second blood sample which was taken before feeding, triglycerides and cholesterol check were requested again. The abnormality of chylomicronemia was recognized for the patient through repetition. Enzymatic and genetic studies were not performed for definitive diagnosis due to high cost. The patient was subjected to a special milk diet, MCT Oil and Omega3. During hospitalization, the patient had a drop in hemoglobin which leads to a PBS request again. Heart signs at consultation and Echo were normal and had an EF of 55%. During patient's monitoring for a week, the experiments had a downward trend.

- Chol: 159 mg/dl
- TG: 678 mg/dl
- WBC: 19900(Neu: 62% Lym: 20%)
- Hb: 10 (mcv: 75)
- PLT: 454000

A blood sample was also taken from patient's mother and father for checking triglycerides and cholesterol which both of them had a normal level.

Discussion

Hyperchilomicronemia leads to impaired laboratory test results. Because of thinning the membrane of red blood cells, hemolysis occurs more rapidly that leads to hemolytic anemia. In the mentioned case, there was high leukocytosis and uric acid and an increase in LDH according to the signs and laboratory results. At first, the patient was approached with suspected malignancy. Malignancy was rejected due to the normal bone marrow result after necessary examinations. The patient was treated for hyperchilomicronemia according to the results of tests during the hospitalization and diagnosis. After the treatment, the results of tests and hemolytic anemia were improving during the follow-up care. According to the results of the treatment, it can be concluded that an increase in blood triglyceride leads to disorders such as hemolytic anemia and leukocytosis and an increase in uric acid in laboratory studies. A similar result is reviewed in another report case.[11]

Conclusion

High triglyceride can cause symptoms similar to hemolytic anemia and leukocytosis. Ruling out the malignancy, treatment can be started and normalization of blood indices has prospected in the follow-up.

Conflict of Interests

Authors have no conflict of interests.

Acknowledgments

The authors would like to thank the patient's family for their cooperation in this study.

How to Cite: Ordooei M, Binesh F, Ashrafi A, Ferdosian F, Saeida-Ardekani M. A Case Report of Familial Chylomicronemia Syndrome. World J Peri & Neonatol 2020; 3(2): 90-3.
DOI: 10.18502/wjpn.v3i2.6160

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