Case Report

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Idiopathic Hypertriglyceridemia in Thalassemia Major: A Case Report

Pathology Section

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ABSTRACT

Thalassemia major is a severe hereditary haemolytic anaemia and is usually associated with normal serum lipid profile. But there are few reports in literature that hypertriglyceridemia can have an idiopathic association with β-thalassemia major. We report a case of hypertriglyceridemia associated with β-thalassemia major, in a seven month old female baby.

Keywords: Haemolytic anemia, Hyperbilirubinemia, Lipid profile

CASE REPORT

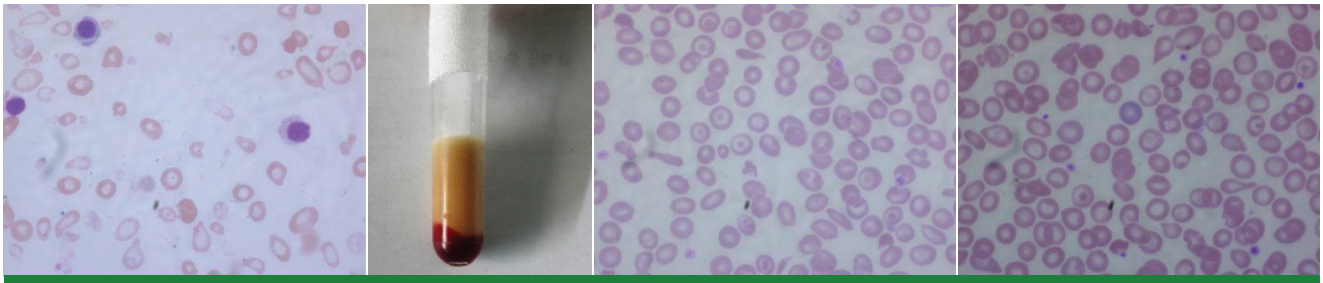
A 7- month-old female baby, first child of non-consanguineous parents was referred to us from a local hospital with complaints of pallor and weight loss. She was delivered by caesarean section at 33 weeks due to premature rupture of membranes and cephalopelvic disproportion. Birth weight was 2.35kg and following neonatal hyperbilirubinemia (non immune) on day 3, phototherapy was given for 6 days. She was exclusively breast fed and immunized for age. Later, parents noticed delay in achieving milestones and poor weight gain for which they visited a local hospital.

On physical examination, she had significant pallor, mild jaundice and hepatosplenomegaly. Weight (5.4kg) and height (62cm) were below 3rd centile for age and sex with a head circumference at the 75th centile. There was no skin rash, oedema, lymphadenopathy, ascites or bleeding manifestations.

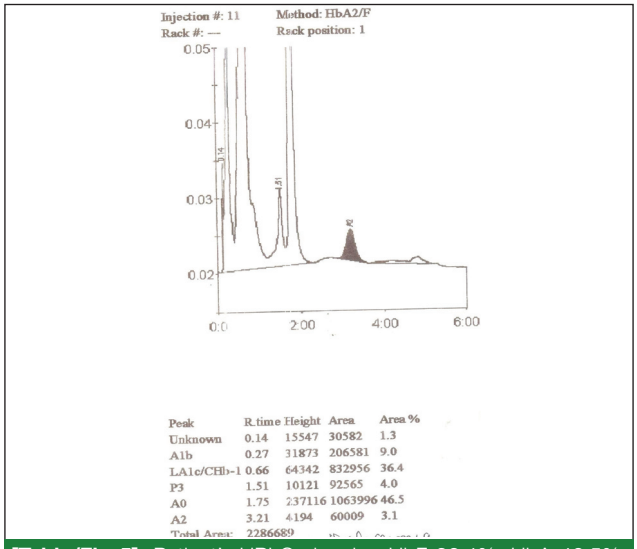
A complete blood count showed a WBC 12x10^3/μL, Hb 6.6 g/dL, HCT 16.9%, MCV 70.5 fL, MCH 47.1 pg, MCHC 33.2 g/dL and platelet count 430×l0^3/μL. Reticulocyte count was 12% and serum LDH was 1114 U/L. Peripheral blood smear revealed microcytic hypochromic anaemia, marked polychromasia with nucleated erythrocytes, target cells and RBC inclusions (basophilic stippling, Howell-Jolly bodies and Cabot rings) [Table/Fig-1]. Her serum was noted to be grossly lipaemic [Table/Fig-2]. Hence, a serum lipid profile was obtained. The fasting triglyceride level was 871mg/dl (Normal for breast fed infants of this age is 150 mg/dl) [1]. Total cholesterol and other lipid fractions were normal. Signs of hypertriglyceridemia like tonsillar hypertrophy, corneal arcus or xanthomas were absent in the baby. Osmotic fragility testing revealed a reduction in fragility when compared to age matched control and her bone marrow examination showed marked erythroid hyperplasia. Both parents blood counts and smears showed microcytic hypochromic anaemia with polychromasia and occasional target cells [Table/Fig-3,4] and their serum lipid fractions were within normal limits.

High Performance Liquid Chromatography (HPLC) carried out in EDTA blood samples of baby [Table/Fig-5] and parents [Table/Fig-6,7] confirmed baby as β-thalassemia major and both parents as thalassemia minor/trait.

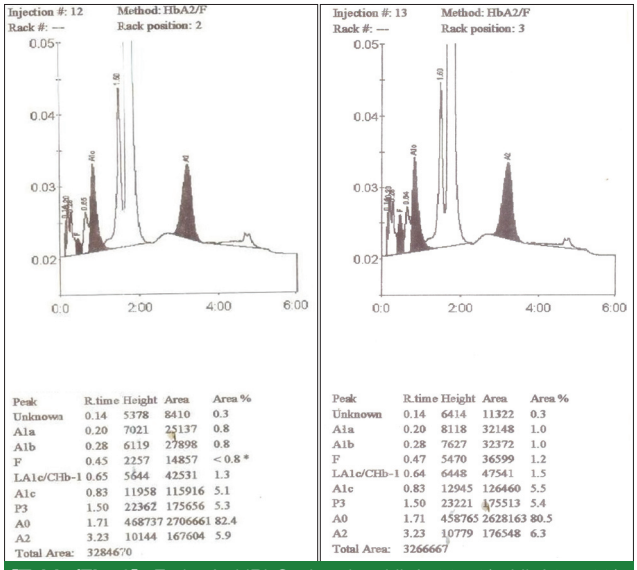
After all necessary hematological and biochemical



[Table/Fig-1]: Blood smear of patient showing anisopoikilocytosis, hypochromia, target cells, and nucleated RBCs. Leishman's stain X1000. **[Table/Fig-2]:** Blood sample from patient with thalassemia major showing lipaemia of plasma. **[Table/Fig-3]:** Blood smear of father showing hypochromia and target cells. Leishman stain X1000. **[Table/Fig-4]:** Blood smear of mother showing hypochromia and target cells. Leishman stain X1000.



[Table/Fig-5]: Patient's HPLC showing HbF 36.4%, HbA 46.5%, HbA2 3.1% (X-axis: Elution time in minutes; Y- axis: Concentration of Hb fraction).



[Table/Fig-6]: Father's HPLC showing HbA 82.4%, HbA2 5.9%.
[Table/Fig-7]: Mother's HPLC showing HbA 80.5%, HbA2 6.3%.

investigations, the baby was diagnosed as beta thalassemia major associated with hypertriglyceridemia and both parents as thalassemia minor. The child is on regular follow-up with red cell transfusion therapy for the past one and a half years and her serum triglyceride level has returned to normal level during follow-up evaluation. A written consent was obtained from the parents for writing the case report.

DISCUSSION

Beta- thalassemia major is an inherited haemolytic anaemia usually presenting within the first year of life with pallor, failure to thrive, hepatosplenomegaly and a positive family history. Beta- thalassemias are caused by various point mutations or deletions in the beta globin gene on chromosome 11, leading to diminished (beta+) or absent (beta 0) production of the beta chains of haemoglobin resulting in ineffective erythropoiesis. In β0 thalassemia major, the major red cell haemoglobin is HbF and the red cells completely lack HbA whereas in β+ thalassemia major, HbF levels are generally normal or slightly increased. HbA2 level is normal or low in beta thalassemia homozygotes and it is elevated (4 to 8%) in beta thalassemia minor. About ten percent of the total world thalassemics are born every year in India [2].

Patients with thalassemia major usually present after six months of life with severe anaemia, failure to thrive, jaundice, hepatosplenomegaly or poor weight gain and require follow-up with regular red blood cell transfusions. Our case presented with pallor, jaundice, hepatosplenomegaly, poor weight gain and delayed milestones at seven months of age with microcytic hypochromic indices and marked degree of anisopoikilocytosis on peripheral blood smear and with HbF levels of 36.4% on HPLC.

Severe hypertriglyceridemia has been rarely reported in infants with thalassemia major, an association known as hypertriglyceridemia–thalassemia syndrome [3]. The pathogenesis of this rare association is not clear. It is important to recognize this rare association for the proper diagnosis and subsequent management of infants with thalassemia, especially in areas with low prevalence of thalassemia.

There are few case reports from North and West India pointing this association but there is no much data available on this from Kerala [4,5]. Children in these reports presented at an age ranging from five months to two years; this case presented at seven months of age. In our case, serum lipid fractions of parents were normal and associated findings like tonsillar hypertrophy, corneal arcus, tendon and tuberous xanthomas were absent excluding primary hypertriglyceridemia. Secondary hypertriglyceridemia is observed in children in association with diabetes mellitus, obesity, nephrotic syndrome and uraemia which were ruled out by proper history and appropriate investigations [5].

This case is presented here as hypertriglyceridemia can have an impact on the prognosis of thalassemic children by adding on to its morbidity by increasing the risk of developing early atherosclerosis and related complications. Follow-up for spontaneous resolution is also advised suggesting a self limited mechanism which may not require therapy [6]. To the best of our knowledge, this is the first case reported from Kerala on this rare association.

CONCLUSION

Screening of all children with thalassemia and other haemolytic anemias is strongly suggested to exclude underlying hypertriglyceridemia and such children should be followed-up more cautiously for proper management.

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