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# Thalassemia Major with Idiopathic Hypertriglyceridemia: A Case Report

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**Abstract:** ***β-Thalassemia major is a common type of hereditary hemolytic anemia. It is usually associated with a normal serum lipid profile. But there have been few reports in literature that β-Thalassemia can have an association with hypertriglyceridemia. We report a case of β-thalassemia major associated with idiopathic hypertriglyceridemia, in a 6-month-old male child.***

**Keywords:** hypertriglyceridemia, thalassemia major, hypertriglyceridemia-thalassemia syndrome

## 1. Introduction

Thalassemia refers to a group of disorders of globin chain production leading to imbalance between the α and β chain production. β-Thalassemia major is a common variant and usually present during the 2nd 6 months of life, with growth failure, pallor, hepatosplenomegaly, and bony deformities. β-Thalassemia generally presents with a normal serum lipid profile, but there have been few reports where it can have an association with hypertriglyceridemia. We report a case of β-thalassemia major associated with idiopathic hypertriglyceridemia, in a 6-month-old male child.

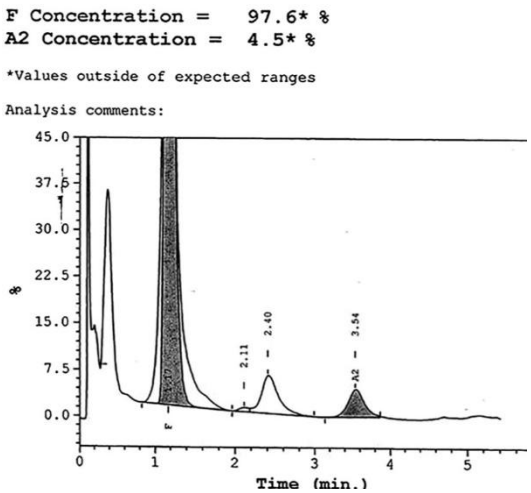
## 2. Case Report

A 6-month-old male baby, first born of non-consanguineous parents was referred to us from a local hospital with complaints of decreased feeding and breathlessness. He was delivered at term via LSCS. Birth weight was 3.2kg and child had an uneventful neonatal period. He was exclusively breast fed and immunized for age. For about 2 weeks parents noticed that the child was having decreased activity and feeding, after which the child also started to develop rapid breathing.

On examination, he had severe pallor, and hepatosplenomegaly. Weight (5.4kg) is below 1st centile and height (60.5cm) is below 3rd centile for age and sex with a head circumference at the 50th centile. There was no rash, icterus, edema, lymphadenopathy, or bleeding manifestations. He presented to our hospital with severe distress and signs of cardiac failure.

Since there was significant pallor, few investigations for the workup for anemia were drawn and the patient was given an emergency blood transfusion. A complete blood count showed a WBC count of 35, 200 cell/mm$^3$, RBC count – 2.25 million/mm$^3$, Hb 5.5 g/dL, HCT 15.9%, MCV 73.5 fL, MCH 23.9pg, MCHC 32.3 g/dL and Platelet count 4, 68, 000/µL. Reticulocyte count was 4.2% with correct reticulocyte count being 1.3% (normal). The peripheral blood smear revealed microcytic hypochromic RBCs, with marked polychromasia, anisopoikilocytosis, with nucleated RBC, target cells and ovalocytes.

His serum sample was noted to be white in color (Figure 1). Consequently, a serum lipid profile was also sent. The triglyceride level was found out to be **534 mg/dl** (normal for this age is <100 mg/dl [1]). Total cholesterol and other lipid fractions were normal. There were no signs of hypertriglyceridemia like xanthomas, tonsillar hypertrophy, corneal arcus in the child. The parents' lipid profile was obtained and was within normal limits. High Performance Liquid Chromatography (HPLC) carried out in EDTA blood samples of baby and was suggestive of βThalassemia (Figure 2).



After all necessary hematological and biochemical analysis, the baby was diagnosed as β thalassemia major associated with hypertriglyceridemia. The child was started on regular packed red cell transfusion therapy and put on regular follow up. No intervention was done for the high serum triglyceride levels. Now the child is 11 months old, and the serum triglyceride level has returned to normal level (96) during follow-up evaluation.

## 3. Discussion

Thalassemia refers to a group of genetic disorders of globin chain production leading to imbalance of α and β chain production. β-Thalassemia syndromes are more common, and result from a decrease in β-globin chain synthesis. They are due to mutations in the β-globin gene on chromosome 11. In β0-thalassemia there is complete absence of β-globin chain, making HbF the major hemoglobin, without any HbA. Whereas in β+-thalassemia, HbF levels are either normal or slightly increased, with decreased HbA. β0-thalassemia syndromes are generally more severe, but there can be significant variability between genotype and phenotype. About ten percent of the total world thalassemics are born in India, every year [2]. It is especially common in the tribal areas.

Children with β-Thalassemia major usually present during the 2nd 6 months of life, with growth failure, pallor, hepatosplenomegaly, bony deformities, and a positive family history. They require regular follow up and need to be on chronic transfusion therapy. Our case, a 6-month-old child presented with severe pallor, hepatosplenomegaly, poor weight gain, signs of cardiac failure and hypertriglyceridemia.

Our patient had a markedly elevated triglyceride level while the rest of the lipid profile was normal. Since the child had no physical findings associated with primary hyperlipidemias like tendon or tuberous xanthomas, tonsillar hypertrophy and corneal arcus, and the lipid profile of the parents was normal, primary hyperlipidemias were ruled out. Hyperlipidemia could also be seen secondary to conditions like, diabetes mellitus, nephrotic syndrome, hypothyroidism, biliary atresia, infection, and chronic renal failure. With history and appropriate biochemical investigations these were ruled out.

There have been few case reports of severe hypertriglyceridemia in infants, in association with thalassemia major. The exact pathogenesis of hypertriglyceridemia is not known. It has been observed that hypertriglyceridemia may accompany acute massive hemolysis [3], which could possibly be the underlying cause. This association of hypertriglyceridemia in thalassemia major has been termed as Hypertriglyceridemia-thalassemia syndrome [4, 5], by some authors in India. Few case reports of this association have been published from North and West India mainly, but no data is available from Andhra Pradesh.

The hypertriglyceridemia can have an impact on the prognosis of these thalassemic children by adding on to its morbidity, with increased risk of developing early atherosclerosis, myocardial infarction, and pancreatitis. Careful follow up for spontaneous resolution of hypertriglyceridemia, and appropriate management is advised.

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**Legend for the Images:**
Figure 1 – Blood sample of the patient which was white in color
Figure 2 – HPLC of the baby (suggestive of β thalassemia)