Case Report

Extremely High Triglycerides in a Beta Thalassemia Minor Patient: A Case Report

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Abstract

Introduction: Hypertriglyceridemia in pediatric population whether familial or acquired is associated with Diabetes Mellitus type 1, uremia, hypothyroidism, nephrotic syndrome etc. abnormal serum lipid levels are associated with premature atherosclerosis. It is also seen that there is an association of hypertriglyceridemia with beta thalassemia in some cases which indicates the poor prognosis.

Materials and methods: In this context here, we are reporting a case of beta thalassemia minor with extremely high triglyceride level. A 2-year-old male child with history of failure to thrive and mild to moderate anemia was investigated for complete blood count (CBC), hemoglobin typing, lipid profile, renal function test, liver function test, thyroid profile and the electrolytes.

Result: The Hb typing showed elevated HbA_2, CBC showed microcytic hypochromic anemia, triglyceride level was 2010 mg/dl. His father and mother are both beta thalassemia minor patient

Conclusion: As hypertriglyceridemia in beta thalassemia is reversible, care should be taken to prevent persistent hypertriglyceridemia which increases chance of atherosclerosis and pancreatitis

Key words: hypertriglyceridemia, thalassemia, atherosclerosis

Introduction

Hypertriglyceridemia is defined as conditions with plasma triglycerides carrying lipoproteins levels > 150 mg/ dl. Hypertriglyceridemia can be familial or acquired, reflecting increased hepatic synthesis or decreased catabolism. In pediatric patients, acquired hypertriglyceridemia has been found to be associated with diseases like Diabetes Mellitus Type I, uremia with or without dialysis, hypothyroidism, nephrotic syndrome, obesity, certain drugs use etc. Literatures have revealed strong association of abnormal serum lipid levels [low total cholesterol (TC) and HDL-cholesterol (HDLc), high triglycerides (TG) and

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TC: HDL ratio] with premature atherosclerosis in children with Beta- thalassemia major, though rare, often high triglyceride in patients being referred to as Hypertriglyceridemia – thalassemia syndrome [1]. Though the association of hypertriglyceridemia with the beta thalassemia is not so much well established, and its effect in the thalassemia is not clearly understood, but according to some recent litreatures the effects of dyslipidemia have poor prognostic consequences. [2] In this context, we present a case report of rare association of extremely high triglyceride level in a Beta thalassemia minor patient to underscore the necessity of estimation of lipid profile even in Beta thalassemia minor cases for prevention of complication and timely management.

Case Report

A 2-year-old male child born out of consanguineous marriage was referred to the pediatric OPD in College of Medicine and Sagore Dutta Hospital with chief complaint of pallor, weight loss & learning difficulties for estimation of Complete Blood Count, Hemoglobin HPLC, lipid profile, liver function test & renal function test. He was delivered by normal vaginal delivery. Birth weight being 2.15 kg and following neonatal hyperbilirubinemia (non-immune) on day 3, phototherapy was given for 2 days and he recovered uneventfully. He was exclusively breast fed and immunized for age. Later the parents found the child to have poor growth & weight gain. The child was having learning disabilities due to lack of concentration as well.

On physical examination, he was found to have mild pallor, however there was no icterus. Weight of the child was 7.5kg and height was 70.5cm were below 3rd & 5th percentile for age and sex respectively with a head circumference at the 75th percentile (49.1 cm). There was no feature of skin rash, edema, lymphadenopathy, ascites or bleeding manifestations. All systemic examinations were within normal limit excepting a mild hepatosplenomegaly.

The complete hemogram showed Hemoglobin was moderately reduced (7.8 g/dl), reduced hematocrit (22.93%), mean corpuscular volume (MCV) (58.8fL) and mean corpuscular hemoglobin (MCH) 19.5 pg level. The WBC count and platelet were within normal limit. The differential count shows relative lymphocytosis (Neutrophil 23% and Lymphocyte 72%).

Peripheral blood smear showed reactive lymphocytes along with hypochromic, microcytic RBC pictures along with presence of target cells.

The hemoglobin typing was done by HPLC method which revealed HbA0 77%, elevated HbF (8.2%) and elevated HbA2 (6.0%)which clearly indicated the patient was suffering from beta thalassemia minor.

Both his parents on screening with Hemoglobin HPLC (Figure 2 & 3) revealed them to be beta thalassemia minor as well as their Peripheral blood smear showed microcytic hypochromic / polychromatic RBC with and occasional target cells.

However, serum obtained for assaying the other biochemical parameters was found to extensively lipemic. Serum sample on processing gave extremely high Triglyceride (2,010 mg/dl). Extensively high triglyceride interfered with estimation of serum HDL, LDL, Alanine Aminotransferase.

(ALT); Aspartate Aminotransferase(AST), sodium, potassium, uric acid, urea (BUN), creatinine by SIEMENS ADVIA CHEMISTRY Auto- Analyzer. Triglyceride being above the cut off level as indicated by the manufacturer the results of sodium, potassium uric acid, urea (BUN), creatinine was not given due to possibility of risk of missing clinically significant abnormality.

The child had no evidences of tonsillar hypertrophy, corneal arcus, tendon or tuberous xanthomas which ruled out primary hypertriglyceridemia. Post transfusion the child came back to us after 4 weeks when his lipid profile showed consequent changes and this time liver function test parameters, thyroid profile, renal function test could be reported a appropriate clinical history taking could negate Secondary hypertriglyceridemia observed in children in association with diabetes mellitus, obesity & nephrotic syndrome.

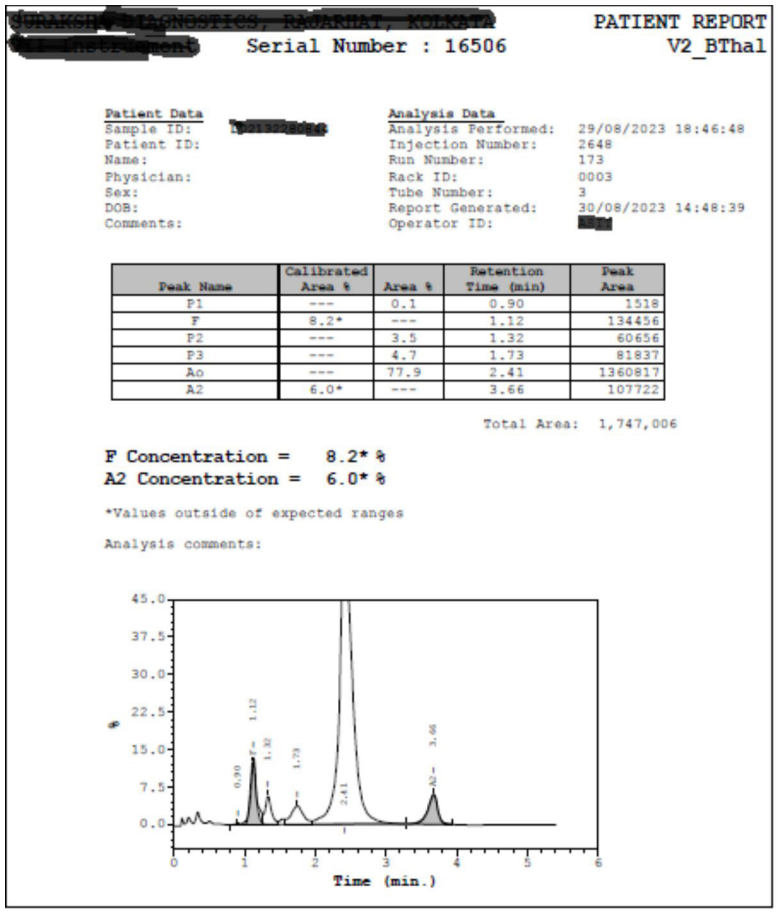

Fig 1: hemoglobin typing report of the baby

Table 1: Biochemical parameters of the patient:

Parameters	Value (Day 1)	Value (4 weeks)	Bio Ref. Interval
Total cholesterol	223mg/dl	286mg/dl	Desirable: < 200 mg/dl Borderline high: 200-239 mg/dl High: > or =240 mg/dl
Triglyceride	2010mg/dl	422mg/dl	Normal: < 150 mg/dlBorderline High: 150-199 mg/dl High: 200-499mg/dl Very High >500mg/dl
HDL	5 mg/dl	41 mg/dl	< 40mg/dl - Low 40-59 mg/dl- Optimum 60mg/dl - High
LDL	35	160	optimal: <100 mg/dl, Near optimal:100-129 mg/dl, Borderline high: 130-159 mg/dl, High: 160-189 mg/dl, Very high: >=190mg/dl

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VLDL	183	84 mg/dl	< 40 mg/dl
UREA	*	8.6mg/dl	15-43mg/dl
CREATININE	*	0.16 mg/dl	0.19-0.49mg/dl
SODIUM	*	135 mEq/l	132-146 mEq/l
POTASSIUM	*	3.5 mEq/l	3.5-5.5 mEq/l
URIC ACID	*	3.2mg/dl	2-5mg/dl
TSH	*	0.815µIU/L	0.67–4.16 µIU/L
T3		1.84ng/ml	1.05–2.07ng/ml
T4		12.5µg/dl	5.5–12.1µg/dl
TOTAL BILIRUBIN	*	1.0mg/dl	0.3-1.2 mg/dL
SGOT	*	19U/L	8-60 U/L
SGPT	*	20 U/L	7-55 U/L
ALKALINE PHOSPHATASE	*	310 U/L	142-335 U/L
TOTAL PROTEIN	*	8.1g/dl	6.0-8.0 g/dL
ALBUMIN	*	4.7g/dl	3.8-5.4 g/dL

Discussion

Hemoglobinopathies are the commonest inherited disorder in India out of which Beta thalassemia has always been a concern considering the burden on the healthcare system of the country. Data on Prevalence of Beta thalassemia amongst different ethnic groups in India has been scarce. Literatures have shown that the prevalence of β-thal trait in Central India ranges between 1.4 and 3.4%, whereas 0.94% β-Thalassemia Major was reported among the anemic patients screened. In South India, however, the prevalence of β-thal trait was found to be between 8.50 and 37.90% and β-Thalassemia Major was between 2.30 and 7.47%. Northern and Western India have shown a higher thalassemic burden while in Eastern India, data have showna higher burden of β-thal trait (0.00-30.50%), β-Thalassemia Major (0.36-13.20%) amongst tribal populations than nontribal populations mostly due to the prevalence of endogamy or consanguinity in marriage [3]

Considering the huge burden of the disease on the society, it has been extremely essential to reach out to the patients and prevent any health complication that may affect the survival. Beta thalassemia Major has been found to be associated with Reversible Hypertriglyceridemia which has been evidenced in several literatures as hypertriglyceridemia–thalassemia syndrome. However, literatures on Beta thal trait patients have been found not to show association much with reversible hypertriglyceridemia comparable to normal population.

However, in our case we revealed a very high triglyceride in the patient's sample which interfered with the biochemical analysis of other parameters like HDL, LDL, VLDL, urea, creatinine, Thyroid profile , electrolytes , liver function tests. Lipemia causes interference in measurement by several mechanisms, non-specific interactions by Lipoproteins can interfere with antigen-antibody reaction by blocking binding sites on antibodies even with antibodies bound to a solid surface resulting into falsely elevated or falsely decreased result depending on the nature of interaction. [4] Not only that, in grossly lipemic sample where the proportion of lipid phase increases up to 25% (over 17 mmol/L of triglycerides), analytes distributed in the aqueous part of the sample (i.e. electrolytes), accounts for only 75% of the sample. Thus, Methods like indirectpotentiometry, measuring concentration of electrolytes in the total plasma volume (including the lipid phase), will report falsely

decreased concentration of electrolytes because of the high dilution prior to analysis. Multiplying the obtained result after the measurement to the full plasma volume, will provide an erroneous electrolyte concentration. [5] Not only that

Lipoprotein particles in the sample can absorb light, the intensity of which is inversely proportional to the wavelength and decreases from 300 to 700 nm, with no specific absorption peaks. Thus, the highest absorbance being in the lowest wavelengths of the spectra, methods using lower wavelengths are more affected by lipemia. Methods for assays like ALT, AST, using reaction NAD(P)+ ↔ NAD(P)H + H+ as an indicator reaction (the change of absorbance measured at 340 nm,) for determining concentration or activity of the analyte, are mostly interfered by lipemia. [5]

Not only the interference, literatures have shown increasing hyperlipidemia (particularly hypertriglyceridemia) is associated with increased hemolysis which may be attributed to the mechanism of increased lipid concentrations altering the composition of the erythrocyte membrane, causing to increased erythrocyte fragility, followed by leakage of cellular content such as hemoglobin which happens overwhelmingly while sample collection due to sheering stress through needles & gel pores.

Malnutrition is known to contribute in causing hypertriglyceridemia, mechanism behind being release of interferons, TNF alpha and other cytokines associated with wasting syndromes. This needs to be taken into account considering the socioeconomic status of our population.[6]

While Hypertriglyceridemia in thalassemia trait being reversible, care should be taken to prevent persistent hypertriglyceridemia which may cause long-term complications with increased risk of atherosclerosis and acute pancreatitis. Thus, our case points towards increased necessity for beta thalassemia trait patients as well to undergo routine investigations alike beta thalassemia major patients for prevention of increased burden of diseases due to deranged lipid profile.

Conclusion

As hypertriglyceridemia in beta thalassemia is reversible, care should be taken to prevent persistent hypertriglyceridemia which increases chance of atherosclerosis and pancreatitis

Ethical clearance: taken from Institutional Ethical committee, College of Medicine and Sagore Dutta Hospital, Kolkata, vide memo no CMSDH/IEC/2023/246 dated 030/9/2023

Informed consent: informed consent was taken from the father of the baby for publication of information in a journal

Conflict of interest: Nil

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