VOLUME - 15, ISSUE - 06, JUNE - 2026 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Paediatrics

CLINICAL PROFILE AND GROWTH PATTERNS IN PEDIATRIC PATIENTS WITH BETA THALASSEMIA MAJOR ATTENDING A HEMATOLOGY CLINIC: A CROSS-SECTIONAL STUDY

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ABSTRACT

Background - Beta thalassemia major is a severe transfusion-dependent hemoglobinopathy associated with chronic anemia, iron overload, growth failure, and multisystem complications. Despite advances in transfusion support and iron chelation therapy, morbidity remains high in developing countries. **Aim -** To study the clinical profile, growth pattern, and biochemical parameters of pediatric patients with beta thalassemia major attending a hematology clinic. **Methods -** This cross-sectional descriptive study was conducted in the Pediatric Hematology Unit of a tertiary care hospital over 15–18 months. Thirty-six children aged 1–18 years diagnosed with beta thalassemia major by HPLC and receiving regular transfusions were included. Clinical history, anthropometric measurements, transfusion details, chelation compliance, and biochemical investigations including serum ferritin and liver function tests were analyzed. **Results -** The mean age of patients was 8.36 ± 4.30 years. Males constituted 58.3% of the study population. Early diagnosis (<1 year) was observed in 88.9% patients. Consanguinity was present in 66.7% families. Hepatomegaly and splenomegaly were observed in 88.9% and 83.3% patients respectively, while facial dysmorphism was present in 77.8%. Growth abnormalities were highly prevalent, with 63.9% children showing abnormal anthropometric indices. Stunting was present in 38.9%, underweight in 50%, and thinness in 27.8% children. Mean transfusion burden was 65.3 ± 33.6 transfusions per child. Only 13.9% patients were compliant with chelation therapy, while 86.1% showed irregular compliance. Mean serum ferritin level was 3413.3 ± 1962.8 ng/mL. Elevated ferritin levels correlated with growth abnormalities and hepatic dysfunction. **Conclusion-** Growth failure, iron overload, and poor chelation compliance are major concerns in pediatric beta thalassemia major patients. Regular monitoring of anthropometry, optimization of chelation therapy, and multidisciplinary care are essential to improve long-term outcomes.

KEYWORDS : Beta Thalassemia Major, Serum Ferritin, Growth Failure, Chelation Therapy, Iron Overload, Pediatric Hematology

INTRODUCTION

Beta thalassemia major is an autosomal recessive hemoglobin disorder caused by defective synthesis of beta globin chains, resulting in severe anemia, ineffective erythropoiesis, and lifelong transfusion dependency. Chronic blood transfusions improve survival but lead to progressive iron overload affecting the liver, heart, endocrine glands, and growth. India carries a significant burden of beta thalassemia, with approximately 10,000 affected children born annually. Growth retardation, delayed puberty, hepatic dysfunction, and endocrine abnormalities remain major complications despite improved treatment modalities. The present study was undertaken to evaluate the clinical profile, growth abnormalities, transfusion burden, and biochemical derangements in pediatric patients with beta thalassemia major attending a tertiary care hematology clinic.

MATERIALS AND METHODS

Study Design: Cross-sectional descriptive study.
Study Setting: Pediatric Hematology Unit/Day Care Centre of a tertiary care teaching hospital.
Study Duration: 15–18 months.
Study Population: Children aged 1–18 years diagnosed with beta thalassemia major by HPLC and receiving regular blood transfusions.
Sample Size: 36 patients.

Inclusion Criteria

* Diagnosed beta thalassemia major
* Age 1–18 years
* Receiving regular transfusions
* Consent obtained from parents/guardians

Exclusion Criteria

* Other chronic hemolytic anemias
* Sickle-thalassemia syndromes
* Severe comorbid conditions affecting growth

Data Collection: Detailed demographic, clinical, anthropometric, and laboratory data were collected using a structured proforma.

Anthropometric Assessment: Growth parameters were assessed using WHO growth charts: * Height-for-age * Weight-for-age * BMI-for-age

Laboratory Investigations * Complete blood count (CBC) * Serum ferritin * Liver function tests * Renal function tests * Viral markers (HBsAg, HCV, HIV)

Statistical Analysis

Data were analyzed using SPSS version 17.0. Descriptive statistics were expressed as mean ± standard deviation and percentages. Correlation analysis was performed between serum ferritin and growth parameters.

RESULTS

Demographic Profile

The mean age of the study population was 8.36 ± 4.30 years. Male predominance was noted (58.3%).

Variable	Findings
Mean age	8.36 ± 4.30 years
Male	58.3%
Female	41.7%
Diagnosed before 1 year	88.9%
Consanguinity present	66.7%

Clinical Feature	Percentage
Anemia	100%
Hepatomegaly	88.9%
Splenomegaly	83.3%
Jaundice	72.2%
Facial dysmorphism	77.8%
History of splenectomy	5.6%

Growth and Nutritional Status

Growth abnormalities were observed in 63.9% patients.

Growth Parameter	Percentage
Stunting	38.9%
Underweight	50%
Thinness (Low BMI)	27.8%
Any growth abnormality	63.9%

Children with growth abnormalities had significantly higher serum ferritin levels compared to children with normal anthropometric indices.

Transfusion and chelation status.

Parameter	Findings
Mean transfusions	65.3 ± 33.6
Regular chelation	13.9%
Irregular chelation	86.1%
Mean serum ferritin	3413.3 ± 1962.8 ng/mL

Poor chelation compliance was strongly associated with elevated ferritin levels and hepatic dysfunction.

Biochemical Parameters

Liver enzymes were elevated in multiple patients, suggesting hepatic involvement secondary to iron overload. Renal parameters remained largely within normal limits.

Viral marker screening showed:

* Negative: 94.4%
* Positive: 5.6%

DISCUSSION

The present study highlights the significant burden of growth failure and iron overload in children with beta thalassemia major. The mean age of 8.36 years reflects prolonged exposure to transfusion-related iron overload and its complications.

Male predominance observed in this study may reflect sociocultural healthcare-seeking patterns rather than true disease prevalence. A high rate of parental consanguinity (66.7%) reinforces the strong genetic basis of the disease and emphasizes the need for premarital and antenatal screening programs.

Growth impairment was one of the most important findings, with nearly two-thirds of children showing abnormal anthropometry. Similar findings have been reported in previous Indian studies. Chronic anemia, nutritional deficiencies, endocrine dysfunction, and iron toxicity contribute to poor growth outcomes.

The mean serum ferritin level was markedly elevated, reflecting substantial iron burden. Poor compliance to chelation therapy remains a major challenge due to cost, accessibility, and side effects. Elevated ferritin levels showed association with stunting, underweight status, and abnormal BMI, indicating the impact of iron overload on growth and metabolism.

Hepatic dysfunction was more evident than renal involvement, suggesting earlier hepatic injury due to iron deposition.

Limitations :

* Single-center study with limited sample size
* Cross-sectional design prevented long-term follow-up assessment
* Endocrine evaluation and MRI T2* iron quantification were not performed
* Socioeconomic and detailed chelation regimen data were not comprehensively analyzed

CONCLUSION

Beta thalassemia major continues to impose a substantial clinical and nutritional burden in paediatric patients. Growth abnormalities, iron overload, and poor chelation compliance are highly prevalent despite regular transfusion support. Routine growth monitoring, improved adherence to iron chelation therapy, early endocrine evaluation, and comprehensive multidisciplinary care are crucial for improving long-term outcomes and quality of life in these children.

Acknowledgement: The authors would like to acknowledge the Department of Pediatrics, SMIMER, Surat, and all patients and caregivers who participated in the study.

Conflict of Interest: None declared.

Funding: No external funding.

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