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Clinical Profile of Beta Thalassemia and Iron Overload Status with Special Reference to Serum Ferritin - A Cross Sectional Observational Study in a Tertiary Care Hospital

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ABSTRACT

Introduction: Beta-thalassemia is a hereditary blood disorder characterized by defective hemoglobin synthesis, leading to chronic anemia and its associated complications. It is especially prevalent in malaria-endemic regions, including India. Patients often require frequent blood transfusions, predisposing them to iron overload, a significant cause of morbidity and mortality. This study focuses on understanding the clinical presentation of beta-thalassemia and evaluating iron overload status using serum ferritin levels. **Objective:** To assess the clinical profile of patients diagnosed with beta-thalassemia and determine their iron overload status with special reference to serum ferritin concentrations. **Methods:** This hospital-based, cross-sectional observational study was conducted in the Department of Medicine at Tezpur Medical College and Hospital from August 2023 to July 2024. A total of 40 patients aged above 12 years, presenting with anemia and diagnosed with thalassemia, were included. Exclusion criteria were applied to rule out confounding causes of anemia. Clinical data were collected using structured questionnaires, and laboratory investigations included hemoglobin analysis via HPLC and serum ferritin estimation**. Results:** The majority of patients (62.5%) were male, with the highest incidence in the 12–20 year age group. Weakness, pallor, and splenomegaly were the most common symptoms. The frequency of blood transfusion varied widely, with most patients receiving transfusions every 26–30 days. Serum ferritin levels ranged from 251–500 ng/mL in most cases, with 10 patients exhibiting levels >1000 ng/mL, indicating significant iron overload. **Conclusion:** Thalassemia predominantly affects adolescents and males. Frequent transfusions contribute to iron overload, necessitating regular monitoring of serum ferritin to guide chelation therapy.

INTRODUCTION

Cooley and Lee first described in 1925, a severe form of anemia that was associated with splenomegaly and bone changes and Whipple coined the phrase thalassic anemia and then condensed it into thalassemia.[1] Thalassemia syndromes are heterogeneous group of disorders caused by inherited mutations that results in decreased synthesis of either the α-globin or β- globin chains of adult haemoglobin Hb (α2β2), leading to anemia, tissue hypoxia and red cell hemolysis related to the imbalance in globin chain synthesis.[2] Two main classes of thalassemia are α-thalassemia and β-thalassemia which results from decreased synthesis of α and β globin chains respectively. β-thalassemia is prevalent mediter-ranean countries, Middle East, Central Asia, India, Southern China, and the north coast of Africa and in South America. It is common in malaria endemic areas in the world. About 40,000 β-thalassemia patients are born yearly [2,3].

Mutations that lead to complete absence of β-globin chain are known as β0 thalassemia. β+ thalassemia mutation cause mild to moderate reductions in β-globin chain synthesis. β-thalassemia major and intermedia are now categorized as transfusion-dependent and non-transfusion-dependent based on the number and frequency of transfusions required to sustain a good quality of life. β-thalassemia can also be associated with other hemoglobin abnormalities e.g.; HbS/β-thalassemia, HbC/β-thalassemia, HbE /β-thalassemia [3].

α-thalassemia traits are found in population with high incidence of malaria as it is thought to be having a protective effect against malaria [3]. Carriers of the most common α-thalassemia chromosomes are found in 5-80% population in tropical and subtropical regions of Africa, the Middle East, India, Southern China. HbH disease, the chief clinically important variant, is most commonly found in Southern China and Southern Asia [3].

Normal diploid individuals contain 4 α-globin genes. Deletion or poor expression of one or two genes results in formation of thalassemia trait [3]. Three gene deletion or malfunction results in HbH disease [3]. If there is no functional gene, Hb Bart's hydrops fetalis result. Besides these, α-thalassemia/intellectual disability syndromes (ATR-16)(ATR-X), α-thalassemia with myelodysplasia (ATMDS) are also included in the classification. hundreds of different sized deletions and rarer point mutations affect the production of α globin and the magnitude of imbalanced globin synthesis and because of this many different variations of the common α-thalassemia syndromes are found [3].

Relative excess of α- chains in β-thalassemia damages red cells and their precursors leading to profound anemia. But in α-thalassemia the excess β-chains form β4 molecules (haemoglobin H), which is soluble and does not precipitate in marrow. However it is unstable and precipitates in older red cells. These syndromes lead to diverse clinical profile that results from both the disease itself and also as consequences of the treatment, such as iron overload from repeated blood transfusions.

In patients who receive inadequate treatment, severe anemia leading to bone marrow expansion; hepatosplenomegaly; iron accumulation in liver, heart, and endocrine organs; pulmonary hypertension and thromboembolic disease can occur [3]. Mothers of infants with Hb Bart's hydrops fetalis have history of stillbirth and may develop preeclampsia, polyhydramnios, and antepartum hemorrhage and have difficult labor and delivery [3].

AIMS AND OBJECTIVES

- To study the clinical profile of thalassemia in patients presenting with anemia, attending department of Medicine, Tezpur Medical College and Hospital.
- To estimate the iron overload status in thalassemia patient with reference to serum ferritin level.

MATERIALS AND METHODS

Place of study: Department of medicine, Tezpur Medical College and Hospital

Study Design: Hospital Based Observational study.

Study Population: All patients attending TMCH following the inclusion and exclusion criteria.

Sample size: In a period from 1st August 2023 to 31st July 2024, a total of 40 patients attending OPD and Emergency (including admitted patients in medicine wards) were diagnosed with Thalassemia.

The current study followed the WHO criteria for laboratory diagnosis of anemia and included anemia cases with haemoglobin level of less than 12 g/dL in males between 12-14 years of age, less than 13 g/dL in males above 14 years of age and less than 12 g/dL in females [4].

Inclusion criteria:

1) Patients presenting with anemia and diagnosed to have thalassemia or follow up cases of thalassemia attending OPD and Emergency (including admitted patients in medicine wards) at TMCH

2) Patients above 12 years of age.

Exclusion criteria:

1) Age less than 12 years

2) Pregnant females.

3) Patients having anemia due to chronic kidney disease, chronic liver disease, malignancy and chronic inflammatory diseases like Crohn's disease , HIV/AIDS, Rheumatoid arthritis, etc.

4) Patients who have been transfused blood in the past 3 months except those who are pre diagnosed with hemoglobinopathies as evidenced by previous records.

A previously set questionnaire is filled after thorough history taking, general and systemic examination, with informed consent from the patient/next of kin. Relevant investigations (biochemical, cytological and radiological) were done to exclude CKD, CLD and other causes leading to anemia. Cation –exchange HPLC is the method of choice for initial screening of Hb variants and for accurate quantification of HbA2 and HbF concentrations. The Bio- Rad D- 10 is an automated cation exchange HPLC instrument that has been used to quantify HbA2, HbF, HbA along with screening haemoglobin variants like α and β thalassemia ,HbS, HbD, HbE and HbC in a single , highly reproducible system.

Normal

HbA - 95% - 98%

HbA2 - 1.5% - 3.5%

HbF - < 1%

RESULT

During the study it was seen that 12-20 years of age had the maximum number of patients (25) followed by age group of 20-30 years(10).It was seen that frequency of blood transfusion was more during 26-30 days (13) followed by 31-45 days (9).In 13 patients serum ferritin level was seen between the range of 251-500 ng/ml and in 10 number of patients ferritin level was >1000 ng/ml despite treatment. Mean serum ferritin levels was 938ng/ml seen in the 17-21 years of age followed by 884ng/ml in 22-26 years of age group. Generalized weakness was found to be the most common complaint in patients (30). On examination pallor was the most common finding (20) followed by splenomegaly (12).

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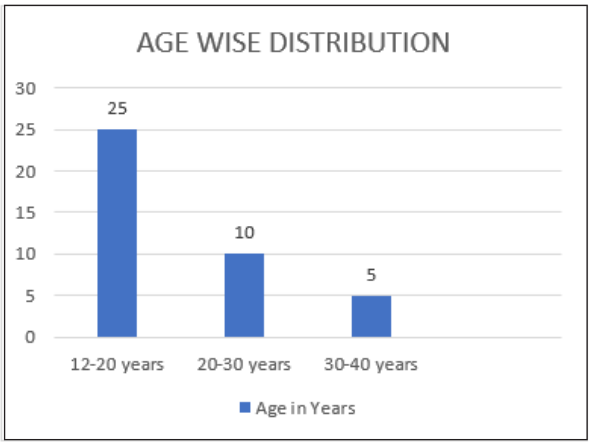

Figure 1: Demographic Profile

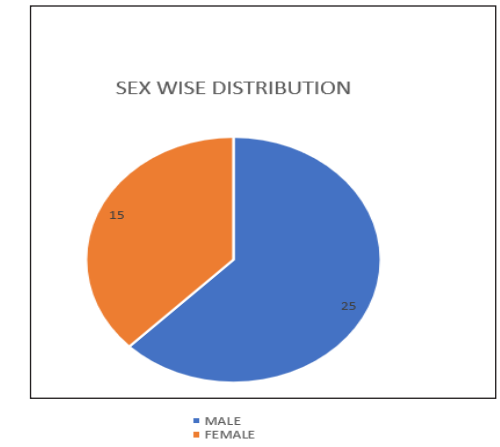

Figure 2: Male preponderance during the study -25 males(62.5%) vs 15 females(37.5%).

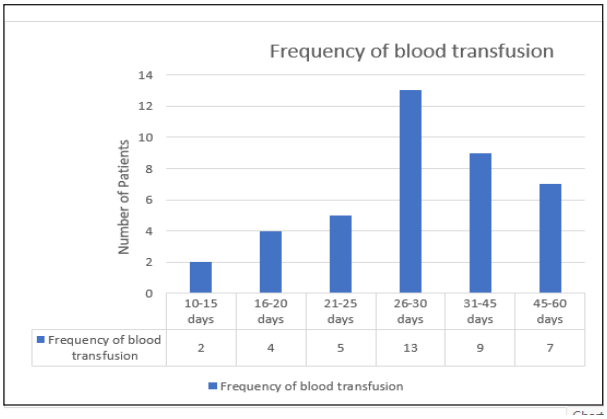

Figure 3: Frequency of Blood Transfusion

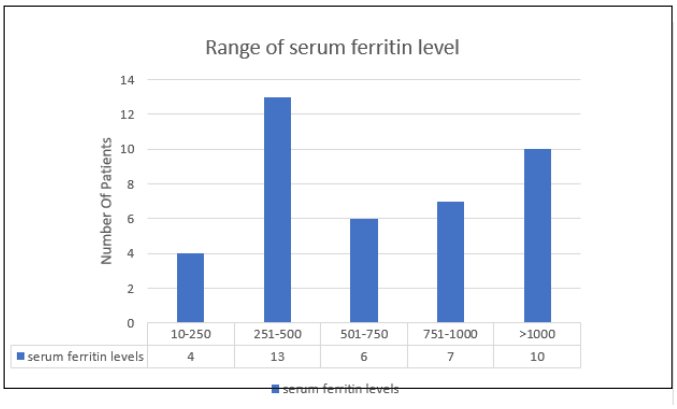

Figure 4: Range of Serum Ferritin Level

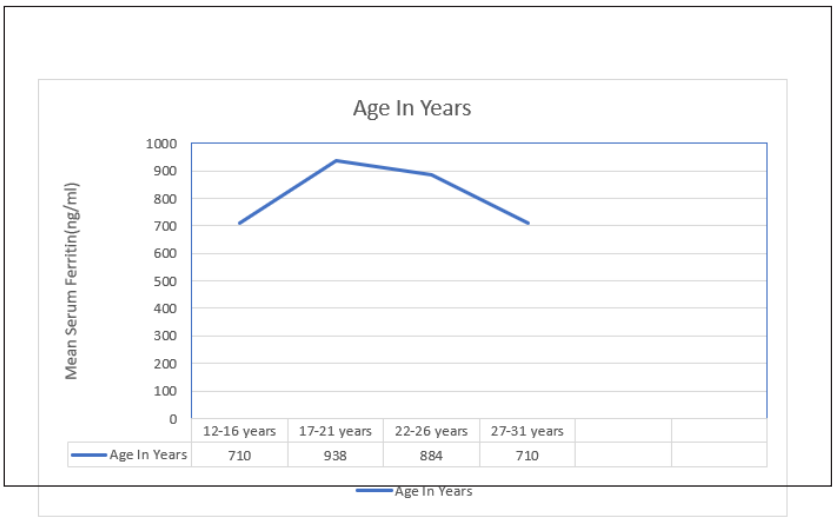

Figure 5: Distribution of Ferritin Level Among Different Age

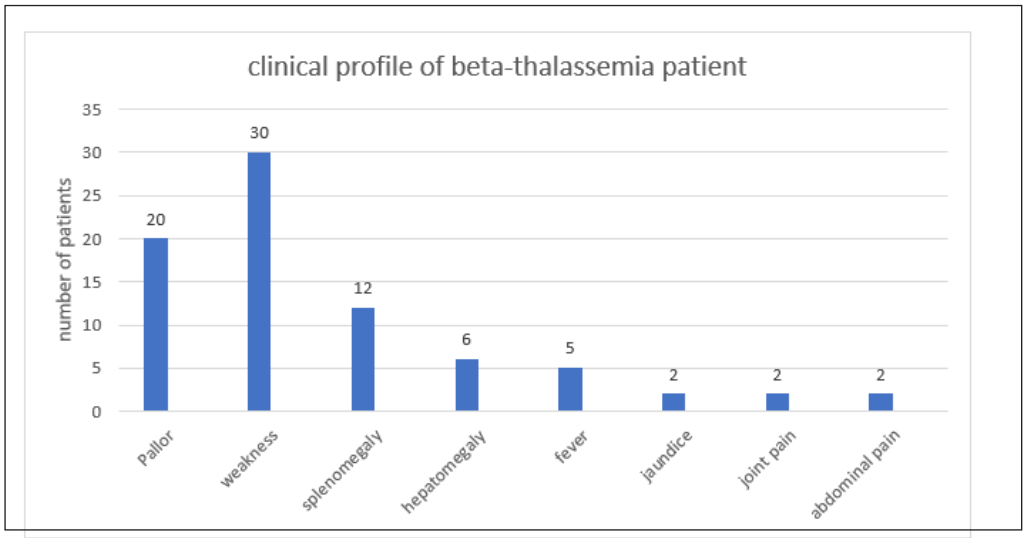

Figure 6: Clinical Profile of Beta Thalassemia Patients

DISCUSSION

The objective of our study is to evaluate the clinical profile of Beta-thalassemia and assess iron overload status, with a particular emphasis on serum ferritin levels, using a cross-sectional observational approach. This study offers a comprehensive overview of thalassemia patients, highlighting their demographics, treatment patterns-especially the frequency of blood transfusions-and key biochemical markers such as serum ferritin levels.

In our study, the clinical profile of thalassemia in patients presenting with anemia revealed that the majority of individuals (25) were in the 12–20 years age group, followed by 10 in the 20–30 years group, and only 5 in the 30-40 years group, indicating a decreasing trend in representation with increasing age. This pattern suggests that thalassemia patients tend to present predominantly during adolescence, with fewer cases observed as age progresses. A similar trend was observed by Trehan et al (2015), who reported that the mean age of children with thalassemia was 13.2 ± 9.7 months, highlighting that symptoms and diagnosis frequently occur early in life. Furthermore, Fucharoen S et al found that most patients exhibited clinical symptoms by 10 years of age.

While our study captures a slightly older demographic, it aligns with previous findings in emphasizing the importance of early manifestation and diagnosis in thalassemia. These studies underscore the progressive burden of thalassemia across different age groups, where early-onset cases, if untreated or inadequately managed, may lead to continued medical complications into adolescence and early adulthood, as observed in our study cohort [5,6].

In our study, the sex-wise distribution of patients shows that 62% were male and 38% were female, indicating a higher prevalence among males. Similarly, Palit S et al.(2012) reported a distribution of 60.4% male and 39.6% female, Hassanzadeh J et al observed that majority of thalassemia patients were male,reinforcing the trend that thalassemia tends to be more prevalent among males across different populations [7,8].

In our study, it was observed that the frequency of blood transfusions varied among patients, with the highest number (13) receiving transfusions every 26–30 days.

A moderate number (9) received transfusions every 31–45 days, while 7 patients had transfusions every 45–60 days. Fewer patients required transfusions at shorter intervals, with 2 patients receiving transfusions every 10–15 days and 4 patients every 16–20 days, indicating variability in transfusion needs based on individual clinical conditions. Additionally, the study found that the majority of patients (13) had serum ferritin levels in the 251–500 ng/ml range, followed by 10 patients with levels exceeding 1000 ng/mL, indicating significant iron overload in some cases. Fewer patients were recorded in the 10–250 ng/ml range (4), 501–750 ng/ml range (6), and 751–1000 ng/ml range (7), demonstrating variability in iron storage across the group. Furthermore, when analyzing serum ferritin levels across different age groups, the highest levels were observed in the 17–21 years age group, peaking at 938 ng/ml, followed by a decline to 884 ng/ml in the 22–26 years group and further decreasing to 710 ng/ml in the 27–31 years group. Similarly, Belhoul KM et al (2012) and Hashemi A et al (2011) also reported fluctuations in ferritin levels among thalassemia patients, consistent with the variations observed in our study. These findings emphasize that both blood transfusion frequency and iron overload, as reflected by ferritin levels, are highly variable among thalassemia patients, influenced by factors such as age, clinical condition, and treatment patterns [9,10].

In our study, the clinical profile of beta-thalassemia patients revealed that weakness was the most common symptom, affecting 30 patients, followed by pallor in 20 patients and splenomegaly in 12 patients. Less frequent symptoms included hepatomegaly (6), fever (5), and rarer occurrences of jaundice, joint pain, and abdominal pain (2 each). Similarly, Kumar D et al (2019) and Parakh N et al(2019) also reported moderate to severe anemia. emphasizing that weakness is the most prevalent symptom among thalassemia patients, consistent with the findings of our study [11,12].

CONCLUSION

This study emphasizes the burden of thalassemia on patients, especially concerning transfusion frequency and iron overload. The data suggest a need for enhanced management protocols, including regular monitoring of serum ferritin levels and timely administration of chelation therapy to prevent long-term complications. Moreover, the young age of most patients signals the need for early intervention strategies and possibly greater investment in public health campaigns for thalassemia awareness and genetic counseling.

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