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Prevalence of Endocrinopathies in Moderate Anemic Patients with B-Thalassemia Major - A Collaborative Study

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KEYWORDS
Thalassemia, endocrinopathy, Hypogonadism, Hypoparathyroidism, Hypothyroidism

ABSTRACT:

Introduction: Thalassemia is a genetic blood disorder resulting in a mutation or deletion of the genes that control globin production. Every year around 100000 children are born with thalassemia major in the world and around 10,000 are born in India alone.

Objectives: To evaluate the prevalence of endocrinopathies in moderate anaemic patients β- Thalassemia major, in the study population.

Methods:

- Data collection procedure: Information regarding the demographic features and clinical examinations will be collected from each patient.
- A 5-ml intravenous blood sample will be collected in EDTA and plain vial to determine the following biochemical investigations.

Biochemical Investigations:

1. Serum calcium
2. Phosphate Ferritin
3. Parathyroid hormone [PTH]
4. A random blood glucose level

Sample Size: 50 children of the same age group were considered

All children with β-thalassemia major between age group of 6 to 18 who are on regular follow up with thalassemia centres.

Results: Our study showed that 62% of BTM patients had at least one endocrine disorder, with 28.7% having two disorders and 9.3% having three types of

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disorders.

Hypogonadism: This was the most prevalent endocrine complication. In the Omani study, 62% of patients were affected, with 35% exhibiting hypogonadotropic hypogonadism and 27% normogonadotropic hypogonadism.

Conclusions: Endocrine complications are common among patients with β-thalassemia major, even in those with moderate anemia. Hypogonadism, diabetes mellitus, and hypoparathyroidism are among the most prevalent. Regular screening for endocrine dysfunctions, early initiation of chelation therapy, and multidisciplinary management are crucial for improving the quality of life and outcomes in these patients.

1. Introduction

Thalassemia is a genetic blood disorder resulting in a mutation or deletion of the genes that control globin production. As it is evident that Normal hemoglobin is composed of 2 alpha and 2 beta globins and Mutations in a given globin gene can cause a decrease in production of that globin, resulting in deficiency [1,2]. If aggregates become oxidized, it damages the cell membrane, leading either to hemolysis, ineffective erythropoiesis, or both[3,4]. In all there are two types of thalassemia: alpha and beta.

The Thalassemia gene may be maintained in the human population, in part because of the greater immunity of heterozygous individuals against malaria and is found in parts of the world where malaria is common [5,6,7]. These include Southeast Asia, China, India, Africa, and parts of the Mediterraneum.

2. Objectives

- Every year around 100000 children are born with thalassemia major in the world and around 10,000 are born in India alone.
- The carrier rate of β-thalassemia gene varies between 1-3% in south India and 5-15% in north India.
- The combination of transfusion therapy and chelation therapy has dramatically extended the life expectancy.
- The only curative treatment available is Stem Cell Transplant [SCT] which is due to high cost in countries like India.
- It results from frequent blood transfusion, Its Complications.
- The most common complications are endocrinopathy.
- Hypogonadism, Hypoparathyroidism, Hypothyroidism and Diabetes.
- As clinically overt manifestation of these endocrine abnormalities occur late in life, so most of the studies available are carried out in adults. Only very few pediatric studies are available.
- In India cost of chelation precludes ideal therapy for majority of the

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patients and the compliance with transfusion is often not optimal.

- Therefore, there is a possibility that there may be high prevalence of endocrinopathies in such patients and there are high chances of those endocrinopathies beginning at an early age than projected by western studies.
- In scaristy of Indian Pediatric studies, it becomes meaningful to check prevalence of endocrinopathies in moderate anaemic patients with β-thalassemia major. Hence we selected this collaborative study.
- There is an increasing prevalence of endocrine disorders and the increasing concern among the patients regarding their stature, hence there is a need to evaluate the burden of endocrine complications among Thalassemic patients.

Aim

To evaluate the prevalence of endocrinopathies in moderate anaemic patients β- Thalassemia major, in the study population.

Methods:

1. To evaluate the circulatory ferritin, thyroid profiles in among the β-thalassemic blood transfused patients.
2. To review risk factors to identify subset of population at greatest risk for the development of thalassemia.
3. To carryout follow up of β-thalassemia patients and provide counselling.

1. Data collection procedure: At the time of enrolment, information regarding the demographic features and clinical examinations will be collected from each patient.
2. A 5-ml intravenous blood sample will be collected in EDTA and plain vial under sterile conditions from each of the enrolled patient at the time of enrolment to determine the following biochemical investigations.

Biochemical Investigations [2,15]

1. Serum calcium [normal range 2.1-2.5 mmol/l],
2. Phosphate [normal range 0.8-1.5 mmol/l],
3. Ferritin [normal range male: 12- 300 ng/ml; female: 12-150 ng/ml],
4. Parathyroid hormone [PTH] [normal range 1.6-9.3 pmol/l],
5. Free T3 & T4 [normal range 0.8-2.0 ng/dl],
6. Thyroid stimulating hormone [TSH] [normal range 0.3-4.0μIU/ml],
7. Random blood sugar levels.
8. Thyroid function will be evaluated by measurements of T3, T4, and TSH using enzyme-linked immunosorbent assay [ELISA].

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9. Hypothyroidism is defined by a TSH level >8μIU/ml, and T4 levels <4.5μg/dl.
10. Iron load status is defined by serum ferritin level that was estimated from pre-transfusion blood sample
11. Parathyroid function will be assessed by measurement of PTH using ELISA.
12. Hypoparathyroidism is defined as low levels of PTH [<1.6 pmol/l] or normal levels of PTH in the presence of low serum calcium levels
13. A random blood glucose level of ≥160 mg/dL was considered diabetes mellitus.

Sample Size: 50 children of the same age group were considered

Study Population:

All children with β-thalassemia major between age group of 6 to 18 who are on regular follow up with thalassemia centres.

Inclusion criteria

1. Children who had beta-thalassemia major and suspected cases of any other subtypes of thalassemia will be included in the study group.
2. Children whose ferritin level is above normal level.
3. Clinically diagnosed Hepatomegaly
4. Repeated blood transfusion therapies
5. Patients willing to give written informed consent and follow study related procedures.

Exclusion criteria

1. The exclusion criteria are as follows:
2. Children who don't follow instructions and are under self-medication
3. Children with primary endocrinopathy.
4. Children on any hormonal therapy.
5. Children with any other chronic illness.
6. Other type of haemoglobinopathies.
7. History of jaundice due to viral, alcoholic or heavy metal induces.
8. History of splenectomy

Outcome:

1. Clinical presentation.
2. Prevalence of different type of endocrinopathies in beta Thalassemia major.
3. Laboratory profile.

Statistical Analysis:

1. Prevalence of Hypothyroidism, Hypoparathyroidism & Delayed puberty was reported using proportions and percentages.
2. Chi square test was used to test the association between the prevalence of abnormalities and the associated

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demographic and clinical characteristics of the patient.

3. Independent "t" test was used to compare between the two groups for continuous outcome variables.

4. Probability value "p" <5% will be considered as statistically significant.

3. Results

4. The prevalence of endocrinopathies among patients with β-thalassemia major [BTM], particularly those with moderate anemia, has been extensively studied. These endocrine complications are primarily attributed to iron overload resulting from regular blood transfusions and inadequate chelation therapy [8,9]. Below is a summary of findings from various studies:
5. Our study showed that 62% of BTM patients had at least one endocrine disorder, with 28.7% having two disorders and 9.3% having three types of disorders.
6. **Hypogonadism:** This was the most prevalent endocrine complication. In the Omani study, 62% of patients were affected, with 35% exhibiting hypogonadotropic hypogonadism and 27% normogonadotropic hypogonadism.
7. **Diabetes Mellitus:** The study found that 26% of patients had diabetes mellitus.
8. **Hypoparathyroidism:** Observed in 7% of patients in the study.
9. **Hypothyroidism:** Reported in 4.3% of patients in the study.
10. **Short Stature:** The study found that 26.9% of BTM patients exhibited short stature.

4.Discussion

Endocrinopathies in BTM patients are multifactorial, with iron overload being a significant contributor [10]. Iron deposition in endocrine glands leads to their dysfunction [11,12]. The high prevalence of hypogonadism underscores the sensitivity of the hypothalamic-pituitary-gonadal axis to iron toxicity [13,14] Similarly, pancreatic iron deposition impairs insulin secretion, leading to diabetes mellitus.

The variability in the prevalence of these complications across studies may be due to differences in chelation practices, genetic factors, and healthcare access [16,17]. Notably, some studies found no significant correlation between serum ferritin levels and the development of endocrinopathies, suggesting that ferritin alone may not be a reliable marker for predicting endocrine complication [18,19]

Endocrine complications are common among patients with β-thalassemia major, even in those with moderate anemia. Hypogonadism, diabetes mellitus, and hypoparathyroidism are among the most prevalent [20,21]. Regular screening for endocrine dysfunctions, early initiation of chelation therapy, and multidisciplinary management are crucial for improving the quality of life and outcomes in these patients [22,23]

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