

PREVALENCE AND PROFILE OF BETA-THALASSEMIA: A PROSPECTIVE ANALYSIS AT A TERTIARY CARE CENTER

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ABSTRACT

Objective: The objective of the present investigation was to identify the hematological profile of β -thalassemia trait.

Methods: The cross-sectional investigation, which included 186 patients, was conducted in a tertiary care hospital's hematology unit of the pathology department from November 2012 to May 2014.

Results: About 73.11% of the study population was in the 21–50 years age group followed by 18.27% in the age group 0–20 years. About 8.62% of the study population was in between 51 and 75 years of age. Eighty-six (46.23%) men and 100 (53.77%) women made up the study's sample. In our study, India had a 19% overall prevalence of the thalassemia trait. In the research, we conducted we found that those with the beta-thalassemia trait (BTT) had much lower levels of red blood cells (RBC) parameters except RBC count which is slightly raised than those without the trait.

Conclusion: There is a significant prevalence of BTT in our population. This information will prove helpful to health policy managers in making informed decisions on how to design accessible screening programs for BTT.

Keywords: Beta-thalassemia, Hemoglobinopathies, Hematological parameters, Prevalence.

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INTRODUCTION

Hemoglobinopathies are clinical syndromes resulting from disorders of hemoglobin synthesis [1]. Hemoglobinopathies are common and pose greater health and economic problems in our society. They are also a psychological burden to families. They commonly present with the symptoms of anemia, hepatosplenomegaly, and profound weakness. Many like sickle cell disease and thalassemia major present with severe obvious clinical symptoms. Hence, diagnosed early and treated. However, some such as β -thalassemia trait and hemoglobin E (HBE) are asymptomatic or present with anemia. Many such patients are not aware of their carrier state.

Beta-thalassemia, a genetic disorder, is one of the most common hemoglobinopathies. Mutation in beta-globin chains at chromosome 11 leads to abnormal hemoglobin synthesis. In India, 30 million people carry the defective gene (12% of the world), and approximately 65,000–67,000 patients with beta-thalassemia are thought to exist [2]. According to reports, 3.3% of Indians have the beta-thalassemia trait (BTT) [3]. The mean prevalence is 5.5% among school-going children. Beta-thalassemia is more prevalent among some Indian communities, including Gujaratis, Sindhis, Bengalis, and Punjabis; the frequency ranges from 1% to 17% [4]. The incidence of the BTT varies from 1% to 3% in southern India to between 3% and 15% in northern India.

The range of β -thalassemias is broad, from thalassemia minor, which results in moderate hypochromic microcytic anemia without overt clinical symptoms, to β -thalassemia major (β -TM) [5], which is marked by severe anemia from an early age and requires blood transfusions. A term β -thalassemia intermedia, which was created to represent people with symptoms that are neither too severe to be designated thalassemia major nor too moderate to be considered β -thalassemia minor, is in the middle. TI is a member of the non-transfusion-dependent thalassemia group of thalassemias, which also comprises mild-to-moderate HbE/thalassemia and α -TI (HbH disease) [6].

When beta-globin chains are absent, alpha-chains combine with gamma- and delta-chains instead, resulting in a beta-thalassemia phenotype. This causes a rise in hemoglobin A2 (HbA2) (a kind of blood that contains delta-globins) and frequently HbF as well (which contains gamma). For those with risk factors, an increase in HbA2 is sufficient to reliably identify the beta-thalassemia trait (BTT), and a HbA2 above 3.5% is sufficient to do so. It is not always possible to diagnose BTT using an elevated HbA2 level; for example, HbA2 may be normal in some patients with BTT (for example, because iron deficiency suppresses HbA2 levels), and a falsely increased HbA2 level can occur in certain people who do not have the BTT (e.g., megaloblastic anemia, sickle cell trait, hyperthyroidism, or antiretroviral therapy) [7].

The key to preventing this significant health issue is the reliable test method for identifying asymptomatic carriers. Given the iron deficiency anemia high prevalence in India, it is crucial to clinically distinguish among iron deficiency anemia and BTT due to their divergent clinical manifestations [8]. Thus, the objective of the present investigation was to identify the clinical profile of thalassemia.

METHODS

The cross-sectional study consisted of 186 cases and was carried out in the hematology section of the department of pathology in a tertiary care hospital from November 2012 to May 2014.

Inclusion criteria

The following criteria were included in the study:

1. Routine patients coming to outpatient department (OPD) without any major illness such as chronic diseases or malignancies
2. Routine patients coming to OPD without hepatosplenomegaly
3. Routine patients coming to OPD without any history of major trauma or surgery
4. Family members of patients having thalassemia.

Exclusion criteria

The following criteria were excluded from the study:

1. Hemolyzed samples
2. Clotted samples
3. Patient was transfused in the past 4 weeks
4. Un-cooperative and unwilling patients
5. Pregnant females.

Patient consent was taken. Fresh blood samples were taken in the OPD of our tertiary care hospital and placed in an EDTA vacutainer. Samples were kept at 4–8 °C and examined by high-performance liquid chromatography (HPLC) in batches within 5 days (Bio-Rad Variant). The components of a mixture are separated, each component is identified, and each component is quantified using the analytical chemistry method known as HPLC. HbA2 levels were calculated, and then, the samples were divided into two groups; one group with HbA2 3.5–7% and another group with HbA2 <3.5%.

Statistical analysis

For statistical analysis, data have been entered into a Microsoft® Excel workbook and exported into SPSS v21.0 (IBM, USA). Frequency and percentages were used to express categorical variables. The mean and standard deviation of quantitative variables were used to compare them using independent t-tests. Statistics were judged significant at $p < 0.05$.

OBSERVATIONS AND RESULTS

The study consisted of 186 cases (with no history of blood transfusion, no hepatosplenomegaly, and no major illness). HPLC was performed on them to identify the HbA2 levels. The cases were further divided based on HbA2 in the groups with HbA2 level of 3.5–7% and a level of <3.5%. These groups were further subdivided based on various laboratory parameters into two groups. The cutoff values for laboratory parameters were derived from Indian studies and are as follows: Hb value of 11.5 g/dL, red blood cells (RBC) count of $5 \times 10^6/\mu\text{L}$, MCV of 76 fL, MCH of 27 pg, MCHC of 31.8 g/dL, and red cell distribution width (RDW) of 13.6%. The results were recorded in a tabulated form as follows.

Table 1 profile reveals that 73.11% of the study population was in the 21–50 years age group followed by 18.27% in the age group 0–20 years. About 8.62% of the study population was in between 51 and 75 years of age (Fig. 1).

The population under research for this study consisted of 100 females (53.77%) and 86 males (46.23%) in total (Fig. 2).

Above Table 2 shows mean RBC count (4.84×10^6 cells/ μL) is significantly more in the group having HbA2 3.5–7% (β -thalassemia trait) when compared to the control (4.25×10^6 cells/ μL), while mean hemoglobin (10.4 g/dL), mean MCV (72.14 fL), mean MCH (22.38 pg), and mean MCHC (30.73 g/dL) were significantly less in the group having HbA2 3.5–7% (β -thalassemia trait) when compared to control group. RDW (14.9%) in the group having HbA2 3.5–7% (β -thalassemia trait) was mildly decreased when compared to the control (15.81%) and the difference was not significant.

DISCUSSION

The hemoglobinopathies, in particular the β -thalassemias, as well as their interaction with hemoglobin S and HBE, are a serious public health issue in India and are a major contributor to morbidity and mortality rates. Hemoglobinopathies are most common in ethnic populations from Africa. However, due to migration, it is now seen in other parts of the world too. The different hemoglobinopathies prevalence varies in different regions.

It is believed that roughly 7% of people throughout the world are carriers of hemoglobinopathy (the number might range anywhere from 1% to 20%). There have been reports of incidences of beta-thalassemia

Table 1: Profile of age and sex distribution

Age group (years)	Male	Female	Total (%)
0-20	14	20	34 (18.27)
21-50	65	71	136 (73.11)
51-75	7	9	16 (8.62)
Total	86	100	186 (100)

Table 2: Mean pattern of laboratory parameters between groups having trait and control

Parameters	Mean \pm SD		p
	Trait	Control	
Hb (g/dL)	10.4 \pm 1.37	11.5 \pm 2.16	0.004
RBC ($\times 10^6$ cells/ μL)	4.84 \pm 0.80	4.25 \pm 0.89	0.000
MCV (fL)	72.14 \pm 10.9	81.40 \pm 12.7	0.000
MCH (pg)	22.38 \pm 5.46	27.97 \pm 7.37	0.000
MCHC (g/dL)	30.73 \pm 3.32	33.50 \pm 3.71	0.000
RDW (%)	14.9 \pm 3.59	15.81 \pm 2.74	0.096

SD: Standard deviation, RBC: Red blood cells, Hb: Hemoglobin, RDW: Red cell distribution width

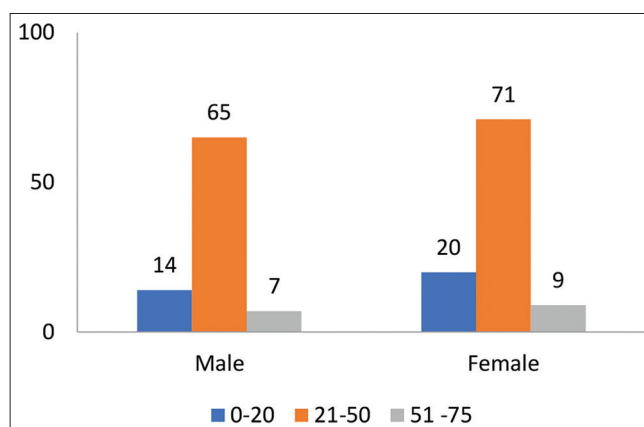


Fig. 1: Age-wise distribution

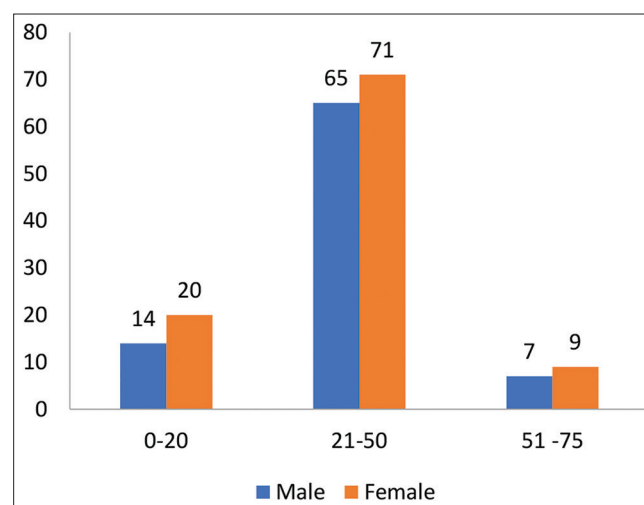


Fig. 2: Gender-wise distribution

in India ranging from <1 to 17%, with an average prevalence of 3.3% in the general population. The most frequent form of the disease is the BTT. There is a significant disparity in the prevalence of BTT throughout India's various regions: 6.5% in the state of Punjab, 8.4% in the state of Tamil Nadu, 4.3% in southern India, and 3.5% in the state of Bengal [9].

In different Indian states, the prevalence of the BTT ranged from 1.48% to 3.64%, totaling 2.78% [10]. According to the findings of our research, the prevalence of the BTT over the entire country of India is 19%.

In our study, the BTT had significantly lower levels of RBC parameters except for RBC count than the non-trait. The findings are in concordance with the study of Mondal and Mandal [11]. Kiss *et al.*, developed an algorithm that calculates the likelihood of finding beta-thalassemia carriers by taking into account both a low MCV and the ethnic background of the subject. In their database, there were 789 patients with MCVs lower than 80 fl, but only 31 patients were identified as being carriers for beta-thalassemia. They came to the conclusion that it was sufficient to discover beta-thalassemia carriers by employing MCV in conjunction with ethnic background [12].

In the study by Roth *et al.*, between β -thalassemia carriers and non-carriers, there were notable differences in each of the RBC parameters. However, there was not a significant difference in the RDW value fluctuations among the two groups of people who had Hgb levels of <90 g/L. This is likely because both groups contained mixed thalassemia carriers and IDA [13].

CONCLUSION

Hemoglobinopathies are a widespread concern all over the world, but they are particularly prevalent in our population since the incidence rate is so high. The health policy managers will find that having access to this information is helpful when it comes to making educated judgments about how to build programs for screening the cases of β -thalassemia trait and reduce the prevalence of β -TM.

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AUTHORS CONTRIBUTIONS

Study conception and design: Sheetal Mahajan, Sachin Dhule, R.V. Taksande, Data collection: Sheetal Mahajan, Analysis and interpretation of data: Sheetal Mahajan, R.V. Taksande, Drafting of manuscript: Sheetal Mahajan, Sachin Dhule.

CONFLICTS OF INTEREST

There are no conflicting interests disclosed by the authors.

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