Study on Prevalence and Severity of Thalassemia in Amravati District of Maharashtra

Keywords: thalassemia, hemoglobinopathies, prevalence, severity, distribution, frequency

ABSTRACT

Objective: The present study was undertaken to determine the prevalence and severity of thalassemia belonging to Amravati district. Methodology: All the previously registered subjects were enrolled in the study who were already screened for the RBC indices, pattern of mutation with its percentage of thalassemia sufferers. A patient interview form was prepared with informed consent. The questionnaire was prepared with a view to collect data on the disease status of the sufferers only. Results: In case of thalassemia, age cannot be regarded as significant factor. From the gender-wise distribution, the thalassemias are more common in males than in females. In the community-wise distribution, the prevalence of thalassemia in Kunbi, Muslim, Boudh, Mali communities is high. Thalassemia disorder are inherited but not always due to consanguinity. The frequency of blood transfusion depends upon the severity of the disease. The hemoglobin status can be managed with respect to the blood transfusion and the treatment adherence. As per Rh blood group typing, the frequency of beta-thalassemia is high in O Rh+ and B Rh+. 

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Submission: 23 July 2018
Accepted: 30 July 2018
Published: 30 August 2018
ABBREVIATIONS

α- alpha

β- beta

HbE- hemoglobin E

SC- schedule caste

ST- schedule tribe

INTRODUCTION

Thalassemia syndromes are the inherited disorders of alpha or beta-globin biosynthesis. Alpha-thalassemia is a quantitative deficiency of alpha globin production while beta-thalassemia is a quantitative deficiency of beta-globin production and usually, occur due to DNA mutations of the beta-globin gene cluster. The beta-globin gene families are clustered on chromosome 11 and are arranged over approximately 60,000 nucleotide bases [3]. The reduced supply of globin diminishes production of hemoglobin tetramers, causing hypochromia and microcytosis [14]. Because of the imbalance in chain synthesis, an excess of freed alpha-globin chains accumulates within erythroid cells. Aggregation, denaturation, and degradation of these chains leads to the formation of insoluble precipitates as well as hemichromes, which damage cell membranes. Membrane damage leads to ineffective erythropoiesis within the bone marrow, hemolysis of red cells within the circulation, and binding of immunoglobulin and complement components to red cell membranes, triggering loss of red cells in the spleen. The resulting anemia leads to diminished tissue oxygenation, an increase in erythropoietin levels, and further stimulation of the bone marrow. Bone marrow expansion causes skeletal deformities and osteopenia. Substances released from degenerating red cells increase iron absorption, which contributes to iron overload [16].
Beta-thalassemia is more common in Mediterranean countries, the Middle East, Central Asia, India, Southern China and countries along the north coast of Africa and in South America. The inherited disorders of hemoglobin are prevalent largely in tropical countries including India. The inherited genetic diseases of hemoglobin are controlled by a single gene that transmits from parents to offspring from one generation to another affecting more than a million people throughout the world. Sickle cell anemia (SCA) and thalassemia (βT) are such genetic disorders caused by point mutation, which are of major concern from the point of view of public health policy [2,3]. Beta-Thalassemia is a highly prevalent major monogenic single gene autosomal recessive disorder characterized by the reduced or absent expression of the beta-globin gene, leading to an imbalance of alpha and beta-globin chains [2] and it is estimated that around 300,000 to 400,000 babies with a severe hemoglobin disorder are born each year [6]. Earlier studies have shown that the overall prevalence of beta-thalassemia is 3–4 % with an estimate of around 8,000 to 10,000 new births with the major disease each year [1]. Few studies done earlier have shown that certain communities like the Sindhis, Kutchhi Bhanushalis and Punjabis from Western and Northern India have a high prevalence of beta-thalassemia (5–15%) and some population groups from the northeastern regions have a high prevalence of HbE (5–50%) [1,8]. In the case of genetic diseases, treatments are expensive, time-consuming and it may not be possible to cure the defect permanently. Although, Over the past three decades, regular blood transfusions along with folic acid supplementation and iron chelation (Desferox) have dramatically improved the quality of life and transformed

Figure 1 Flowchart of pathophysiology of beta-thalassemia [16]
thalassemia from a rapidly fatal disease in early childhood to a chronic disease compatible with prolonged life. Life expectancy of patients suffering from thalassemia varies between 25-55 years, depending on the compliance with medical treatment. Despite increased life expectancy, complications may arise [7]. Thus the prevention of beta-thalassemia is an important health issue as more and more number of children born every year in India with beta-thalassemia and they need treatment, which is expensive. Prevention can be done by public awareness, carrier screening, genetic counseling, prenatal diagnosis and selective termination of affected fetuses. Most of these children have a severe clinical presentation but are managed sub-optimally due to lack of financial resources in the majority of the families. Thus preventing the birth of affected children is the best option for India. A prerequisite for this is the knowledge of the prevalence of beta-thalassemia and other hemoglobinopathies in different regions of the country and in particular in different ethnic groups. However, there is not much information about the incidence and prevalence of thalassemia in Vidarbha region of Maharashtra. Realizing the need for awareness, this study was conducted as it may help to enlighten the future perspectives of the disease in this particular region.

MATERIALS AND METHODS

This prospective study was carried out in Amravati District of Vidarbha in Maharashtra, for the period of one year. The data of 49 subjects were recorded in a one year period on the basis of patient interview form which was prepared by considering all etiological factors with informed consent. The sample size of this study was small as the data collected was only from those subjects who were previously registered as thalassemia sufferers in the Day Care Centre of District General Hospital, Amravati. The questionnaire of thalassemia was prepared to collect data on the disease status of the sufferers only. The collected data of the sufferers was organized and arranged using MS-excel. The diseased data was further analyzed and represented graphically.

Disease screening:

All the previously registered subjects were already screened for the RBC indices, the pattern of mutation with its percentage.
RESULTS AND DISCUSSION

Following tables represent data for one year period April 2015 to March 2016 under the thalassemia management program. The data was collected from the Day Care Center of District General Hospital, Amravati.

1. Age-wise distribution:

In case of thalassemia, age cannot be regarded as a significant factor, because detection of thalassemia depends on the onset of a symptom regardless of age where family history is unknown. But all the cases belonged to the age group of 0-15 years of age (Fig 2). A similar study can be seen in Bangladesh and Maharashtra were the majority of the hemoglobinopathies belonged to neonatal to childhood period (0–15 years), followed by reproductive age group (16–45 years) [10,11].

![Age-wise distribution](image)

*Figure 2 Age-wise distribution*

2. Gender-wise distribution:

A total of 49 patients were enrolled in the study where 27 were males and 22 were females who were admitted to the district general hospital during the period 2015-2016 (Fig 3). All of the subjects were directly linked to beta-thalassemia. This data stated that in this study the male patients were more in number than the female patients. Similar data was also found earlier by Wasi et al (1985), Yagnik (1997) and Balgir (1996) and reported 65.5, 56 and 62.1 % of male patients respectively [9]. Though the difference in percentage was not significantly high, it might indicate that thalassemias are more common in males than in females.

Figure 3 Gender-wise distribution

Community-wise distribution:

In the given Fig 4 Kunbi, Muslim, Boudh, Mali communities which belong to SC, ST category exhibited the highest prevalence of beta thalassemia, while all other communities were found to be with the lowest prevalence. Similar results were seen in the previous study in north Maharashtra were thalassemia was most prevalent in ST category (79%), followed by OBC category (8%) [11].
3. Family history:

It was found that in the case of 10 out of 49 patients, their parents were close family relatives who were related by blood (Fig 5.a). Also, among 43 patients, both parents were found to be carriers (Fig 5.b). This is in compliance with the fact that like in most of the genetic diseases, thalassemia is a genetically inherited disease, but may not always due to consanguinity. But, since the study involves less number of cases along with the previous study were the incidence of beta- thalassemia was the highest in outcome of first cousin marriages (69.66 %) and the least in outcome of distant relation marriages (1.0 %), we can state that consanguinity is important in family history[12].

4. Blood group distribution:

As per Rh blood group typing, 91.83% of the subjects were Rh positive while 8.16% were unknown and no subjects were Rh negative. With respect to both ABO and Rh grouping system, the prevalence of blood group O Rh+ve and B Rh+ve was highest (53.6% and 28.57% respectively), while A Rh+ve and AB Rh+ve showed the lowest prevalence (6.12% and 4.08% respectively). The frequency of beta-thalassemia with respect to ABO and Rh systems can be shown as $\text{O}^+ > \text{B}^+ > \text{A}^+ > \text{AB}^+$ (Fig 6). Similar results were seen in the previous study of the distribution of ABO blood groups in β-thalassemia patients were the most common blood group in β-thalassemia patient is O+ve [13].

5. The frequency of Blood transfusion:

Out of 49 patients, 11 patients came for blood transfusion after every 15 days, 31 patients came on a monthly basis; whereas 2 patients came bi-monthly and 5 patients came every tri-monthly respectively (Fig 7). As the treatment of thalassemia major is blood transfusions to maintain the hemoglobin level, the frequency of blood transfusion depends upon the severity of the disease; also it depends upon the patient adherence to the treatment measures as per counseling is given.

Figure 6 Blood group distribution
6. Hemoglobin (Hb) status:

The data indicates that most of the patients had the Hb value less than normal levels. 22 patients had Hb value below 6 gm % and required the blood transfusion. Rest 22 patients had the Hb range between 6 to 8 gm % and could be managed with the prophylactic treatment. Hemoglobin status of five patients was unknown (Fig 8).
7. Treatment:

The present study reveals that 44 patients were receiving Folic acid whereas 43 were receiving Desferox along with Folic acid. One patient was on insulin treatment along with Folic acid and Desferox. The rest of the 5 patients did not receive any treatment. No patient was on antibiotic treatment (Fig 9). Previous studies stated that Diabetes Mellitus is more prevalent among the thalassemic patients (10.1 %) [14] but, a number of patients enrolled in this study was less, we could not determine the exact prevalence. During the study period, patients showed better treatment compliance with the chelation therapy.

![Figure 9 Treatment](image)

**CONCLUSION**

The small sample size limits the outcome of the present study. The data collected from 49 beta-thalassemia positive patients was presented. In the case of thalassemia, age cannot be regarded as a significant factor. From the gender-wise distribution, the thalassemias are more common in males than in females. In the community-wise distribution, the prevalence of thalassemia in Kunbi, Muslim, Boudh, Mali communities is high. Thalassemia disorder is inherited but not always due to consanguinity. The frequency of blood transfusion depends upon the severity of the disease. The hemoglobin status can be managed with respect to the blood transfusion and the treatment adherence. As per Rh blood group typing, the frequency of beta-thalassemia is high in O Rh+ and B Rh+.
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