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Prevalence of Thalassemia in and Around Mumbai District Maharashtra

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Abstract

Background : *Thalassemia is a heterogeneous group of disorders resulting from abnormalities of globin chain synthesis. Which in turn results in abnormal hemoglobin synthesis. Thalassemia minor screening would help to reduce the thalassemia major cases.* ***Methods:*** *The present study includes a total number of 287 patients in and around Mumbai District, Maharashtra, during the period from August 2021 to February 2022.* ***Results:*** *This original article showed a prevalence of thalassemia to be around 6.2% in the total number of admitted patients in the pediatric ward. Maximum students presented with anemia, few showed thrombotic complications, and very few presented with bony deformities associated with osteoporosis.* ***Conclusion*** *The outcome for patients in the form of blood transfusion and drug therapy is much better and the life pattern of the patient is more comfortable. More emphasis should be given to health education in society the result of consanguineous marriages and how to prevent disease from occurring in the community.*

Keywords*: Thalassemia, Prevalence, Consanguineous marriages, anemia*

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Introduction

India is a country with many religions which are divided into many castes. There are around 3000 castes and 25,000 subcastes in India. Consanguineous marriage remains the choice of an estimated 10.4% of the global population. [1] There are more consanguineous marriages taking place in India and premarital checkup is not under process in India. which leads to many genetic disorders. Many Diseases are passed on from parents to their children. Thalassemia is one of them. Thalassemia is present almost in every community. Thalassemia is caused by a genetic inability to make normal amounts of hemoglobin. [2] Thalassemias are a heterogeneous group of disorders of hemoglobin synthesis resulting from the reduced rate of synthesis of one or more globin chains of hemoglobin. Decreased synthesis of α chain produces α thalassemia, while decreased synthesis of β chains produces β thalassemia. Thalassemia is among the most common genetic disorders worldwide, occurring more frequently in the Mediterranean region, the Indian subcontinent, Southern Asia, and West Africa. [3] If there is a reduced output of α chains, any excess of γ chains or β chains produced will give rise to molecules with the formula γ4 (Hb Bart's) and β4 (Hb H), respectively. Hence α thalassemia's usually associated with the presence of Hb Bart's in infancy and Hb H in adult life. [4] Thalassemia is a major health problem, placing an immeasurable emotional, psychological, and financial burden [5, 6] Screening can be done for thalassemia in four different ways. They are prenatal, newborn,

premarital, and random total population screening. Thalassemia minor screening would help to reduce the thalassemia major cases. Screening can be done using different methods. Initially, a complete hemogram is done in which the MCV less than 80fl would be further screened using Naked Eye Single Tube Red Cell Osmotic Fragility Test (NESTROFT). The person who shows positivity for NESTROFT would be checked for HbA2 level using HPLC or Hb electrophoresis. Mutations can be checked by PCR and an automated sequencer. [7]

Materials and Methods

The present study was carried out in the Department of Clinical Hematology. The study included all anemic patients from the pediatric ward. They are from the district and adjoining areas. These are the patients who come from these different places. A 3 ml blood samples were obtained after voluntarily collecting information consent papers from the kids' parents as well. Osmotic fragility (OF) and dichlorophenol indophenol precipitation (DCIP) tests were run after a complete blood count (CBC) in order to check for thalassemia. Hemoglobin type and -thalassemia were determined using high-performance liquid chromatography (HPLC) and real-time quantitative polymerase chain reaction (qPCR), respectively. At the significance level of = 0.05, the connections between variables were found using the t-test, chi-square, and logistic regression. The 10 total patients were found with thalassemia in the total n=287 samples during the study period.

Results

In the present study 10 patients are detected in the above period. The distribution of patients is shown in the following tables.

Table No 1 shows the total number of students with age distribution.

Sex	Total Number	Maximum age	Minimum Age
Female	10	34	17

Table No 2 shows the average and minimum hemoglobin in the cases studied.

Sex	Average	Minimum
Female	19.63	17

287-Total cases, Prevalence of thalassemia trait 3.87 %. Maximum cases presented with anemia, few showed thrombotic complications, and very few presented with bony deformities associated with osteoporosis.

Discussion

Ten percent of the total world thalassemia cases are born in India every year. [8] It has been estimated that the prevalence of pathological hemoglobinopathies in India is 1.2/1,000 live births, with approximately 27 million births per year. [9, 10] This would suggest the annual birth of 32,400 babies with a serious hemoglobin disorder. The first case of Thalassemia, described in a non-Mediterranean person, was from India. Subsequently, cases of thalassemia were documented in all parts of India. [11] Thalassemia treatment is costly. To get an ideal treatment for one thalassemic child it costs around Rs. 1,25,000/annum. Hence for 50,000 children, the cost would be nearly Rs.620 crores. This staggering cost is beyond the reach of our country, India; moreover, this cost is expected to increase due to additional children being born. [12] (12). Thalassemias are a major health problem, and approximately 1 in 14 of the population are carriers for one of the subtypes. Over the past three decades, regular blood transfusions and iron chelation have dramatically improved the quality of life and transformed thalassemia from a rapidly fatal disease in early childhood to a chronic disease compatible with prolonged life. [13, 14] Today life expectancy varies between 25-55 years, depending on compliance with medical treatment. [15,16] Despite increased life expectancy, complications keep arising. Health education is an important component of preventive genetic programs. This requires proper health education and adequate sensitization to the individual, family, or community to accept these preventive remedial measures. The high cost of treatment repeated blood transfusion and chelation therapy, and the economic burden on family resources, all suggest that prevention is better than cure. Thus, a joint venture of antenatal and inductive screening seems to be the most fruitful strategy for beta-thalassemia in India. With improving environmental and socio-economic conditions, better public health care and medical facilities,

and better nutrition, children suffering from thalassemia and hemoglobinopathies can be better managed and rehabilitated in India. [17-20] Prevention of thalassemia. The need for prevention of thalassemia is obvious due to the high frequency of the condition, the great expense and difficulties in providing optimal treatment for patients, and the innumerable fatalities from untreated β-thalassemia. Prevention would not only be a good public health practice, as envisioned in the Alma Ata declaration, but it would also be cost-effective, as the ratio of the cost of treatment to prevention is 4:1, as shown in a study from Israel would help tremendously in reducing the burden of the disease for patients, families and the health services. The strongest argument for prevention is that it would ensure the best possible care for the affected, by curbing the increase in their number. The chief elements of a control program were developed in the 1970s by a team of experts at the World Health Organization, led by Dr. B. Modell These are (i) Political and financial support, (ii) Improving curative services; (iii) Prenatal diagnosis in couples who have given birth to an affected child, as well as those identified to be at risk, (iv) Prospective antenatal screening, (v) Community carrier screening, (vi) Counselling and prenatal diagnosis, and (vii) Network of centers, and National/Regional working groups. The outcome for patients in the form of blood transfusion and drug therapy is much better and the life pattern of the patient is more comfortable. More emphasis should be given to the health education of society on the result of consanguineous marriages and how to prevent the disease from occurring in the community.

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