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Article

Thalassemia Unveiled: An In-Depth Analysis of Characteristics, Public Perception, and Medical Approaches

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Abstract: Thalassemia, a genetic blood disorder, presents a significant healthcare burden worldwide, particularly in regions like India. This article provides an in-depth analysis of thalassemia in India, examining its features, public perception, and medical approaches. Through a comprehensive review, the study elucidates the epidemiological aspects, genetic predisposition, clinical manifestations, and socioeconomic impacts of thalassemia in the Indian population. Furthermore, the article explores public awareness and perception of thalassemia, shedding light on prevailing misconceptions, stigmas, and barriers to prevention and treatment. Additionally, the review outlines existing medical approaches for thalassemia management in India, including blood transfusion therapy, iron chelation therapy, and hematopoietic stem cell transplantation. By integrating diverse perspectives from healthcare professionals, patients, and policymakers, this article aims to inform strategies for improving thalassemia prevention, diagnosis, and treatment in India.

Keywords: Thalassemia; Awareness; Autosomal Recessive

1. Introduction

The Greek words "thalas," which means "the sea," and "emia," which means "blood," are the sources of the name "thalassemia" [1]. Patients with thalassemic disease are thought to number 15 million globally, accounting for 4.4 out of every 10,000 births. The development of aberrant antibodies is a major problem in individuals receiving continuous transfusions [2]. One of the most prevalent autosomal recessive genetic disorders is beta thalassemia, which affects 5000 children worldwide and around 3% of the global population each year. Approximately twenty-five thousand children are registered with the India Thalassemia Federation. Since many patients who reside in remote areas are not registered with any thalassemic centers, the initial number is still unknown.

The most effective thalassemia prevention approach, implemented by Cyprus and Turkey, is preventing the birth of children who are thalassemic. In contrast, Iran has mandated premarital examinations in order to prevent the birth of children with thalassemy. The least expensive and most successful thalassemia preventive method is to determine the premarital genetic status of the parents or other carriers of the disease and to provide appropriate counseling and assistance regarding the implications. Thus, appropriate screening programs to identify carriers can help lower the likelihood of new thalassemia births [5,6]. Due to financial constraints, the majority of thalassemic patients—roughly a million people in India do not receive appropriate care or blood transfusions. This is because these procedures can cost up to Rs. 8,000 per month, which is a significant expense for most households. In order to offer these patients equipment and treatments on par with those in other countries, India's health budget is unable to provide more than Rs. 900 million a month and Rs. 7.2 billion annually. Since such facilities cannot be provided, the only option left is to prevent the birth of a thalassemic kid, which can only be done if people are informed about the disease's causes and effects.

Anemia caused by beta thalassemia involves chromosomes. It interferes with hemoglobin's B globin chain synthesis. Thus, it is either diminished or eliminated, resulting in an early aberrant red blood cell production.

Table 1. Showing the Patient's Data

Factors to be studied	Analysis	%ages
Does patient have any family history of Thalassemia?	Yes	54%
	No	46%
	No	26%
Gender	Male	66.7%
	Female	31.4%
Area	Urban	80%
	Rural	20%

Anemia is caused by the mononuclear phagocytic system (MPS), which promptly destroys red blood cells (RBC), mostly by the spleen. Regular blood transfusions are essential for survival in patients with thalassemia. However, individuals who get blood transfusions frequently develop a number of potentially fatal side effects, including endocrine dysfunction, liver disorders, and cardiomyopathy. In high-income nations, only 50–65% of patients live past the age of 35 [2,4]. The purpose of this study is to raise public awareness of thalassemia, a chronic illness, by highlighting the financial and social challenges that parents of affected children experience.

2. METHODOLOGY

2.1. Study Area

The current study was conducted in District Ladakh, India. It is India's 12th most populated and one of the most important industrialized area of India.

2.2. Study Design

Patients at DHQ Ladakh and Sundas Foundation Ladakh were given a self-administered questionnaire to complete as part of a thalassemia survey. Patients, physicians, and parents, if the patient was a minor, gave their consent to participate in the study. Two sections made up the designed questionnaire. The first component contained information on the sociodemographic and economic characteristics of the patients, such as their present age, sex, weight, and place of residence (rural or urban), as well as any dietary restrictions and treatments they are receiving. The purpose of the study's second half was to gauge the general public's awareness of thalassemia among Ladakh people.

3. Statistical Analysis

The data were analyzed statistically using SPSS.

4. RESULTS

Of the 50 patients, 72.5% (n=37) stated that thalassemia and intermarriages are common in their family. The majority of patients were from rural areas, and Table 1 shows that the rate of thalassemia was substantially (P<0.001) higher in males (66.7%) and 31.4% in females.

145 students took part in the study's second section. The majority of them were in the age range of 18 to 23. Of the pupils, 41.4% were 18–19 years old, 35.2% were 20–21 years old, and 23.4% were 22–23 years old.

Of the 145 people in the survey, 86.9% (n=126) knew nothing about thalassemia, and 13% (n=19) had no idea. Of the students in the former group, 75.9% (n=110) believe it to be a genetic condition, and 79.5% (n=115) believe that families with a high incidence of cousin marriages are the ones with the highest prevalence of thalassemia disorder. Furthermore, 41.4% (n=60) of the public consider that dietary restrictions are necessary for treating thalassemia, whereas 58.6% (n=85) disagree. Of the population, 62.1% (n = 89) believe thalassemia is totally curable, while 29.7% (n = 39) believe environmental factors are the cause.Additionally, 51.7% (n=75) of the public believe that a family history of malnutrition is the cause of thalassemia, whereas 72.4% (n=105) of the people believe that thalassemia is a birth condition. Conversely, 59.3% (n=86) of the population stated that thalassemia is treated with medication, and 79.9% (n=131) stated that genetic counseling is required for a genetic disorder Table 2.

Table 2. Relative knowledge of students about thalassemia

Factors	Categorization	Frequencies
Age	18-19 years	41%
	20-21 years	35%
	22-23 years	23%
Knowledge about thalassemia	Yes No	87%
		13%
Genetic disorder	Yes	76%
	No	24%
Cousin marriage	Yes	79%
	No	21%
Malnutrition	Yes	52%
	No	48%
Dietary restriction	Yes	41%
	No	59%
Environmental factor	Yes	32 %
	No	68%
By birth	Yes	72%
	No	28%
Curable	Yes	62%
	No	38%
Genetic counseling	Yes	87%
	No	13%
Medicine	Yes	51%
	No	49%

5. DISCUSSION

One of the most significant issues facing the globe now is thalassemia. According to studies, there are approximately 60,000 thalassemic births worldwide each year. Since there is currently no known cure for thalassemia or its cause, many nations have found that the only practical approach is to avoid having children who are thalassemic. There were more successful males (66.7%) than females (31.4%) in the initial study that was designed as it is now. In their respective fields of study, a study [5,6] also revealed that men were substantially more likely than women to have thalassemia.

Low knowledge of the illness and a high percentage of illiteracy Due to a lack of awareness of the elevated risk of thalassemia, the majority of thalassemic patients (78.1%) were from rural areas. It was found that cousin marriages were less common in urban regions than in rural ones, and as a result, more halassemic kids were born to parents in rural areas. This is because urban areas have a considerably higher literacy rate than rural ones. A study that was done produced findings that were comparable. The majority of the patients' families were impoverished and very anxious about the financial strain they were carrying. The majority of patients require blood transfusions once or twice a month, and most parents find it challenging to make frequent blood appointments. Because of low family income and high treatment charges, they fail to provide sufficient treatment facilities to their children. Financial burdens eventually increase the stress level of parents [7].

The general public's awareness of pupils in various age groups was noted in the second phase of the planned study. It was noted that pupils' knowledge of the illness was lacking. A small portion of the population (13.1%) stated they were unaware of thalassemia, whilst the majority of students (86.9%) stated they had only a slight introduction to the condition. There were a lot of prevalent misconceptions among the populace. In addition, almost 51.7% of participants thought that starvation was the primary cause of thalassemia, whereas 48.3% disagreed.While most people (68.3%) recognized thalassemia wasn't caused by environmental factors, some pupils (31.1%) thought it was. Their reasoning was flawed. The majority of participants (62.1%) thought that thalassemia could be cured. The idea of pre-marital screening is gradually disappearing as a result of these misconceptions, which are the result of a lack of awareness efforts. In both educational and non-educational institutions, appropriate awareness campaigns ought to be planned in light of the disease's rising prevalence. Though free medications and blood transfusion services are provided by the government and certain NGOs to treat thalassemic patients, these initiatives are limited to large cities and have not yet shown any significant side effects.Appropriate counseling for the families affected by the sickness might eliminate the anxiety or superstitions that are typically present in the illiterate population with relation to it. Universities, colleges, and schools should all have appropriate screening procedures. It is necessary to raise awareness through social media and seminars. It is important to remember that the only way to prevent or completely eradicate thalassemia in the nation is to cease having thalassemic children [8,9].

6. CONCLUSION

The majority of people, the study found, are ignorant of the causes and effects of thalassemia. Worldwide, the usage of extremely costly medical procedures and pharmaceuticals results in the deaths of numerous children. To reduce thalassemia in the nation, awareness campaigns such as seminars and educational lectures must be developed on genetic counseling of individuals.

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