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FREQUENCY OF HEART COMPLICATION IN BETA THALASSEMIA MAJOR CHILDREN AT TERTIARY CARE HOSPITAL

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ABSTRACT

Objective: to evaluate the prevalence of cardiac problems, including myocardial iron deposition, pericarditis, and valvular abnormalities, among people with β-thalassemia and to shed light on the distribution of β-thalassemia and hemoglobin levels within these populations. **Methods:** Hemoglobin levels, β-thalassemia status, and cardiac problems were measured in two groups of people: those who attended school and those who did not. The information in the data contained the β-thalassemia status (Yes/No) and hemoglobin levels grouped into specified ranges (e.g., <8.0 g/dl, 8-8.9 g/dl). information was documented for both groups on cardiac problems, such as arrhythmias, cardiomyopathies, pericarditis, myocardial iron deposition, heart failure, and valvular abnormalities. The frequency and percentages of people with β-thalassemia, hemoglobin levels, and cardiac problems within each group were ascertained by analyzing the collected data. **Result:** The investigation produced some interesting results about hemoglobin levels, cardiac problems, and the prevalence of β-thalassemia in people who attend school and those who do not. Of those attending school, most had hemoglobin levels between 9 and 10.9 g/dl, and β-thalassemia impacted a sizable portion of them. Cardiac problems were common in school-age β-thalassemia patients, especially myocardial iron deposition and pericarditis. Likewise, β-thalassemia was shown to be more prevalent in non-school-going individuals, exhibiting a spectrum of hemoglobin levels and cardiac problems. **Conclusion:** the significance of early identification, surveillance, and treatment approaches for β-thalassemia-associated consequences, especially heart-related problems including pericarditis and myocardial iron accumulation. Furthermore, the research highlights the necessity of all-encompassing care and customized therapies to enhance outcomes and quality of life for β-thalassemia patients, with a specific emphasis on the avoidance and handling of cardiac issues.

Keywords: β-thalassemia, Hemoglobin levels, Cardiac abnormalities, Children.

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INTRODUCTION

Several genetic disorders are inherited and can be multi-genetic. These disorders include alpha, beta, and delta beta thalassemia's, among others. Since thalassemia is an inherited condition, it requires the presence of at least one carrier parent. A child has to inherit one defective gene from each parent in order to be diagnosed with the condition. It is brought on either by a hereditary disease or by specific major gene segments being deleted. Molecular flaws cause incorrect hemoglobin synthesis in a cluster of the beta-globin gene on chromosome 11,3 and the 16 chromosomes on the alpha-globin gene cluster **[1-3]**. A spectrum of severity is used to describe thalassemia diseases with a variety of clinical symptoms, phenotypes, and treatment choices. There are two forms of thalassemia: TDT (transfusion-dependent thalassemia) and NTDT (non-transfusion-dependent thalassemia). Iron excess is linked to increased morbidity in both transfusion-dependent and non-transfusion-dependent thalassemia. An excessive buildup of intestinal iron, as indicated by insufficient erythropoiesis, results in iron overload. Numerous essential organs are harmed by excess iron deposition, which starts the first year of regular blood transfusions. Iron overload, or hemochromatosis, is a condition marked by abnormal iron buildup in an organ's functional components, which causes organ failure and damage. Iron is mostly stored by human bodies as ferritin **[4-6]**. Ferritin is secreted into the bloodstream in small amounts. When there is no inflammation, the blood ferritin content is measured and positively corrected by the body's total iron storage. Age and sex differences exist in ferritin standard concentrations. Around the age of one year, ferritin concentration starts to rise, and it continues to rise throughout adulthood. However, compared to females, males have higher levels of concentration values. The human body lacks a

physiological way to eliminate excess iron load brought on by blood transfusions. Elemental iron levels in each transfused unit of packed red blood cells range from 200 to 250 mg. For TDT patients, transfusion iron typically amounts to 0.3 to 0.6 mg/kg per day, with an estimated average transfusion volume of 2 to 4 units of red blood cells each month. This extra iron results in irreparable damage and disrupts the functioning of the important organs **[7, 8]**. Heart failure that is common and avoidable is cardiomyopathy, which is frequently brought on by iron excess. Iron buildup in the cardiac tissue facilitates non-homogeneous electrical conduction and repolarization in atrial and ventricular tachyarrhythmias. The disease thalassemia is autosomal recessive, meaning that for a child to inherit it, both parents must either have the condition or be carriers. It is brought on by mutations in the Hb genes, which induce either an underproduction or absence of alpha or beta chains. The causes of thalassemia are known to involve more than 200 mutations. The loss of the alpha- and beta-globin genes, respectively, results from a point mutation on chromosome 11 in the beta-globin gene's splicing site and promoter regions, which causes alpha-thalassemia and beta-thalassemia **[9, 10]**.

MATERIALS AND METHOD

following ethics clearance, the study was carried out from 1 January 2023 to 31 December 2023 at the Lady Reading Hospital in Peshawar, Pakistan. Serving a diverse population from Peshawar and its neighboring areas, Lady Reading Hospital is a tertiary care hospital and one of the biggest healthcare facilities in the area. Research on β-thalassemia and its consequences can be conducted in an optimal environment because to its extensive resources and expert divisions.

For patients receiving care at Lady Reading Hospital, this length of time allowed for thorough data collection, analysis, and interpretation of findings about the prevalence of β-thalassemia, hemoglobin levels, and related cardiac problems. The study attempted to take into consideration any seasonal fluctuations and temporal trends in disease presentation and care by collecting data over the course of a year.

A retrospective cohort design was utilized in the study to evaluate the frequency of β-thalassemia and related cardiac problems among patients who were treated at Lady Reading Hospital throughout the designated timeframe. Data on hemoglobin levels, cardiac problems, β-thalassemia status, and demographic variables were gathered by an examination of patient medical records and computerized databases. From the time of the patients' first hospital presentation until the conclusion of the research, they were monitored to monitor the development of cardiac issues and determine whether they were related to hemoglobin levels and β-thalassemia.

thalassemia was encompassed under the investigation. A molecular genetic study or hemoglobin electrophoresis test was one of the diagnostic criteria for β-thalassemia. Individuals who had comprehensive medical records with details on their β-thalassemia status, hemoglobin levels, and heart issues were qualified for participation. The study excluded those with cardiac problems or β-thalassemia status. Patients who had a history of substantial comorbidities or other medical disorders that would complicate the relationship between cardiac issues and β-thalassemia were also removed from the study. To maintain the homogeneity of the study population, individuals with acute or chronic diseases requiring immediate or specialized care, as well as pregnant women, were removed from the analysis.

RESULTS AND DISCUSSION

An interesting contrast of hemoglobin (HB) levels, β-thalassemia status, and cardiac problems between persons who attend school and those who do not is presented by the data. In **Table 1**, most school-age individuals have HB levels between 9 and 10.9 g/dl, with 68.18% of them falling into this range. It's interesting to note that although while fewer people have anemia below the crucial threshold of 8 g/dl, a sizable minority of people—9.09%—still do. The populations to be screened include teenagers in high school and college for the purpose of determining their β-thalassemic status, providing them with information about the condition, counseling before and after marriage, and education. It is crucial to screen women for βTT early in pregnancy and, if necessary, to screen their spouses for prenatal diagnosis in order to lower the number of homozygotes born. By screening only 13% of the population, extended family screening for thalassemic enables identification of the vast majority of the population at risk **[11, 12]**. Furthermore, the percentage of school-age individuals affected by β-thalassemia ranges from 18.18% to 68.18%, suggesting a high prevalence of the condition. **Table 2** shows a similar pattern among those who do not attend school. Notably, 40.85% of this group of people have HB levels between 9 and 9.9 g/dl. Although β-thalassemia is not as common as in the school-going population, it nonetheless affects a considerable proportion of non-school-going people (7.04% to 40.85%). It's interesting to note that although the proportion of people in this category with HB levels below 8 g/dl is smaller, it still makes up 14.08%, suggesting a worryingly high incidence of severe anemia among people who do not attend school. Screening programs are important because they give the screened population and the linked population—parents, teachers, friends, siblings, and employees—a

forum for better understanding and education about thalassemia. Screening could be required or optional. Automated hematology cell counts and specialist HPLC systems, when well-calibrated, can now identify β-thalassemia carriers within a family. However, due to a number of circumstances, the preventative program in India, which includes early screening for antenatal diagnosis and termination of a homozygote baby in pregnant women and spouses of thalassemic pregnant women, has been reluctant to take off. These include the dearth of widely available facilities for prenatal diagnosis and screening as well as the tardiness in reporting pregnancies **[13, 14]**.

Table 3 shows that β-thalassemia-related cardiac problems can affect people who attend school as well as those who do not. Myocardial iron deposition is found to be a common problem in both groups, impacting 16 persons in the group that attends school and 8 individuals in the group that does not. Another serious worry is pericarditis, of which there have been 21 cases documented in students and 15 cases in non-students. The aforementioned results emphasize the significance of preemptive surveillance and management tactics aimed at reducing the likelihood of cardiac problems in β-thalassemia patients. Special attention should be paid to measures that target myocardial iron accumulation and avert issues linked to pericarditis. There has been ongoing discussion on whether thalassemia awareness campaigns and student screening in high school and college will be successful in Kyber pukhtoon khwa. It's also unclear if high school students who receive screening and counseling will be sensitive to the information about thalassemia and if they will recall their status when they get married. For this large and ethnically varied nation, a multipronged strategy that includes antenatal diagnosis, premarital screening, screening of the extended family of thalassemic, and screening of high school and college students is necessary. In order to lower morbidity and mortality as well as the financial and sociopsychological load on thalassemic families, there is an urgent need to expedite education and knowledge of thalassemic among medical professionals, paramedics, the thalassemic community, and the general public **[15]**.

Table 1: School going HB (g/dl) variables.

HB(g/dl)	Male	female	frequency	Beta-thalassemia	percentages
<8.0	1	3	4	Yes	9.09%
8-8.9	0	2	2	No	4.55%
9-9.9	13	17	30	Yes	68.18%
10-10.9	17	9	26	Yes	59.09%
11-11.9	4	8	12	Yes	27.27%
12-12.9	6	12	18	Yes	40.91%
13-13.9	2	6	8	Yes	18.18%
>14	3	11	14	yes	31.82%

Table 2: Non-school going HB(g/dl) variables.

HB(g/dl)	Male	female	frequency	Beta-thalassemia	percentages
<8.0	3	7	10	Yes	14.08%
8-8.9	5	3	8	No	11.27%
9-9.9	12	17	29	Yes	40.85%
10-10.9	1	4	5	Yes	7.04%
11-11.9	14	5	19	Yes	26.76%
12-12.9	6	14	20	Yes	28.17%
13-13.9	9	8	17	Yes	23.94%
>14	1	3	4	yes	7%

Table 3: Cardiac complications.

Cardiac complications	School going	Non-school going
Hear failure	5	7
Arrythmias	3	0
Cardiomyopathies	7	9
Pulmonary hypertension	2	4
Myocardial iron deposition	16	8
Pericarditis	21	15
Valvular abnormalities	4	3

CONCLUSION

Data regarding hemoglobin levels, related cardiac problems, and the prevalence of β-thalassemia in those who attend school and those who do not are provided by the data. The results show that there are notable numbers of people with β-thalassemia in both categories, with different hemoglobin level distributions. Significantly, heart problems such pericarditis and myocardial iron deposition are common in people with β-thalassemia, underscoring the high prevalence of heart illness in this group. These results highlight the significance of proactive approaches to management, monitoring, and screening in order to address the complex issues associated with β-thalassemia and reduce the risk of cardiac problems. Targeted therapies that improve hemoglobin levels and manage iron overload are also necessary to improve the quality of life and long-term results for those who are impacted.

Limitation: A number of variables, including sample size, representativeness, and possible biases in data gathering, may have an impact on how broadly the data can be applied. Improving patient care techniques and expanding our knowledge of β-thalassemia need addressing these constraints with rigorous study design, long-term follow-up, and thorough clinical assessment.

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