Title: Prevalence of beta thalassemia, heart failure and valvular abnormalities in children at tertiary care hospital

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Abstract

Objective: to evaluate, stratified by age and educational attainment, the distribution of pediatric beta-thalassemia, heart failure, and valve anomalies to find any appreciable variations in the prevalence rates.
Methods: A study using a cross-sectional design was carried out on 115 youngsters. There were two age categories of participants: 43.5 percent were between the ages of 2 and 9 and 56.5% were those over 10. Children's educational status was divided into two categories: those who attend school (8.7%) and those who do not (91.3%). Medical records were used to gather information on the prevalence of valvular anomalies, heart failure, and beta-thalassemia. P-values and other statistical analyses were used to assess the significance of variations in prevalence rates among different age and educational groups.
Result: 17.4% of participants in the research had beta-thalassemia; there were no significant variations in this condition between age groups (p=0.101) or educational attainment (p=0.820). Children who attended school had a higher prevalence of heart failure (69.6%) than children who did not attend school (34.4%), however this difference was not statistically significant (p=0.625). Children who did not attend school (38.2%) had considerably higher rates of valve anomalies than children who attended school (8.6%); a p-value of less than 0.001 indicated a highly significant difference.
Conclusion: The study finds no significant changes in the prevalence of beta-thalassemia or heart failure across age and educational status, but it does show a substantial difference in the prevalence of valvular anomalies between children who attend school and those who do not. These results highlight how crucial it is to address socio-economic and educational gaps to improve health outcomes for communities that are already at risk.
Keywords: β-thalassemia, heart failure, valvular abnormalities, children, education status, age distribution.

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:

From January 1 to December 31, 2023, a cross-sectional study was carried out at the Lady Reading Hospital in Peshawar. The purpose of the study was to examine the prevalence of heart failure, valve anomalies, and beta-thalassemia in children, stratified by age and educational attainment. Children who visited the pediatric department between the ages of 2 and 15 were included in the study. Two age groups were created for the research population: those over 10 and those between 2 and 9 years old. Medical records and in-person interviews with parents or guardians were used to gather information, with a focus on health issues and educational attainment. The study inclusion criteria included children between the ages of 2 and 15 who were either inpatients or outpatients at Lady Reading Hospital and had medical records pertaining to their ailments. Included were children having a verified diagnosis of cardiac failure, valvular anomalies, or beta-thalassemia. Participants also needed to have unambiguous records of their educational status, either as attending school or not. Before being included in the study, parents or legal guardians gave their informed consent. Children under the age of two or older than fifteen, as well as those without official medical records for the health disorders being studied, were excluded from the study. Additionally eliminated were kids with unclear educational statuses and kids whose parents or guardians declined to let them participate. Statistical software 24 version was utilized to evaluate the data and ascertain the prevalence rates of various health disorders across various age and educational categories. P-values were utilized to evaluate the significance of variations and detect any discrepancies in health outcomes according to age and educational attainment.

Result:

There were no discernible differences seen between the age groups (2–9 years and more than 10 years) or educational status (non–school-going and school-going) among the 115 pediatric participants who tested positive for beta-thalassemia (17.4%). Children who attended school had a higher prevalence of heart failure (69.6%) than children who did not attend school (34.4%), however this difference was not statistically significant (p=0.625). On the other hand, there was a substantial difference in the prevalence of valvular anomalies depending on educational status, with non-school-going children having a significantly higher frequency of these conditions (38.2%) compared to school-going children (8.6%). This difference was highly significant, with a p-value of less than 0.001. These results imply that valvular anomalies are much higher in children who do not attend school, yet there is no significant difference in beta-thalassemia or heart failure according to age or education.

Table: 1 age and education wise distribution

distribution		frequency	percentages
age	2-9	50	43.5%
	>10	65	56.5%
education	Non-school going	105	91.3%
	School going	10	8.7%

Table 2: beta-thalassemia, heart failure and valvular abnormalities status

distribution		frequency	percentages
Beta thalassemia	yes	20	17.4%
	no	95	82.6%
Heart failure	Non-school going	35	34.4%
	School going	80	69.6%
Valvular abnormalities	Non-school going	34	29.6%
	School going	81	70.4%

Table 3: different variable wise stratification of cardiac complication

variables		Beta thalassemia		P-value
		Yes	no	
Age group	2-9	12(24%)	38(76%)	0.101
	>10	8(12.3%)	57(87.7%)	
education	Non-school going	18(17.1%)	87(82.9%)	0.820
	School going	2(20%)	8(80%)	
Heart failure	Non-school going	7(20%)	28(80%)	0.625
	School going	1316.2(%)	67(83.8%)	
Valvular abnormalities	Non-school going	13(38.2%)	21(61.8%)	<0.001
	School going	7(8.6%)	74(91.4%)	

Discussion:

The study provides information on the distribution of age and education levels, as well as the prevalence of heart failure, valve anomalies, and beta-thalassemia in a pediatric population. Table 1 reveals that 56.5% of participants are over 10 years old, while 43.5% are between the ages of 2 and 9. About education, only 8.7% of the youngsters attend school, while a sizable majority (91.3%) do not. This high percentage of children who don't attend school suggests that there may be differences in this population's access to education, which may have an impact on their general health and wellbeing. 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, counselling 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 to lower the number of homozygotes born. By screening only 13% of the population, extended family screening for thalassemic enables identification of most of the population at risk[11-12]. The prevalence of heart failure, valve anomalies, and beta-thalassemia is seen in Table 2. 17.4% of the subjects have beta-thalassemia, suggesting a high frequency of this genetic condition in the general population. It's interesting to note that children who attend school seem to have a higher prevalence of heart failure (69.6%) than children who do not (34.4%). Similarly, children who attend school (70.4%) have valvular anomalies at a higher rate than children who do not attend school (29.6%). These results imply that, despite their smaller number, school-age children may be more likely to suffer from specific cardiac diseases, which calls for more research into the potential causes of this trend. 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 several 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]. A categorization of cardiac problems according to various factors is shown in Table 3. The prevalence of beta-thalassemia is higher in children 2–9 years old (24%) than in older children (12.3%), with a p-value of 0.101 suggesting no significant difference. Upon stratifying by education, there is no statistically significant difference seen between the 17.1% of children who do not attend school and the 20% of children who do (p=0.820). With a p-value of 0.625, heart failure rates do not significantly differ between children who do not attend school (20%) and those who do (16.2%). These findings imply that the prevalence of beta-thalassemia and heart failure in this population is not substantially impacted by age or educational attainment. 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 counselling 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 multi-pronged strategy that includes antenatal diagnosis, premarital screening, screening of the extended family of thalassemic, and

screening of high school and college students is necessary. 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 3's noteworthy discovery is the variation in valvular abnormality prevalence. With a highly significant p-value of <0.001, 38.2% of children who do not attend school had valvular anomalies, compared to only 8.6% of children who attend school. This striking disparity implies that children who do not attend school have a significantly increased risk of valvular abnormalities. Numerous factors, including children attending school, early detection and treatment of health concerns, and dietary status, may be to blame for this.

Conclusion: Among children who attend school and those who do not, the study reveals a notable difference in the occurrence of valvular anomalies; in contrast, illnesses like beta-thalassemia and heart failure do not exhibit significant differences based on age or educational attainment. These results highlight how critical it is to overcome educational and socio-economic gaps to enhance health outcomes for communities that are already at risk. Future studies ought to concentrate on figuring out the fundamental reasons behind these differences and creating focused strategies to lessen the dangers connected to low educational attainment and unfavorable health outcomes.

Limitation: The limited sample size and narrow focus on a specific region, among other limitations, may restrict the generalizability of the results even with our study comprehensive nature. Furthermore, bias may be introduced when certain variables are based solely on self-reported data. It is suggested that further research with more center and bigger, more diverse populations corroborate these findings and go deeper into the discrepancies that have been found.

Conflict: none

Funds: none

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