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Rising Trend of Thalassemia Minor Among Women of Reproductive Age Residing in Rural Areas of Sindh

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ABSTRACT

Introduction: A class of hereditary blood illnesses known as beta thalassemias (β thalassemias) are characterized by decreased or nonexistent synthesis of the beta chains of hemoglobin, leading to a range of phenotypes from severe anemia to clinically asymptomatic persons. Patients with beta thalassemia trait (BTT) typically have no symptoms and are unaware that they are carriers unless they are tested for the condition. The purpose of the current study was to determine the prevalence of thalassemia minor among women in rural areas who are of reproductive age. **Objectives:** To determine the thalassemia minor among women of reproductive age residing in rural areas of Sindh. **Study Design:** Descriptive study. **Study duration:** August 2024 to December 2024. **Materials & Methods:** There were 167 anemic women between the ages of 18 and 45. Patients who did not have beta thalassemia major or any other hemoglobinopathy were not included. Following the collection of pertinent medical history, hemoglobin electrophoresis was performed on cellulose acetate paper strips at a pH of 8.5. The presence or absence of beta thalassemia trait was noted, and Hb A2 was estimated by measuring the elute's absorbance using a spectrophotometer. **Results:** The study's participants ranged in age from 18 to 45, with a mean age of 30.38 ± 4.34 years. Ninety-one (54.49%) of the patients were in the 31–45 age range. According to this study, 10.18% of women of reproductive age had thalassemia minor. **Conclusion:** The study emphasizes that women of reproductive age have a higher prevalence of beta-thalassemia trait.

INTRODUCTION

One of the autosomal recessive hereditary illnesses, beta thalassemia is characterized by extremely low hemoglobin or fewer erythrocytes (RBCs) in the blood. Weakened bones, hypoxia, pallor, and jaundice are the most prevalent symptoms. Additional related abnormalities seen in β thalassemia patients include hepatomegaly, growth retardation, splenomegaly, and bone deformation.[1] The Middle East, South-East Asia, and Mediterranean regions have the highest rates of the disease.[2] The primary cause of the hereditary condition is a substitution mutation in the β-globin gene that results in either diminished or nonexistent beta-globin chain synthesis.[3] If treatment is not received, patients with beta thalassemia may only live for 30 years. Patients with beta thalassemia accumulate excess iron as a result of frequent blood transfusions. The body uses iron chelation treatment (ICT) to get rid of extra iron.[4] Bone marrow transplant therapy, which necessitates HLA (Human Leukocyte Antigen) matching between donor and recipient blood, is the most successful therapeutic method used against B.T. aside from blood transfusions. But because it is more expensive, families and the health sector bear a heavy financial burden that must be lessened by implementing innovative preventative medicines.[5]

It must be a top priority to put cost-effective screening methods into place in order to reduce the prevalence of BTM (Beta-Thalassemia Major) in society.[6] A number of anti-thalassemia screening initiatives around the world were crucial in lowering the prevalence of beta-thalassemia. Nonetheless, the prevention and treatment of β-thalassemia in Pakistan require some coordinated national measures. It is estimated that approximately 1.5% of people globally have beta-thalassemia, while 60,000 kids are born with the condition each year. Between 5 and 8% of people in Pakistan have beta thalassemia mild.[7]

Regarding common mutations, IVS 1-5 is the most widespread B.T. mutation, with over 218 beta thalassemia mutations documented globally. Thus far, it has been determined that 20 mutations account for approximately 90% of the beta-globin gene disease.[8] The most frequent mutations among Pakistani people, according to the literature, are IVS 1-5 (G-C) (with an overall incidence rate

of 37.7%), and F.Sc 8/9 (+G) (21.1%), 619bp (12.9%), and IVS 1-1 (9.5%). Additionally, with an overall prevalence of 9.17%, additional frequently reported mutations include CD 16, CD 30, IVS 1-1 (G-T), and CD41/42.[9]

One important metric for the screening of Beta Thalassemia Trait is the Complete Blood Count (CBC), a frequently conducted laboratory test. However, the final diagnosis of BTT is made by Hb electrophoresis.[10] In order to give patients the information they need to make an informed decision about continuing their pregnancy, the prenatal test is performed during the 12th week of pregnancy. The purpose of the current study was to determine the prevalence of thalassemia minor among women in rural areas who are of reproductive age.

METHODOLOGY

This cross-sectional study was conducted at the People's University of Medical and Health Sciences for Women in Shaheed Benazirabad, Sindh, between August and December of 2024. All women between the ages of 18 and 45 who were anemic (hemoglobin levels <11 g/dl at presentation) were included. Because this study was descriptive, the necessity to obtain informed consent was waived in accordance with our institutional protocol. The research was conducted in accordance with ethical guidelines. In order to prevent missing carrier states because of blood dilution and the potential for transfusion from HbE carriers, which could alter HbA2 levels, women with a history of recent blood transfusions (within 6 months) were excluded. Due to possible effects on folate metabolism, haematological parameters, and HbA2 levels, women who had recently lost blood, known epilepsy patients on antiepileptic medications, drinkers, and HIV patients taking medication were also disqualified.

Two milliliters of blood were drawn in an EDTA tube and three milliliters in a red top tube under aseptic conditions following informed permission. On the same day, a Coulter automated cell counter was used to assess the EDTA sample for Complete Blood Count (CBC). On the day of collection, the Naked Eye Single Tube Red Blood Cell Osmotic Fragility Test (NESTROFT) was also conducted with 0.36% buffered saline. Within four days of collection, HbA2 was determined using a Biorad D10 system and High-Performance Liquid Chromatography (HPLC). For standard tests such as blood sugar, urea, and creatinine, the blood collected in the red top tube was centrifuged for 15 minutes at 2000–2500 rpm after being allowed to clot. The serum was isolated right away and stored at -20°C for the Electrochemiluminescence Immunoassay (ECLIA) ferritin measurement. Peripheral smear analysis and HPLC detection of HbA2 were additional laboratory tests.

The computer program SPSS 25.0 will be used to evaluate the collected data. For quantitative values, such as age, the mean and standard deviation will be computed. For qualitative variables, such as parity (primiparous/multiparous), monthly income (<30,000/30000-60000/>60000), and beta thalassemia minor (present/absent), frequency and percentage will be computed. Using stratification and post-stratification, effect modifiers like as age, parity (primiparous/multiparous), and monthly income (<30,000/30000-60000/>60000) will be managed. To determine their impact on the prevalence of the beta thalassemia trait, chi square will be used. A P-value of less than 0.05 will be deemed significant.

RESULTS

The study's participants ranged in age from 18 to 45, with a mean age of 30.38 ± 4.34 years. Table I shows that the majority of the patients, 91 (54.49%), were between the ages of 31 and 45. Table II displays the patient distribution by parity. Table III displays the patient distribution by monthly income.

According to Figure I, the prevalence of thalassemia minor among women living in this research who were of reproductive age was 10.18%. Table IV displays the stratification of thalassemia minor according to age, parity, and monthly income.

Table I

Age distribution of patients (n=167).

Age (in years)	No. of Patients	%age
18-30	76	45.51
31-45	91	54.49
Total	167	100.0

Table II

Distribution of patients according to parity (n=167).

Parity	No. of Patients	%age
Primiparous	46	27.54
Multiparous	121	72.46
Total	167	100.0

Table III

Distribution of patients according to monthly income (n=167).

Monthly income	No. of Patients	%age
<30000	76	45.51
30000-60000	54	32.34
>60000	37	22.16
Total	167	100.0

Figure I

Frequency of beta thalassemia minor among women of reproductive age (n=167).

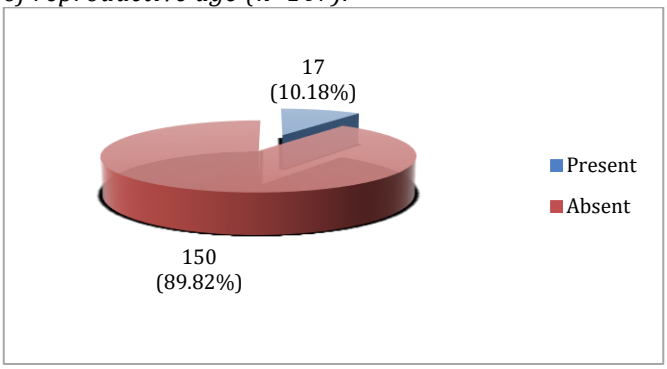

Table IV

Stratification of thalassemia minor with respect to age, parity and monthly income.

		Yes (n=17)	No (n=150)	P-value
Age (years)	18-30	12 (15.79%)	64 (84.21%)	0.029
	31-45	05 (5.49%)	86 (94.51%)	
Parity	Primiparous	04 (8.70%)	42 (91.30%)	0.696
	Multiparous	13 (10.74%)	108 (89.26%)	

Monthly income	<30000	05 (6.58%)	71 (93.42%)	0.154
	30000-60000	09 (16.67%)	45 (83.33%)	
	>60000	03 (8.11%)	34 (91.89%)	

DISCUSSION

A hereditary condition called thalassemia causes faulty hemoglobin synthesis. The homozygous state is known as Thalassemia major, and the heterozygous state is known as Thalassemia trait. Blood transfusions are necessary for the rest of a person's life. Eventually, they have iron overload, which damages multiple organs. This raises these patients' rates of morbidity and death. Globally, thalassemia is common. The thalassemia mutation is thought to be present in about 3% of the global population. Every year, 60,000 newborns are born with thalassemia, with Asian nations bearing the majority of this burden (80%).[11] Every year, 5,000 instances of thalassemia are diagnosed in Pakistan. In our nation, the carrier rate ranges from 5 to 8%.[12] The incidence of thalassemia in Taiwan has been decreased thanks to prenatal screening. The number of thalassemia cases dropped from 20 year to three after seven years. For eleven years, a similar program was implemented in China's Guangdong province. Prenatal thalassemia diagnosis is used in many other nations, including Cyprus, Greece, Italy, Bahrain, India, Iran, Saudi Arabia, United Arab Emirates, Malaysia, Indonesia, Maldives, Singapore, Thailand, and the United Kingdom.[13]

The purpose of this study is to ascertain the prevalence of thalassemia minor among reproductive-age women living in rural Sindh. The study's participants ranged in age from 18 to 45, with a mean age of 30.38 ± 4.34 years. Ninety-one (54.49%) of the patients were in the 31–45 age range. According to this study, 10.18% of women of reproductive age had the beta thalassemia trait. In one study, Qadir M et al.[14] found that 56.70% of pregnant anemic patients had the beta thalassemia trait; in another, Kadija et al.[15] found that the prevalence was 40.0%. According to a different local study, 7.5% of pregnant anemic individuals have the beta thalassemia trait.[16]

Of 210 pregnant women, 18 (8.5%) were determined to be thalassemia carriers by Kulkarni et al. (2015).[17] An 8.5% carrier prevalence was also discovered in a study by Rizwan et al. on pregnant women.[18] Beta thalassemia characteristics were shown to be 4.9% prevalent by Mustafa et al. (2018).[19] In a research conducted in Lahore, Hafeez M et al. found that 53.1% of pregnant women had been diagnosed with beta thalassemia trait.[20] 108 (51.6%) of the 209 pregnant anemic women investigated by Sarda H et al. had the beta thalassemia phenotype.[21] Likewise, there have been reports of research in our area that have findings similar to ours.

In 2010, Sukrat et al[22] conducted a study on both anemic and non-anemic pregnant women. In the anemic pregnant instances, 39.7% of the females were determined to be thalassemia carriers; in the other group, the incidence of thalassemia trait was 24.4%. In 2016, Sur D et al. screened 1,083 women for thalassemia, and the prevalence of thalassemia carriers was 4.61%.[23]

194 expectant mothers who came to the Khyber Teaching Hospital's gynecology and obstetrics unit in Peshawar were assessed by Qadir and Amir. Thalassemia trait was identified in 56.7% of the patients. In comparison to our studies, this figure is rather high.[24] The ß Thalassemia trait was shown to be more common in other Asian countries. Sinha et al. reported that in a province in India, 50% of the patients (n = 120) had the ß Thalassemia trait.[25] In a similar vein, Mohanty D. et al. discovered that 55.9% of the pregnant women carried the illness.[26] In Indore, India, Baxi et al. discovered that just 2.78% of pregnant women (n = 1006) were carriers.[27] According to Wanapirak et al., 25.4% of pregnant women had thalassemia.[28] In a similar vein, Sarda H. et al. discovered that 51.6% of anemic pregnant women were carriers.[29] The varied values indicate that the carrier state is not evenly distributed throughout the nation.

A considerable number of thalassemia cases are born in Pakistan each year as a result of declining literacy rates, an increase in consanguineous marriages, and a failure to implement preventive initiatives. Given the severity of the thalassemic problem, prevention is the most effective strategy. Offering couples premarital exams and heightened awareness programs is advised. Other areas have successfully used this tactic.[30] Additionally, it is advised that partner screening be finished before 12 weeks of pregnancy and prenatal screening be finished by 11 weeks of gestation.[31] According to a study by Haq et al., 92% of parents who married cousins had no prior knowledge of thalassemia.[32] Therefore, if the couple undergoes prenatal screening in the early stages of pregnancy, they may be advised to terminate the pregnancy if a chorionic villous sample reveals a fetus with Thalassemia Major.

Our study included a number of limitations. First, hemoglobin electrophoresis, a traditional screening method, was employed. Even if more recent and precise methods have been developed, they are still costly and not generally accessible. Second, only one hospital was chosen for the study, making it a hospital-based investigation. An improved understanding of the prevalence of this condition in a community could have been obtained by a population-based study or by including the other local hospitals.

CONCLUSION

The study emphasizes that women of reproductive age have a higher prevalence of beta-thalassemia trait. A prospective, multicenter study on the same subject to allow for the generalization of findings. Confirmation requires molecular genetic testing. Programs for targeted screening, particularly for those of reproductive age.

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