Original Article

Frequency of Beta Thalassemia Trait Among Anemic Pregnant Women Presenting to Tertiary Care Hospital

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Abstract

Objectives: This study aimed to assess the frequency of β Thalassemia trait among pregnant patients presenting to a tertiary care hospital.
Methodology: A descriptive cross-sectional study was conducted at the Department of Gynaecology and Obstetrics, Shaikh Zayed Hospital, Lahore. Using a non-probability, consecutive sampling strategy, 200 pregnant women with anemia (hemoglobin < 11 g/dL) in their first and second trimesters were enrolled. Written consent was obtained from all participants. Detailed medical histories, physical examinations, and blood tests, including complete blood count and serum ferritin levels, were performed. Women with serum ferritin levels below 15 µg/dl were classified as having iron deficiency anemia. The presence of β Thalassemia trait was determined by HbA2 levels greater than 3.5%.
Results: The average age of the participants was 26.00 ± 5.34 years. The mean hemoglobin level was 7.68 ± 1.80 g/dl, the mean corpuscular volume (MCV) was 63.26 ± 9.40 fl, and the mean corpuscular hemoglobin (MCH) was 18.54 ± 4.54. Among the 200 women, 38 (19%) had mild anemia, 88 (44%) had moderate anemia, and 74 (37%) had severe anemia. Eighteen (9%) women were found to have the β Thalassemia trait. Specifically, 1.5% of those with mild anemia, 4% with moderate anemia, and 3.5% with severe anemia were diagnosed with the β Thalassemia trait.
Conclusion: The β Thalassemia trait was identified in 9% of the anemic pregnant women in this study. These findings highlight the need for routine screening and genetic counseling to manage and prevent β Thalassemia, particularly in populations with high prevalence and risk factors such as consanguineous marriages.
Keywords: Beta Thalassemia, Pregnant, Morbidity, Anemia, Screening, Genetic Counseling

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Introduction

The most frequent causes of microcytosis and hypochromia are beta thalassemia (β-thal) trait and iron deficiency anemia.[1] β-thal is a heterogeneous genetic disorder brought on by mutations in genes that produce beta chains, resulting in reduced or absent beta chains, which lowers erythropoiesis, anemia, and the amount of haemoglobin in erythrocytes.[2] Thalassemia is inherited as an autosomal recessive trait. Males and females with thalassemia are equally distributed, with a global incidence of 4.4 per 10,000 live births. In Pakistan, iron deficiency anemia is the most common kind among expectant mothers.[3-7]

In Pakistan, the trait frequency of β-thal ranges from 5.0 to 7.0%, meaning that more than ten million individuals are carriers. Approximately 5000 children are diagnosed with β-thal major (β-TM) each year, but only 10-15% of them receive the best possible care.[8] A single thalassemic youngster requires yearly treatment costs of Rs. 6000$.[9] The majority of patients cannot afford bone marrow transplantation, the only treatment now available. Therefore, the focus should be on preventing these kinds of births. Using a thorough

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screening program to find carriers and offering illness counseling is the most efficient way to reduce the burden of disease in society. In Pakistan, thalassemia is highly prevalent, and screening is a crucial step in preventing the disease from being passed down through the generations. Some investigations have shown that thalassemia-related pregnancy complications include low birth weight, restricted intrauterine development, and premature labor.[10,11] The implications of thalassemia characteristics on various obstetric problems and their pathogenesis are still unknown. Pregnancy-related physiological changes, such as a 30% increase in red blood cell volume and a 40% increase in total blood volume, precipitate anemia in women with thalasemia trait, despite the fact that these women typically do not experience any symptoms before getting pregnant.[12]

Methodology

This descriptive cross-sectional study was conducted at Shaikh Zayed Hospital Lahore's Department of Gynaecology and Obstetrics. The method used was a non-probability, consecutive sampling strategy. This study included 200 pregnant women who met the inclusion criteria. In all cases, written approval was obtained. Detailed medical histories and physical examin0ations were conducted on all females. Blood drawn in EDTA (an anticoagulant) was analysed for a complete blood count. The Sysmex KX2100, a three-part haematology analyzer, was used to check the haemoglobin and RBC indices, among other haematological parameters. Additional tests included the use of peripheral blood smears. All samples were examined for serum ferritin. Serum ferritin levels below 15 µg/dl indicate iron deficient anaemia. Women were found to be carriers of beta thalassemia if their HbA2 levels were more than 3.5%.

Anemic pregnant women (hemoglobin < 11 g/dL) in first and 2nd trimester presenting to the hospital for antenatal care were included in the study**.** Patients already diagnosed with haemoglobinopathies were excluded.

Results

The average age of the cases was 26.00 ± 5.34. A total of 200 females were enrolled in this study. The mean Hb was 7.68 ± 1.80 g/dl, the mean corpuscular volume (MCV) was 63.26 ± 9.40 fl, and the mean corpuscular hemoglobin (MCH) was 18.54 ± 4.54. Mean serum ferritin was 12.61 ± 2.99 ug/dl. In this study 38 (19%) females were found with mild anemia, 88 (44%) with moderate anemia and 74 (37%) with severe anemia. From 200 patients 18 (9%) found to have beta thalassemia trait with HB A2 > 3.5%. (Table I).

Table I: Demographics and clinical parameters.

Demographics and clinical parameters	
Age (Mean ± S.D)	26.00 ± 5.34
HB g/dl (Mean ± S.D)	7.68 ± 1.80
MCV fl (Mean ± S.D)	63.26 ± 9.40
MCH g/dl(Mean ± S.D)	18.54 ± 4.54
Serum Ferritin µg/dl	12.61 ± 2.99
Anemia	
Mild (Hb >10-11)	38 (19%)
Moderate (Hb 7-10)	88 (44%)
Severe (Hb < 7)	74 (37%)
Electrophoresis Pattern	
Normal	182 (91%)
Trait (HB A2 > 3.5%)	18 (9%)

In this study in mild anemia group 3 (1.5%) females diagnosed with beta thalassemia while in moderate anemia group 8 (4%) and severe anemia group 7 (3.5%) females found with beta thalassemia. (Figure 1)

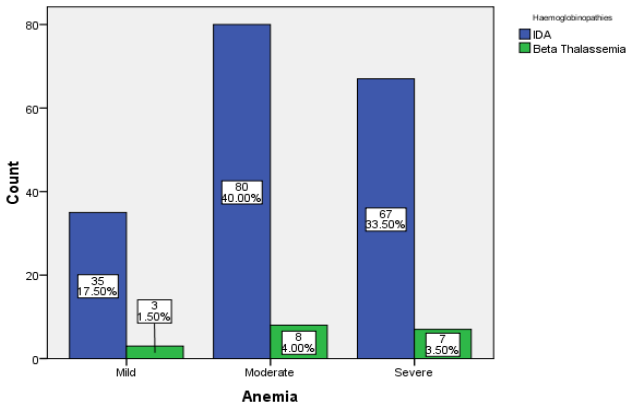

Figure 1. Frequency of thalassemia trait in anemia groups.

Discussion

Thalassemia is defined by a decrease or absence in the synthesis of one or more globin chains. β-thal is considered to be one of the most prevalent monogenic disorders globally. Most newborns with thalassemia major are born in developing countries as a result of insufficient governmental prevention measures.[13-14] If both parents are carriers, there is a 50% probability that the kid would be born with β thalassemia minor, a 25% probability of inheriting β-thal major, and a 25% probability of being born without the condition. Individuals diagnosed with beta thalassemia minor usually encounter a moderate form of anemia and do not exhibit any symptoms. The majority of clinicians

incorrectly diagnose it as iron deficiency anemia and suggest hematinic therapy. Microcytosis, hypochromia, and mild anemia are all shown by the blood test.

In this study the average age of the cases was 26.00 ± 5.34. The mean age of the cases in a published study by Iqbal et al, (2018) was recorded as 26.4 ± 4.3 years.[15] The mean Hb was 7.68 ± 1.80 g/dl, the mean corpuscular volume (MCV) was 63.26 ± 9.40 fl, and the mean corpuscular hemoglobin (MCH) was 18.54 ± 4.54. Similar findings were recorded in a previous study i.e., mean hb 8.5±0.5g/dl, mean MCV 64.32±1.56fl, and mean MCH 21±0.56 g/dl.[15]

In this study 38 (19%) females were found with mild anemia, 88 (44%) with moderate anemia and 74 (37%) with severe anemia. Likewise, in another study 20 (17%) females had mild anemia 85 (71%) had moderate anemia and 15 (12%) had severe anemia.[15]

In our study from 200 patients 18 (9%) found with beta thalassemia trait. Iqbal et al. (2018) found that 7.5% of pregnant anemic patients were β thalassemia carriers.[15] Kulkarni et al. (2015) found that 18 (8.5%) of 210 pregnant women were thalassemia carriers.[16] Similarly, Rizwan et al investigation on pregnant women found an 8.5% carrier prevalence.[17] Mustafa et al. (2018) discovered a 4.9% prevalence of the beta thalassemia traits. [18]

In Pakistan, decreased literacy rates, a rise in consanguineous marriages, and a failure to execute preventive programs all contribute to a significant number of thalassemia cases born each year. Prevention is the most efficient approach to address the magnitude of the thalassemic issue. It is recommended to offer heightened awareness programs and premarital screenings for couples. This strategy has effectively been employed in other regions, such as Saudi Arabia, resulting in a 70% reduction in the occurrence of beta-thalassemia within a span of six years. [19]

Conclusion

The β Thalassemia trait was identified in 9% of the anemic pregnant women in this study. These findings highlight the need for routine screening and genetic counseling to manage and prevent β Thalassemia, particularly in populations with high prevalence and risk factors such as consanguineous marriages.

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