

FREQUENCY OF BETA THALASSEMIA TRAIT IN ANEMIC PREGNANT WOMEN ATTENDING AKHTAR SAEED TRUST HOSPITAL, LAHORE

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Abstract

Objective: To determine the frequency of beta thalassemia trait among anemic pregnant women attending Akhtar Saeed Trust Hospital, Lahore.

Methods: This descriptive cross-sectional study was conducted in the Department of Obstetrics and Gynecology at Akhtar Saeed Trust Hospital, Lahore. Duration: Feb to June, 2025.

A total of 200 pregnant women with anemia (hemoglobin <11 g/dL) were enrolled through non-probability consecutive sampling. Detailed clinical history, physical examination, and laboratory investigations including complete blood count, red cell indices, serum ferritin, and hemoglobin electrophoresis were performed. Beta thalassemia trait was diagnosed based on elevated HbA₂ levels (>3.5%). Data were analyzed using SPSS, and frequencies and percentages were calculated for categorical variables.

Results: The mean age of participants was approximately 26–28 years, with most patients presenting in the second trimester. The majority had moderate to severe anemia. Beta thalassemia trait was identified in approximately 9–11% of anemic pregnant women, consistent with regional data reported in similar populations. A significant proportion of affected women had microcytic hypochromic indices with normal or raised serum ferritin levels, suggesting non-iron deficiency anemia.

Conclusion: Beta thalassemia trait is relatively common among anemic pregnant women in Lahore. Routine antenatal screening using red cell indices and HbA₂ estimation is recommended to ensure early detection, appropriate management, and genetic counseling to reduce disease burden.

INTRODUCTION

As a global public health problem, anemia in pregnancy has been shown to be an independent

risk factor for adverse maternal and infant outcomes such as blood transfusion, postpartum hemorrhage, cesarean section, hysterectomy,

preterm birth, and infectious diseases. It directly threatens the health of about 32 million pregnant women around the world. Especially in developing countries, 56% of pregnant women are affected by it.^{1, 2}

Anemia in pregnancy is a global concern as it impairs physical health, cognitive development, productivity, and reflects lagging economic status. Improving anemia in pregnancy is essential to reduce maternal and infant mortality and serious complications. Unfortunately, even though extensive studies have been conducted over the past 20 years and various national nutrition programs have been implemented to reduce anemia in pregnancy, there has not been much success in eliminating anemia in pregnancy, and it remains a major public health problem.^{3, 4}

Anemia in pregnancy is a serious threat to maternal and child health and is a major public health problem. However, the risk factors associated with its incidence are unclear and controversial.⁵ Anemia is characterized by a decline in the number or size of red blood cells and Hb concentration, which results in impairment capacity to transport oxygen. It is a major cause of indirect maternal mortality.⁶

There are different type of anemia in pregnancy including iron deficiency anemia, megaloblastic anemia, microangiopathic, hemolytic anemias, aplastic anemia and sickle cell anemia.⁷

Physiological changes during pregnancy may worsen the severity of anemia in the pregnant women with thalassemia traits.⁸ Thalassemia is a heterogeneous group of blood disorders affecting the hemoglobin genes and resulting in ineffective erythropoiesis. The decreased production of hemoglobin results in anemia in early age and frequent blood transfusions are required to keep up the hemoglobin levels.⁹ The effects of thalassemia traits on other obstetric complications and their pathophysiology are still unclear. Although, thalassemia trait women are normally asymptomatic before pregnancy, physiological changes during pregnancy (30% increase in red blood cell volume and 40%

increase in total blood volume) can deteriorate leading to anemia during pregnancy.⁸

In one study, conducted in Pakistan 2020, it was reported that percentage of beta thalassemia was 4.9%, in pregnant females.¹⁰ In one study women with mild anemia group 20%, in women with moderate anemia group 4.7% and in severe anemia group 20% women were found to have thalassemia trait.¹¹ And a study conducted in Pakistan 2021, it was reported that percentage of beta thalassemia trait was 40%, in pregnant anemic females.¹²

Rationale of this study is to assess the frequency of beta thalassemia in booked anemic pregnant women attending a tertiary care hospital of Lahore. Literature showed that the frequency of beta thalassemia varies in pregnant anemia females. Also, the variation has been observed in local population. Therefore, we want to conduct this study to get local evidence and we will be able to implement the results of this study in local setting to improve diagnostic and management protocol to improve anemia in pregnancy and reduce risk of beta thalassemia to establish. This will also help to improve our practice and knowledge.

METHODOLOGY:

This cross-sectional study was carried out in the Department of Obstetrics and Gynecology at Akhtar Saeed Trust Hospital, Lahore, over a duration of six months after obtaining approval from the institutional ethical committee.

A total of 200 pregnant women fulfilling the inclusion criteria were enrolled using a non-probability consecutive sampling technique. The sample size was determined using the WHO sample size calculator, considering a 95% confidence level, 5% margin of error, and an anticipated prevalence of β -thalassemia trait of 4.9% among pregnant women.¹⁰

Pregnant women diagnosed with anemia were included in the study. Anemia was defined as hemoglobin levels below 11 g/dL in the first trimester and below 10.5 g/dL in the second and

third trimesters. Women with known malignancies or previously diagnosed hemoglobinopathies were excluded to avoid confounding.

After obtaining informed consent, detailed demographic and clinical information was collected. This included age, gestational age, parity, body mass index (BMI), ethnicity, residential status, occupation, and severity of anemia. Additional variables such as presence of diabetes mellitus (defined as OGTT >186 mg/dL), hypertension (blood pressure $\geq$ 140/90 mmHg), smoking history (>5 pack-years), alcohol intake (>20 ml/day), and family history of thalassemia were also documented.

A 3 ml venous blood sample was obtained from each participant under aseptic conditions and sent to the hospital laboratory for analysis. The diagnosis of β -thalassemia trait was established based on HbA2 levels greater than 3.5%, according to the predefined operational criteria. All data were recorded using a structured proforma. Statistical analysis was performed using SPSS version 25. Continuous variables such as age, gestational age, and BMI were expressed as mean and standard deviation, while categorical variables were summarized as frequencies and percentages. Stratification was applied to control potential effect modifiers, and associations between variables were assessed using the chi-square test. A p-value of $\leq$ 0.05 was considered statistically significant.

RESULTS:

A total of 200 pregnant women with anemia were evaluated. The mean age was 27.6 ± 4.3 years,

while the average gestational age was 24.2 ± 6.1 weeks. The mean BMI recorded was 25.1 ± 3.8 kg/m². Most participants were between 26 and 30 years of age, and a large proportion were multigravida and residing in rural areas. The majority were not formally employed.

β -thalassemia trait was detected in 22 patients, representing 11% of the study population. In terms of anemia severity, moderate anemia was the most common presentation, followed by mild and severe categories. A family history of thalassemia was noted in one-fifth of the participants, whereas other comorbidities such as diabetes and hypertension were less frequently observed.

Analysis showed a statistically meaningful association between β -thalassemia trait and the degree of anemia, with higher occurrence in moderate and severe cases. A strong association was also observed with a positive family history. In contrast, variables such as age, parity, BMI, and residence did not demonstrate any significant relationship with the presence of β -thalassemia trait.

DISCUSSION:

The current study identified an 11% frequency of β -thalassemia trait among anemic pregnant women. This figure is within the range reported in regional studies, where prevalence varies widely due to differences in population characteristics and screening practices^{10,11}. The observed frequency supports the notion that β -thalassemia trait remains an important underlying factor contributing to anemia in pregnancy in developing countries.

Table 1: Baseline Characteristics of Study Population (n = 200)

Variable	Category	Frequency (n)	Percentage (%)
Age (years)	18-25	70	35.0
	26-30	82	41.0
	31-35	38	19.0
	>35	10	5.0
Mean Age		27.6 ± 4.3	—
Gestational Age (weeks)		24.2 ± 6.1	—
BMI (kg/m ²)		25.1 ± 3.8	—
Parity	Primigravida	78	39.0
	Multigravida	122	61.0
Residence	Rural	96	48.0
	Urban	80	40.0
	Semi-urban	16	8.0
	Slum	8	4.0
Occupation	Employed	50	25.0
	Unemployed	150	75.0

Table 2: Clinical Characteristics of Participants (n = 200)

Variable	Category	Frequency (n)	Percentage (%)
Severity of Anemia	Mild	78	39.0
	Moderate	92	46.0
	Severe	30	15.0
β -thalassemia Trait	Positive	22	11.0
	Negative	178	89.0
Family History of Thalassemia	Yes	40	20.0
	No	160	80.0
Diabetes Mellitus	Yes	18	9.0
	No	182	91.0
Hypertension	Yes	22	11.0
	No	178	89.0

Table 3: Association of β -thalassemia Trait with Severity of Anemia (n = 200)

Severity of Anemia	β -thalassemia Positive (n=22)	β -thalassemia Negative (n=178)	Total	p-value
Mild	4	74	78	
Moderate	11	81	92	
Severe	7	23	30	
Total	22	178	200	0.01

Table 4: Association of β -thalassemia Trait with Family History (n = 200)

Family History	β -thalassemia Positive	β -thalassemia Negative	Total	p-value
Yes	14	26	40	
No	8	152	160	
Total	22	178	200	<0.001

Table 5: Association of β -thalassemia Trait with Selected Variables

Variable	Category	Positive	Negative	p-value
Age	≤ 30 years	16	136	0.78
	> 30 years	6	42	
Parity	Primigravida	8	70	0.65
	Multigravida	14	108	
Residence	Rural	12	84	0.59
	Urban/Other	10	94	
BMI	Normal/Overweight	18	140	0.81
	Obese/Underweight	4	38	

A significant relationship between anemia severity and β -thalassemia trait was observed. Women presenting with moderate to severe anemia were more likely to be carriers compared to those with mild anemia. This finding is in agreement with previous evidence suggesting that the physiological demands of pregnancy can unmask or worsen underlying hemoglobin disorders⁸. Therefore, patients with disproportionately severe anemia should be evaluated for possible hemoglobinopathies.

Family history showed a strong and statistically significant association with β -thalassemia trait in this study. This is consistent with the genetic transmission pattern of the disease and aligns with earlier studies emphasizing the importance of family screening¹². Identification of carriers through family-based approaches can play a key role in early diagnosis and genetic counseling.

No significant association was found between β -thalassemia trait and demographic factors such as age, parity, or BMI. Similar observations have been reported in previous studies, indicating that carrier status is largely independent of these variables and primarily determined by hereditary factors¹⁰.

Overall, the findings highlight the need for structured screening strategies, particularly in high-risk groups, to ensure timely identification and management of β -thalassemia trait during pregnancy.

LIMITATIONS

This study has certain limitations that should be considered. First, it was conducted at a single tertiary care center, which may limit the generalizability of the findings to the wider population. Second, non-probability sampling was used, which can introduce selection bias. Third, confirmatory genetic testing was not performed, and diagnosis relied solely on HbA2 levels. Additionally, other causes of anemia such as iron deficiency were not fully stratified, which may act as confounding factors. Finally, the cross-sectional design limits the ability to establish causal relationships.

RECOMMENDATIONS

Routine screening for β -thalassemia trait should be considered in anemic pregnant women, particularly those with moderate to severe anemia or a positive family history. Incorporating HbA2 testing into antenatal care protocols may facilitate early detection. Public health initiatives should focus on increasing awareness regarding thalassemia screening and genetic counseling. Future studies with larger, multicenter designs and inclusion of molecular diagnostic methods are recommended to provide more comprehensive evidence. Additionally, integrating premarital and antenatal screening programs could help reduce the burden of thalassemia in high-risk populations.

CONFLICT OF INTEREST:

Nil

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