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PREVALENCE OF THALASSEMIA IN A TERTIARY CARE HOSPITAL IN MULTAN, PAKISTAN

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Abstract

Thalassemia is a significant inherited hemoglobin disorder that contributes to chronic anemia and poses a public health burden in South Asia. This cross-sectional study assessed the prevalence and hematological patterns of thalassemia among 150 patients in a tertiary care hospital in Multan. Hemoglobin fractions and red cell indices were analyzed using standard laboratory methods. The majority of participants were male (81.3%) with a mean age of 35.26 ± 17.72 years. Reduced MCV (67.60 fL) and MCH (20.05 pg) indicated microcytic anemia. Elevated HbA2 (>3.5%), suggestive of β-thalassemia trait, was observed in 61.3% of patients, while 17.3% showed increased HbF (>30%). Hemoglobin levels varied significantly across age groups ($p = .035$), with the lowest levels in children. A strong positive correlation was found between Hb and HbA1 ($r = .744$), and a negative correlation with HbF ($r = -.786$, $p < .001$). The findings highlight β-thalassemia trait as the most prevalent condition and emphasize the need for early screening and preventive strategies. The findings confirm that β-thalassemia trait is the most prevalent hemoglobin disorder in the studied population. Reduced red cell indices align with previous studies identifying microcytic anemia as a hallmark feature. Elevated HbA2 levels serve as a reliable diagnostic indicator of β-thalassemia carriers. The strong inverse relationship between Hb and HbF suggests increased disease severity with higher fetal hemoglobin levels. Age-related differences further highlight the vulnerability of pediatric populations.

INTRODUCTION

Thalassemia is one of the most common inherited hematological disorders worldwide, characterized by defective hemoglobin synthesis leading to chronic hemolytic anemia of varying severityincludes alpha (α) and beta (β) thalassemia, with β-thalassemia being the most clinically significant form due to its frequent requirement for lifelong blood transfusions. Clinical presentation ranges from asymptomatic carriers to severe transfusion-dependent thalassemia (TDT),

which imposes a substantial clinical and economic burden on affected individuals and healthcare systems.

Repeated blood transfusions in TDT patients often lead to iron overload, resulting in multi-organ complications such as cardiac failure, liver dysfunction, endocrine abnormalities, and impaired growth and development. If left untreated, severe β-thalassemia can lead to early mortality, particularly in low-resource settings where access to specialized care remains limited.

Globally, thalassemia represents a major public health concern. According to the World Health Organization, thousands of children are born annually with β-thalassemia major, with a disproportionately high burden in South Asia, the Middle East, and Mediterranean regions. Pakistan lies within the "thalassemia belt," where high carrier frequency, consanguineous marriages, and limited premarital screening contribute significantly to disease prevalence and transmission risk.

Despite its increasing burden, accurate hospital-based epidemiological data remain limited in many developing regions, including Southern Punjab. Most existing data are derived from fragmented registries or small-scale studies, making it difficult to estimate the true prevalence and distribution of thalassemia in clinical populations.

Therefore, this study aims to determine the prevalence of thalassemia in a tertiary care hospital in Multan, Pakistan, and to analyze its distribution across demographic variables and risk factors. The findings are expected to contribute to improved screening strategies, early diagnosis, and preventive genetic counseling programs.

1.1 Objectives of the Study

1. To determine the distribution of thalassemia according to age and gender.
2. To estimate the prevalence of thalassemia in the study population.
3. To identify the frequency of thalassemia carriers.
4. To assess family history as a significant risk factor for thalassemia.

1.2 Research Questions

1. What is the distribution of thalassemia among different age groups and genders?
2. What is the prevalence of thalassemia in the study population?
3. What is the frequency of thalassemia carriers?
4. Is family history a significant risk factor for thalassemia?

1.3 Significance of the Study

This study will help identify high-risk demographic groups for targeted screening and early diagnosis. It will also provide evidence for the burden of thalassemia in a tertiary care setting, supporting healthcare planning, resource allocation, and strengthening genetic counseling and prevention programs. Additionally, identifying carrier frequency and familial risk patterns will contribute to reducing future disease transmission.

Existing literature highlights thalassemia as a persistent global health burden with significant variability in prevalence across regions. Studies indicate that β-thalassemia major is associated with severe clinical complications including iron overload, growth retardation, endocrine dysfunction, and increased mortality, particularly in low-income settings with limited access to chelation therapy and regular transfusions (Esmaeilzadeh et al., 2021).

Research also shows that early screening and preventive strategies are more cost-effective than long-term treatment of affected individuals, emphasizing the importance of carrier detection and premarital screening programs (Vanness et al., 2021; Iyevhobu et al., 2023). However, screening coverage remains limited in many high-burden countries, including Pakistan, where awareness of genetic risks and preventive services is still low.

In South Asia, thalassemia prevalence remains high due to consanguineous marriages, lack of genetic counseling, and inadequate screening systems. Studies conducted in Pakistan and neighboring regions report substantial carrier frequencies and significant variation in disease burden across populations (Pal et al., 2025; Rao et al., 2024).

Furthermore, literature emphasizes that hospital-based prevalence studies are essential for understanding the actual disease burden, as population-level estimates often underestimate clinical cases due to underreporting and lack of registries. Variability in diagnostic approaches and inconsistent data collection methods further limit accurate epidemiological estimation (Bellis & Parant, 2021).

Overall, previous studies strongly support the need for localized hospital-based research to determine prevalence patterns, identify high-risk groups, and guide prevention strategies. This study addresses this gap by examining thalassemia prevalence in a tertiary care hospital in Multan, Pakistan.

Methodology

Study Design and Sample Characteristics

This study employed a cross-sectional design to determine the prevalence and hematological profile of thalassemia among patients attending a tertiary care hospital in Multan, Pakistan. A total of 150 patients were included in the analysis. The study aimed to examine the distribution of thalassemia based on age and gender, estimate its prevalence, identify carrier frequency, and assess family history as a risk factor.

Research Design: Cross-sectional study
Sample Size: 150 patients
Setting: Tertiary care hospital, Multan
Data Collection: Blood samples analyzed for hemoglobin fractions (HbA1, HbA2, HbF) and hematological indices (Hb, MCV, MCH, MCHC)
Statistical Analysis: One-way ANOVA, Chi-square test, Pearson correlation, and independent t-test
Ethical consideration were followed.

Results

4.1 Demographic and Hematological Characteristics

Table 1 presents the demographic and baseline hematological profile of the study population. A total of 150 patients were included. The mean age was 35.26 ± 17.72 years, indicating representation across pediatric and adult populations. The sample was predominantly male (81.3%). Hematological indices showed reduced mean hemoglobin (11.18 ± 2.96 g/dL), low MCV (67.60 ± 6.19 fL), and low MCH (20.05 ± 2.81 pg), suggesting a high prevalence of microcytic hypochromic anemia consistent with hemoglobinopathies such as thalassemia.

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Table 1: ***Demographic and Hematological Characteristics of the Study Sample (N = 150)***

Variable	Mean ± SD / n (%)	Range
Age (years)	35.26 ± 17.72	5–70
Gender		
Male	122 (81.3%)	—
Female	28 (18.7%)	—
Hemoglobin (g/dL)	11.18 ± 2.96	3.5–16.8
MCV (fL)	67.60 ± 6.19	55.4–85.8
MCH (pg)	20.05 ± 2.81	14.1–29.0
MCHC (g/dL)	29.43 ± 2.28	22.2–34.2

Note. SD = Standard Deviation; MCV = Mean Corpuscular Volume; MCH = Mean Corpuscular Hemoglobin; MCHC = Mean Corpuscular Hemoglobin Concentration.

4.2 Age Distribution and Hemoglobin Levels

Table 2 shows the distribution of patients across age groups with corresponding mean hemoglobin levels. The majority of patients belonged to the young adult group (44%). Children (≤12 years) showed the lowest hemoglobin levels (8.64 ± 3.37 g/dL), indicating greater severity of anemia in early age presentation.

Table 2: ***Age Group Distribution and Mean Hemoglobin Levels***

Age Group	n (%)	Mean Hb ± SD (g/dL)
Child (≤12 years)	10 (6.7%)	8.64 ± 3.37
Adolescent (13–18 years)	10 (6.7%)	11.81 ± 1.88
Young Adult (19–35 years)	66 (44.0%)	11.68 ± 2.86
Middle Age (36–60 years)	49 (32.7%)	10.89 ± 2.83
Elderly (>60 years)	15 (10.0%)	11.25 ± 3.43
Total	**150 (100%)**	**11.18 ± 2.96**

4.3 Prevalence of Thalassemia (Hemoglobin Pattern Distribution)

Table 3 represents the distribution of hemoglobin patterns used to identify thalassemia cases and carriers. The most common pattern was elevated HbA2 (>3.5%), observed in 61.3% of patients, indicating a high prevalence of beta-thalassemia trait. High HbF levels were observed in 17.3%, suggesting possible beta-thalassemia major or hereditary persistence of fetal hemoglobin.

Table 3: *Distribution of Hemoglobin Patterns in the Study Population*

Hemoglobin Pattern	n	%
Elevated HbA2 (>3.5%)	92	61.3%
High HbF (>30%)	26	17.3%
Normal Pattern	21	14.0%
Moderate HbF (10–30%)	11	7.3%
Total	**150**	**100%**

Note. Elevated HbA2 is strongly suggestive of beta-thalassemia trait (carrier state).

4.4 Inferential Analysis of Group Differences

Table 4 summarizes statistical comparisons across gender, age groups, and hemoglobin patterns. A significant difference in hemoglobin levels was observed across age groups ($p = .035$). However, no significant association was found between gender and hemoglobin pattern ($p = .123$). Only MCV showed a statistically significant gender difference.

Table 4: ***Inferential Statistics for Group Comparisons***

Analysis	Test	Statistic	p-value	Interpretation
Gender vs Hematological parameters	t-test	MCV: $t = -2.209$	0.028*	Significant
Gender vs Hemoglobin pattern	Chi-square	$\chi^2 = 5.777$	0.123	Not significant
Age group vs Hemoglobin	ANOVA	$F = 2.660$	0.035*	Significant
Hb (gender comparison)	t-test	$t = 1.904$	0.059	Not significant

*Note. $p < .05$ indicates statistical significance.

4.5 Correlation Between Hemoglobin and Hematological Parameters

Table 5 presents Pearson correlation analysis. Hemoglobin showed a strong positive correlation with HbA1 and MCHC, and a strong negative correlation with HbF. These findings confirm that elevated fetal hemoglobin is associated with reduced total hemoglobin, a characteristic pattern in thalassemia.

Table 5: ***Pearson Correlation Between Hemoglobin and Hematological Variables***

Variable Pair	r	p-value	Strength
Hb vs MCV	0.253	0.002**	Weak positive
Hb vs MCH	0.440	<.001**	Moderate positive
Hb vs MCHC	0.655	<.001**	Strong positive
Hb vs HbA1	0.744	<.001**	Strong positive
Hb vs HbA2	0.188	0.021*	Weak positive
Hb vs HbF	-0.786	<.001**	Strong negative

*Note. *p < .05, *$p < .01$.

Summary of Key Findings (Aligned with Objectives)

- The prevalence of abnormal hemoglobin patterns (thalassemia-related markers) was high, with **61.3% showing elevated HbA2**, indicating a high burden of **beta-thalassemia trait**.
- Males were more frequently affected than females (81.3%).
- Age was significantly associated with hemoglobin levels (p = .035), supporting its role as a modifying factor.
- No significant relationship was found between gender and hemoglobin pattern (p = .123).
- Strong correlations between Hb, HbF, and HbA2 confirm classical thalassemia hematological patterns.

Discussion

The present study investigated the prevalence of thalassemia and associated hematological patterns in a tertiary care hospital in Multan, with a focus on age and gender distribution, frequency of affected individuals, carrier patterns, and potential risk factors such as family history. The findings provide important insight into the burden of hemoglobinopathies in a clinical population within a high-risk region.

Prevalence of Thalassemia and Hemoglobin Pattern Distribution

The most significant finding of this study was that **61.3% of patients exhibited elevated HbA2 (>3.5%)**, which is a well-established diagnostic marker of **beta-thalassemia trait**. This high proportion indicates a substantial burden of thalassemia carriers within the study population. Additionally, **17.3% of patients showed elevated HbF (>30%)**, which is commonly associated with beta-thalassemia major or hereditary persistence of fetal hemoglobin (HPFH)**.**

These findings are consistent with previous hospital-based studies in Pakistan, where carrier frequencies are often elevated due to high rates of consanguineous marriages and limited premarital genetic screening programs. Similar prevalence patterns have been reported in tertiary care settings across South Asia, where beta-thalassemia trait remains the most frequently identified hemoglobinopathy among anemic patients (Khanzada et al., 2024). The predominance of elevated HbA2 in this study reinforces the continued diagnostic importance of hemoglobin fraction analysis (HPLC) in identifying silent carriers in clinical populations.

Age and Gender Distribution of Thalassemia

The study found that the majority of patients were **young adults (44.0%)**, followed by middle-aged individuals (32.7%). Hemoglobin levels varied significantly across age groups ($p = 0.035$), with children (≤12 years) showing the **lowest mean hemoglobin (8.64 g/dL)**. This pattern suggests that disease severity or early clinical manifestation is more pronounced in pediatric cases, which is consistent with the clinical presentation of beta-thalassemia major in early childhood.

These findings align with Muriuki (2021), who reported that thalassemia-related anemia is often more severe in children due to early genetic expression and increased physiological demand for hemoglobin during growth. The statistically significant ANOVA result further supports the need for age-stratified diagnostic interpretation in hemoglobinopathy screening.

In terms of gender distribution, the study population was predominantly male (81.3%). However, no statistically significant association was found between gender and hemoglobin pattern ($p = 0.123$). This suggests that thalassemia and related hemoglobin disorders are **biologically independent of gender**, and the observed male predominance is more likely due to **health-seeking behavior, referral bias, or hospital-based sampling patterns** rather than true epidemiological differences. Similar findings have been reported in other regional studies, where gender imbalance in hospital samples reflects access-to-care differences rather than disease prevalence (Al-Maaroof & Al Sharifi, 2025).

Hematological Correlations and Pathophysiological Insights

The correlation analysis provided important insight into the hematological behavior of hemoglobin disorders. A **strong negative correlation between Hb and HbF ($r = -0.786$, $p < 0.001$)** indicates that higher fetal hemoglobin levels are associated with lower total hemoglobin concentration. This is a characteristic feature of beta-thalassemia major and related conditions, where defective beta-globin synthesis leads to compensatory but insufficient gamma-chain production.

Conversely, a **strong positive correlation between Hb and HbA1 ($r = 0.744$, $p < 0.001$)** suggests that preservation of normal adult hemoglobin is associated with better overall hematological status. These findings are in agreement with established literature indicating that increased HbF is a compensatory response but does not fully restore oxygen-carrying capacity in thalassemia patients (Weatherall & Clegg, 2021).

Moderate and weak correlations between Hb and other red cell indices (MCV, MCH, and HbA2) further support the classical microcytic hypochromic pattern observed in thalassemia syndromes. The significantly reduced MCV and MCH values across the sample reinforce the diagnostic profile of hemoglobinopathies in this population.

Clinical and Public Health Implications

The findings highlight a **high burden of beta-thalassemia trait in a tertiary care setting**, which reflects the broader genetic risk in the population of southern Punjab. The observed prevalence emphasizes the urgent need for **early screening, premarital genetic counseling, and public awareness programs** to reduce disease transmission.

Countries such as Iran and Cyprus have successfully reduced thalassemia incidence through mandatory premarital screening and national prevention programs, demonstrating that structured public health interventions can significantly reduce disease burden (Daniels et al., 2025). Similar strategies could be effectively adapted in Pakistan, particularly in high-prevalence regions such as Multan.

Recommendations

- Implement nationwide screening programs
- Promote premarital genetic counseling
- Increase awareness regarding consanguinity risks
- Strengthen early diagnostic facilities

Limitations of the Study

Despite its important findings, this study has certain limitations. First, the **hospital-based cross-sectional design** introduces selection bias, limiting generalizability to the broader community population. Second, the absence of **molecular genetic testing** restricts confirmation of specific thalassemia mutations. Third, the predominance of male participants may affect representativeness. Finally, family history data was not deeply explored in statistical modeling, limiting analysis of hereditary risk factors.

Conclusion of Discussion

Overall, this study demonstrates a **high prevalence of beta-thalassemia trait (61.3%)** among patients in a tertiary care hospital in Multan, with significant age-related variation in hemoglobin levels and strong hematological correlations consistent with classical thalassemia pathology. These findings underscore the clinical importance of early detection, laboratory screening, and preventive genetic counseling. The results also highlight the urgent need for structured public health interventions to reduce the burden of thalassemia in high-risk populations of Pakistan.

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