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DEMOGRAPHIC AND CLINICAL PROFILE OF THALASSEMIC PATIENTS: A RETROSPECTIVE STUDY

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Abstract

The clinical, demographic, and genetic risk factors linked to transfusion-dependent beta-thalassemia in a Pakistani cohort were examined in this retrospective case-control study. In October 2024 and January 2025, 100 patients and 100 age- and gender-matched healthy controls were surveyed. SPSS v27 was used for statistical analysis, which included multivariate logistic regression, chi-square, and t-tests. Compared to controls, thalassemia patients were significantly younger (mean age 11.87 vs. 22.24 years, p<0.0001) and had lower BMI, height, and weight (p<0.0001). The associations between thalassemia and consanguinity (58% vs. 16%) and family history (94% vs. 22%) were significant (p<0.00001). Consanguinity (OR=2.12), age (OR=1.24), low height (OR=0.75), and family history (OR=7.54) were all found to be independent risk factors by regression analysis. Clinically, 64% received a diagnosis prior to the age of six; 67% had splenomegaly, and 72% had iron overload. The serum ferritin levels of older patients were significantly higher (1580.7 ng/ml vs. 1300.5 ng/ml, p=0.014). Younger patients needed more frequent transfusions, while older patients were more likely to receive iron chelation therapy (OR=3.12). According to the findings, the transmission and severity of disease are influenced by genetic inheritance, especially in consanguineous marriage.

Keywords: *Beta-thalassemia major; Consanguinity; Iron overload; Growth impairment; Serum ferritin; Transfusion-dependent anemia.*

Introduction

Thalassemia is group of inherited autosomal recessive blood disorders that leads to abnormal or no synthesis of one or more globular chain of hemoglobin (Shafique *et al*., 2021). The protein hemoglobin is present in red blood cells and is responsible for transportation of oxygen from the lungs to different regions of the body (Fucharoen & Winichagoon, 2002). Due to defective hemoglobin, the RBCs produced lose the ability to carry oxygen hence leading to anemia. The hemoglobin protein is made up of four alpha-globin and two beta-globin protein chains (Sanchez-Villalobos *et al*., 2022). The type of thalassemia, either alpha or beta, depends on which of the chain is affected (Muncie Jr & Campbell, 2009). The mutations in α (*HBA1/HBA2*) and β globin (*HBB*) lead to abnormal and structurally defective hemoglobin molecule induces impaired erythropoiesis (Ribeil *et al*., 2013). The degree of mutation calculates the severity of thalassemia (Angastiniotis & Lobitz, 2019). Beta-thalassemia, also called Cooley's anemia is the most severe form of major thalassemia (Iyevhobu *et al*., 2023). It requires regular and lifelong blood transfusions and iron chelation therapy to manage iron overload (Weiner *et al*., 1978).

Thalassemia is known to be one of the most prevalent inherited blood disorders worldwide, affecting populations in the Middle East, South Asia, and the Mediterranean region (Modell &

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Darlison, 2008; Weatherall & Clegg, 2001). In 2021, the worldwide number of thalassemia cases was 1,310,407 with an age-standardized prevalence rates (ASPR) of 18.28 per 100,000 persons (Tuo et al., 2024). In Middle and Southeast Asia, India and parts of Africa, thalassemia gene is most common *(Colah et al., 2010; Fucharoen & Winichagoon, 1992; Li, 2017).* In Pakistan, thalassemia is a serious public health hazard and its estimation among the population is about 5-7% carriers (Khaliq, 2022). The financial and social burden of managing thalassemia, including frequent medical visits, regular blood transfusions and iron chelation therapy is high *(Shafie et al., 2021; Kaur, n.d.)*. Despite progress in medical treatment, thalassemia remains a major healthcare challenge, with patients combating with chronic impairment such as anemia, iron overload and bone deformities (Piriyakhuntorn *et al*., 2020).

This retrospective analysis studies the diverse clinical and demographic factors of thalassemic patients depending on the data collected using structured performas. These not only included detailed information on patient demographics like age, weight, height but also their diagnostic intervals, family history, iron chelation, the frequency of blood transfusions and consequences. A particular attention is given to the role of consanguinity, specifically cousin marriages, as a major contributing factor in prevalence of disease. This study highlights higher risk of autosomal recessive disorders in specific regions where cousin marriages are common. Although multiple awareness campaigns by government or non-government organizations have promoted genetic literacy among people and contributed in controlling the genetic spread of disease (Tariq *et al.*, 2021). Behavioral change has been observed over the years as more couples opt for genetic counseling and voluntarily undergo premarital and prenatal screening. While, some underdeveloped regions still remain under the hues of illiteracy and need more public awareness to combat the disease.

Moreover, the study centers particularly on the high frequency of infections that thalassemic patients acquire through frequent blood transfusions. Transfusion transmitted diseases (TTIs) such as hepatitis C, hepatitis B, and, occasionally HIV, remain to be a serious medical risk, regardless of the implementation of rigorous donor laboratory testing or significant advancements in blood screening procedures. Insufficient facilities in certain blood banks, window periods of infection, and discrepancies in safety protocols among regions are primary root causes of this persistent issue *(Ansari et al., 2024; Lal et al., 2018; Porter, 2009)*. Even a single transfusion safety mishap can result in long-term medical consequence for thalassemia patients who need life-long regular transfusions to maintain their hemoglobin levels (Ricerca, Di Girolamo, & Rund, 2009). Thus, the enduring nature of chronic infections has an enormous effect on the prognosis and overall life quality of challenged patients, besides rendering the management of disease more complicated. To ensure the safest transfusion protocols, more rigorous screening protocols, use of advanced diagnostic techniques and improved surveillance systems ae required.

This retrospective study develops a comprehensive understanding of the multifaceted posed by thalassemia by assessing these multiple aspects, such as demographic profiles, family history, patterns of consanguinity, diagnostic timelines, treatment regimens, transfusion practices, and the incidence of transfusion-related infections. A thorough investigation of the ways in which inherent and acquired risk factors jointly impact patient outcomes is made possible by the integration of both genetic and clinical information. The recognition of susceptible risk factors, such as the elevated frequency of cousin marriages in certain populations and inadequacies in

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transfusion safety protocols, reveals significant data and concentrated attention may have substantial benefits. Besides aiding to access disease's existing impacts on patients and healthcare systems, this multidimensional approach provides an evidence-based framework for effective preventative measures. Promoting public awareness initiatives, boosting genetic counseling services, establishing and implementing laws regulating premarital screenings, and improving the safety and effectiveness of blood transfusion services are just a few instances of such approaches. The ultimate objective of this study's findings is to contribute in the creation of long-lasting, socially appropriate, and analytically sound regulations that minimize the overall incidence of thalassemia and elevate the existing standard of care for those who are impacted by the disease.

Materials and Methods

This retrospective case-control study aimed to assess the relationship between the prevalence of beta-thalassemia and consanguineous marriages, as well as to examine the relationship between patient age, serum ferritin levels, blood transfusion frequency, and family history. Data was collected from Aleem Daar Foundation, Noor Foundation, Ume-Nusrat Foundation and Children Hospital for the study between October 2024 and January 2025 in Lahore, Pakistan. The study included 200 participants, 100 of whom were healthy controls and 100 of whom had been diagnosed with transfusion-dependent beta-thalassemia. Patients had to meet the inclusion requirements for the thalassemia group, which included having a regular transfusion history and a verified diagnosis of transfusion-dependent beta-thalassemia with supporting clinical records. These individuals also had to be free of co-infections, including syphilis, hepatitis B, hepatitis C, and HIV. To ensure that these conditions were ruled out, negative serological reports were examined. The study did not include patients who had a history or currently had any of these infections. Healthy people without a history of blood disorders, chronic illnesses, or genetic diseases made up the control group. In order to minimize confounding variables, participants in this group were chosen to closely resemble the patient group in terms of age and gender distribution. Prior to being enrolled in the study, informed written consent was collected from all participants or their legal guardians.

A comprehensive family history and medical background, including any history of consanguineous marriage within the family, were also gathered in the consent form. Protocols were conducted in compliance with medical research ethics, and ethical approval was acquired. To protect the privacy of the participants, all data were anonymized and kept completely private. The most recent laboratory reports from the individual thalassemia centers were used to determine each patient's serum ferritin levels. Iron overload is frequently seen in patients receiving repeated blood transfusions, and these levels were a sign of it. Additionally, information about the patient's age, gender, monthly blood transfusion frequency, parental consanguinity, and a thorough family history of thalassemia were gathered. To perform statistical analyses, IBM SPSS Statistics version 27 was used. To summarize clinical and demographic features, descriptive statistics were employed. Age, serum ferritin levels, and the frequency of blood transfusions were compared using Pearson correlation coefficients. Variables between groups were compared using independent sample t-tests and one-way analysis of variance (ANOVA). Using parametric tests that were appropriate for the data distribution, correlation analysis was carried out. Statistical significance was defined as a p-value (less than 0.05). To ensure accuracy and statistical integrity, only complete and verifiable datasets were included in

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the final analysis; missing data was not imputed. By using a thorough methodological approach, we were able to examine the clinical, demographic, and genetic factors affecting the severity and complications of beta-thalassemia in a representative sample of the Pakistani population.

Results

The objective of the current study was to assess and contrast a number of clinical, anthropometric, genetic, and demographic characteristics between thalassemia patients and healthy controls. There were 200 participants in all, split equally between a control group and thalassemia patients. To find significant group differences and related risk factors, statistical analyses such as multivariate logistic regression, independent t-tests, and chi-square tests were carried out using SPSS version 22. Strong correlations between thalassemia and a number of risk factors, such as age, genetic background, consanguinity, and clinical complications, were demonstrated by the results. The age distribution showed a notable difference, according to the baseline demographic data (Table 1). While 72% of the control group consisted of adults aged 18 or older, 51% of thalassemia patients were children under the age of 12. The statistical significance of this difference ($p < 0.0001$) suggests that younger people were disproportionately affected by thalassemia. In addition, the mean height and weight of thalassemia patients were significantly lower than those of controls. They were 134.9 cm taller on average than the control group (167.94 cm), and they weighed 28.49 kg instead of 48.5 kg. It appears that thalassemia had a significant effect on physical development because these differences were highly significant ($p < 0.0001$). According to the BMI classification, 40% of patients with thalassemia were underweight, while the control group's underweight percentage was 10% (Table 2).

Both groups were comparable in terms of gender and ethnicity, with 97% identifying as Muslim and an equal gender distribution that was not statistically significant ($p = 0.39$). Hereditary patterns differed widely, though. Compared to 22% in the control group, 94% of patients had a positive family history of thalassemia ($p < 0.00001$). Furthermore, none of the controls had thalassemia-affected siblings, whereas 30% of patients did. In the patient group, maternal cousins also had a significantly higher incidence of thalassemia (16% vs. 2%, $p = 0.0005$). 58% of patient families reported consanguineous marriages, compared to only 16% of control families ($p < 0.00001$), confirming the disease's genetic history (Table 3). A multivariate logistic regression analysis revealed a number of thalassemia predictors that were statistically significant. There was an inverse relationship between height and BMI ($p = 0.0001$ and $p = 0.03$), and age was linked to an increased risk (OR = 1.24, $p = 0.007$). Consanguinity (OR = 2.12, $p = 0.027$), siblings with thalassemia (OR = 2.35, $p = 0.004$), and family history (OR = 7.54, $p = 0.018$) were found to be significant predictive factors. These findings highlighted how consanguineous marriage and genetic factors contribute to the spread of thalassemia (Table 4).

The clinical characteristics of patients with thalassemia were described in Table 5. A diagnosis was made for the majority of patients (64%) at or before age six. Iron overload affected 72% of patients, and 67% of patients had splenomegaly. 54% of patients needed weekly transfusions, and 87% received iron chelation treatment, indicating a high frequency of blood transfusions. A characteristic consequence of thalassemia major, significant iron accumulation due to repeated transfusions was indicated by the significantly elevated mean serum ferritin level i.e., 1450.3 ± 520.7 ng/ml in this case (Table 5). When patients were categorized by age (≤15 and >15 years), older patients had a slightly higher prevalence of splenomegaly (35% vs. 32%, $p = 0.02$). Younger patients were more likely to receive weekly transfusions ($p = 0.003$), and transfusion

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frequency also varied. There was a statistically significant difference ($p = 0.014$) in serum ferritin levels between older patients (1580.7 ng/ml) and younger patients (1300.5 ng/ml). Despite nearly equal use of iron chelation therapy in both groups (87%), iron overload was more common in the older age group (77%). The multivariate logistic regression analysis confirmed the association between thalassemia complications and age groups. Iron overload also showed increased odds in older people (OR = 1.90, $p = 0.045$), and the chance of splenomegaly was significantly higher in patients older than 15 years (OR = 2.75, $p = 0.006$). Despite the fact that the older group had a higher prevalence of elevated serum ferritin (>1500 ng/ml), this difference was not statistically significant ($p = 0.625$). The odds of older patients receiving iron chelation therapy were also higher (OR = 3.12, $p = 0.002$), most likely because of their higher iron burden and longer duration of disease. The analysis concluded that thalassemia was primarily a childhood-onset condition that was significantly linked to hereditary risk factors, particularly consanguinity. The condition manifested as growth retardation and clinical complications, including iron overload and splenomegaly (Table 7). The study emphasized the importance of early diagnosis, genetic counseling, and community-level screening to reduce disease prevalence and improve management outcomes.

Table 1: Baseline characteristics of study subjects (*n* = 200)

Sr. No.	Characteristics		Thalassemia patients (*n* =100)	Controls (*n* = 100)
1	Age groups	Children (< 12 years)	51	4
		Adolescents (12-17 years)	29	24
		Adults (≥18 years)	20	72
2	Ethnicity	Muslims	97	97
		Christians	3	3
3	Height	3-4 ft	58	-
		4-5 ft	30	78
		>5 ft	12	22
4	Gender	Male	53	47
		Female	47	53
5	Weight	≤ 50 kg	92	52
		> 50 kg	8	48

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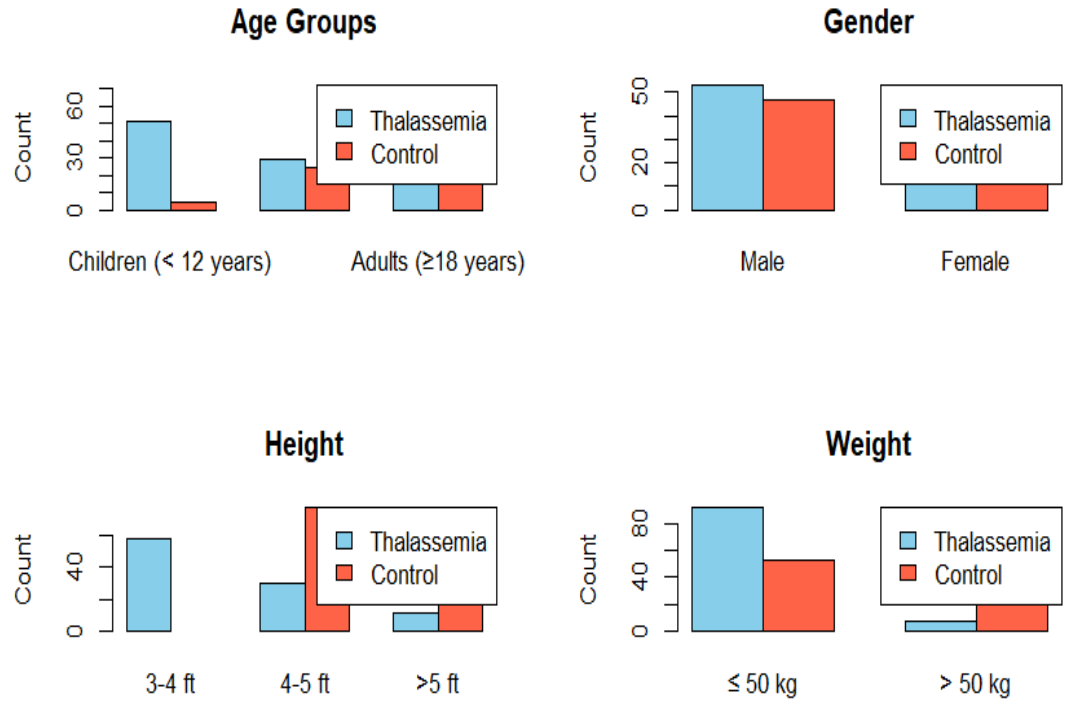

Fig 1: Comparative analysis of demographic and anthropometric variables in thalassemia patients and controls

These graphs show the key anthropometric and demographic features of thalassemia patients (n = 100) and healthy control (n = 100). **Age Groups:** 51% were children below 12 years in patients while in control majority (72%) were above 18 years. **Gender:** 53% were male with male dominance among thalassemia patients. **Height:** 58% patients were in the range of 3-4ft. In comparison, most controls were taller with a large portion in the 4-5 ft and >5ft categories. **Weight:** Majority of patients (92%) weighed ≤50 kg most common issue in affected individuals.

Table 2: Comparative analysis of demographic and anthropometric variables in thalassemia patients and controls

Sr. No.	Variables		Thalassemia patients (*n* =100)	Controls (*n* =100)	p-value
1	**Age (years), Mean ± SD**		11.87 ± 6.77	22.24 ± 6.73	0.0001*
2	**Gender, n (%)**	**Male**	53	47	0.39
		Female	47	53	
3	**Height (cm), Mean ± SD**		134.9 ± 24.97	167.94 ± 6.99	0.0001*
4	**Weight (kg), Mean ± SD**		28.49 ± 13.64	48.5 ± 12.33	0.0001*
5	**BMI**	**Underweight**	40	10	<0.00001*
		Normal	6	82	
		Overweight	46	5	
		Obese	6	3	

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Table 3: Influence of hereditary and marital risk factors in thalassemia and control groups

Sr. No.	Variables		Thalassemia patients	Controls	p-value
1	**Family history**	**Yes**	94	22	<0.00001*
		No	6	78	
2	**Siblings with thalassemia**	**Yes**	30	-	<0.00001*
		No	70	100	
3	**Maternal cousins with thalassemia**	**Yes**	16	2	0.0005*
		No	84	98	
4	**Paternal cousins with thalassemia**	**Yes**	14	15	0.84
		No	86	85	
5	**Consanguineous marriages**	**Yes**	58	16	<0.00001*
		No	42	84	

Table 4: Multivariate logistic analysis of risk factors associated with thalassemia

Sr. No.	Variables	OR (95% CI)	p-value
1	Age	1.24 (1.06-1.45)	0.007*
2	BMI	0.79 (0.26-2.43)	0.03*
3	Height	0.75 (0.67-0.85)	0.0001*
4	Weight	0.98 (0.92-1.04)	0.562
5	Family history	7.54 (2.39-11.26)	0.018*
6	Siblings with thalassemia	2.35 (1.32-4.21)	0.004*
7	Maternal cousins with thalassemia	0.67 (0.34-1.29)	0.228
8	Paternal cousins with thalassemia	1.12 (0.63-1.98)	0.702
9	Consanguineous marriages	2.12 (1.73-5.14)	0.027*

Table 5: Clinical characteristics of thalassemia patients

Sr. No.	Variables	Category / Unit	*n* / Mean ± SD
1	Age at diagnosis (years)	≤ 6	64
		> 6	36
2	Splenomegaly	Yes	67
		No	33
3	Blood transfusion frequency	Weekly	54
		Monthly	43
		Bimonthly	3
4	Iron chelation therapy	Yes	87
		No	13
5	Iron overload issue	Yes	72
		No	28
6	Serum ferritin	ng/ml	1450.3 ± 520.7

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Table 6: Age-stratified analysis of clinical parameters in thalassemia patients

Sr. No.	Clinical factors		Age of patients (years)		p-value
			≤ 15 (*n* = 69)	> 15 (*n* = 31)	
1	Splenomegaly	Yes	22	11	0.02*
		No	47	20	
2	Age at diagnosis	≤6	47	17	0.201
		>6	22	14	
3	Blood transfusion frequency	Weekly	35	15	0.003*
		Monthly	31	12	
		Bimonthly	3	4	
4	Iron chelation therapy	Yes	60	27	0.984
		No	9	4	
5	Iron overload issue	Yes	48	24	0.04*
		No	21	7	
6	Serum ferritin (ng/ml)	Mean ± SD	1300.5 ± 450.2	1580.7 ± 530.1	0.014*

Table 7: Multivariate logistic analysis of age group with thalassemia-related complications

Clinical Outcome	Age of patients		p-value	OR (95% CI)
	≤ 15 (*n* = 69)	> 15 (*n* = 31)		
Splenomegaly	32%	35%	0.006*	2.75 (1.32-5.72)
Serum ferritin >1500 ng/ml	44%	70%	0.625	0.85 (0.44–1.64)
Iron chelation therapy	87%	87%	0.002*	3.12 (1.51–6.43)
Iron overload issue	70%	77%	0.045*	1.90 (1.01–3.56)

Discussion

This study aims at providing a thorough investigation of the clinical, anthropometric, demographic and genetic traits of thalassemia patients in comparison with healthy controls. The findings highlighted the complexity of thalassemia and the significant impact that early diagnosis, persistent complications, and genetic predisposition have on patient.

Demographic and Anthropometric Findings

A notable finding was the substantial age difference between the two groups. While control group consisted of 72% adults aged 18 or older whereas 51% of patients were the children under 12. This fact is consistent with the literature confirming that thalassemia is mostly a childhood-onset disease, with early detection normally before six years age (De Sanctis *et al*., 2023). The extent of the disease's early signs and symptoms, which demands immediate medical care, could be the reason of thalassemia being more prevalent in younger individuals (Piel & Weatherall, 2015). According to anthropometric measurements, the average weight and height of thalassemia patients were significantly less than those of controls. The mean height for thalassemia patients were 134.9 cm tall, whereas controls had an average of 167.94 cm heights. Similarly, 48.5 kg

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was the mean weight of controls and 28.49 kg was that for thalassemia patients. The discrepancies being statistically significant ($p < 0.0001$) shows the influence of chronic anemia and iron overload on physical growth and development. Chronic hypoxia, iron-induced endocrine dysfunction, and delayed puberty are some of widely recognized contributors to growth retardation in thalassemia (De Sanctis *et al*., 2023; Skordis & Kyriakou, 2011).

Genetic and Family History Associations

The study demonstrated a significant difference in the occurrences of positive family history among control and thalassemia patients. Only 22% of the controls claimed having a positive family history, as opposed to 94% of thalassemia patients. Moreover, 58% of patients reported cousin marriages. In contrast, affected siblings were reported in only 16% in the control group, and 30% of thalassemia patients. These results highlight the crucial role of consanguinity in the emergence of thalassemia and are in accordance with the autosomal recessive inheritance pattern of the disease (De Sanctis *et al*., 2023). The need of carrier screening programs and genetic counseling is highlighted by the close association between consanguinity and thalassemia, especially in regions where cousin marriages are common. Consanguinity is an integral factor contributing to the substantial prevalence of thalassemia in Pakistan (Khalid *et al*., 2019). According to literature, 77% of thalassemia patients had a consanguinity pattern in their families, and 74% of the patients turned out to be product of cousin marriages (Khattak *et al*., 2023). According to another survey, 76.7% of thalassemia cases in Punjab were linked with first cousin marriages (Masih *et al*., 2023). These statistics highlight the integral role of consanguinity in thalassemia transmission and the necessity of targeted public health campaigns.

Clinical Complications and Management

Clinically, thalassemia patients had a high rate of disease-related complications. Iron excess affected 72% of patients, while splenomegaly was observed in 67%. These challenges are typical in thalassemia major and are essentially the result of a long-term transfusion therapy, which causes excessive iron accumulation in key organs (De Sanctis *et al*., 2023). This study's increased mean blood ferritin level of 1450.3 ± 520.7 ng/ml indicates substantial iron overload. The study also discovered that 54% of patients needed weekly transfusions, while 87% underwent iron chelation therapy. Despite the high rate of chelation therapy, older patients (> 15 years) had higher serum ferritin levels and a greater prevalence of iron overload, indicating that the cumulative effect of iron deposition over time. The medical effects varied according to age. Older patients were more probably to develop iron overload (OR = 1.90, $p = 0.045$) and splenomegaly (OR = 2.75, $p = 0.006$). These findings are consistent with thalassemia's progressive personality, in which cumulative illness burden increases the risk of problems with age (Bonifazi *et al*., 2017). The higher chances of receiving iron chelation therapy in elderly patients (OR = 3.12, $p = 0.002$) may reflect the increased recognition of overproduction of iron and the necessity for intervention as patients age.

Limitations and Future Directions

While this study provides valuable insights into the demographic, clinical, and genetic characteristics of thalassemia patients, several limitations must be acknowledged. The retrospective design and reliance on hospital records may introduce selection bias and limit the generalizability of the findings. Additionally, the study was conducted at a single center, which may not be representative of broader populations. Future multicenter, prospective studies are needed to validate these findings and explore the long-term outcomes of thalassemia patients.

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Furthermore, the study did not assess the impact of socioeconomic factors, access to healthcare, and patient adherence to treatment protocols, all of which can influence disease progression and outcomes. Incorporating these variables into future research could provide a more comprehensive understanding of the factors affecting thalassemia patients.

Conclusion

To summarize, this study emphasizes the complicated nature of thalassemia, with considerable contributions from genetic predisposition, early start, and chronic problems. The high incidence of consanguinity and positive family history highlights the importance of focused genetic counselling and carrier screening programs. The observed growth retardation and medical implications highlight the necessity of timely identification, regular monitoring, and proper management techniques for better patient outcomes. Addressing these issues via public health efforts and healthcare policy changes will help to improve management and reduce disease burden in thalassemia-affected communities.

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Authors Contribution:

Warisha Nabeel**:** Data collection, visualization, and initial manuscript drafting.
Bisma Naeem: Data collection, Literature review and data interpretation.
Nageen Hussain**:** Project supervision, conceptual guidance, and manuscript revisions.
Hassan Saeed**:** Statistical analysis, data visualization, figure preparation, and final manuscript writing.
Kashifa Tul Khair: Assistance in Survey administration and Report Collection

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