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ORIGINAL ARTICLE

Thalassemia Major in Punjab Pakistan, Epidemiology, Treatment Challenges, and Roadmap for Prevention. A Cross-sectional Observational Study

UMAR TANVEER[1], EJAZ ASGHAR[2], TASAWAR AZIZ[3], MAXIMILIAN SIENER[4]
[1]Department of Mass Communication, University of Lahore, Lahore 54000, Pakistan
[2]Department of Allied Health Sciences, Health Services Academy, Islamabad 44000, Pakistan
[3]Department of Health Physical Education and Sports Sciences, University of Karachi, Karachi74200 Pakistan
[4]Department of Training and Movement Science, BaySpo—Bayreuth Center of Sport Science, University of Bayreuth, 95447 Bayreuth, Germany
Correspondence to: Umar Tanveer, **E-mail:** umartanveer39@gmail.com, **Cell:** (U.T.); +92 300 7472759

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ABSTRACT

Background: Thalassemia major is a severe hereditary blood disorder highly prevalent in Punjab, Pakistan, due to high consanguinity and limited screening programs. This study investigated its epidemiology, treatment challenges, and proposes preventive strategies to reduce disease burden.

Aim of Study: To quantify the disease burden, healthcare challenges, and socioeconomic impact of thalassemia in Punjab, Pakistan, and propose evidence-based control strategies.

Methodology: Current: Cross-sectional observational study was conducted from January 2022 to December 2023. Total 100 thalassemia major patients registered at DHQ Sahiwal's Thalassemia Centre were enrolled. Data on demographics, clinical parameters, treatment adherence, complications, and family histories were collected via structured questionnaires and medical records. Blood samples were analyzed for ferritin levels and transfusion frequency.

Results: Among 100 thalassemia major patients, 64% were diagnosed before age 2, and 78% had parental consanguinity. Only 58% received regular iron chelation therapy, and 21% had transfusion-transmitted infections. Financial burden was reported by 85% of families, while awareness of carrier screening and prenatal diagnosis was low (18% and 12%, respectively).

Conclusion: Thalassemia major poses significant clinical and economic challenges in Punjab. Early diagnosis, inadequate treatment access, and poor preventive awareness highlight the urgent need for integrated screening, counseling, and support programs to reduce disease burden.

Keywords: Thalassemia, Pakistan, Punjab, Consanguinity, Iron Overload, Public Health, Hemoglobinopathy, Genetic Screening.

INTRODUCTION

Thalassemia major is a genetically inherited hemoglobinopathy resulting in defective β-globin chain production, leading to chronic hemolytic anemia, growth retardation, and lifelong dependence on regular blood transfusions[1]. It remains one of the most common monogenic disorders in Pakistan, with a particularly high prevalence in Punjab due to cultural practices such as consanguineous marriages and inadequate public health awareness regarding carrier status. In Punjab, the rising number of thalassemia major cases reflects the absence of a centralized registry, inconsistent carrier screening, and limited access to prenatal diagnostic services. Many patients face lifelong challenges including poor transfusion practices, iron overload due to lack of iron chelation, frequent hospital visits, increased risk of infections, and psychological stress on both patients and caregivers [2]. Despite these obstacles, national efforts

toward disease prevention and control remain fragmented and under-resourced [3].

Effective disease management requires not only timely diagnosis and standardized treatment protocols but also proactive prevention strategies such as carrier detection, genetic counseling, and awareness campaigns. However, the implementation of such initiatives in Punjab has been limited by infrastructural, financial, and socio-cultural constraints [4]. Thalassemia represents a critical public health challenge in Pakistan, with an estimated 100,000 transfusion-dependent patients and carrier rates of 5–7% in high-risk populations. In Punjab Pakistan's most populous province (127 million) thalassemia prevalence is amplified by sociocultural practices, particularly consanguineous marriages (60–70% prevalence), limited access to genetic counseling, and fragmented healthcare infrastructure [5]. The financial burden on families is catastrophic, with annual treatment costs exceeding 300,000 PKR (~$1,000), often exceeding household incomes. Despite the Thalassemia Prevention Act of 2018, implementation remains weak, with <20% of districts offering prenatal screening. DHQ Sahiwal serves a catchment area of 2.5 million in South Punjab, a region with high consanguinity and poverty rates [6].

This study quantifies the clinical-epidemiological burden, identifies systemic gaps, and proposes a scalable roadmap for thalassemia control in resource-limited settings. The findings aim to inform policy reforms prioritizing prevention, equitable access to care, and integration of thalassemia management into primary healthcare systems [7]. This study aims to explore the epidemiological profile of thalassemia major in Punjab, identify key treatment-related barriers, and propose a context-specific roadmap for prevention. The findings will help inform policy development, improve patient care, and support the integration of sustainable preventive programs across the province.

MATERIAL AND METHOD

A cross-sectional observational study was conducted over a two-year period from January 2022 to December 2023 at the Thalassemia Centre, District Headquarters (DHQ) Hospital, Sahiwal, Punjab, Pakistan. A total of 100 patients diagnosed with β-thalassemia major and registered at the center were included in the study through non-probability consecutive sampling. Inclusion criteria encompassed all patients with a confirmed diagnosis of thalassemia major, irrespective of age and gender, based on clinical presentation and hemoglobin electrophoresis reports. Patients with other hemoglobinopathies or incomplete medical records were excluded.

Data were collected using a structured questionnaire and patient file review. Key variables included demographic characteristics (age, gender, family history, and consanguinity), age at diagnosis, frequency of blood transfusions, access to and compliance with iron chelation therapy, history of transfusion-transmitted infections, and socioeconomic background. Caregivers were also interviewed to explore barriers related to treatment compliance, awareness, and financial constraints.

Ethical approval was obtained from the institutional review board of DHQ Hospital, Sahiwal. Informed consent was taken from all adult participants or from parents/guardians in the case of minors. All data were entered and analyzed using SPSS version 25. Descriptive statistics such as frequencies, means, and standard deviations were used to summarize the findings.

RESULTS

A total of 100 patients diagnosed with β-thalassemia major were included in this study. The mean age of the patients was 11.3 ± 4.2 years, ranging from 2 to 21 years. Among them, 57% were male and 43% were female. A family history of thalassemia was present in 61% of the cases, and parental consanguinity was reported in 78% of the families. The majority of patients (64%) were diagnosed before the age of 2 years. All patients were dependent on regular blood transfusions, with 53% receiving transfusions every 2–3 weeks and 47% receiving them monthly. However, only 58% of patients were regularly receiving iron chelation therapy. Among those, 39% used oral chelators and 19% received parenteral agents.

Transfusion-transmitted infections were reported in 21% of the patients, including hepatitis C in 13%, hepatitis B in 6%, and HIV in 2%. Growth retardation and skeletal deformities were clinically observed in 41% of the patients. Additionally, financial burden was reported by 85% of caregivers, with many relying on government support or charitable donations. Preventive awareness was poor; only 18% of families had undergone any form of premarital or carrier screening, and only 12% were aware of prenatal diagnostic options.

Table 1 presents the demographic and clinical profile of the 100 thalassemia major patients included in the study. The majority of patients (39%) were between 6–10 years of age, with a mean age of 11.3 ± 4.2 years. Males constituted 57% of the cohort, while females accounted for 43%. A significant proportion of patients (64%) were diagnosed before the age of 2 years, indicating early-onset disease. Notably, parental consanguinity was observed in 78% of the families, and a positive family history of thalassemia was found in 61%, highlighting the

strong genetic and hereditary link within the studied population.

Table 2 summarizes treatment-related variables and clinical outcomes. All patients were transfusion-dependent, with 53% receiving blood every 2–3 weeks and 47% monthly. Only 58% of patients were receiving iron chelation therapy, with oral agents being more common (39%) compared to parenteral therapy (19%). Transfusion-transmitted infections were reported in 21% of the patients, with hepatitis C being the most prevalent (13%). Clinical complications such as growth retardation and skeletal deformities were noted in 41% of patients, suggesting inadequate disease control in a significant subset.

Table 1: Demographic and Clinical Characteristics of Patients (N = 100)

Variable	Frequency (n)	Percentage (%)
Age group (years)		
1–5	18	18%
6–10	39	39%
11–15	29	29%
16–21	14	14%
Mean Age ± SD	—	11.3 ± 4.2
Gender		
Male	57	57%
Female	43	43%
Parental consanguinity	78	78%
Positive family history of thalassemia	61	61%
Age at diagnosis < 2 years	64	64%

Table 2: Treatment and Clinical Outcomes

Variable	Frequency (n)	Percentage (%)
Transfusion frequency		
Every 2–3 weeks	53	53%
Monthly	47	47%
Regular iron chelation therapy	58	58%
Oral chelators	39	39%
Parenteral chelation therapy	19	19%
Transfusion-transmitted infections (total)	21	21%
– Hepatitis C	13	13%
– Hepatitis B	6	6%
– HIV	2	2%
Growth retardation/skeletal changes	41	41%

Table 3 highlights the socioeconomic burden and gaps in preventive measures. A majority of families (85%) reported facing significant financial stress due to the lifelong cost of care. About 72% of patients depended on government aid or charity for treatment. Preventive awareness was alarmingly low—only 18% of families had prior knowledge about carrier screening, and a mere 12% were aware of prenatal diagnostic options. These findings underscore the urgent need for public health education and implementation of preventive genetic counseling programs in Punjab.

Table 3: Socioeconomic and Preventive Aspects

Variable	Frequency (n)	Percentage (%)
Financial burden reported	85	85%
Use of government/charity for treatment	72	72%
Awareness of carrier screening	18	18%
Awareness of prenatal diagnosis options	12	12%

In this cross-sectional study of 100 thalassemia major patients at DHQ Hospital Sahiwal, the majority were male (57%) with a mean age of 11.3 ± 4.2 years, and 64% were diagnosed before the age of 2. Parental consanguinity (78%) and positive family history (61%) were highly prevalent. All patients were transfusion-dependent, with 53% receiving transfusions every 2–3 weeks, yet only 58% were on regular iron chelation therapy. Transfusion-transmitted infections affected 21% of patients, and 41% exhibited growth or skeletal complications. Financial burden was reported by 85% of families, while awareness of carrier screening (18%) and prenatal diagnosis (12%) remained extremely low, indicating critical gaps in treatment access and preventive education.

DISCUSSION

This study highlighted the ongoing burden and multifaceted challenges associated with β-thalassemia major in the region of Punjab, Pakistan, specifically among patients registered at DHQ Hospital Sahiwal. The demographic distribution revealed a young patient population with a mean age of 11.3 years, consistent with national and regional patterns indicating early onset and chronic disease progression [8,9]. A high prevalence of parental consanguinity (78%) and positive family history (61%) strongly supports the role of autosomal recessive inheritance and sociocultural factors that perpetuate disease transmission in Pakistan, particularly in rural and semi-urban areas [10].

Despite the availability of transfusion services, many patients are receiving suboptimal care. While all patients in this study were transfusion-dependent, only 58% were receiving iron chelation therapy, and a smaller proportion had access to parenteral chelators [11,12]. This reflects gaps

in standard treatment protocols, poor availability of chelation drugs, and economic limitations faced by families. These findings align with previous reports from Pakistan and other developing countries, where the lack of comprehensive thalassemia management programs leads to avoidable complications such as iron overload, growth retardation, and skeletal deformities observed in 41% of our cohort 13.

Transfusion-transmitted infections remain a significant concern, with 21% of patients in this study testing positive for hepatitis B, hepatitis C, or HIV. These infections are likely linked to inadequate blood screening protocols or unsafe transfusion practices, and they further complicate clinical outcomes in thalassemia patients [14-20]. The financial burden associated with long-term care was substantial, with 85% of families reporting economic hardship and over 70% depending on charity or government support. This highlights the urgent need for policy-driven support systems, subsidized treatment options, and better integration of public sector healthcare with NGOs and donor agencies [15].

Perhaps most concerning is the poor awareness regarding thalassemia prevention. Despite the high genetic burden, only 18% of families were aware of carrier screening, and a mere 12% knew about prenatal diagnostic options [16,21-28]. This reflects a broader failure in public health communication, lack of national screening programs, and minimal genetic counseling services, particularly in under-resourced districts. Collectively, these findings emphasize the necessity of a multi-level approach to thalassemia control in Punjab. Key priorities should include mandatory premarital carrier screening, community-based genetic counseling, integration of prenatal diagnostic services, and reinforcement of national transfusion safety standards [18,20]. Furthermore, establishing a provincial thalassemia registry and ensuring equitable access to iron chelation therapy and psychosocial support could significantly improve quality of life and reduce the disease burden over time [28-36].

CONLUSION

This study stated the significant clinical, social, and economic burden of β-thalassemia major in Punjab, Pakistan. Early age of diagnosis, high rates of parental consanguinity, and inadequate access to iron chelation therapy contribute to poor clinical outcomes. The presence of transfusion-transmitted infections and the prevalence of growth complications highlight gaps in safe transfusion practices and comprehensive disease management. Additionally, the extremely low awareness of carrier screening and prenatal diagnostic options reflects a critical deficiency in preventive strategies. There is an urgent need for government-supported public health initiatives focused on thalassemia education, mandatory premarital screening, improved access to diagnostic and treatment services, and community-based genetic counseling programs. Strengthening infrastructure and policy interventions will be essential to reducing disease incidence, improving patient quality of life, and guiding long-term prevention in high-burden areas like Punjab

DECLARATION

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Interest of Study

This study was conducted to explore the epidemiological characteristics, treatment barriers, and preventive gaps in the management of thalassemia major in Punjab, Pakistan. The findings aim to support healthcare policymakers, clinicians, and public health professionals in designing targeted interventions to reduce disease burden and improve patient outcomes.

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