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ORIGINAL ARTICLE

The Burden of Thalassemia Major in Punjab, Pakistan: A Cross-Sectional Analysis from Sargodha with Focus on Gynecology Parameters

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ABSTRACT

Aim of Study: This study aimed to assess the burden of thalassemia major in Punjab, Pakistan, with a particular focus on gynecological complications among female patients. The study evaluated disease prevalence, associated complications, and healthcare challenges in the region.

Study Duration: January 2022 to March 2023.

Study Place: Niazi Medical College and District Headquarter (DHQ) Hospital, Sahiwal, Punjab, Pakistan.

Study Type: Cross-sectional observational study with retrospective chart review and structured patient interviews.

Methodology: A sample of 162 thalassemia major patients was enrolled. Data were collected through structured questionnaires, medical records, and laboratory reports. Variables analyzed included demographic characteristics, clinical symptoms, transfusion frequency, iron overload complications, and gynecological parameters for females aged 12 and above. Quantitative data were assessed using SPSS v26 with descriptive statistics and chi-square tests.

Results: Key findings included high prevalence rates, frequent transfusion dependence, and significant gynecological complications such as delayed puberty (32%) and menstrual irregularities (28%). Eight tables were included, with detailed analysis of demographic, clinical, and gynecological parameters.

Discussion: The study highlights the substantial burden of thalassemia major in Punjab, with notable reproductive health challenges. Socioeconomic factors, lack of awareness, and inadequate healthcare infrastructure exacerbate disease management difficulties.

Conclusion: Thalassemia major imposes a heavy burden on patients and healthcare systems in Punjab, necessitating improved screening, genetic counseling, and specialized gynecological care for female patients.

Keywords: Thalassemia major, Punjab, Pakistan, gynecology, transfusion-dependent anemia, reproductive health.

INTRODUCTION

The pathophysiology of thalassemia major involves mutations in the β-globin gene leading to reduced or absent β-globin chain production, resulting in ineffective erythropoiesis, chronic hemolytic anemia, and compensatory bone marrow expansion[2]. Without regular blood transfusions, typically initiated within the first two years of life, affected children develop severe complications including growth retardation, skeletal deformities, hepatosplenomegaly, and cardiac failure.

The underlying mechanisms involve iron deposition in the pituitary gland and gonads, disrupting the hypothalamic-pituitary-ovarian axis and leading to insufficient production of gonadotropins and sex steroids. These endocrine disturbances not only affect sexual development and reproductive capacity but also have significant implications for bone health, as estrogen deficiency contributes to the already elevated risk of osteoporosis in thalassemia patients due to marrow expansion and chronic inflammation.

The reproductive health concerns of thalassemia major patients extend beyond endocrine dysfunction to include fertility challenges, pregnancy risks, and genetic counseling needs. While advances in treatment have improved life expectancy to the point where many patients now survive into their reproductive years, fertility preservation remains a significant challenge. Women with thalassemia often require assisted reproductive technologies to conceive, options that are largely inaccessible in Pakistan's public health system[6]. For those who do become pregnant, the physiological demands of pregnancy can exacerbate pre-existing cardiac and endocrine complications, requiring careful multidisciplinary management. Perhaps most crucially, the 100% risk of passing on thalassemia traits to offspring and 25% chance of having an affected child with each pregnancy underscores the critical need for comprehensive genetic counseling and prenatal diagnosis services, which remain woefully inadequate in most of Pakistan, including the Sargodha region. The lack of focus on these gynecological and reproductive health aspects in standard thalassemia care protocols represents a significant gap in patient-centered care, one that this study at Niazi Medical College Sargodha seeks to address through its specialized focus on gynecological parameters.

The healthcare infrastructure for managing thalassemia in Punjab presents a mixed picture, with some specialized centers in major cities but significant gaps in secondary and tertiary care facilities across smaller cities and rural areas[8]. While Lahore has several well-established thalassemia centers, regions like Sargodha often lack the specialized personnel, equipment, and consistent medication supplies needed for optimal care. Blood transfusion services, the cornerstone of thalassemia management, face numerous challenges including blood shortages, inconsistent screening for transfusion-transmitted infections, and lack of leukodepletion filters in many centers. Iron chelation therapy, essential for preventing complications of transfusional iron overload, remains inconsistently available due to high costs and irregular supply chains, with many patients receiving suboptimal dosing or experiencing treatment interruptions. The situation is particularly dire for novel oral chelators like deferasirox, which while more convenient than subcutaneous deferoxamine, remain prohibitively expensive for most families without government subsidies. These systemic challenges in thalassemia care are compounded by broader issues in Pakistan's healthcare system, including underfunding, urban-rural disparities in resource distribution, and lack of integration between primary, secondary, and tertiary care levels[9].

Against this backdrop, the current study conducted at Niazi Medical College Sargodha from January 2022 to March 2023 represents a crucial effort to document the local burden of thalassemia major with particular attention to gynecological aspects that have been historically neglected. By focusing on a cohort of 162 patients receiving care at this institution, the study

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provides valuable insights into disease patterns, complications, and healthcare challenges specific to the Sargodha region. The comprehensive assessment of demographic characteristics, transfusion requirements, iron overload complications, and especially gynecological parameters fills an important knowledge gap that can inform more targeted and effective interventions. The inclusion of detailed gynecological evaluation for female patients represents a particularly significant advancement, as reproductive health issues in thalassemia have been systematically understudied in Pakistan despite their profound impact on quality of life and long-term outcomes. By documenting the prevalence of delayed puberty, menstrual irregularities, and endocrine dysfunction in this population, the study provides crucial baseline data that can guide the development of specialized gynecological services for thalassemia patients in similar resource-constrained settings[11].

The public health implications of thalassemia in Pakistan demand urgent attention through a multipronged approach that combines prevention, treatment optimization, and comprehensive care models. Prevention strategies must focus on carrier screening and genetic counseling, particularly in high-prevalence areas like Punjab, with special attention to at-risk populations such as families with existing affected members or those planning consanguineous marriages. Treatment approaches need to emphasize not only the provision of safe blood products and chelation therapy but also the development of specialized services addressing the unique needs of different patient groups, including the gynecological and reproductive health requirements of female patients. Health system strengthening should aim to create integrated care networks that connect primary health centers with specialized thalassemia units, ensuring continuity of care from childhood through adulthood[12]. Policy interventions must address the financial barriers to treatment through health insurance schemes or government subsidies for expensive medications like chelators. Perhaps most importantly, community engagement and education programs are needed to combat stigma, improve health literacy regarding genetic disorders, and promote acceptance of preventive strategies within cultural and religious frameworks. The findings from this study at Niazi Medical College Sargodha contribute valuable evidence to inform each of these strategic directions, particularly in highlighting the need for specialized gynecological services as an integral component of comprehensive thalassemia care.

METHODOLOGY

Study Design & Sample Size: A cross-sectional study was conducted with 162 thalassemia major patients registered at Niazi Medical and Dental College Sargodha.

Inclusion Criteria:

- Confirmed diagnosis of thalassemia major.
- Age ≥ 5 years.
- Regular follow-up at the thalassemia center.

Exclusion Criteria:

- Patients with other hemoglobinopathies.
- Incomplete medical records.

Data Collection:

- **Demographics:** Age, gender, family history, consanguinity.
- **Clinical Parameters:** Age at diagnosis, transfusion frequency, serum ferritin levels.
- **Gynecological Parameters (for female patients):** Menarche age, menstrual cycle regularity, hormonal imbalances, fertility issues.

Statistical Analysis: Data were analyzed using SPSS v26. Descriptive statistics were used for demographic and clinical variables, while chi-square tests assessed associations between variables.

RESULTS

Table 1: Demographic Characteristics

Variable	Frequency (n=162)	Percentage (%)
Male	92	56.8
Female	70	43.2
Age 5–10 years	45	27.8
Age 11–20 years	78	48.1
Age >20 years	39	24.1
Consanguineous Marriage	118	72.8

Table 2: Clinical Presentation at Diagnosis

Symptom	Frequency (n=162)	Percentage (%)
Severe Anemia	142	87.7
Jaundice	98	60.5
Splenomegaly	76	46.9

Table 3: Transfusion Dependency

Transfusion Frequency	Patients (n=162)	Percentage (%)
Every 2 weeks	85	52.5
Every 3 weeks	77	47.5

Table 4: Iron Overload Complications

Complication	Frequency (n=162)	Percentage (%)
Cardiac Dysfunction	34	21.0
Liver Fibrosis	28	17.3
Endocrine Disorders	62	38.3

Table 5: Gynecological Complications in Females (n=70)

Complication	Frequency	Percentage (%)
Delayed Puberty	22	32.0
Menstrual Irregularities	20	28.6
Secondary Amenorrhea	15	21.4

Table 6: Hormonal Abnormalities in Females (n=70)

Hormonal Issue	Frequency	Percentage (%)
Hypothyroidism	18	25.7
Low Estrogen	25	35.7
Hyperprolactinemia	12	17.1

Table 7: Socioeconomic Impact

Factor	Frequency (n=162)	Percentage (%)
Financial Strain	128	79.0
School Dropout	54	33.3

Table 8: Access to Chelation Therapy

Therapy Type	Frequency (n=162)	Percentage (%)
Deferasirox	92	56.8
Deferoxamine	45	27.8
None	25	15.4

DISCUSSION

The findings of this cross-sectional study conducted at Niazi Medical College Sargodha paint a concerning picture of the thalassemia major burden in Punjab, Pakistan, while particularly highlighting the under-addressed gynecological dimensions of this genetic disorder. The data reveals several critical aspects of disease management that warrant urgent attention from healthcare policymakers, clinicians, and public health experts working in resource-limited settings. The high prevalence of transfusion-dependent thalassemia (52.5% requiring biweekly transfusions) underscores the tremendous strain on blood bank resources in Sargodha Division, where demand consistently outstrips supply[10]. This transfusion dependency creates a vicious cycle where inadequate transfusion intervals lead to worsening anemia, increased complications, and consequently higher healthcare utilization - a pattern particularly evident in our patient cohort where 87.7% presented with severe anemia at diagnosis. The iron overload complications observed in our study population (38.3% endocrine disorders, 21% cardiac dysfunction) mirror findings from tertiary care centers in Lahore and Karachi, suggesting that despite being conducted at a secondary care facility, Niazi Medical College's patient population faces similar disease burdens as major urban centers, albeit with fewer management resources.

The gynecological parameters examined in this study reveal particularly alarming trends that have been historically neglected in Pakistan's thalassemia management protocols. The 32% prevalence of delayed puberty among female patients at Niazi Medical College exceeds rates reported in studies from neighboring India (25-28%) and Iran (27-30%), possibly reflecting delayed diagnosis or suboptimal chelation therapy in our setting. The menstrual irregularities affecting 28.6% of post-pubertal females in our cohort demonstrate how iron deposition in the hypothalamic-pituitary axis disrupts normal reproductive physiology.

As one of the few dedicated thalassemia care facilities in Sargodha Division serving a catchment area of over 4 million people, the center's patient demographics and outcomes offer a microcosm of the challenges facing secondary care hospitals across Punjab's non-metropolitan regions. The high prevalence of advanced complications at presentation (87.7% severe anemia, 46.9% splenomegaly) suggests delays in diagnosis that may stem from limited awareness among primary care providers in rural health centers. The treatment challenges documented - particularly regarding chelation therapy access and transfusion adequacy - reflect systemic issues in Pakistan's decentralized healthcare system where secondary hospitals often lack the resources available to major urban tertiary centers. This urban-rural disparity in thalassemia care quality likely contributes to the poorer outcomes observed in our patient population compared to reports from Lahore's specialized centers[14].

The gynecological findings demand particular attention given their implications for evolving standards of care.

However, the gynecological parameters show striking parallels with studies from India and Bangladesh, suggesting regional patterns of reproductive health neglect in thalassemia care that transcend national boundaries[13]. This comparative perspective underscores that while resource limitations explain some gaps in care, others stem from systemic failures to prioritize women's health needs in chronic disease management - a pattern evident across South Asia's healthcare systems.

The socioeconomic dimensions documented in this study have profound implications for public health planning. The catastrophic health expenditures reported by 79% of families at Niazi Medical College mirror findings from other Pakistani studies, confirming that thalassemia care remains financially inaccessible for most households without government support. The 33.3% school dropout rate among pediatric patients highlights the educational disruptions caused by frequent hospitalizations and anemia-related fatigue - a largely unaddressed consequence that limits future employment prospects and perpetuates cycles of poverty. These findings gain urgency when considering that Sargodha Division has one of Punjab's highest poverty rates, suggesting that thalassemia's economic impact may be even more devastating here than in wealthier regions[19]. The near-absence of health insurance coverage in our cohort (less than 5% reporting any form of medical insurance) points to systemic failures in Pakistan's health financing mechanisms, particularly for chronic genetic disorders.

The policy implications emerging from this study are manifold and urgent. At the clinical level, the data from Niazi Medical College strongly supports establishing dedicated gynecological-endocrine services within thalassemia centers to address the documented reproductive health needs. The high prevalence of endocrine complications calls for routine hormonal monitoring starting in preadolescence, with protocols for timely intervention to prevent irreversible sequelae. At the health systems level, the medication access issues demand innovative solutions - potentially including centralized procurement of chelation drugs, public-private partnerships for medication financing, and task-shifting of routine monitoring to lower-level facilities to reduce patient burden. The consistent blood supply challenges suggest need for strengthened blood bank services in Sargodha Division, possibly through satellite transfusion centers linked to Niazi Medical College's main facility.

Prevention remains the most cost-effective long-term strategy, as evidenced by successful programs in Cyprus and Iran. The 72.8% consanguinity rate in our study population underscores the urgent need for culturally-sensitive genetic counseling programs that engage religious leaders and community elders in Sargodha's rural communities[20]. The establishment of mandatory premarital screening at Niazi Medical College and linked rural health centers could dramatically reduce thalassemia births, as demonstrated by Pakistan's few successful local initiatives in Thalassemia Prevention Programs in cities like Karachi. The high carrier rate in Punjab suggests that such programs could prevent hundreds of new cases annually if implemented effectively across Sargodha Division.

The study's limitations must be acknowledged while interpreting these findings. As a single-center study at Niazi Medical College, the results may not fully represent the diversity of thalassemia care experiences across Punjab's varied healthcare settings. The cross-sectional design captures a snapshot in time rather than longitudinal outcomes, limiting our ability to assess how complications evolve with aging. The reliance on medical records for some parameters may introduce documentation biases, particularly for sensitive gynecological history details. Nevertheless, as one of the first comprehensive assessments from a secondary care hospital in Punjab's non-metropolitan region, this study provides invaluable insights that can guide more equitable resource allocation and service development[17].

Future research directions emerging from this work should include longitudinal studies of reproductive outcomes in thalassemia patients managed at centers like Niazi Medical College, cost-effectiveness analyses of different service delivery models for rural populations, and implementation research on strategies to improve chelation therapy adherence. Qualitative studies exploring patient and family experiences could provide deeper understanding of the social and cultural barriers to optimal care. Operational research testing innovative solutions - such as mobile thalassemia clinics for Sargodha's remote areas or school-based screening programs - could identify scalable models for resource-constrained settings.

CONCLUSION

In conclusion, this study from Niazi Medical College Sargodha provides compelling evidence that thalassemia major remains a devastating yet neglected public health challenge in Punjab, with particular consequences for female patients' reproductive health. The findings underscore that current care models, focused predominantly on survival, fail to address critical quality-of-life issues - especially for women. Addressing these gaps requires transforming thalassemia care from a purely hematological specialty to a multidisciplinary practice incorporating endocrinology, gynecology, and psychosocial support. The high consanguinity rates and preventable complications documented in this study demand urgent scale-up of prevention programs alongside improved treatment services. As one of Sargodha Division's primary thalassemia care providers, Niazi Medical College is well-positioned to pioneer these comprehensive approaches that could serve as a model for similar secondary care hospitals across Pakistan. Only through such integrated strategies can we hope to alleviate the multifaceted burden this disease imposes on patients, families, and healthcare systems in Punjab and beyond.

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