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# Prevalence of Thalassemia in Relation to Blood Group Distribution in Shaheed Benazir Abad, Sindh

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**Abstract**

**Background:** There are millions of children affected by $\beta$-thalassemia, making it one of the most prevalent genetic disorders globally. An estimated 50,000–60,000 new instances of $\beta$-thalassemia are diagnosed annually, and around 1.5% of the global population, or 80–90 million individuals, carry the gene for it. **Objectives:** To determine the prevalence of thalassemia across different blood groups in Shaheed Benazir Abad city, Sindh. **Methods:** This cross-sectional study was conducted at the Department of Molecular Biology & Genetics, Shaheed Benazir Bhutto University, after obtaining ethical approval. Of the 253 patients diagnosed with thalassemia who were receiving blood transfusions at the Thalassemia Centre, Nawabshah, 114 (45.1%) were enrolled. Socio-demographic and clinical data, along with blood group distribution, were recorded through structured interviews and questionnaires. **Results:** The prevalence of thalassemia was 114 (45.1%) out of 253 individuals, while blood group distribution revealed that the majority had group O (46.5%), followed by group A (29.8%), group B (19.3%), and group AB (4.4%). **Conclusion:** Thalassemia prevalence in Shaheed Benazir Abad was 45.1%, with blood group O being the most common among affected patients. These findings highlight a significant association of thalassemia with specific blood group distributions in the region.

**Keywords:**
Thalassemia, SBBU, Blood groups, Nawabshah, Prevalence

## 1. Introduction

Among the most prevalent hereditary disorders, $\beta$- thalassemia impacts millions of children globally.[1] There are around 50,000–60,000 new instances of $\beta$-thalassemia every year, and about 1.5% of the global population is a carrier for the condition. Asia, the Indian subcontinent, Mediterranean nations, Africa, and the Middle East have the highest prevalence of $\beta$-thalassemia.[3–5] Carriers account for 5 to 7% of the Pakistani population, making $\beta$-thalassemia one of the most prevalent genetic illnesses in the country.[2] Blood transfusion and iron chelation have increased the longevity of $\beta$-thalassemia patients. Presently, there are around 100,000 individuals on the registry, but the illness burden is growing; 5,000 to 9,000 infants are born with the ailment every year.[6]

In Pakistan, thalassemia patients die from blood-borne illnesses at a rate that is second to none.[2] Patients with thalassemia who have regular blood transfusions put themselves at a greater risk of acquiring the HCV virus infection, particularly in cases when proper viral screening of blood donors has not been done. Because people with thalassemia are more likely to receive blood transfusions, their infection risk is a good indicator of the general population's risk of transfusion-transmitted infections. A negligible risk to the general population may be inferred from the low infection rate in $\beta$-thalassemia patients.

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One of the most prevalent illnesses transmitted by blood is hepatitis C. There is a substantial risk of illness and death among Pakistan's more than 10 million HCV-infected people.[7] Pakistan is a developing nation; out of 189 nations and territories ranked by the UN's Human Development Index, it ranks 150th.[8] All nations strive for a level of health care that is equal to or better than Pakistan's. Consequently, a significant risk factor for the propagation of HCV is still contaminated blood transfusion. This is because paid blood donors are often used without screening.[9] There is a large discrepancy in the reported prevalence of various blood groups among the several studies that have examined this topic in Pakistani thalassemia patients. This was the first research we were aware of to examine the frequency of thalassemia in various blood types at the Shaheed Benazir Abad Thalassemia Center.

# 2. Materials and Methods

## 2.1. Ethical Approval and Study Design

This study was conducted following approval from the Departmental Ethical Committee of the Department of Molecular Biology & Genetics, Shaheed Benazir Bhutto University, Shaheed Benazirabad (SBBU), Sindh, Pakistan. Written informed consent was obtained from all participants or their guardians before enrollment. A total of 253 patients visited the Thalassemia Centre in Nawabshah for thalassemia screening between June 2024 and August 2024. A total of 114 individuals were diagnosed with thalassemia via clinical evaluation and confirmatory laboratory testing, with all diagnoses verified by physicians at the thalassemia care center. Standard laboratory tests, like a complete blood count (CBC), a hemoglobin profile (like Hb electrophoresis or HPLC), and metabolic tests done for patient care, confirmed the clinical diagnosis. The biochemical tests included serum creatinine, liver function tests (like SGPT), and serum ferritin levels. People with thalassemia get these tests done on a regular basis to check on their organs and see if they have too much iron. The researchers did not conduct any additional tests; all laboratory results were obtained from patient records at the thalassemia center.

## 2.2. Data Collection

We used standardized questionnaires made in Google Forms and interviews with patients or their families to get demographic and clinical data. The data included the person's age, gender, where they lived, how often they had blood transfusions, and their family background.

## 2.3. Blood Sample Collection

Following the use of 70% alcohol swabs to disinfect the puncture site, 3.0 mL of aseptically collected venous whole blood was transferred to sterile syringes for laboratory testing. We let the samples sit at room temperature until they clotted. After that, to remove them from the serum, they were spun at 4000 rpm for three minutes.

## 2.4. Blood Group Determination

Using the conventional slide agglutination technique, the blood was categorized. Anti-A, anti-B, and anti-D antisera were sourced from Spinreact, a Spanish manufacturer. A drop of each antiserum was placed on a clean glass slide, and then a drop of the patient's blood was added. The liquid was meticulously mixed using a sterile applicator stick, and within two minutes, it was observed under bright light for any signs of clumping. The presence or absence of agglutination was used to determine the ABO and Rh blood groups. A skilled lab worker double-checked all tests three times to guarantee accurate findings.

## 2.5. Study Population

One hundred forty-four individuals with confirmed thalassemia who were seen at the Nawabshah Thalassemia Healthcare Centre for biweekly blood transfusions and regular follow-ups made up the final research study.

## 2.6. Statistical Analysis

Excel 365 from Microsoft and SPSS Statistics 26 from IBM were utilized for data entry and analysis. Demographic, clinical, and genetic factors were defined using descriptive statistics. We used percentages and frequencies to show information about gender, age ranges, places of residency, blood types, family history, paternal consanguinity, and transfusion needs. The distribution of the research population was demonstrated by assembling the data.

# 3. Results

Table 1 shows the demographic details of the people who took part in the study. Thalassemia was diagnosed in 114 of 253 individuals (45.1%). There were 114 people in all: 68 men (59.6%) and 46 women (40.4%). The biggest group of people who took part were 6 to 10 years old (48.2%). The next biggest group was 1 to 5 years old (36.8%), then 11 to 15 years old (8.8%), and finally 16 to 20 years old (6.1%).

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**Table 1**: Demographic characteristics of study participants

| Variables | Frequency | Percentage |
|---|---|---|
| **Gender** | | |
| Male | 68 | 59.6 |
| Female | 46 | 40.4 |
| **Age groups (Years)** | | |
| 1–5 | 42 | 36.8 |
| 6–10 | 55 | 48.2 |
| 11–15 | 10 | 8.8 |
| 16–20 | 7 | 6.1 |
| **Residency** | | |
| Rural | 85 | 74.6 |
| Urban | 29 | 25.4 |

Table 2 summarizes the clinical and genetic characteristics of the enrolled thalassemia patients. Blood group distribution revealed that the majority had group O (46.5%), followed by group A (29.8%), group B (19.3%), and group AB (4.4%). A positive family history of thalassemia was noted in 48 (42.1%) cases, whereas parental consanguinity was observed in 91 (79.8%) participants. Regarding transfusion requirements, most patients required 2 transfusions per month (68.4%), while smaller proportions received 1 (11.4%), 3 (10.5%), 4 (7.0%), or 5 (1.8%) transfusions.

**Table 2**: Clinical and Genetic Characteristics of Thalassemia Patients

| Clinical and Genetic Characteristics (Total = 114) |
|---|
| **Blood group:** A — 34 (29.8%); B — 22 (19.3%); AB — 5 (4.4%); O — 53 (46.5%) |
| **Family history:** 48 (42.1%) |
| **Parental consanguinity:** 91 (79.8%) |
| **Number of transfusions per month:** 1 — 13 (11.4%); 2 — 78 (68.4%); 3 — 12 (10.5%); 4 — 8 (7.0%); 5 — 2 (1.8%) |

## 4. Discussion

A total of 114 (45.1%) individuals were diagnosed with thalassemia. Previous studies reported a high prevalence of thalassemia. Thalassemia was found in 58.75% of the population, according to research.[10] Siblings of patients accounted for 62.2% of the total number of $\beta$-thal trait carriers, according to recent cross-sectional research out of an urban thalassemia treatment and prevention facility in Karachi, Pakistan.[11] The $\beta$-thal subtype of thalassemia is the most common in Pakistan. One descriptive analysis found that 85 (4.05% of the total) of 2010 people tested positive for the $\beta$-thal trait, residing in both urban and rural settings.[12] On Thalassemia Day, 192 adult couples, some married and others single, participated in a research study conducted in Karachi, Sindh Province. We did not include eleven couples because of their past relationships. Just 10 out of 181 couples (5.5%) were discovered to be bearers of the $\beta$-thal trait.[13] In different cross-sectional research, 62 participants, whose ages varied from three months to sixty years, were included.[14]

In our study, the majority of participants were aged 6–10 years (48.2%), followed by 1–5 years (36.8%), 11–15 years (8.8%), and 16–20 years (6.1%). A previous study aligns with our results: Kandhro et al. reported that the distribution of thalassemia trait cases (n = 584) across age groups shows that the majority are diagnosed in adulthood, while fewer are identified in childhood. Only 36 cases (6.17%) were recorded in the 2–4-year age group, which shows that very few were found in early life. This number goes up a lot during school age (5–11 years), with 110 cases (18.83%). This shows that doctors are more likely to notice these cases during this time. There were 84 cases (14.38%) in teenagers aged 12 to 14, which means the burden is still there but is a little less. The majority of cases, 353 individuals (60.41%), were adults, which shows that most diagnoses happen later, usually during the reproductive years.[15]

In this study, there were 114 people in total, 68 of whom were men (59.6%) and 46 of whom were women (40.4%). The preceding study corroborated our findings, indicating that the cohort of 584 individuals with thalassemia trait comprised 323 cases (55.31%) in males and 261 cases (44.69%) in females. This distribution shows that thalassemia trait is more common in males than females in this group, even though it affects both sexes.[15]

Pakistan has a high carrier ratio because of a number of problems, such as not knowing enough about the disease, not being able to read or write, marrying relatives, and not having enough resources for health care, especially in rural areas where most people live. In Pakistan, where intermarriage is prevalent, the thalassemia gene is primarily found in afflicted families rather than randomly spread across the population. Weddings between close relatives account for 80% of weddings in rural Pakistan.[16] The rising number of thalassemic patients is a direct result of the lack of consistent national strategies and goals.

In the current study, blood group distribution revealed that the majority had group O (46.5%), followed by group A (29.8%), B (19.3%), and AB (4.4%). This aligns with previous studies. Blood type O is more prevalent among thalassemia patients in the central area of Saudi Arabia compared to the control sample, according to Laghari, Z. A. et al. (2018).[17] Among thalassemia patients, blood type O is the most prevalent, according to research by Sinha, P. A. et al. (2017).[18]

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In the present study, parental consanguinity was observed in 91 (79.8%) participants. Previous literature aligns with our results; one study reported a 78.5% rate of consanguinity.[19] Among first cousin marriages, another research found that 73% of the children would be born with thalassemia.[20]

## 5. Conclusions

Thalassemia prevalence in Shaheed Benazir Abad was 45.1%, with blood group O being the most common among affected patients. These findings highlight a significant association of thalassemia with specific blood group distributions in the region.

## 6. List of abbreviations

| | |
|---|---|
| **CBC** | Complete blood count |
| **HPLC** | high-performance liquid chromatography |
| **NGS** | Next-Generation Sequencing |
| **SBBU** | Shaheed Benazir Bhutto University |
| **SPSS** | Statistical Package for the Social Sciences |

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## 8. Authorship

All authors are accountable for this work, meet ICMJE authorship criteria, and have approved the final version for publication.

## 9. Authors' Contributions:

W.A.M Supervision, conceptualized and designed the study, Writing – Original Draft. M.Z Collected and analyzed the data, Writing – Review & Editing, Formal analysis, Data curation.
All authors reviewed and approved the final manuscript.

## 10. Conflicts of interest

None

## 11. Funding

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## 12. Consent for publication

Not Applicable

## 13. AI Use Disclosure

ChatGPT assisted with portions of the writing to improve language clarity. The authors reviewed and edited the content and are responsible for the final manuscript.

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