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Investigate the Mortality, Morbidity and Factors Associate in Thalassemia Patients in Faisalabad, Pakistan

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ABSTRACT

Introduction: Thalassemia represents a genetic blood disorder that people obtain from their parents through hereditary materials since their bodies produce abnormal types of hemoglobin. The red blood cell destruction occurs in high numbers because of this disorder thus producing anemia.It is a prevalent disease, and in our society, its prevalence is 75%, with 35% of cases in females and 40% in males. Most of the cases are of beta thalassemia major. **Objective:** We are conducting this research so that in the future, we can correlate the disease with comorbidity in order to reduce the prevalence and so to improve treatment. **Method:** The study design of this research was retrospective cross-sectional study. We made a questionnaire provided to participants for gathering data.This was followed by a peripheral blood smear to assess cell morphology. Iron deficiency tests were then conducted to rule out other causes of anemia. The questionnaire was carefully designed to gather detailed information on patient and family history, lifestyle, awareness levels, and genetic background Patients were monitored through a follow-up process. **Result:** The study found that 75% of participants were affected by thalassemia, with 40% males and 35% females, and most cases were beta thalassemia major. Blood tests like CBC confirmed genetic anemia, not due to iron deficiency. The majority of participants lacked awareness about the disease and its hereditary nature, highlighting the need for screening and education programs. **Conclusion:** In this review we will discuss about Investigate the Mortality, Morbidity and factors associate on Thalassemia patients in Faisalabad 2025Pakistan. Its prevalence rate is high about 75% which is 35% females and 40% males related to our research. **Future Perspective:** This study's findings will inform evidence-based healthcare policies and interventions to reduce mortality and morbidity in thalassemia patients in Faisalabad, Pakistan, ultimately improving patient outcomes and quality of life. The insights gained will also contribute to the development of targeted prevention strategies and optimized management protocols for thalassemia care in resource-constrained settings. Future research can build upon these findings to explore innovative solutions and best practices for thalassemia management in Pakistan.

INTRODUCTION

Thalassemia represents a genetic blood disorder that people obtain from their parents through hereditary materials since their bodies produce abnormal types of hemoglobin. The red blood cell destruction occurs in high numbers because of this disorder thus producing anemia. It is a prevalent disease, and in our society, its prevalence is 75% with 35% cases in females and 40% in males. Most of the cases are of beta thalassemia major. We are conducting this research so that in future, we can reduce the occurrence of disease.

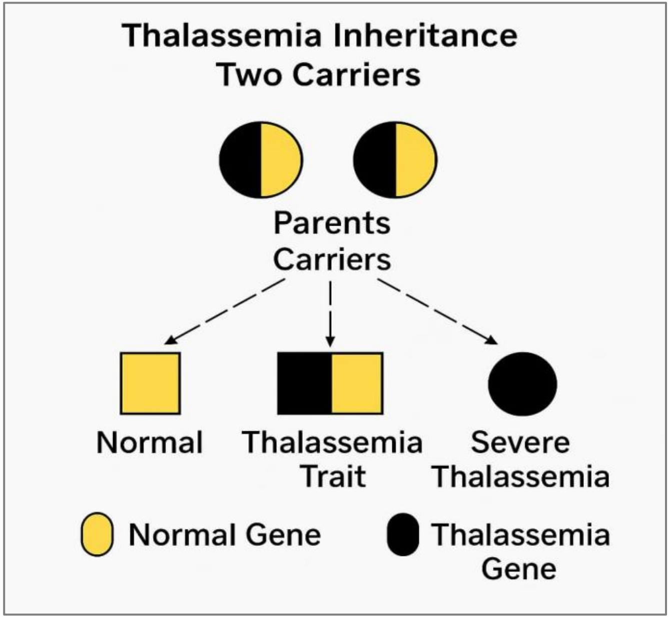
In 1925, Dr. Cooley described a severe type of hypochromic microcytic anemia primarily affecting children of Mediterranean origin. This condition later became known as Cooley's anemia. Around the same period, Italian scientists identified a milder variant of the same illness. By the 1940s, researchers had confirmed that the disease had a hereditary component through the discovery of asymptomatic patients. While the full genetic explanation was still unclear at the time, it was understood that these conditions shared a common genetic basis. Given its high occurrence among Mediterranean populations, the disorder became widely known as thalassemia, specifically beta-thalassemia major.

The term "thalassemia" was first used by George Whipple in 1932. It comes from the Greek word *thalassa*, which means "sea," because the disease was commonly found in areas near the sea. In 1959, Ingram and Stratton suggested that the disease stemmed from a reduced production of beta-globin or alpha-globin chains, leading to a form of anemia that requires regular blood transfusions. Major breakthroughs in the 1970s uncovered specific mutations in hemoglobin genes responsible for the condition. Today, thalassemia is recognized as a group of inherited blood disorders marked by abnormal or reduced hemoglobin production, with a global impact. Beta-thalassemia affects approximately 3% of the global population, with around 60,000 babies born with the condition each year. Common symptoms include anemia, pale skin, an enlarged spleen (splenomegaly), fatigue, difficulty breathing, deformities in bone structure, an enlarged liver, and excess iron in the body.

Transmissibility

Thalassemia is an inherited blood disorder marked by reduced hemoglobin production, causing chronic anemia. It is a carrier state in which carry gene from parents to their offspring's. It is an inherited autosomal recessive disorder affecting the production of either the alpha or beta chains of hemoglobin.

Figure 1



Types of Thalassemia

Alpha Thalassemia

Caused by deletion or mutation in alpha-globin genes (HBA1, HBA2). Humans have four alpha genes. Severity depends on how many genes are affected: 1 gene: Silent Carrier (No symptoms) 2 genes: Alpha Thalassemia Trait (Mild anemia) ,3 genes: Hemoglobin H Disease (Moderate to severe anemia),4 genes: Hydrops Fetalis (Usually fatal before or shortly after birth)

Beta Thalassemia

Caused by mutation in the beta-globin gene (HBB). Humans have two beta genes i.e. Beta Thalassemia Minor (Trait): One defective gene; mild/no anemia. Beta Thalassemia Major (Cooley's anemia): Both genes severely mutated; severe anemia from early life.

Early diagnosis

Early diagnosis of thalassemia is crucial to manage the disease effectively, reduce complications, and improve quality of life.

a. Prenatal Diagnosis

Chorionic Villus Sampling (CVS) (10–12 weeks gestation)**,** Amniocentesis (15–20 weeks gestation)**,** Preimplantation Genetic Diagnosis (PGD) – used with IVF to select embryos without the disease**.** These tests detect mutations in the HBB (beta) or HBA1/HBA2 (alpha) genes.

b. Newborn Screening

Performed in many countries as part of routine neonatal screening**.** Hemoglobin electrophoresis or high-performance liquid chromatography (HPLC) is used to detect abnormal hemoglobin variants.

c. Carrier (Trait) Screening

Especially important in high-risk populations**.** It is done by Complete Blood Count (CBC) – shows microcytic hypochromic anemia. Hemoglobin electrophoresis – detects abnormal hemoglobin variants (e.g., HbA2 elevation in beta-thalassemia trait)**.** DNA analysis – confirms specific gene mutations

Who Should Be Screened?

People with a family history of thalassemia**.** Individuals from high-prevalence regions Couples planning to have children (especially if both are carriers)

Hemoglobin Electrophoresis

Hemoglobin electrophoresis is a specialized laboratory technique used to identify and quantify different types of hemoglobin in a blood sample. This test plays a crucial role in the diagnosis of various hemoglobin disorders, particularly thalassemia, as well as other conditions like sickle cell disease and hemoglobin C disease.

The principle behind hemoglobin electrophoresis is based on the movement of charged hemoglobin molecules in an electric field. Hemoglobin's vary slightly in their structure and electrical charge, so when an electric current is applied to a gel or capillary containing a blood sample, the different types of hemoglobin migrate at different speeds and directions. By analyzing the resulting banding pattern or peaks, laboratory professionals can determine which types of hemoglobin are present and in what proportion.

β-TM patients need periodic blood transfusions and iron chelation treatment starting in early childhood to stay alive due to autosomal recessive blood disorder conditions. The projected β-TM affected newborn population in Pakistan spans from 4,000 to 9,000 annually. Medical experts predict that Pakistan faces 5,250 β-TM cases annually because of its birth rate of 1.3 children per 1,000 births. The carrier frequency extends between 5 and 8% while the disorder occurs in every population ethnicity. Patients with β-TM undergo growth retardation along with bone pains and fragility fractures because of excessive erythropoiesis and hormonal imbalances and bone hypoxia and hemochromatosis and chelation therapy. The body needs strict regulation of Iron and P metabolism to develop healthy bones. Studies on thalassemia patients' calcium homeostasis and vitamin D deficiency grew prominent during the last decade while phosphate homoeostasis remained sparsely researched by scholars.

The proper treatments and quality care options for β-TM enhance a patient's chances of living longer. The developed nations demonstrate monitoring techniques but Pakistan maintains a significant gap in medical care level. The country of Pakistan exists as a developing state that surpasses 200 million permanent residents. Healthcare limitations together with non-communicable diseases pose the greatest challenges to Pakistan's healthcare system. The WHO now leads cross-institutional programs which develop genomic approaches for public health applications. Public health treatment implementations aren't established under the current legislation system because Pakistan lacks monetary support and trained personnel. Blood safety represents a major problem in Pakistan because the country experiences high frequencies with hepatitis B and hepatitis C being the most prevalent.

Patients with thalassemia major experience psychosocial problems as a result of their illness. Stuttering and mold-like features of the face together with hepatosplenomegaly and growth impediments along with yellowed skin and distinctive physical structures characterize the illness. The illness produces noticeable face wrinkles which match its features. People's self-esteem and confidence This body image causes severe damage to patients which isolates them from others. Their presence in social interactions becomes limited because of this condition so patients remain inactive. Detrimental effect on their mental and quality of life. Unlike other forms of treatments, thalassemia is treated with innovative yet therapeutically options like regular transfusions, gene therapy, iron chelation therapy and bone marrow transplanting. This condition is widespread in areas around the Mediterranean Sea extending to South Asia, Middle East as well as Africa.

In Pakistan the public awareness program regarding thalassemia disease is very poor so parents have little information about this deadly disease. The effective way for the prevention of thalassemia is through seminars, trainings of practitioners, print media and electronic media, social workers, obstetricians etc. For the successful prevention program it is mandatory that all the stakeholders, electronic and print media, thalassemia families, nongovernmental organization, government should play their vital role. We designed this survey to identify how many β-thalassemia major patients with multiple blood transfusions also presented short stature to enable proper identification and management of their growth velocity.

Bone marrow transplantation remains the only definitive cure currently available. Individuals with thalassemia intermedia may require splenectomy, folic acid supplementation, treatment of extra medullary erythropoietic masses and leg ulcers, prevention and therapy of thromboembolic events. Prognosis for individuals with beta-thalassemia has improved substantially in the last 20 years following recent medical advances in transfusion, iron chelation and bone marrow transplantation therapy. However, cardiac disease remains the main cause of death in patients with iron overload.

Statement of the Problem

Thalassemia is an inherited blood disorder marked by reduced hemoglobin production, causing chronic anemia. Despite treatment advances, it remains a major health challenge, especially in high-prevalence regions. Patients often suffer from complications like iron overload, organ damage, and frequent transfusions. Mortality rates are still high, particularly where access to care is limited.Factors such as socioeconomic status, healthcare access, genetics, and treatment adherence settings. This study aims to explore morbidity, mortality, and associated risk factors to improve patient outcomes.

MATERIAL AND METHODS

This research was conducted using a retrospective cross-sectional study design. A structured questionnaire was provided to participants to gather relevant data. It included questions about patient and family history, lifestyle, awareness of thalassemia, and genetic background. After collecting the questionnaire data, a peripheral blood smear was performed to examine red blood cell morphology. This helped identify typical features of thalassemia. Iron deficiency tests were also carried out to rule out other potential causes of anemia. The combined diagnostic approach ensured accurate identification of the disease. Patients were then monitored through a follow-up process to assess ongoing health conditions. This method provided a comprehensive understanding of thalassemia and its effects on the studied population.

Antibodies

Thalassemia is a genetic disorder, not an autoimmune disease, so it is not directly associated with autoantibodies in the way autoimmune conditions are. However, antibodies do become relevant in thalassemia management, especially in the context of blood transfusions, which are a common treatment for moderate to severe forms like beta-thalassemia major.

Here is a detailed explanation of how antibodies are involved in thalassemia care:

A. Alloantibodies and Autoantibodies in Transfusion Therapy

Patients with thalassemia, especially those requiring frequent blood transfusions, are at risk of developing alloantibodies and, less commonly, autoantibodies:

Alloantibodies

These are antibodies formed against antigens on transfused red blood cells (RBCs**)** that the patient’s immune system recognizes as foreign. Common alloantibodies seen in transfused thalassemia patients include: Anti-Kell (K), Anti-D, Anti-C, Anti-E (Rh system),Anti-Jka (Kidd system) Anti-Fya (Duffy system) .Formation of these antibodies can lead to delayed hemolytic transfusion reactions and difficulty in finding compatible blood.

Autoantibodies

These are antibodies that attack the patient's own red blood cells, leading to autoimmune hemolytic anemia. They are usually detected through a direct ant globulin test (DAT or Coombs test).

B. Antibody Screening and Crossmatching

Before any transfusion, especially in thalassemia patients: Antibody screening is done to detect any pre-existing alloantibodies.Crossmatching ensures donor blood is compatible and won't trigger a reaction. Extended red cell phenotyping or genotyping is recommended early on to match donor blood as closely as possible to prevent alloimmunization.

C. No Diagnostic Antibody for Thalassemia

It's important to note:
There are no specific diagnostic antibodies for detecting thalassemia. Thalassemia is diagnosed using genetic tests, hemoglobin electrophoresis, and CBC, not through antibody detection.

D. Monoclonal Antibodies in Research

In recent years, monoclonal antibodies and immune-modulating therapies are being explored in thalassemia-related complications (e.g., targeting ineffective erythropoiesis or iron overload), but these are mostly experimental or adjunctive and not yet standard care.

Thalassemia Diagnosis

Diagnosing thalassemia involves a step-by-step approach combining hematological tests, biochemical analysis, and genetic studies. Below it is a detailed description of the standard test procedure are used to diagnose thalassemia which include

Complete Blood Count (CBC)

Purpose

To assess red blood cell (RBC) indices and screen for anemia.

Procedure

Fresh venous blood (2–3 ml) was Collected from the thalasemia patients in an EDTA tube and analyzed using an automated hematology analyzer. Its look for: Low hemoglobin (Hb), Microcytosis (↓ MCV < 80 fL), Hypochromic (↓ MCH < 27 pg)

Peripheral Blood Smear

Purpose

To examine the shape and appearance of red blood cells.

Procedure

To prepare a thin blood smear on a glass slide. Stain with Wright-Giemsa stain. Examine under a microscope for: Microcytosis, Hypochromia, Target cells and Basophilic stippling

Hemoglobin Electrophoresis

Purpose

To identify and quantify types of hemoglobin.

Procedure (Electrophoresis)

Place a hemolysate of the patient's blood on a gel or cellulose acetate strip. Apply an electric current. Hemoglobin moves based on their charge; observe and compare the pattern.

Procedure (HPLC)

Inject blood sample into HPLC machine. Detect and quantify: HbA, HbA2, HbF and other variants (e.g., HbS, HbE)

Interpretation

In beta-thalassemia trait, hemoglobin electrophoresis typically shows an increased HbA2 level (greater than 3.5%) along with a mild increase in HbF. In contrast, beta-thalassemia major is characterized by a marked decrease or complete absence of HbA, with a significant increase in HbF levels. Alpha-thalassemia, however, often presents with normal results on hemoglobin electrophoresis, making it difficult to detect through this method alone; therefore, DNA testing is usually required for accurate diagnosis.

Genetic Testing (DNA Analysis)

Purpose

To confirm the specific mutation(s) in the alpha or beta globin genes.

Procedure

Collect whole blood in an EDTA tube. Extract genomic DNA from white blood cells.
Use techniques like: PCR (Polymerase Chain Reaction), Reverse dot blot hybridization, DNA sequencing like detect deletions or point mutations in HBB gene (beta-thalassemia) HBA1, HBA2 genes (alpha-thalassemi)

RESULT

Demographic information:

The table above presents the age distribution of patients divided into four groups: 6-10 years, 11-15 years, 16-20 years, and 21-25 years. The frequency of patients in each group is as follows: 54 patients (36.0% of the total) for the 6-10 years group, 46 patients (30.7% of the total) for the 11-15 years group, 24 patients (16.0% of the total) for the 16-20 years group, and 26 patients (17.3% of the total) for the 21-25 years group.

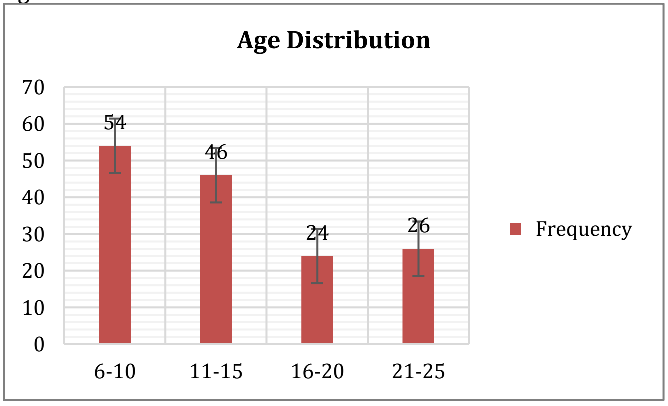
Table

Age Distribution

Age	frequency	valid percentage
6-10	54	36%
11-15	46	30.7%
16-20	24	16%
21-25	26	17.3%
Sum	150	100%

Graph 2.1

Age Distribution



Gender Distribution

This table illustrates the gender distribution among the 150 participants in a survey or study, consisting of 88 males and 62 females. These groups represent 58.7% and 41.3% of the total valid responses, respectively.

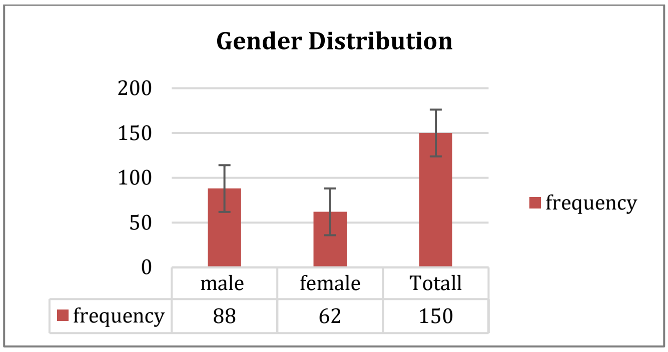
Table 2.2

Gender Distribution

Gender	Frequency	Valid Percentage
male	88	58.7%
female	62	41.3%
sum	150	100%

Graph 2.2

Gender distribution



	male	female	Totall
frequency	88	62	150

Type of Thalassemia

The graph presented above illustrates the frequency of various types of thalassemia. Our findings indicate that beta major thalassemia is the most prevalent type compared to others. Out of 150 patients, 122 were diagnosed with beta thalassemia, while approximately 18 had beta minor thalassemia. Additionally, 10 patients were identified as having alpha thalassemiathalassemia.

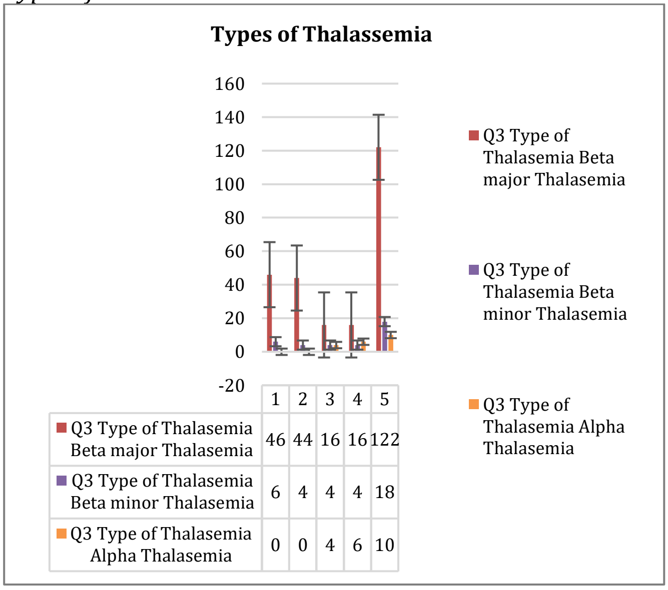
Table 2.3

Types of Thalassemia

age	B.Th major	B.Th minor	A.Th	valid percentage
6-10	46	8	0	36%
11-15	44	2	0	30.7%
16-20	16	4	4	16%
21-25	16	4	6	17.3%
sum	122	18	10	100%

Gable 2.3

Types of Thalassemia



	1	2	3	4	5
Q3 Type of Thalasemia Beta major Thalasemia	46	44	16	16	122
Q3 Type of Thalasemia Beta minor Thalasemia	6	4	4	4	18
Q3 Type of Thalasemia Alpha Thalasemia	0	0	4	6	10

Requited Blood Transfusion

The graph presented above illustrates the frequency of transfusion intervals among patients with thalassemia. It indicates that 28 patients with 19 % received transfusions every 10-20 days (weekly), reflecting the severity of their condition. Meanwhile, 122 patients with 81.3% had transfusion intervals of 30-45 days (monthly)

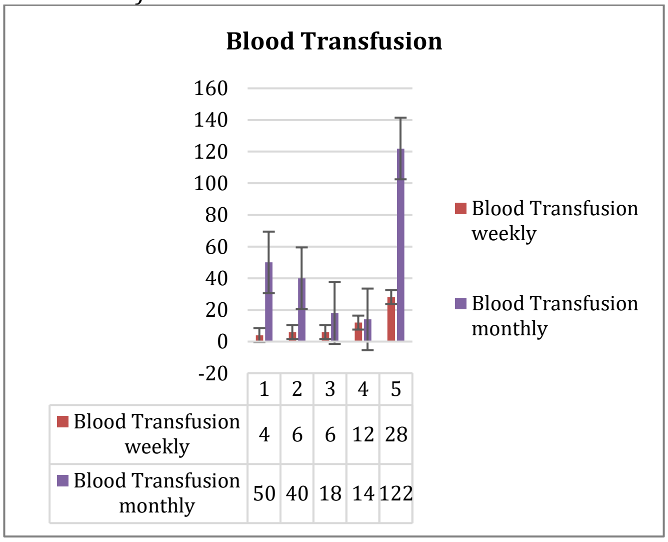
Table 2.4

Blood Transfusion

Age	weekly	monthly	valid percentage
6-10	4	50	36%
11-15	6	40	30.7%
16-20	6	18	16%
21-25	12	14	17.3%
sum	28	122	100%

Graph 2.4

Blood Transfusion



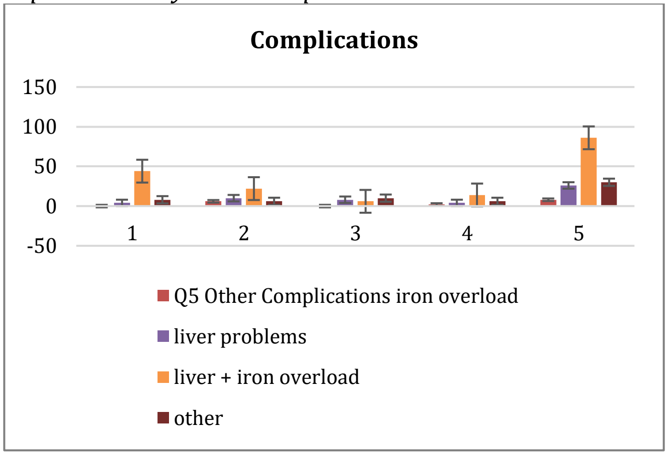
	1	2	3	4	5
Blood Transfusion weekly	4	6	6	12	28
Blood Transfusion monthly	50	40	18	14	122

Experienced any other complications due to thalassemia

The graph presented above illustrates the frequency of other complications due to thalassemia among patients with Thalassemia.The "valid percentage" indicates that the 6-10 age group constitutes 37.3% of the valid records, observed by means of the 11-15 age group at 29.3%, then jthe 21-25 age group at 17.3%, and the 16-20 age organization at 16%. This suggests that the majority of the information comes from more youthful age companies.

Graph 2.5

Experienced any other complications due to thalassemia



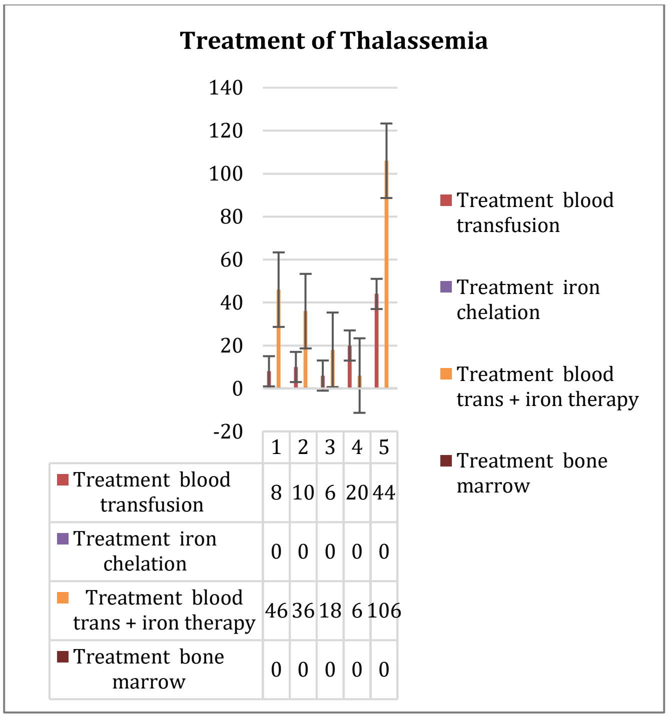
Treatment

The table reflects the distribution of medical treatment in various age groups. Age Group 6–10 attained 46 examples of this treatment, followed by 36 examples in the age group of 11–15, 18 examples in the age group of 16-20 and 6 in the age group of 21-25. With a total of 44 examples in all age groups, blood transfusion was also administered. In particular, "Iron Chelost" and "Bone Marrow" do not represent treatment data. The valid percentage column adds up to 100%, showing that the data is widespread.

Table 2.6
Treatment

Age	blood transfusion	iron chelation	blood trans + iron therapy	bone marrow	valid percentage
6-10	8	0	46	0	36%
11-15	10	0	36	0	31%
16-20	6	0	18	0	16%
21-25	20	0	6	0	17%
sum	**44**	**0**	**106**	**0**	**100%**

Graph 2.6
Treatment



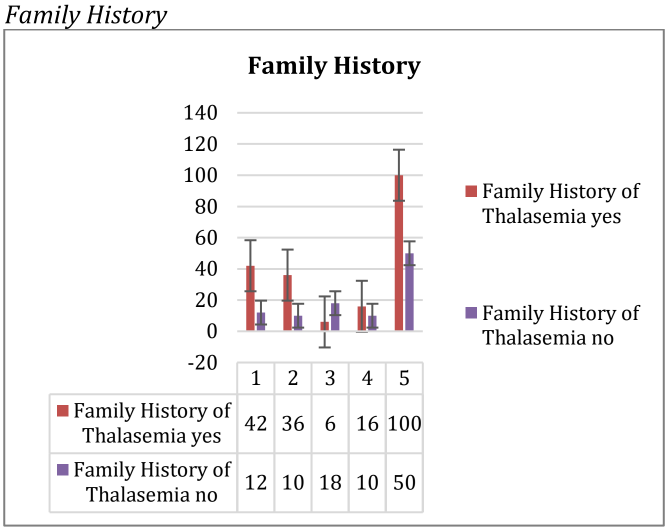
Family History

Table and bar graphs show family history data in groups between 6–10, 11–15, 16–20 and 21–25. Most individuals with family history are 6–10 (42) and 11–15 (36) age groups. The data indicates the high proliferation of family history among groups of young age.

Table 2.7
Family History

Age	yes	no	valid percentage
6-10	42	12	36%
11-15	36	10	30.7%
16-20	6	18	16%
21-25	16	10	17.3%
Sum	**100**	**50**	**100%**

Graph 2.7
Family History



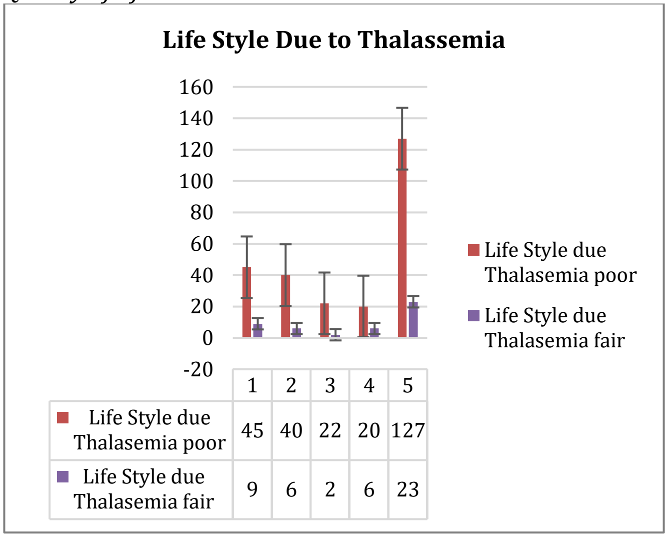
Quality of life due to Thalassemia

The chart shows the great of lifestyles of thalassemia sufferers throughout one of a kind age corporations. Most sufferers suggested a bad pleasant of lifestyles, especially inside the 6–10 and 11–15 age agencies. The range of humans reporting a truthful satisfactory of life is plenty lower in all age organizations. Universal, 127 patients had poor quality, 23 had fair nice, and the information covers a hundred% of legitimate responses.

Table 2.8
Quality of life due to Thalassemia

Age	poor	fair	valid percentage
6-10	45	9	36
11-15	40	6	30.7
16-20	22	2	16
21-25	20	6	17.3
sum	**127**	**23**	**100**

Graph 2.8
Quality of life due to Thalassemia



Family member passed away due Thalassemia

A bar chart indicate a records categorized by way of age businesses, with responses to whether or not a family member passed away because of Thalassemia (sure or no), along with the valid percentage. Age companies: 6–10, 11–15, sixteen–20, 21–25general responses: one hundred fifty

(58 "yes", ninety two "no") maximum effect: Age group 6–10 had the best percent (36%) reporting a family demise because of Thalassemia. Observed by eleven–15 (30.7%).

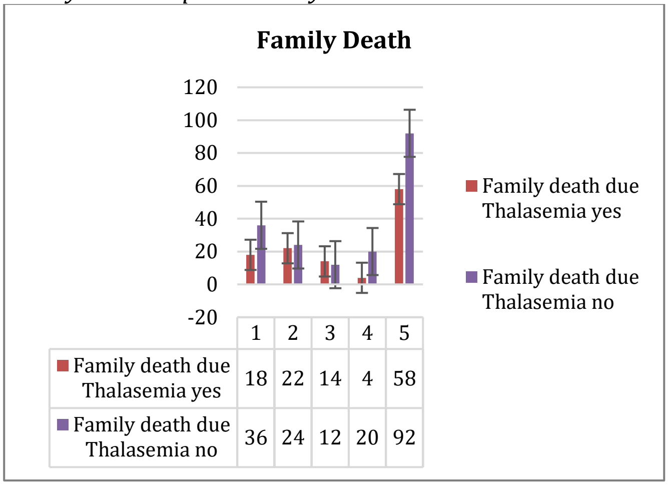
Table 2.9

Family member passed away due Thalassemia

Age	yes	no	valid percentage
6-10	18	36	36
11-15	22	24	30.7
16-20	14	12	17.3
21-25	4	20	16
Sum	58	92	100

Graph 2.9

Family member passed away due Thalassemia



Aware of genetic counseling for thalassemia carriers

This graph indicates that awareness will increase with age maximum conscious: 21–25 years (22 stated "yes") Least aware: 6–10 years (most effective nine stated "yes") general: fifty seven aware, ninety-three are unaware. This indicates that older age agencies are extra informed, probably due to better schooling and publicity.

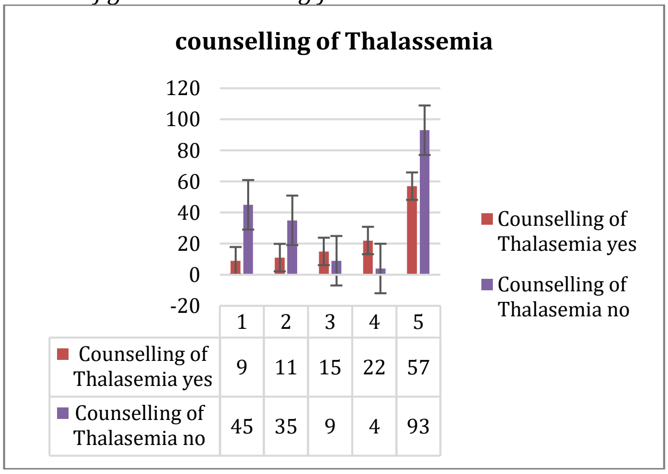
Table 2.10

Aware of genetic counseling for thalassemia carriers

Age	yes	no	valid percentage
6-10	9	45	36%
11-15	11	35	31%
16-20	15	9	16%
21-25	22	4	17%
sum	57	93	100%

Graph 2.10

Aware of genetic counseling for thalassemia carriers



ANOVA outcomes

between businesses SS (Sum of Squares) = 16575.14 This looks as if the result of an ANOVA (evaluation of Variance) evaluating a couple of express companies across some established variable(s), in all likelihood associated with Thalassemia traits or outcomes

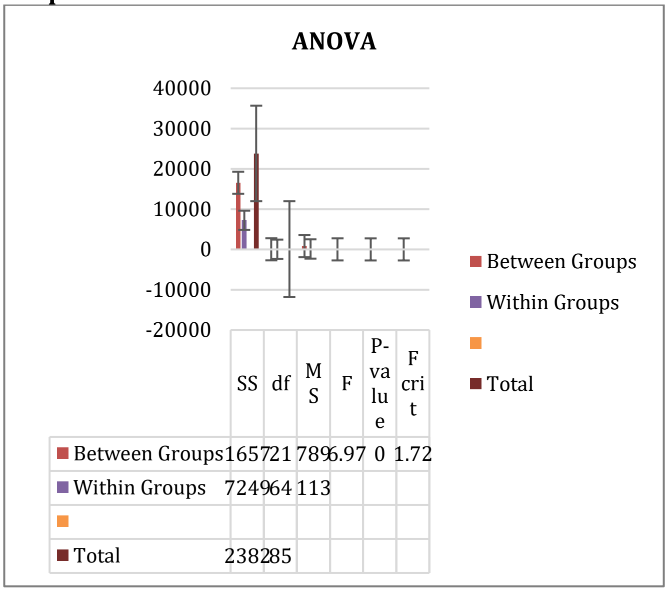
Table 2.23

ANOVA Outcomes

ANOVA

Source of Variation	*SS*	*df*	*MS*	*F*	*P-value*	*F crit*
Between Groups	16575.13953	21	789.2923588	6.968507513	7.67522E-10	1.723277736
Within Groups	7249	64	113.265625			
Total	23824.13953	85				

Graph 2.23



ANOVA outcomes

between businesses SS (Sum of Squares) = 16575.14 inside businesses SS = 7249 general SS = 23824.14 tiers of Freedom (df): not explicitly stated, but appears popular (e.g., df among = quantity of corporations - 1) ,F-value = 789.2924 P-fee = 7.68E-10 Fcrit = 1.723278

Interpretation

P-cost < 0.05: Indicates statistically significant differences between group means. < Since the F-valueFcrit, we reject the null Hypothesis — the way aren't all identical; as a minimum one group differs notably. There are big differences among the numerous Thalassemia-related conditions and factors (e.g., type, headaches, own family history, counseling).agencies like "Beta major Thalassemia" and "Counseling (No)" show high variance and averages, which likely make a contribution most to between-institution variant.

MORBIDITY & MORTALITY RATE

As our work on Investigate the Mortality, morbidity and factors associate on Thalassemia patients Faisalabad Pakistan 2025.

- **Morbidity Rate:**

Morbidity Rate= $\left(\frac{150}{150}\right) \times 100 = 100\%$

- **Interpretation:**

The horribleness rate in this test is 100%, as all members were influenced by Thalassemia. Typically anticipated since the test particularly focused on Thalassemia patients.

- **Mortality Rate :**

Mortality Rate =$\left(\frac{3}{150}\right) \times 100 = 20$ deaths per 1,000

Number of Deaths: 3 Sample size: 150 Number of Death based upon the thesis title and study duration (means 4month of study)

- **Interpretation**

The disease-specific mortality rate among Thalassemia patients in this test was 20 per 1,000. This demonstrates that 2% of the patients kicked the bucket amid the watched period.

DISCUSSION

The study was designed to investigate the mortality, morbidity and factors associate on thalassemia patient in Faisalabad. Thalassemia was a genetically inherited blood disorder marked by body's inability to produce inadequate hemoglobin. The Clinical findings that indicate the thalassemia remains a significant health concern in the region, with a considerable number of patients (including 150 patients) was found to be influenced by several key factors, including the Frequency of blood transfusions, iron overload, lack of iron chelation therapy, and socioeconomic conditions. Thalassemia is a hereditary blood disorder marked by ineffective erythropoiesis and chronic hemolytic anemia due to mutations in the globin genes. In this study, we investigated the mortality, morbidity, and associated hematological factors in 150 patients with thalassemia, whose data were collected from two specialized treatment centers in Pakistan — the Sundas Foundation and the Ali zaib Foundation. We created a questionnaire and conducted a survey of different types of people. We collected data from them and kept our sample size between 6 and 50. The design of this study was a retrospective cross-sectional study. It was conducted in various hospitals located in Faisalabad, specifically including Sundas Foundation and Ali Zaib Foundation, where data was collected from patients. The inclusion criteria for participants in this study were as follows: individuals aged between 6 and 50 years who were diagnosed with thalassemia (both genotype and phenotype). The study included teenagers, adult males and females, as well as pregnant women. Participants could be thalassemia major, intermediate, minor, or carriers. Additionally, people with a previous history of Type I or Type II diabetes were also included in the study. The exclusion criteria for this study included individuals who were not diagnosed with thalassemia, as well as patients with a history of cancer. Participants suffering from other blood disorders or abnormalities, and those with infectious diseases such as HIV and Hepatitis, were also excluded from the study. The sample size selected for this study was 150 patients, as detailed in Table 1. Data was collected from two major thalassemia care centers in Faisalabad: 25 patients from Sundas Foundation and 125 patients from Ali Zaib Foundation. The duration of the study was approximately 3 to 4 months, which began after receiving approval for the research synopsis. For the sampling technique, the 50 required sample size was calculated using the WHO sample size calculator. Based on a 5% margin of error and a 90% confidence level, it was determined that at least 150 participants were needed. However, the study included 150 participants to enhance the robustness of the findings. The data collection tools included a structured questionnaire that was administered to participants. The data collection process followed a stepwise medical diagnostic approach, starting with routine blood tests such as Complete Blood Count (CBC), which included measurements like RBCs, MCV, HCT, and MCHC. This was followed by a peripheral blood smear to examine red blood cell morphology. Iron deficiency tests were conducted to exclude other causes of anemia. In addition, participants were monitored through a follow-up process to ensure continuous and accurate evaluation. Data for this study was gathered from various healthcare institutes in Faisalabad, notably from organizations actively involved in the treatment and management of thalassemia, such as Sundas Foundation and Ali Zaib Foundation. The structured questionnaire was used as a standardized tool to ensure consistency and accuracy in data collection. After collecting the data, we used SPSS and Microsoft Excel as our primary data analysis tools. While SPSS was used for some statistical procedures, most of the data organization and initial analysis were carried out using Excel. In Excel, we created a data sheet that categorized participants based on gender (male and female). We then identified and recorded various factors that were affecting their daily lives. These factors included physical, emotional, and lifestyle-related aspects that influence the quality of life of thalassemia patients. We specifically focused on evaluating their overall life quality, assessing whether it was good or poor. Indicators used in this assessment included the presence of stress, anxiety, and whether the patients were maintaining a proper and balanced diet. By analyzing these variables, we aimed to understand how thalassemia impacts not just the physical health of individuals but also their mental well-being and everyday functioning. As part of our study, we also examined the morbidity and mortality rates among thalassemia patients. Specifically, we recorded the number of deaths reported in 2025 so far. According to the available data, three deaths have been reported among patients diagnosed with thalassemia major. Upon further analysis; we observed that the majority of these cases involved young children under the age of 15. This indicates a higher mortality risk in pediatric thalassemia major patients, highlighting the urgent need for improved medical management, early intervention, and consistent follow-up care in this vulnerable age group. We applied the ANOVA (Analysis of Variance) statistical test as part of our data analysis. The main objective of using ANOVA was to determine whether there were any statistically significant differences between the different groups we had created based on various demographic and clinical characteristics. By applying ANOVA, we aimed to identify whether the differences observed between groups were meaningful or due to random chance. This helped us examine the relationships between the factors we studied through the questionnaire — such as stress levels, dietary habits, mental health, and overall quality of life — and assess whether these factors actually apply to the population of thalassemia patients. Through this analysis, we found that many of the observed factors, particularly those affecting quality of life, were indeed statistically significant. Moreover, one of the key findings was that genetic factors played a major role in influencing these outcomes. This supports the understanding that thalassemia, being a hereditary genetic disorder, has effects that are deeply rooted in the patient's genetic background, which in turn impacts both the clinical presentation and the social and emotional challenges faced by the patients. The study revealed critical Finding. Many thalassemia patients in Faisalabad had died, especially when they didn't receive regular treatment. Many patients also had other health problems like too much iron in the body, liver and heart issues, slow growth, and frequent infections. These problems were often caused by not taking iron-removing medicine properly, not getting enough blood transfusions, not knowing enough about the disease, and had low income. Morbidity with also prevalent, with complication included iron overloaded, heart disease, growth retardation and recurrent infection being observed. The results showed that patients who got regular blood transfusions but not enough iron-removing treatment (chelation therapy) had a higher chance of having too much iron in their bodies. This extra iron could damage organs and lead to more health problems and even death. It was also found that patients from poor families or those living in villages had less access to good healthcare and important treatments. The data showed that older thalassemia patients had more health problems, likely because of the long-term effects.

CONCLUSION

Thalassemia major is a chronic inherited disorder with serious physical consequences. In Pakistan, it represents a growing public health challenge due to limited awareness, inadequate healthcare infrastructure, and insufficient resources. The disease not only compromises physical health but also significantly affects the mental and emotional well-being of patients. Social stigma and isolation further diminish their quality of life. High rates of morbidity and mortality are often the result of delayed diagnosis and limited access to effective treatment. Rural and underserved populations are especially vulnerable due to the lack of medical facilities. Addressing these challenges requires urgent governmental reforms and sustained collaboration between the public and private sectors. Promoting awareness, expanding screening programs, and ensuring equitable access to care are essential steps toward reducing the overall burden of thalassemia in Pakistan.

REFFERENCES

1. Al_Hamadiny, S. Q., Alqaisi, A. A., Al-Sarray, R. G., Ali, S. A., Trad, H. Q., & Abood, A. J. (2024). Psychosocial factors associated with major thalassemia. *European Journal of Medical Genetics and Clinical Biology*, *1*(9), 10-15. https://doi.org/10.61796/jmgcb.v1i9.917
2. Ansari, A. H., Ansari, S. H., Salman, M. J., Ansari, M. U., & Jabeen, R. (2024). A scoping review on the obstacles faced by beta thalassemia major patients in pakistan- Matter of policy investment. *AIMS Public Health*, *11*(4), 1105-1124. https://doi.org/10.3934/publichealth.2024057
3. Asif, N., & Hassan, K. (2016). Management of thalassemia in Pakistan. *Journal of Islamabad Medical & Dental College*, *5*(4), 152-153. https://mail.jimdc.org.pk/index.php/JIMDC/article/view/118
4. Baig, S. M., Azhar, A., Hassan, H., Baig, J. M., Aslam, M., Ud Din, M. A., Qureshi, J. A., & Zaman, T. (2006). Prenatal diagnosis of β-thalassemia in southern Punjab, Pakistan. *Prenatal Diagnosis*, *26*(10), 903-905. https://doi.org/10.1002/pd.1523
5. Batool, I. R. A. M., Ishfaq, K. A. M. R. A. N., & Bajwa, R. S. (2017). Psychosocial burden among thalassemia major patients: An exploratory investigation of south Punjab, Pakistan. *Pakistan Paediatr J*, *41*(3), 158-62.
6. Benz, E. J. (2023). Introduction to the thalassemia syndromes. *Hematology/Oncology Clinics of North America*, *37*(2), 245-259. https://doi.org/10.1016/j.hoc.2022.11.001
7. Galanello, R., & Origa, R. (2010). Beta-thalassemia. *Orphanet Journal of Rare Diseases*, *5*(1). https://doi.org/10.1186/1750-1172-5-11
8. Jabeen, R., Ansari, I., Durrani, B., Salman, M. J., Mazhar, L., Ansari, M. U., Ansari, A. H., Kabani, S., & Ansari, S. H. (2024). Exploring the perceptions and experiences of female's with S-thalassemia major in a tertiary care private hospital in Pakistan. *Transfusion Clinique et Biologique*, *31*(4), 244-252. https://doi.org/10.1016/j.tracli.2024.06.008
9. Jafri, L., Jameel Farooqui, A., Moiz, B., Sheikh, A., Majid, H., Nadeem, S., Quddus, R., Khan, S., Khan, Q., & Habib Khan, A. (2025). Factors associated with phosphate homeostasis in children with beta-thalassemia major: An analytical cross sectional study from Pakistan. *PLOS ONE*, *20*(2), e0316566. https://doi.org/10.1371/journal.pone.0316566
10. Marsella, M., Pepe, A., & Borgna-Pignatti, C. (2010). Better survival and less cardiac morbidity in female patients with thalassemia major: A review of the literature. *Annals of the New York Academy of Sciences*, *1202*(1), 129-133. https://doi.org/10.1111/j.1749-6632.2010.05588.x
11. Mashon, R. S., Mandrelle, K., & Kakkar, N. (2024). Experience of antenatal thalassemia screening and prenatal diagnosis from a tertiary care teaching hospital in Punjab. *Indian Journal of Hematology and Blood Transfusion*, *41*(1), 96-101. https://doi.org/10.1007/s12288-024-01785-z
12. Meri, M. A., Al-Hakeem, A. H., & Al-Abeadi, R. S. (2022). Overview on thalassemia: A review article. *Medical Science Journal for Advance Research*, *3*(1), 26-32. https://doi.org/10.46966/msjar.v3i1.36
13. Prathyusha, K., Venkataswamy, M., Goud, K. S., Ramanjaneyulu, K., Himabindu, J., & Raj, K. S. (2019). Thalassemia-A blood disorder, its cause, prevention and management. *Research Journal of Pharmaceutical Dosage Forms and Technology*, *11*(3), 186. https://doi.org/10.5958/0975-4377.2019.00033.8

14. Soliman, A., Yassin, M., Alyafei, F., Alaaraj, N., Hamed, N., Osman, S., & Soliman, N. (2023). Nutritional studies in patients with β-thalassemia major: A short review. *Acta Bio Medica: Atenei Parmensis*, *94*(3), e2023187. https://doi.org/10.23750/abm.v94i3.14732
15. Ghafoor, M., Sabar, M. F., & Sabir, F. (2020). Prevention programs and prenatal diagnosis for beta thalassemia in Pakistan: A narrative review. *Journal of the Pakistan Medical Association*, 1-17. https://doi.org/10.47391/jpma.665
16. Ganaee, M. A., & Iqbal, M. J. (2018). Development of IIUI e-Theses collection: a case study. *Pakistan Library & Information Science Journal*, *49*(4). https://openurl.ebsco.com/EPDB%3Agcd%3A5%3A32711925/detailv2?sid=ebsco%3Aplink%3Ascholar&id=ebsco%3Agcd%3A134723605&crl=c&link_origin=scholar.google.com
17. Arshad, B., Iqbal, T., Jamil, A., & Asan, I. J. H. (2014). Hematological and biochemical changes in male patients with thalassemia major. *Pak. j. life soc. Sci*, *12*(1), 31-35. https://www.pjlss.edu.pk/pdf_files/2014_1/31-35.pdf
18. Aziz, A., Tehreem, H., Aslam, R., Khan, W. A., Javed, M. U., Ali, M. S., Riaz, A., Tariq, M. N., Gilani, S. S., Noor, S., & Noor, E. (2024). Consanguineous marriages and thalassemia major in Pakistan: A cross-sectional study on awareness and prevalence. *The American Journal of Medical Sciences and Pharmaceutical Research*, *6*(7), 49-61. https://doi.org/10.37547/tajmspr/volume06issue07-07
19. Malik, M., Gul, Z., Bohair, J., Naeem, S. K., & Sadiq, N. (2024). Family planning practices among the parents of beta thalassemia major patients in Makran Division, Balochistan: A cross-sectional study. *Pakistan Journal of Medical Sciences*, *41*(1), 230-234. https://doi.org/10.12669/pjms.41.1.10263
20. Daar, S., Al-Naamani, K., De Sanctis, V., Al Rahbi, S., Zedjali, S., Khan, H., ... & Al-Khabori, M. (2023). Mortality and complications in Omani patients with beta-thalassemia major: a long-term follow-up study. *Acta Bio Medica: Atenei Parmensis*, *94*(4), e2023191. https://doi.org/10.23750/abm.v94i4.14856
21. Faranoush, M., Faranoush, P., Heydari, I., Foroughi-Gilvaee, M. R., Azarkeivan, A., Parsai Kia, A., Sadighnia, N., Elahinia, A., Zandi, A., Rezvany, M. R., Hashemi-Madani, N., Ziaee, A., Nekouian, R., & Rohani, F. (2023). Complications in patients with transfusion dependent thalassemia: A descriptive cross-sectional study. *Health Science Reports*, *6*(10). https://doi.org/10.1002/hsr2.1624
22. Hussein, S. Z. (2022). Evaluation of thyroid hormones and ferritin level in patients with β- thalassemia. *Medicine and Pharmacy Reports*, *95*(2), 152-157. https://doi.org/10.15386/mpr-2053
23. Bizri, M., Koleilat, R., Akiki, N., Dergham, R., Mihailescu, A. M., Bou-Fakhredin, R., Musallam, K. M., & Taher, A. T. (2024). Quality of life, mood disorders, and cognitive impairment in adults with β-thalassemia. *Blood Reviews*, *65*, 101181. https://doi.org/10.1016/j.blre.2024.101181
24. Jewad, A. M., & Shwayel, A. J. (2024). Evaluation of some nonroutine cardiac biomarkers among adults and children with beta-thalassemia major. *Laboratory Medicine*, *55*(5), 559-565. https://doi.org/10.1093/labmed/lmae007
25. Khaliq, S. (2022). Thalassemia in Pakistan. *Hemoglobin*, *46*(1), 12–14. https://doi.org/10.1080/03630269.2022.2059670
26. Ansari, A. H., Ansari, S. H., Salman, M. J., Ansari, M. U., & Jabeen, R. (2024). A scoping review on the obstacles faced by beta thalassemia major patients in pakistan- Matter of policy investment. *AIMS Public Health*, *11*(4), 1105-1124. https://doi.org/10.3934/publichealth.2024057
27. Ali, S., Mumtaz, S., Shakir, H. A., Tahir, H. M., & Mughal, T. A. (2019). Current perspective of beta thalassemia and its treatment strategies. https://doi.org/10.20944/preprints201907.0277.v1
28. Ansari, A. H., Ansari, S. H., Salman, M. J., Ansari, M. U., & Jabeen, R. (2024). A scoping review on the obstacles faced by beta thalassemia major patients in pakistan- Matter of policy investment. *AIMS Public Health*, *11*(4), 1105-1124. https://doi.org/10.3934/publichealth.2024057
29. Tantiworawit, A., Kamolsripat, T., Piriyakhuntorn, P., Rattanathammethee, T., Hantrakool, S., Chai-Adisaksopha, C., Rattarittamrong, E., Norasetthada, L., Fanhchaksai, K., & Charoenkwan, P. (2024). Survival and causes of death in patients with Alpha and beta-thalassemia in northern Thailand. *Annals of Medicine*, *56*(1). https://doi.org/10.1080/07853890.2024.2338246
30. Hussain, S., Ashfaq, M., Nisa, B., & Ally, K. (2025). Frequency of cardiac complications in beta-thalassemia patients at National Institute of child health, Karachi, Pakistan. *The Professional Medical Journal*, *32*(03), 309-314. https://doi.org/10.29309/tpmj/2025.32.03.8811
31. Batool, N., Saleem, Z., Saeed, H., Yasmeen, S., Anwar, R., Ahmad, F., Tauqeer, F., & Mahboob, M. (2022). Factors affecting health-related quality of life (HRQoL) in Pakistani children with thalassemia. *Family Medicine & Primary Care Review*, *24*(1), 37-42. https://doi.org/10.5114/fmpcr.2022.113012
32. Musallam, K. M., Rivella, S., Vichinsky, E., & Rachmilewitz, E. A. (2013). Non-transfusion-dependent thalassemias. *Haematologica*, *98*(6), 833-844. https://doi.org/10.3324/haematol.2012.066845
33. Borgna-Pignatti, C., & Gamberini, M. R. (2011). Complications of thalassemia major and their treatment. *Expert Review of Hematology*, *4*(3), 353-366. https://doi.org/10.1586/ehm.11.29
34. Cappellini, M. D., Farmakis, D., Porter, J., & Taher, A. (2021). Guidelines for the Management of Transfusion Dependent Thalassaemia (TDT) Thalassaemia International Federation. *Strovolos, Cyprus*.
35. Modell, B., & Darlison, M. (2008). Global epidemiology of haemoglobin disorders and derived service indicators. *Bulletin of the World Health Organization*, *2008*(6), 480–487. https://doi.org/10.2471/blt.06.036673
36. VICHINSKY, E. P. (2005). Changing patterns of thalassemia worldwide. *Annals of the New York Academy of Sciences*, *1054*(1), 18-24. https://doi.org/10.1196/annals.1345.003
37. Cohen, A. R., Galanello, R., Pennell, D. J., Cunningham, M. J., & Vichinsky, E. (2004). Thalassemia. *Hematology*, *2004*(1), 14-34. https://doi.org/10.1182/asheducation-2004.1.14
38. Tan, J. Y., Yeo, Y. H., Chan, K. H., Shaaban, H., & Guron, G. (2024). Causes of death and mortality trends in individuals with thalassemia in the United States, 1999-2020. *Journal of Blood Medicine*, *15*, 331-339. https://doi.org/10.2147/jbm.s470177
39. Dhanya, R., Agarwal, R. K., Sedai, A., Kumari, A., Parmar, L., Hegde, S., ., G., Gowda, A., Gujjal, P., H, P., Ramaiah, J. D., Karri, C., Jali, S., Tallur, N., Shenoy, U. V., Pinto, D., Ramprakash, S., Raghuram, C. P., Trivedi, D., ... Faulkner, L. (2019). Assessment of mortality and its associated risk factors in patients with transfusion dependent thalassemia in India. *Blood*, *134*(Supplement_1), 973-973. https://doi.org/10.1182/blood-2019-125151

40. Datta, S. S., & Sinha, A. (2024). Current challenges of blood transfusions in patients with thalassemia in India and future perspectives. *Transfusion Clinique et Biologique*, *31*(3), 162-166. https://doi.org/10.1016/j.tracli.2024.03.004
41. Yasmeen, H., & Hasnain, S. (2019). Epidemiology and risk factors of transfusion transmitted infections in thalassemia major: A multicenter study in Pakistan. *Hematology, Transfusion and Cell Therapy*, *41*(4), 316-323. https://doi.org/10.1016/j.htct.2019.03.008
42. Ishfaq, K., Shabbir, M., Naeem, S. B., & Hussain, S. (2015). Impact of thalassemia major on patients. *The Professional Medical Journal*, *22*(05), 582-589. https://doi.org/10.29309/tpmj/2015.22.05.1271
43. Nawaz, K., Khan, S. N., Bashir, A., Rehman, A., Tariq Masood Khan, M., Mir, A., & Ahmad, S. (2024). Unraveling impact of hemoglobin F and A2 levels: Correlation with disease severity and treatment response in transfusion-dependent beta-thalassemia. *Cureus*. https://doi.org/10.7759/cureus.52002
44. Sadiq, I. Z., Abubakar, F. S., Usman, H. S., Abdullahi, A. D., Ibrahim, B., Kastayal, B. S., Ibrahim, M., & Hassan, H. A. (2024). Thalassemia: Pathophysiology, diagnosis, and advances in treatment. *Thalassemia Reports*, *14*(4), 81-102. https://doi.org/10.3390/thalassrep14040010
45. Khan, S. U., Khan, M. U., Khan, R., Iqbal, M., Khan, S., & Ullah, J. (2024). A survey of acute transfusion reactions in thalassemic patients in Pakistan: A single centre experience. *Journal of Islamabad Medical & Dental College*, *13*(3), 458-463. https://doi.org/10.35787/jimdc.v13i3.1044
46. Khan, H., Ghafoor, T., Shahbaz, N., Khattak, T. A., Rahim, U., & Awan, M. N. (2025). Outcome of Haematopoietic Stem Cell Transplant in Beta-Thalassemia Major: Single Centre Experience from a Low-and Middle-Income Country. *Journal of the College of Physicians and Surgeons--Pakistan: JCPSP, 35*(4), 513-518. https://doi.org/10.29271/jcpsp.2025.04.513
47. Etemad, K., Mohseni, P., Aghighi, M., Bahadorimonfared, A., Hantooshzadeh, R., Taherpour, N., Piri, N., Sotoodeh Ghorbani, S., Malek, F., Kheiry, F., Khodami, A., Valadbeigi, T., & Hajipour, M. (2021). Quality of life and related factors in β-thalassemia patients. *Hemoglobin*, *45*(4), 245-249. https://doi.org/10.1080/03630269.2021.1965617
48. Tan, J. Y., Yeo, Y. H., Chan, K. H., Shaaban, H., & Guron, G. (2024). Causes of death and mortality trends in individuals with thalassemia in the United States, 1999-2020. *Journal of Blood Medicine*, *15*, 331-339. https://doi.org/10.2147/jbm.s470177
49. Musallam, K. M., Vitrano, A., Meloni, A., Pollina, W. A., Karimi, M., El-Beshlawy, A., Hajipour, M., Di Marco, V., Ansari, S. H., Filosa, A., Ricchi, P., Ceci, A., Daar, S., Vlachaki, E., Singer, S. T., Naserullah, Z. A., Pepe, A., Scondotto, S., Dardanoni, G., … Maggio, A. (2021). Survival and causes of death in 2,033 patients with non-transfusion-dependent β-thalassemia. *Haematologica*, *106*(9), 2489-2492. https://doi.org/10.3324/haematol.2021.278684
50. Iam-arunthai, K., Suwanban, T., Thungthong, P., Chamnanchanunt, S., & Fucharoen, S. (2024). Predicting factors of survival rates among alpha- and beta-thalassemia patients: A retrospective 10-year data analysis. *Frontiers in Hematology*, *3*. https://doi.org/10.3389/frhem.2024.1339026
51. Maryam, M., Rahman, A., & Haider, S. (2024). Investigation of the relationship between thalassemia and depression to predict a base for rehabilitation measures. *Journal of Disability Research*, *3*(5). https://doi.org/10.57197/jdr-2024-0045
52. Khan, M. A., Fazlul Aziz Mian, Subaat Basit, Bilqees, Laiba Sarfraz, & Shahjahan Wahid. (2025). Structural and hemodynamic changes on echocardiography in beta-thalassemia major patients. *Insights-Journal of Health and Rehabilitation*, *3*(3 (Health & Allied)), 222-230. https://doi.org/10.71000/3ts9cm74
53. Kattamis, A., Kwiatkowski, J. L., & Aydinok, Y. (2022). Thalassaemia. *The Lancet*, *399*(10343), 2310-2324. https://doi.org/10.1016/s0140-6736(22)00536-0
54. Lorey, F., Charoenkwan, P., Witkowska, H. E., Lafferty, J., Patterson, M., Eng, B., Waye, J. S., Finklestein, J. Z., & Chui, D. H. (2001). HB H hydrops foetalis syndrome: A case report and review of literature. *British Journal of Haematology*, *115*(1), 72-78. https://doi.org/10.1046/j.1365-2141.2001.03080.x
55. Lorey, F., Cunningham, G., Vichinsky, E. P., Lubin, B. H., Witkowska, H. E., Matsunaga, A., Azimi, M., Sherwin, J., Eastman, J., Farina, F., Waye, J. S., & Chui, D. H. (2001). Universal newborn screening for HB H disease in California. *Genetic Testing*, *5*(2), 93-100. https://doi.org/10.1089/109065701753145538
56. Weatherall, D. J., & Clegg, J. B. (2001). Inherited haemoglobin disorders: an increasing global health problem. *Bulletin of the World Health Organization*, *79*(8), 704-712. https://www.scielosp.org/pdf/bwho/v79n8/v79n8a05.pdf
57. Vichinsky, E. P. (2000). Report of proceedings: 1999 International Conference on E-β thalassemia. *Journal of Pediatric Hematology/Oncology*, *22*(6), 550. https://doi.org/10.1097/00043426-200011000-00020
58. Lorey, F. (2000). Asian immigration and public health in California: Thalassemia in newborns in California. *Journal of Pediatric Hematology/Oncology*, *22*(6), 564-566. https://doi.org/10.1097/00043426-200011000-00024
59. Musallam, K. M., Cappellini, M. D., Wood, J. C., Motta, I., Graziadei, G., Tamim, H., & Taher, A. T. (2011). Elevated liver iron concentration is a marker of increased morbidity in patients with thalassemia intermedia. *Haematologica*, *96*(11), 1605-1612. https://doi.org/10.3324/haematol.2011.047852
60. Vogiatzi, M. G., Macklin, E. A., Fung, E. B., Cheung, A. M., Vichinsky, E., Olivieri, N., Kirby, M., Kwiatkowski, J. L., Cunningham, M., Holm, I. A., Lane, J., Schneider, R., Fleisher, M., Grady, R. W., Peterson, C. C., & Giardina, P. J. (2009). Bone disease in thalassemia: A frequent and still unresolved problem. *Journal of Bone and Mineral Research*, *24*(3), 543-557. https://doi.org/10.1359/jbmr.080505
61. Musallam, K. M., Sankaran, V. G., Cappellini, M. D., Duca, L., Nathan, D. G., & Taher, A. T. (2012). Fetal hemoglobin levels and morbidity in untransfused patients with β-thalassemia intermedia. *Blood*, *119*(2), 364-367. https://doi.org/10.1182/blood-2011-09-382408
62. Limpawattana, P., Juntararuangtong, T., Teawtrakul, N., Wanitpongpun, C., Lanamtieng, T., Phiphitaporn, P., & Chansung, K. (2022). Cognitive impairment in thalassemia and associated factors. *Dementia and Geriatric Cognitive Disorders*, *51*(2), 128-134. https://doi.org/10.1159/000522655
63. Efat, A., Wahb, R., Shoeib, S. A., Dawod, A. A., Abd ElHafez, M. A., Abd ElMohsen, E. A., & Elkholy, A. (2022). GDF-15 is associated with atherosclerosis in adults with transfusion-dependent beta-thalassemia. *eJHaem*, *3*(2), 353-361. https://doi.org/10.1002/jha2.415
64. Shamaoon, M., Nawaz, J., & Ahsan, M. (2018). β-THALASSEMIA MAJOR. *The Professional Medical Journal*, *25*(06), 823-827. https://doi.org/10.29309/tpmj/2018.25.06.265

65. Aslam, M. S., Roshan, E., Iqbal, A., & Shahid, M. (2013). FREQUENCY OF SHORT STATURE IN (Beta)-THALASSEMIA MAJOR PATIENTS. *Pakistan Armed Forces Medical Journal*, (4), 43.
66. Ahmed Kiani, R., Anwar, M., Waheed, U., Asad, M. J., Abbasi, S., & Abbas Zaheer, H. (2016). Epidemiology of transfusion transmitted infection among patients with*β*-thalassaemia major in Pakistan. *Journal of Blood Transfusion*, *2016*, 1-5. https://doi.org/10.1155/2016/8135649
67. Mittal, K., Abrol, P., & Yadav, J. (2016). Prevalence of transfusion transmitted infections amongst multiple blood transfused patients of β-thalassemia major in a tertiary care hospital. *International Journal of Research in Medical Sciences*, *5*(1), 181. https://doi.org/10.18203/2320-6012.ijrms20164545
68. Salama, K., Khaled, H. Z., El Dien, H. M., Afifi, R. A., Shaheen, N. M., & El Wahab, M. A. (2022). Assessment of cardiac functions and arrhythmia in children with beta-thalassemia major and beta-thalassemia Intermedia. *Open Access Macedonian Journal of Medical Sciences*, *10*(B), 890-895. https://doi.org/10.3889/oamjms.2022.9026
69. Jafri, L., Jameel Farooqui, A., Moiz, B., Sheikh, A., Majid, H., Nadeem, S., Quddus, R., Khan, S., Khan, Q., & Habib Khan, A. (2025). Factors associated with phosphate homeostasis in children with beta-thalassemia major: An analytical cross sectional study from Pakistan. *PLOS ONE*, *20*(2), e0316566. https://doi.org/10.1371/journal.pone.0316566
70. Zareef, A., Ansar, F., Khan, A. A., Khan, M. T. M., & Shah, F. (2024). Assessment of growth retardation among transfusion-dependent Thalassemia patients in Peshawar, Pakistan. *Khyber Medical University Journal*, *16*(3), 207-12.

Questionnaire Paper

Investigate the Mortality, morbidity and factors associate on Thalassemia patients Faisalabad 2025 Pakistan

Section 1: Demographic Information

1. **Age: ______**
2. **Gender:**
   - ○ Male
   - ○ Female
   - ○ Other
3. **What is your marital status?**
   - Married
   - Unmarried
   - Widow

Section 2: Disease History and Morbidity

1. **What type of thalassemia do you have?**
   a. Alpha thalassemia
   b. Beta thalassemia minor
   c. Beta thalassemia major
   d. Other (please specify): _____________
2. **At what age were you diagnosed with thalassemia?**
   ______
3. **How frequently do you require blood transfusions?**
   a. Weekly
   b. Monthly
   c. Every 2-3 months
   d. Never

4. **Have you experienced any complications due to thalassemia?**
   - Iron overload
   - Heart problems
   - Liver problems
   - Diabetes
   - Osteoporosis (bone thinning)
   - Other (please specify): _____________

4. **Do you take any medications or treatments for thalassemia?**
   - Blood transfusions
   - Iron chelation therapy
   - Bone marrow transplant
5. **Do you have a family history of thalassemia?**
   - Yes
   - No
6. **How would you rate your quality of life due to thalassemia?**
   1. Poor
   2. Fair
   3. Good

Section 3: Mortality and Life Expectancy Perception

➢ **Do you know someone in your family or community who has passed away due to thalassemia?**
   1. Yes
   2. No

➢ **Are you aware of genetic counseling options available for thalassemia carriers**
   1. Yes
   2. No