

Frequency Of Beta Thalassemia Major Abo Blood Groups In Different Foundation Of District Peshawar

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ABSTRACT

Background: Thalassemia major (TM) is an inherited hemolytic disorder occur due to abnormal hemoglobin production as a result of HBB (Haemoglobin Subunit Beta) gene mutation because HBB gene provides instructions for making of beta globin. The research study aim is to determine the frequency of beta thalassemia major ABO blood group in different foundation of district Peshawar.

Methods: A descriptive cross-sectional study was conducted, involving 100 individuals with Thalassemia major from different foundations in Peshawar, including Hamza Foundation and Fatmid Foundation. The data was analyzed for statistical analysis using SPSS version 22.0 software.

Results: The study analyzed the gender-wise distribution of Thalassemia major patients, revealing that out of the 100 individuals, 43 were female (43%) and 57 were male (57%). Moreover, the study examined the ABO blood types among the participants. The most prevalent blood type among the Thalassemia major patients was B+ (35.0%), followed by O+ (34.0%), A+ (26.0%), AB+ (4.0%), and O- (1.0%).

Conclusion: Based on the results, it can be said that among those with Thalassemia major in the population under study, the B+ blood type is the most prevalent and well-known ABO blood group.

Keywords: Thalassemia major, congenital hemolytic disorder, ABO blood group, Haemoglobin Subunit Beta

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INTRODUCTION

Beta thalassemia major (TM) is a congenital haemolytic disorder characterized by an abnormal haemoglobin production due mutations in haemoglobin subunit beta (HBB).¹ In beta thalassemia major a type of point mutation in the HBB gene located on chromosome 11.² People can inherit the thalassemia trait or disease from their parents through the genes.³ Thalassemia major ABO blood group identification is based on the presence or absence of specific antigens on the surface of red blood cells.⁴ Individuals with beta thalassemia cannot make enough beta globin chains, causing an excess number of alpha chains.⁵ ABO blood groups refer to the classification system of human blood based on the inherited properties of red blood cells.⁶ The ABO blood system is used to determine blood compatibility for transfusions.⁷

According to classification there are two main types of thalassemia that is "Alpha-thalassemia" and "Beta-thalassemia".⁸ Alpha-thalassemia is defined as a deletion of one or both alpha genes.⁹ Alpha thalassemia is classified on basis of number of alpha globin genes that are affected.¹⁰ Alpha thalassemia Silent carrier is the mildest form of alpha thalassemia, where deletion occur in one out of four alpha globin genes.¹¹ Alpha thalassemia trait deletions occur in two of the four alpha globin genes.¹² Hemoglobin H disease is a rare type in which three of the four alpha globin genes deletion occur.¹³ HbH disease include mild jaundice and hepatosplenomegaly.¹⁴ Alpha

thalassemia major is most severe form where complete deletion of all four alpha globin genes occur.¹⁵

Beta thalassemia is defined as an inherited blood disorder caused by mutation in beta chains of hemoglobin occurs.¹⁶ The disease severity depends on mutation of HBB gene responsible of instruction for making beta globin subunit of hemoglobin.¹⁷ (Tasnim *et al.*, 2019). Beta thalassemia also causing a mutation at binding site of SOX6 to γ -globin, thus increasing γ -globin expression.¹⁸ Each individual with thalassemia major carried an abnormal beta globin protein.¹⁹ With passage of time HBB blockage leads to decrease beta-chain synthesis and body failed to build up new beta-chains leads to under production of adult hemoglobin.²⁰ Every year 60,000 infants are born with beta thalassemia major.⁵ Beta thalassemia has been divided into three main forms as beta thalassemia major, beta thalassemia intermedia and beta thalassemia minor.⁵

Beta thalassemia major also known as Cooley's anemia.²¹ Individuals with this type inherits two mutated HBB genes, one from each parent produce little or no beta-globin protein.²² Beta thalassemia intermedia is a milder form and individuals with this type have milder anemia that does not require regular transfusion.¹ Beta thalassemia minor occurs when a person inherits one mutated HBB gene from one parent and one normal HBB gene from the other parent.²³ Beta thalassemia major occurs when individual inherits both mutated HBB genes

frequently displaying severe anemia.²⁴ Beta thalassemia major results in a severe shortage of hemoglobin and causes severe problems including liver damage, heart disease, endocrine dysfunction and cardiovascular disease.²⁵ Most infants with thalassemia major stillborn hydrophs fetalis.²⁶ Beta thalassemia a most common inherited genetic blood disorder affecting millions both in developed and under developed countries.^{27, 28}

According to frequency distribution the most prevalent ABO Blood group of thalassemia major is blood group O.⁴ Number of studies suggests the correlation of ABO blood groups with various diseases.²⁹ However, the association of ABO blood groups with thalassemia has largely been under studied subject.³⁰ Blood group is an important and comparatively known parameter today which exhibits a correlation with some common disease of thalassemia.³¹ A genetic blood disease with a carrier prevalence of 5-8%, beta-thalassemia major is thought to affect 5,000 children in Pakistan each year.³² Beta thalassemia major is the problem of concern for public health in Pakistan.³³ The Beta thalassemia major diagnosis is mainly by typical blood film and presence of more than 20% fetal hemoglobin.³⁴

Modern treatment for thalassemia includes correcting anemia by taking Reblozyl 25mg/vial medicine (FDA approved) available in injection form obtained with prescription under supervision of experienced hematologist.³⁵ By inhibiting the Smad2/3 signaling pathway, which slows down red blood cell development, luspatercept, the active ingredient in reblozyl, controls the maturation of red blood cells.³⁶ The second method is gene therapy, which involves taking patient blood stem cells and genetically modifying them outside of the body to prevent the expression of the BCL11A gene, which typically stops the fetal hemoglobin synthesis soon after birth.³⁷ Thirdly uses of Iron chelating agents that include medication Deferoxamine 500mg/vial also known as desferrioxamine available in injection form,³⁸ Asunra 400mg tablets is also used to treat an excessive accumulation of iron in body due to multiple blood transfusion³⁹ and is used for adults and children above 2 years of age, Kelfer-250

capsule is a medication used to treat chronic iron overload caused due to frequent blood transfusion.⁴⁰ It remove excess iron and also reduce the risk of organ damage. The study purpose was to find out ABO blood groups frequencies of beta thalassemia major patients registered at Thalassemia Care centre, Peshawar.

MATERIALS AND METHOD

A descriptive cross-sectional study was conducted in Fatimid foundation and Hamza foundation of district Peshawar. Duration of the study was 4 months from February to June 2023. A total of 100 samples were collected for this study. A primary data were collected through Sample convenience method. All those individuals who were diagnosed for Beta thalassemia major are included in this study. Individuals diagnosed with any other types of thalassemia (e.g., alpha-thalassemia, Beta thalassemia (minor, intermedia) are excluded from this study.

A 3ml of venous blood was drawn using disposable syringe and placed in EDTA tubes used for CBC and performing blood group. The collected blood were putted on sterile glass slides into three separate drops, the first drop were mixed with antisera A and the second drop were mixed with antisera B and the third drop were mixed with antisera D. The obtained results were recorded, and similar procedure was applied to all collected samples.

The data was analyzed for statistical analysis using SPSS version 22.0 software.

RESULTS

Out of 100 individual participating in this study representing that 57 were male and 43 were female and individual from 1-10 years were mostly affected showing percentage of about 86.0%, while those from 11-20 years showing percentage of about 13.0% and similarly those from 21-30 years showing percentage of about 1.0% (Table 1).

Among 100 individuals with thalassemia major, the most common ABO blood type was B+ (35.0%), followed by O+ (34.0%), A+ (26.0%), AB+ (4.0%) and O-(1.0%). This distribution was different from the general population in Pakistan, where blood type B is the most commonly affected, followed by O, A, and AB (Table 1).

Table 1: Gender, age-wise, and ABO blood group frequencies of Thalassemia patients

Gender wise distribution of thalassemia patients		Age wise distribution of thalassemia patients	Blood groups of thalassemia patients					Total percentage
Gender	Frequency Percentage		A Positive	B Positive	AB Positive	O Positive	O Negative	
Male	57 (57.0%)	1-10 Years	21	28	4	32	1	86.0
Female	43 (43%)	11-20 Years	5	6	0	2	0	13.0
Total	100 (100%)	21-30 Years	0	1	0	0	0	1.0
Total Sum			26	35	4	34	1	100.0

Table 2: Distribution of Thalassemia major patients according to ABO blood group

Blood groups	Frequency (n)	Percent
A Positive	26	26.0
B Positive	35	35.0
AB Positive	4	4.0
O Positive	34	34.0
O Negative	1	1.0
Total	100	100.0

DISCUSSION

A hereditary blood illness known as beta thalassemia major is characterized by aberrant synthesis of hemoglobin,⁴¹ which results in severe anemia, exhaustion, feeding difficulties, weakness, jaundice, irritability, stunted development, enlarged belly, deformed facial bones, and failure to thrive. Thalassemia major manifests clinically between the ages of 6 and 24 months. It is most frequently noted that consanguineous couples have higher rates of thalassemia.⁴²

There is an increased likelihood of producing children with thalassemia major with each pregnancy when both parents carry the gene.⁴³ Babies with beta thalassemia major exhibit aberrant linear development, variable degrees of muscular atrophy, and malnutrition, which impairs growth.⁴⁴

Additionally this study reveals that frequency of thalassemia major among individuals with aged groups between 1-10 years (86.0%), followed by the age groups 11-20 (13.0%) and 21-30 (1.0%). By comparing with other studies conducted in Pakistan suggest that thalassemia major is mostly occurs in children because the life expectancy of thalassemia major is about 1-10 years.⁴⁵

According to our study among the thalassemia major patients the most commonly affected ABO blood group was B+ (35.0%), followed by O+ (34.0%), A+ (26.0%), AB+ (4.0%), and O- (1.0%). This observation agreed with another study published by Ayub Medical College in Pakistan, which found similar trends in ABO blood type frequencies among thalassemia major patients.⁴⁶ This consistency strengthens the reliability of the findings and suggests that B+ blood type individuals are more susceptible to thalassemia major in different foundation of District Peshawar.

CONCLUSION

This study concludes that B+ blood type as the most commonly affected among thalassemia major patients in District Peshawar. This study also concludes that O- blood type as the less commonly affected among thalassemia major patients in District Peshawar. By comparing to all other types of thalassemia individual with thalassemia major survived for a long time with regular transfusion of packed RBCs. Individuals with beta thalassemia major have not been regularly transfused usually die before the second-third decade. The study found that

among thalassemia majors, those between the ages of 1 and 10 were the most often afflicted.

To educate the patients and their families about awareness and how to cope with thalassemia conditions. Also recommend prenatal testing to prevent passing thalassemia to their offspring, beta thalassemia screening for at risk couples, informed decisions about pregnancy, identification of carriers in the family. Growth and development often return to normal once 9.5 to 10.5 g/dL of Hb is maintained with frequent transfusion treatment for a duration of 10 to 12 years. Correction of anemia, suppression of erythropoiesis, and prevention of gastrointestinal iron absorption, which happens in patients who are not transfused—are the objectives of transfusion treatment. The relationship between blood groups A, B, and O and thalassemia major has to be investigated further.

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