



Research Article

# Knowledge, Attitudes, and Perceptions Towards Beta-Thalassemia among Residents of Azad Kashmir, Pakistan

Farheen Shafique<sup>1</sup>, Shaukat Ali<sup>2\*</sup>, Mahreen ul Hassan<sup>3</sup>, Saiqa Andleeb<sup>1</sup>

<sup>1</sup>Department of Zoology University of Azad Jammu and Kashmir Muzaffarabad, Muzaffarabad, Azad Kashmir, Pakistan

<sup>2</sup>Department of Zoology, Government College University Lahore, Lahore, Punjab, Pakistan

<sup>3</sup>School of Bioscience, University of Sheffield, Sheffield, South Yorkshire, United Kingdom

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## Authors' Contributions

FS conducted the research, analysed data and wrote the manuscript. S Ali supervised the research and helped in data validation. MH conceptualized the methodology and helped in data curation. S Andleeb co-supervised the research. S Ali and S Andleeb reviewed and edited the manuscript.

## Keywords

Thalassemia, Knowledge, Genetic disorders, Blood disorders, Pakistan



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**Abstract** |  $\beta$ -Thalassemia is the most common genetic disorder of the time. In Pakistan, around 1–4 per 1000 infants is diagnosed with the disease making it one of the high-risk countries in the world. This study was aimed to quantify the public's awareness, attitude, perception and prevention among the population of Azad Jammu and Kashmir. A descriptive cross-sectional was conducted from Mar-Sep 2021. Participants were selected through random sampling and questioned face to face using a predesigned, pretested questionnaire. The first portion of the questionnaire enquired about basic demographic data; the second half evaluated the participant's knowledge about thalassemia. The third and fourth sections dealt with people's perceptions and attitudes regarding thalassemia. The Statistical Package for Social Sciences (SPSS) Statistics was used to analyse the data. Approximately 75% of the participants (n = 1677) had heard of thalassemia. Around 48% of the people interviewed were unaware that one might be a thalassemia carrier. Only 5.3% of the participants strongly agreed that necessary tests should be run before and after birth to screen a  $\beta$ -thalassemia minor/carrier baby. Thalassemia carriers should not get married and 6.2% thought that premarital screening for the disease was important. Around 69.47% of the respondents strongly agreed that cousin marriages are not a reason behind  $\beta$ -thalassemia in the family. Despite the highest literacy rate, large number of young population and prosperity in the region, awareness about genetic disorders is not sufficient.

**Novelty Statement** | This manuscript presents the first knowledge assessment study on  $\beta$ -thalassemia in Azad Jammu and Kashmir, Pakistan. Our research contributes to the understanding of  $\beta$ -thalassemia in this region and highlights areas where educational interventions may be necessary. This study fills an important gap in knowledge and provides a foundation for future research on  $\beta$ -thalassemia in Azad Jammu and Kashmir.

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## Introduction

Inherited disorders of hemolytic anemia are known as Thalassemia which is the most common genetic disorder of the time It can manifest in various symptoms such as

fatigue, poor appetite, abdominal pain, and bone deformities. Patients with  $\beta$ -thalassemia major require lifelong dependence on blood transfusions. Due to the excess iron in the body from multiple transfusions, individuals with this condition are at risk for iron overload, which can increase the production of free radicals and cause oxidative stress. This can lead to damage and dysfunction in several organs, including the liver, heart, and various glands, such as the parathyroid and thyroid glands (Porter

**Corresponding Author: Shaukat Ali**

dr.shaukatali@gcu.edu.pk

and Viprakasit, 2014; Sevindik, 2018; Sevindik *et al.*, 2018; Sevindik, 2019). Besides these abnormalities, individuals with  $\beta$ -thalassemia major may also experience a variety of bone disorders, including spinal and bone deformations, delayed growth, and osteopenia or osteoporosis (Ali *et al.*, 2019).

With an estimated 5% of the world's population carrying the thalassemia gene, it is one of the most prevalent genetic disorders. Thalassemia is prevalent in regions like the Mediterranean, Middle East, Africa, and Southeast Asia (Morris, 2010).

Thalassemia can be classified into alpha and beta thalassemia (Bernshtein, 2022). Mutations in any of the alpha-globin genes result in an insufficient amount of alpha-globin chains and, hence, to the disease known as alpha-thalassemia. Depending on how many genes are affected, the condition may vary in severity. There are four forms of alpha-thalassemia i.e., silent carrier in which case individuals have one mutated alpha-globin gene and three normal genes. They do not have any symptoms and are unaware of their condition unless diagnosed through genetic testing. The second form is the Alpha-thalassemia trait; individuals with this type of thalassemia have two mutated alpha-globin genes and two normal genes. They may experience mild anemia, but the symptoms are generally not severe. The third type is hemoglobin H disease; patients with this type of thalassemia have three mutated alpha-globin genes and one normal gene. They experience moderate to severe anemia, as well as other symptoms such as jaundice, enlarged spleen, and bone deformities (Piel and Weatherall, 2014). Hydrops fetalis, the most severe kind of alpha-thalassemia, is the fourth type. It develops when all 4 alpha-globin genes are altered, making it impossible for the developing foetus to manufacture any alpha-globin chains. Before or soon after delivery, the condition frequently leads to death (Galanello and Cao, 2011).

Mutations in either of the two genes responsible for making beta-globins lead to beta-thalassemia, a blood disorder characterised by abnormally low or non-existent levels of the beta-globin chain (Galanello and Origa, 2010). There are three forms of beta-thalassemia which includes Beta-thalassemia minor in which affected individuals have one mutated beta-globin gene and one normal gene. They may experience mild anemia, but the symptoms are generally not severe. The other type is beta-thalassemia Intermedia; individuals with this type of thalassemia have mutations in both beta-globin genes, but the mutations are less severe than those in beta-thalassemia major. They may experience moderate to severe anemia, as well as other symptoms such as jaundice, enlarged spleen, and bone deformities. In case of beta-thalassemia major type Individuals have mutations in both beta-globin genes

and require lifelong blood transfusions. They experience severe anemia, as well as other symptoms such as jaundice, enlarged spleen, bone deformities, and an increased risk of infections (Karimi *et al.*, 2015).

Beta thalassemia usually requires frequent blood transfusions (Shafique *et al.*, 2023). The prevalence of beta-thalassemia is high in developing countries. In Pakistan, approximately 1-4 per 1000 infants are diagnosed with the disease. This makes it one of the high-risk countries in the world (Maheen *et al.*, 2015). An estimated 9.8 million people in Pakistan are carriers of beta-thalassemia. Currently, there are roughly 50,000 patients with thalassemia enrolled in treatment clinics throughout the country. Other than random mutations, over population, and traditional customs of consanguineous marriages also contribute to this (Ehsan *et al.*, 2020).

Current study was designed in the light of present scenario of beta-thalassemia in Pakistan particularly in the region of Azad Jammu and Kashmir reported by Shafique *et al.* (2020). It was revealed during the study that several families residing in the area are suffering from thalassemia one way or the other due to caste system and inter marriages despite occurrence of many genetic disorders reported in the region (Ahmed *et al.*, 2019; Ain-ul-Batool *et al.*, 2019; Ali *et al.*, 2019, 2020, 2021; Shafique *et al.*, 2020; Tariq *et al.*, 2019; Umair *et al.*, 2019). Therefore, it was important to determine the exact magnitude and determine the perception of people towards this disease for its timely prevention and eradication from the region.

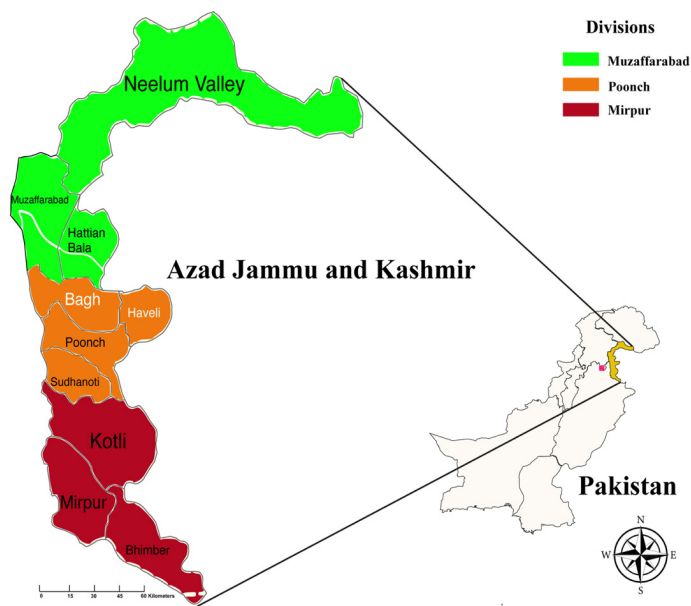
This study was aimed at quantifying the awareness, attitude, and perception of the public about thalassemia and its prevention among the population of Azad Jammu and Kashmir.

## Materials and Methods

### *Study design, location, participants, and sampling*

The study was carried out in Azad Jammu and Kashmir (13,297 sq. Km) which is situated in the northwest of Pakistan and shares its border with the Khyber Pakhtunkhwa and Punjab provinces in the north and south, respectively (Figure 1). The total population of the area is around 4.361 million according to the official portal of the Azad Kashmir government (GOVAJK, 2021). The main communities living in this region are Gujjars, Sudhans, Jutts, Rajputs, Mughals, Abbasids, Dhunds, Sadaats, and Kashmiris (Snedden, 2013). According to the 2019 census, the literacy rate in Azad Kashmir was 79.80% (Dr Parvez, 2022; GoAJK, 2021). There is a very strong family system present in the area. In Pakistani society, it is normal for two people to marry each other due to their blood ties. It is noticeably more frequent in Kashmiri culture (Jaffar *et al.*, 2021). A key risk factor is intermarriage in the context of

high gene frequency and a high birth rate, which are both present in a large population size (Madiani *et al.*, 2019).



**Figure 1: Map of the study is (Azad Jammu and Kashmir) and its major divisions.**

A descriptive, cross-sectional study was conducted during 6 months from March to September 2021 in all three divisions of Azad Jammu and Kashmir (AJK), Pakistan (Figure 1). The STROBE guidelines for survey-based research were followed for this study (Cevallos and Egger, 2014). The study scheme was designed in such a way that the entire investigation could be divided into three phases. In phase 1, a sample population was selected from each division of Azad Kashmir. A questionnaire was designed, and a pilot study was conducted to test the questionnaire then an estimated timeline was set to reach the maximum number of respondents in the area. In the second phase, interviews were conducted through representatives in each division and exclusion criteria were applied. In the last phase all the data was gathered and analysed. To assess the knowledge, various statistical tools were applied. The research was carried out in the area due to the increased number of thalassemia cases reported over the years. The survey was conducted in all divisions, viz. Muzaffarabad, Poonch, and Mirpur. A pre-tested questionnaire was distributed to people in all public places including parks, hospitals, shopping centres and academic institutions. in different cities of Azad Kashmir. The places were chosen because of the presence of a diverse population in the area where people could easily be reached and interviewed.

The data collectors stayed at each site for an average of 2 hours at a time that was randomly chosen. Participants were approached at random and encouraged to participate in the survey during a 2-hour period. Participants with reading or writing difficulties were assisted by an interviewer to collect data.

### *Sample size calculation and eligibility criteria*

The sample size was computed using the Raosoft application (Raosoft, 2016). From each division a sample size of 384 individuals was generated. This research was open to anyone aged 18 and above who was clinically stable and didn't have any mantle disabilities. Participants were eliminated if they were unwilling to engage in the study or if they did not live in the designated area.

### *Design and validation of the questionnaire*

The questionnaire was developed after a thorough literature review. It was written in English, then translated into Urdu for participants, and later translated back to English for analysis. The questionnaire was created under the guidance of two physicians, an expert in the field of public health and communicable diseases, a pharmacist, and a sociologist. A pilot study was conducted on 25 random individuals. By considering other published literature, the modifications suggested by the participants in the questionnaire was adopted prior to distribution among test subjects. The responses obtained from the participants of the pilot study were excluded from the final data. To determine reliability, Cronbach's alpha test was applied. The questionnaire was found to have a reliability coefficient of 0.72.

The questionnaire was divided into three sections with 45 questions. The first part consisted of 10 questions that evaluated demographic data such as age, gender, and ethnicity, income, and literacy, etc. The second section consisted of 14 questions to assess participants knowledge of thalassemia, as well as their ability to recognise its symptoms and possible risk factors. In Section three, participants were evaluated for their attitude towards the disease based on 9 attitude-related statements. In the final section, participants were observed for their perception towards Beta thalassemia by assessing their responses to 12 questions.

### *Data analysis*

All the information gathered through this study was analysed by Microsoft Excel 16.0, and GraphPad version 9.0.0. and SPSS version 27.1.0.

## **Results and Discussion**

The research was carried out in view of an alarming situation in the region where the prevalence of the beta thalassemia trait was recorded as 5.6% according to Ahmed *et al.* (2016), making it a high-risk population. A total of 1677 individuals from all 3 divisions viz Muzaffarabad, Poonch, and Mirpur of Azad Kashmir, Pakistan, participated in the study. Approximately 705 (42%) males and 972 (58%) females participated in this study. Demographic variables are shown in Table 1. Mean age of participants was recorded at  $30.52 \pm 12.4$ . The

population included females, belonging to low-income families (78%) undergraduates, unemployed (48.48%), and without a family history of thalassemia (86.2%).

**Table 1: Percentage of number of respondents for each variable.**

Variables	No. of respondents	Percentage (%)
Mean age $\pm$ SD (years)	30.52 (12.4)	--
Male	705	42.00
Female	752	45.00
Graduates	917	54.68
Family history of thalassemia	248	14.80
Housewives/non-employed	813	48.48
High income status	362	22.00

The research was carried out in all three divisions of Azad Kashmir, Pakistan. Around 586 (35%) people from Muzaffarabad division, 514 (31%) from Poonch division and 577 (34%) men and women took part in the study.

The participants were asked 13 different close-ended questions to assess the knowledge about beta thalassemia. The percentage of correct and incorrect answers regarding knowledge is listed in Table 2. An overall possible score was 0-14, with scores 9 or higher indicating sufficient knowledge and below 9 indicated poor knowledge.

Compared to earlier studies, our findings showed that 1250 (74.54%) people were familiar with the name of the disease, which is quite contrary to the other studies where 53% of the respondents heard the name; according to Balci *et al.* (2014), around 57.7% had previously heard of thalassemia. Around 473 (28.2%) people thought thalassemia was a contagious disease. Majority of

participants (47.3%) did not know that thalassemia can be inherited through genes which is opposite to a finding of a similar study conducted by Ebrahim *et al.* (2019). Surprisingly, 759 (45.3%) knew the disease could not be transmitted through blood transfusion. This is close to the outcome of a study conducted by Islam *et al.* (2021) where 32.1% responded the same.

Around 90% of the participants were aware that the parents of the carriers might have a child with thalassemia, while 1162 (69.3%) individuals didn't believe that a child could be born with the disease if only one of the parents is the carrier. These findings showed that people in the region have some knowledge if not adequate about the transmission of the disease. The results shown here are quite encouraging as compared to the study by Shukla *et al.* (2021), where only 14.33% of the respondents knew the correct answer. The other 39.7% believed that a carrier person is equally responsible for the disease in a family. These myths must be clarified, and knowledge about the different types of thalassemia must be shared to ensure that carriers of thalassemia do not suffer discrimination in society. Interestingly, 75% of the participants believed that the disease is caused by cousin marriages, which was a positive sign that people are aware of the risks of consanguinity in the society. The response percentage is higher than a finding from a previous study in which 41.2% people showed concern about intermarriages (Balci *et al.*, 2014). When asked about the health of thalassemia patients' majority 1024 (61.0%) responded incorrectly. It was revealed during study that 614 (36.6%) of participants didn't think that this disease could be prevented while 424 (25%) individuals were quite optimistic that the disease is completely curable as compared to the study of Islam *et al.* (2021) where 50.1% participants responded the same. Similarly, 801 (47.8%) didn't know that anyone could be a thalassemia carrier.

**Table 2: Knowledge of the participants about thalassemia.**

Question statements	Correct answer (%)	Incorrect answer (%)
Have you heard of the word $\beta$ -thalassemia?	74.54	25.46
Is thalassaemia a contagious disease?	71.8	28.2
Can you inherit thalassemia?	52.7	47.3
Do you think that thalassemia could be transmitted through blood transfusion from a person with thalassemia?	54.7	45.3
Do you think marriage between two carriers can lead to a child with thalassemia major?	90	10
Do you think if one parent is a carrier, the couple has a chance of having a child with thalassaemia Disease?	30.7	69.3
Do you think marriage between close relatives can increase the chance of thalassaemia?	75	25
Do you think that thalassemia carriers are as healthy as normal people?	39	61
Do you think thalassaemia is a preventable disease?	63.4	36.6
Do you think thalassemia is a completely curable disease?	74.7	25.3
Do you know thalassaemia is a blood disorder?	85.4	14.6
Do you know someone who could be a carrier of thalassaemia, including you?	52.2	47.8
Do you know thalassemia can be identified by a blood test?	84.4	15.6

**Table 3: The relationship between demographic variables and participants knowledge towards thalassemia.**

Demographic variables	Knowledge (%)		Odd ratios (95% CI)	p value
	Good knowledge	Poor knowledge		
<b>Age (years)</b>				
18-30	51.6	48.4	1.05 (0.941 to 1.18)	<.37
31-40	47.6	52.4		
>40	54.2	45.8		
<b>Gender</b>				
Male	49.0	51.0	0.84 (0.69, 1.0)	.078
Female	53.3	46.7		
<b>Education</b>				
Non-graduates	51.5	48.5	1.0 (0.82 to 1.2)	.97
Graduates	51.4	48.6		
<b>Employment</b>				
Employed	45.7	54.3	1.61 (1.32, 1.95)	<.001
Unemployed	57.6	42.4		
<b>Monthly income</b>				
Low-income status	53.2	46.8	0.72 (0.57 to 0.91)	.006
High income status	45.3	54.7		
<b>Marital status</b>				
Married	44.7	55.3	1.64 (1.35, 1.99)	<.001
Unmarried	57.0	43.0		
<b>Consanguineous marriage</b>				
Yes	42.8	57.2	1.36 (1.20, 1.55)	<.001
No	46.0	54.0		
<b>Having beta thalassemia in family</b>				
Yws	54.8	45.2	0.85 (0.64, 1.11)	.24
No	51.0	49.0		
<b>Division</b>				
MZD	57.3	42.7	0.76 (0.68, 0.86)	<.001
RWK	53.0	47.0		
MRP	44.2	55.8		

Through this study, a link was developed between various demographic variables and the knowledge of people, as shown in Table 3. People in all age groups were observed to have some knowledge about thalassemia, especially people over 40 years of age, which showed the highest percentage (54.2%) among all age groups. The study revealed that female individuals were more aware (53.3%) of the disease as compared to male participants (49.0). It was also observed that the education has not that much influence on people know how as non-graduate (51.5%) and graduate (51.4%) participants had same knowledge about the disease. Interestingly, 45.7% of the participants working somewhere showed good knowledge while there were 57.6% of the participants who had good knowledge but no job. It is obvious from the study that money plays no or a minimum role in a person's knowledge about anything. The study revealed that participants with less income (53.2%) were well informed than those who

earned more than 50,000 per month (45.3%). Surprisingly, married participants were less aware (44.7%) than unmarried ones. Likewise, people with cousin marriage (42.8%) were not very familiar with the disease. Division-wise, 57.3% of Muzaffarabad residents were aware of the disease, while in Rawalakot 53.0% and in Mirpur divisions 44.2% people have good knowledge about thalassemia.

To evaluate the perception of the participants about beta thalassemia, a few statements were given, and their agreement was recorded with each statement, as shown in Table 4. The evaluation was based on responses i.e., strongly agree, agree, disagree, and strongly disagree. The results showed that most of the participants strongly disagree with the idea that beta thalassemia is caused by a defect in the mother (46.39%) or farther (48.12%). Only 5.31% of the population strongly agreed that a medical test should be performed to detect babies for  $\beta$ -thalassemia before or shortly after birth. Compared to a study by 536 (31.96%) participants strongly rejected the idea that everyone should be bound to go for  $\beta$ -thalassemia screening contrary to a finding by Bassu (2015) where 99.6% of participants agreed that premarital testing should be performed for the presence of the thalassaemia carrier. Similarly, 1534 (91.47%) strongly disagreed with the statement that babies should be screened for  $\beta$ -thalassemia before or after birth. Likewise, 1021 (60.88%) interviewees showed disagreement with the statement that a baby should be aborted when it diagnosed with  $\beta$ -thalassemia major as compare to the study by Ebrahim *et al.* (2019) where only 2.2% were agreed. Whereas in a study on parents of thalassemia patients in Iran by Karimi *et al.* (2010) 91.3% of participants accepted the option of medical abortion. It is observed during the study that most people consider any kind of abortion as a sin. This is due to the lack of information and advance technology in the area where a doctor can diagnose a genetic disorder in the early days of a pregnancy. Thalassemia is prevalent in several countries, including India, Pakistan, Iran, and the Middle East. These countries also have significant religious populations that may be opposed to abortion (Ghanei *et al.*, 1997; Karimi *et al.*, 2007; Tasleem *et al.*, 2007; Ali *et al.*, 2019). However, not all religious groups are against abortion. Some religious scholars have argued that abortion may be permissible in certain circumstances, including cases where the mother's life is at risk or when the fetus has severe abnormalities. These scholars believe that abortion can be a way to prevent suffering and preserve life (Al-Matary and Ali, 2014). Therefore, it is necessary to engage with religious leaders and scholars to create awareness and understanding of thalassemia and its consequences. It is crucial to educate people about the severity of the disease and the impact it can have on individuals and families. By doing so, people may be more willing to consider abortion as a means of preventing the birth of children affected by thalassemia.

**Table 4: Participants perception towards thalassemia disease.**

Statements	Strongly agree (%)	Agree (%)	Disagree (%)	Strongly disagree (%)
$\beta$ -thalassemia is caused because of the defect in mother.	15.86	24.99	12.76	46.39
$\beta$ -thalassemia is caused by a defect in the father.	17.29	23.20	11.39	48.12
A child with $\beta$ -thalassemia makes the family life miserable.	62.61	10.97	3.10	23.32
A medical test must be run to screen babies for $\beta$ -thalassemia before or right after birth.	5.31	2.39	0.83	91.47
It should be mandatory for everyone to go for $\beta$ -thalassemia screening.	53.13	10.73	4.17	31.96
A prenatal diagnosis must be made if you and your spouse are $\beta$ -thalassemia minors/carriers.	79.67	8.65	2.86	8.83
A baby should be aborted when diagnosed with $\beta$ -thalassemia major.	23.38	12.16	3.58	60.88
Necessary blood test should be done before marriage to prevent the birth of a thalassaemic child.	62.19	10.73	4.77	22.30
You should inform others about the potential danger of thalassemia.	91.77	4.29	1.07	2.86
Your necessary steps to ensure blood testing for thalassemia before the marriage of your family members	75.85	8.47	4.71	10.97
Your institution should take the initiative to raise awareness among students about thalassemia.	91.71	5.78	0.54	1.97

**Table 5: The attitude of the participants towards thalassemia disease.**

Statement	Strongly agree (%)	Agree (%)	Disagree (%)	Strongly disagree (%)
Do you believe $\beta$ -thalassemia is caused because of sin committed by parents?	4.05	19.74	7.69	68.52
Do you think family is unlucky where the $\beta$ -thalassemic child is born?	19.26	13.30	4.53	62.91
Is it better to die than to live with a deadly disease like $\beta$ -thalassemia?	25.70	8.83	4.17	61.30
Do you feel discomfort from the neighbor having $\beta$ -thalassemia?	25.76	6.92	2.62	64.70
Do you feel discomfort living with a person suffering from $\beta$ -thalassemia?	22.12	11.63	3.28	62.97
Do you feel discomfort establishing a marriage relation where someone is suffering from $\beta$ -thalassemia?	58.74	13.42	5.37	22.48
Do you feel discomfort accepting a life partner who is $\beta$ -thalassemia minor/carrier?	65.53	8.77	1.67	24.03
would you like to donate my blood for patients with thalassemia?	73.76	6.14	2.15	17.95
would you be happy to befriend a patient with thalassaemia?	69.47	5.84	2.15	22.54

The public health department, in collaboration with Islamic clerics, can play a role in making people aware of Islamic teachings (fatwa) for such ill-fated children, as well as the hazards of carrying a child whose life would be hell with the passage of time, since there is no treatment for this condition. Interestingly, 1539 (91.77%) participants strongly agreed that they should inform others about the potential danger of thalassaemia and 1538 (91.71%) individuals wanted that their institution should take the initiative to generate awareness among students about thalassaemia.

To calculate the attitude of people participating in the study, a 4-point likert scale was used as shown in Table 5. The evaluation was carried out to better understand the false beliefs or myths that circulate in society about this genetic disorder. Overall positive attitude towards thalassemia patients and negative attitude towards the disease was

observed. Approximately 68 (4%) participants believed that believe  $\beta$ -thalassemia is caused by the sin committed by parents, while 323 (13.8%) individuals strongly agreed that a family is unlucky where the  $\beta$ -thalassemic child is born. The number may not be very high, but unfortunately, such people exist and create social issues in the society. About 431 (25.7%) people were of strong believe that it was better to die than to live with  $\beta$ -thalassemia. About 985 (58.7%) respondents strongly disagree with the idea of marrying a person suffering from thalassemia. Similarly, only 22.48% were strongly disagree when asked whether they would feel discomfort accepting a thalassemia carrier as a life partner.

About 73.76% of the participants responded positive when asked if they would donate blood to a patient with thalassemia, while 17.95% strongly disagreed. Despite highest literacy rate in the region and better lifestyle than

rest of the country, the beta-thalassemia is on rise in the study area. The reason may be the strong caste system where people prefer to marry with their cousins rather than marry outside the family.

During the study, it was revealed that regardless of their origin, financial status, or qualification, about 69.47% of people still believed that consanguineous marriages are not the reason behind diseases like beta thalassemia. In a similar study conducted by Ahmed *et al.* (2020), 68.8% participants disagreed that cousin marriage is a major reason behind beta thalassemia. With all the findings in view, the provision of access to genetic counseling and testing services in such regions is inevitable. Genetic counseling can help individuals and couples understand their risk of having a child with thalassemia and make informed decisions about their reproductive options. Genetic testing can help identify carriers of thalassemia and allow them to make informed decisions about their reproductive choices.

## Conclusions and Recommendations

It was concluded that regardless of the highest literacy rate, the large number of young people, and prosperity in the region compared to other provinces of Pakistan, awareness of genetic disorders, particularly thalassemia, is not sufficient. Mass communication and journalism are required to develop an awareness campaign for thalassemia among the public. Increasing general and high risk focused awareness raising activities that are culturally suitable and linguistically modified will play a critical role in the prevention and treatment of thalassemia.

Raising awareness about genetic issues can be challenging, but there are some effective ways to integrate this topic into general education so that even literate people can remain fully aware of complex genetic issues. For example, genetics is a complex subject that requires a strong foundation in biology. Therefore, including genetics in school science curriculums is an effective way to raise awareness about genetic issues among students. Genetic problems can be difficult to understand, especially for those without a scientific background. Using storytelling can help break down complex genetic problems into simpler, relatable terms that people can understand. Multimedia tools such as videos, interactive simulations, and online resources can be used to explain genetic issues in an engaging and interactive way. Encourage discussions and debates on genetic issues can help promote critical thinking and deepen the understanding of complex genetic issues. Bringing in experts and guest speakers who have experience in genetics can provide students with first-hand knowledge and experience, making the subject more relatable and engaging.

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### Availability of data

Data can be provided upon request.

### Consent of publication:

The authors grant their consent for the publication of their work.

### Ethics approval and consent to participate.

Not Applicable.

### Conflicts of interest

The authors have declared no conflict of interest.

## References

- Ahmed, M., Sharif, M.S., Yaqoob, R., Nadeem, M.S.A., Haroon, Z. and Iqbal, T., 2019. Impact of Thalassemia Centre on awareness of parents of Thalassemic patients about the disease: Comparative study in Muzaffarabad and Kotli districts of Azad Kashmir. *Pak. J. Physiol.*, **15**: 11-15.
- Ahmed, M.M., Salaria, S.M., Qamar, S., Soaz, M.A., Bukhari, M.H. and Qureshi, A.H., 2016. Incidence of  $\beta$ -thalassemia carriers in Muzaffarabad, Azad Kashmir. *Ann. Punjab Med. Coll.*, **10**: 11-19.
- Ahmed, N., Khan, B.A., Bukhari, S.W., Khan, K.S., Sabir, T. and Nazir, M.B., 2020. Knowledge, attitude and practices (KAP) of the families of B-thalassemia patients in a thalassemia center of Karachi. *Int. J. Curr. Med. Pharm. Res.*, **6**: 4972-4976.
- Ain-ul-Batool, S., Blasius, K., Kaindl, A. and Ghazanfar, A., 2019. A homozygous c. 1131G> a missense mutation in BBS9 gene manifesting autosomal recessive Bardet-Biedl syndrome in consanguineous Kashmiri family. *Pakistan J. Zool.*, **51**: 1575. <https://doi.org/10.17582/journal.pjz/2019.51.4.sc1>
- Ali, G., Awan, N.B., Khawaja, A.W., Foo, J.N., Khor, C.C., Chang, C.H., Chew, E.G., Kiani, F.R. and Jelani, M., 2020. Identification of a recurrent nonsense mutation in HR gene responsible for atrichia with popular lesions in two Kashmiri families. *J. Gene Med.*, **22**: e3167. <https://doi.org/10.1002/jgm.3167>
- Ali, G., Foo, J.N., Nasir, A., Chang, C.H., Chew, E.G., Latif, Z., Azeem, Z., Ain-Ul-Batool, S., Kazmi, S.A.R., Awan, N.B. and Khan, A.H., 2021. Identification of a novel homozygous missense (c. 443A> T: p.N148I) mutation in BBS2 in a Kashmiri

- family with Bardet-Biedl syndrome. *Biomed. Res. Int.*, **2021**: 1-9. <https://doi.org/10.1155/2021/6626015>
- Ali, G.S., Najeeb, M.K., Jubrail, A.M.S. and Selamoğlu, Z., 2019. A study of vitamin D and calcium level in beta thalassemia major in Duhok province. *Turk. J. Agric. Fd. Sci. Technol.*, **7**: 1924-1927. <https://doi.org/10.24925/turjaf.v7i11.1924-1927.2893>
- Ali, I., Abbassi, A.A., Asghar, U.N.R., Ishaq, K., Aziz, T. and Hussain, M.A., 2021. Molecular analysis of families afflicted with autosomal recessive oculocutaneous albinism from Azad Jammu and Kashmir. *Pure appl. Biol.*, **5**: 107-113. <https://doi.org/10.19045/bspab.2016.50014>
- Al-Matary, A. and Ali, J., 2014. Controversies and considerations regarding the termination of pregnancy for foetal anomalies in Islam. *BMC Med. Ethics*, **15**: 1-10. <https://doi.org/10.1186/1472-6939-15-10>
- Balci, Y.I., Ergin, A., Polat, A., Atilgan, T., Uzun, U. and Koyuncu, H., 2014. Thalassemia premarital screening program: Public view, what has been done, what needs to be done? *Int. J. Hematol. Oncol.*, **24**: 247-252. <https://doi.org/10.4999/uhod.14569>
- Basu, M., 2015. A study on knowledge, attitude and practice about thalassemia among general population in outpatient department at a Tertiary Care Hospital of Kolkata. *J. Prev. Med. Holist. Hlth.*, **1**: 6-13.
- Bernshtein, V., 2022. Types causes and treatment of thalassemia. <https://www.ucsf-ahp.org/types-causes-and-treatment-of-thalassemia/>. UCSF alliance health project. Available at: <https://www.ucsf-ahp.org/> (Accessed: March 7, 2023).
- Cevallos, M. and Egger, M., 2014. Strobe (STrengthening the Reporting of OBservational studies in Epidemiology). Guidelines for reporting health research: A users manual, pp. 169-179. <https://doi.org/10.1002/9781118715598.ch17>
- Dr Pervez, T., 2022. Education spending in AJK. The express tribune. Archived from the original on March 20, 2022. Retrieved December 8, 2022.
- Ebrahim, S., Raza, A.Z., Hussain, M., Khan, A., Kumari, L., Rasheed, R., Mahmood, S., Khatri, M.A., Bijoor, M., Zaheer, R. and Sattar, N., 2019. Knowledge and Beliefs regarding thalassemia in an urban population. *Cureus*, **11**: e5268. <https://doi.org/10.7759/cureus.5268>
- Ehsan, H., Wahab, A., Anwer, F., Iftikhar, R. and Yousaf, M.N., 2020. Prevalence of transfusion transmissible infections in beta-thalassemia major patients in Pakistan: A systematic review. *Cureus*, **12**: e10070. <https://doi.org/10.7759/cureus.10070>
- Galanello, R. and Cao, A., 2011. Alpha-thalassemia. *Genet. Med.*, **13**: 83-88. <https://doi.org/10.1097/GIM.0b013e3181fcb468>
- Galanello, R. and Origa, R., 2010. Beta-thalassemia. *Orphanet J. Rare Dis.*, **5**: 1-15. <https://doi.org/10.1186/1750-1172-5-11>
- Ghanei, M., Adibi, P., Movahedi, M., Khami, M.A., Ghasemi, R.L., Azarm, T., Zolfaghari, B., Jamshidi, H.R. and Sadri, R., 1997. Pre-marriage prevention of thalassaemia: Report of a 100000-case experience in Isfahan. *Publ. Hlth.*, **111**: 153-156. <https://doi.org/10.1038/sj.ph.1900333>
- Goajk Bureau of Statistics, 2021. AJ and K At A Glance-2021. Muzaffarabad: Planning and Development, Govt. of Azad Jammu and Kashmir. <http://www.raosoft.com/samplesize>, <https://www.pndajk.gov.pk/>
- Islam, M.M., Hossain, F., Sakib, N., Zeba, Z., Bhuiyan, A.I., Mamun, M.A., Kaggwa, M.M., Yoshimura, K., Afrin, S., Selim, S. and Hossain, M., 2021. Distribution of  $\beta$ -thalassemia and other hemoglobinopathies in Bangladeshi University Students and Ready-Made Garment Workers. *Risk Manag. Healthc. Policy*, **14**: 2707. <https://doi.org/10.2147/RMHP.S317852>
- Jaffar, N., Khan, L., Ahmed, U.I., Vistro, N.H. and Memon, M.Y., 2021. Barriers to premarital thalassemia screening in Asia. *World Fam. Med.*, **19**: 146-153.
- Karimi, M., Cohan, N. and Pishdad, P., 2015. Hydroxyurea as a first-line treatment of extramedullary hematopoiesis in patients with beta thalassemia: Four case reports. *Hematology*, **20**: 53-57. <https://doi.org/10.1179/1607845414Y.0000000168>
- Karimi, M., Jamalain, N., Yarmohammadi, H., Askarnejad, A., Afrasiabi, A. and Hashemi, A., 2007. Premarital screening for  $\beta$ -thalassaemia in Southern Iran: Options for improving the programme. *J. Med. Screen*, **14**: 62-66. <https://doi.org/10.1258/096914107781261882>
- Karimi, M., Johari, S. and Cohan, N., 2010. Attitude toward prenatal diagnosis for  $\beta$ -thalassemia major and medical abortion in Southern Iran. *Hemoglobin*, **34**: 49-54. <https://doi.org/10.3109/03630260903547690>
- Maheen, H., Malik, F., Siddique, B. and Qidwai, A., 2015. Assessing parental knowledge about thalassemia in a thalassemia centre of Karachi, Pakistan. *J. Genet. Couns.*, **24**: 945-951. <https://doi.org/10.1007/s10897-015-9830-z>
- Mediani, H.S., Tiara, A. and Mardhiyah, A., 2019. Factors related to the needs of parents having school age thalassaemic children. *J. Keperawatan Padjadjaran*, **7**: 175-189. <https://doi.org/10.24198/jkp.v7i2.1119>
- Morris, C.R., 2010. Role of arginase in sickle cell lung disease and hemolytic anemias. *Open Nitric Oxide J.*, **2**: 41-54. <https://doi.org/10.2174/1875042701002020041>
- Piel, F.B. and Weatherall, D.J., 2014. The  $\alpha$  thalassaemias. *N. Engl. J. Med.*, **371**: 1908-1916. <https://doi.org/10.1056/NEJMp1400000>



[org/10.1056/NEJMra1404415](https://doi.org/10.1056/NEJMra1404415)

- Porter, J. and Viprakasit, V., 2014. Iron overload and chelation. In: *Guidelines for the management of transfusion dependent thalassemia (TDT)* (eds. A.C., Cohen, J. Porter, A. Taher and V. Viprakasit). 3<sup>rd</sup> ed. Cyprus: TIF publication pp. 42-74.
- Raosoft.com. 2016. Sample size calculator. [online] Available at: <[http://www.raosoft.com/sample\\_size.html](http://www.raosoft.com/sample_size.html)> [Accessed 28 March 2023]
- Snedden, C., 2013. The untold story of the people of Azad Kashmir, 2012. Kashmir: The Unwritten History. HarperCollins India. ISBN 978-9350298985.
- Sevindik, M., Rasul, A., Hussain, G., Anwar, H., Zahoor, M.K., Sarfraz, I., Kamran, K.S., Akgul, H., Akata, I. and Selamoglu, Z., 2018. Determination of anti-oxidative, anti-microbial activity and heavy metal contents of *Leucoagaricus leucothites*. *Pak. J. Pharm. Sci.* **31**(5Supp.): 2163-2168.
- Sevindik, M., 2018. Heavy metals content and the role of *Lepiota cristata* as antioxidant in oxidative stress. *J. Bacteriol. Mycol. Open Access.*, **6**: 237-239. <https://doi.org/10.15406/jbmoa.2018.06.00211>
- Sevindik, M., 2019. The novel biological tests on various extracts of *Ceriporus varius*. *Fresenius Environ. Bull.*, **28**: 3713-3717.
- Shafique, F., Ali, S., Almansouri, T., Van Eeden, F., Shafi, N., Khalid, M., Khawaja, S. and Andleeb, S., 2021. Thalassemia, a human blood disorder. *Braz. J. Biol.*, **83**: <https://doi.org/10.1590/1519-6984.246062>
- Shafique, F., Ali, S., Andleeb, S., Rauf, A., Kazmi, S.A., Idrees, S., Farooq, F., Khan, F.N., Hassan, M.U., Mumtaz, R.A. and Khalid, S., 2020. Prevalence of hepatitis B and C and assessment of responsible risk factors among the vulnerable  $\beta$ -thalassemic patients of Azad Kashmir, Pakistan. *Pakistan J. Zool.*, **52**: 793-796. <https://doi.org/10.17582/journal.pjz/20180805090822>
- Shukla, V., Mondal, T.K., Ray, K., Dutta, S., Mandal, M.M. and Basu, M., 2022. Screening before marriage is important: A cross-sectional study on thalassemia among eligible couples from a slum of Kolkata. *Med. J. Dr. DY Patil Vidyapeeth*, **15**: 62. [https://doi.org/10.4103/mjdrdypu.mjdrdypu\\_348\\_20](https://doi.org/10.4103/mjdrdypu.mjdrdypu_348_20)
- Tariq, H., Zaigham, K., Kousar, S. and Azhar, A., 2019. Genetic contribution of GJB2 gene to hearing impairment in Pakistan. *Adv. Life Sci.*, **7**: 38-43.
- Tasleem, S., Tasleem, H., Siddiqui, M.A., Adil, M.M. and Rashid, Y., 2007. Prenatal diagnosis of  $\beta$ -thalassaemia by chorionic villous sampling. *J. Pak. Med. Assoc.*, **57**: 528-31.
- Umair, M., Ahamd, F., Bilal, M., Asiri, A., Younus, M. and Khan, A., 2019. A comprehensive review of genetic skeletal disorders reported from Pakistan: A brief commentary. *Meta Gene*, **20**: 100559. <https://doi.org/10.1016/j.mgene.2019.100559>