ORIGINAL ARTICLE

Beta Thalassemia: A Family Affair - Exploring the Interplay Between Disease Awareness, Parental Stress, Child Well-being and Quality of Life in Karachi, Pakistan

Fareeha Shahid[1], Saleem Shahzad[2], Saqlain Anwar[3], Zohab Ahmed[4], Hassan Waqar[3], and Ahmad Hassan[4]

ABSTRACT

Objective: To assess the general public's level of disease awareness and understanding, as well as its impact on various elements of life.
Methodology: This is a cross-sectional study. Total sample size was 275. Study duration was September 2020 –February 2021. Inclusion criteria was all men and women, either married or unmarried, above the age of 18 years, who did not work in healthcare sector and lived in Karachi, Pakistan. A standardized questionnaire was created using Google Forms, and data was collected online. In addition, a consent form was appended to the questionnaire. The study was given approval by the institute'sÊethicalÊreview committee. The data was analyzed using SPSS version 25. Chi square test was used for data comparison. A p-value of <0.05 was considered statistically significant.
Results: Out of the total study population, 81.7% were married and 17.3% were unmarried. The majority (51.4%) were aged 18 to 22 years. A significant association was found between knowledge and quality of life (p-value; 0.018), between the role of consanguineous marriages and disease etiology (p-value; 0.014), between the number of units of blood transfused to the thalassemic patient in one sitting and its effect on quality of life (p-value; 0.006), and between knowledge and the number of hospital visits for blood transfusion (p-value; 0.031).
Conclusion: The study indicated that our sample group lacked sufficient awareness regarding thalassemia and its impact on quality of life for both the patient and the family.

Keywords: Beta-thalassemia major, consanguineous marriages, disease awareness, quality of life.

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INTRODUCTION

Mediterranean, Southeast Asia, Africa, and the Middle East are the regions where thalassemia is most prevalent[1]. Around 4 children worldwide are affected by thalassemia for every 100,000 live births, and 7% of the world's population is a carrier of this illness[2]. Between 50,000 and 100,000 children in third-world nations die from thalassemia major each year[3]. An estimated 9000 babies in Pakistan are born with beta thalassemia each year[4,5]. In the entire population, there are 9.8 million carriers, or an estimated 5-7% carrier rate[5]. If we break down the figures, it means that in an average group of 100 Pakistani citizens, five are disease carriers. The cultural and religious scenario of Pakistan is such that consanguineous marriages are quite common[6]. There is lack of widespread awareness of premarital screening and counseling of individuals with a family history of the disease[7]. Furthermore antenatal diagnoses is not widely available. The concept of termination of pregnancy is an ethical and religious issue in the community[8,9]. With these state of affairs, thalassemia is best treated conservatively with all its inherent complications, eventually resulting in debility.

1 Bahria University Health Sciences Campus, Karachi, Pakistan

2 Jinnah Postgraduate Medical Centre (JPMC), Karachi, Pakistan

3 PNS Shifa Hospital, Karachi, Pakistan

4 Aga Khan University Hospital, Karachi, Pakistan

Correspondence: Dr. Fareeha Shahid, Bahria University Health Sciences Campus, Karachi, Pakistan

Email: drfareeha@live.com

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Thalassemia can lead to physical deformity, growth retardation and delayed puberty[10-12]. Its effect on physical appearance adds to the child's negative self-perception. Due to frequent hospital visits and laboratory testing, families of thalassemic patients are more likely to have behavioral issues. Parents frequently bear an excessive financial load as a result of the intricate and ongoing treatment process. All things considered, thalassemia presents financial, emotional, cognitive, and physical challenges for the sufferer and their family. Inadequate treatment, a lack of psychosocial support, and the absence of public screening programs, all contribute to an increase in the burden of disease[13,14]. Community wide education programs can dramatically increase the population awareness regarding the disease. Understanding the factors that contribute to patients' and their families' quality of life is essential for improving it[15,16]. In order to fill in the knowledge gaps, this study focuses on a less studied area, namely the relationship between emotion and illness. Moreover, we will assess public awareness of thalassemic patients and the variables affecting their health-related quality of life. All of this knowledge will assist us in determining whether the psychological health of the parent affects the quality of life of the child. It might open the door to understanding the area that requires attention in order to improve the outcome of the disease.

METHODOLOGY

IRB/ERC Approval:

The research was conducted using an online, structured, closed-ended questionnaire created with Google Forms, approved by the Institutional Review Board of the Bahria University Health Sciences Campus, Karachi Ref. No. RUHS-IRB # 099/24.

Duration of the study was six months from September 2020-February 2021.The sample size calculation was carried out by statistical software StatCalc sample size calculator tool with 95% confidence level and 5% margin of error. The calculated sample size of the study was 275. Total number of responses received was 301 and finally the valid number of responses among these was 275. Random sampling technique was applied and each respondent was given equal chance of selection. All residents of Karachi, male or female, married or unmarried, above the age of 18 years were included in research and those belonging to any health-care service, and those who did not give consent were excluded from the study. Data was collected using an online questionnaire form, the link of which was distributed amongst non-medical students residing in Karachi, Pakistan to increase the diversity of the sample. A structured, standardized questionnaire was designed using Google Forms. The questionnaire consisted of 22 questions. A consent form was attached before the questionnaire. Each participant was instructed to select the option that best suits their opinions to analyze the level of perception about various factors amongst the participants. Confidentiality of respondents was maintained. This study was impartial and independent. Data analysis was performed using Excel spread sheet along with Statistical Package for Social Science (SPSS) version 25.0 to analyze the data obtained via our response forms. Using this data, the responses were also analyzed into percentages and frequencies using SPSS descriptive tool. A p-value of less than 0.05 was considered statistically significant in all cases. During data analysis total of 24 variables were merged into 7 variables altogether (daily activities, socializing, attitude towards patient, parents mental health, effect on siblings, family gatherings and blood arrangement) to form a new variable which was then labelled as “quality of life (QoL)” in thalassemic children and their families. Five variables (relationship, premarital screening, prenatal screening, termination of pregnancy, costs) were merged to form a new variable which was labelled as “knowledge (Klg) among the participants” of our study. Disparity between categorical variables was checked using Chi square test whereas descriptive statistics were used to report frequencies and proportions for categorical responses. The categorical variables used in this research were age, marital status, consanguineous marriages and relationship.

RESULTS

Basic characteristics: A total of 275 persons were selected and 100% consented to participate in the study, therefore the response rate was also 100%. Confidence level was set at 95%. None of the response forms were excluded from the study as no missing data was found. This was solely since it was made mandatory for the participant to answer all the questions prior to its submission. During data analysis a few changes were made, out of a sum total of 24 variables we merged seven variables altogether (daily activities, socializing, attitude towards patient, parents mental health, effect on siblings, family gatherings and blood arrangement) to form a new variable which was then labelled as “quality of life (QoL)” in thalassemic children and their families. Five variables (relationship, premarital screening, prenatal screening, termination of pregnancy, costs) were merged to form a new variable which was labelled as “knowledge (Klg) among the participants” of our study.

Demographics:

Table-1 shows the socio-demographic characteristics

of our sample population. Our data indicated, out of the total 275 study population, the majority (51.4%) were in the 18 to 22 year age group. The mean age of our participants was 24±2.75 years; with ages ranging from 18-30 years. Furthermore, when inquired about the relationship status of the participants, responses indicated that 81.7% (n=227) of the total participants (n=275) were married and 17.3% (n=48) were unmarried. However, no significant association was found between the socio-demographic data (age, marital status) and the overall knowledge among the participants.

Spread of thalassemia and hospital related variables:

Table-2 shows that the factors related to the spread of thalassemia were covered using three variables namely "etiology", "transmission" and "role of consanguineous marriages" in spread of disease. When asked about the etiology of beta-thalassemia, majority of the participants (64.4%: n= 179/275) already knew that thalassemia is a genetic disease, passed down from parents to their offspring; these numbers were followed by a total number of 34 participants who considered thalassemia an infectious disease.However; a total number of 62 participants (22.3%) considered thalassemia to be a non-infectious or metabolic disease. Less than half of the total number of the participants (48.9% n=136/275) knew about the role of consanguineous marriages in the spread of thalassemia. 139/275 respondents did not believe cousin/consanguineous marriages to be an important factor in the spread of thalassemia.

Moreover, Table-2 shows that a significant association was found between the role of consanguineous marriages in the spread of thalassemia and etiology of the disease (p-value 0.014). However, no significant association was found between 'knowledge" and role of "consanguineous marriages" in the spread of thalassemia.

Upon analyzing the data to assess the knowledge of the participants regarding the transmission of thalassemia, we found that more than half our sample (58.7%) believed the disease is genetic, followed by 63 respondents who believed its transmission is due to blood transfusions, whereas only 7.6% of the sample opted for the "other" option. Additionally, as showed in table-3 our data indicated that 113 out of 275 respondents were clueless about the no of bottles a thalassemic child receives in one sitting, however 60 participants were certain that one bottle/sitting, whereas approximately 84 participants opted for two bottles/sitting. A significant association (p- value; 0.006) was found between the units of blood transfused to the thalassemic patient in one sitting and its effect on (Qol) quality of life which comprises of 7 variables merged together (daily activities, socializing, attitude towards patient, parents mental health, effect on siblings, family gatherings and blood arrangement). Our analysis indicated that the 42, 149 and 84 respondents of our study thought that a thalassemic child has to visit the hospital daily, weekly and monthly respectively.

More than half of our participants (69.1%) did not know about the importance of premarital and prenatal genetic counseling in high risk families which is used as a method of prevention against the disease, while only 83/275 respondents considered genetic counselling as preventable measure against the disease. A higher proportion of participants (54%) considered blood transfusion as the only treatment option available for the thalassemic children whereas only a small percent (23.4%) of the sample already knew that bone marrow transplantation, which if done timely can permanently cure thalassemia and therefore produce better results.

Knowledge:

Knowledge regarding beta-thalassemia major among the participants of our study was evaluated based on a mean score of 5 variables. The unknown part of the information amongst our study population was mainly the poor knowledge regarding the availability of diagnostic tests.

1. Premarital screening: (Mean: 0.77, SD = 0.419) - more than half i.e. 76.6% of the sample not knowing about premarital screening.

2. Prenatal screening: (Mean: 0.77, SD = 0.423) - 75.9% of the respondents were completely clueless about the availability of prenatal diagnostic tests for potential carriers of the disease.

3. Termination of Pregnancy: (Mean: 0.77, SD = 419) - an overwhelming majority (76.6%) of participants were against the termination of pregnancy in case of a positive screening test result. Amongst which a total of 213 participants would not terminate pregnancy at any cost whereas 62 respondents would opt for termination of pregnancy based on a positive screening test result; which amounts to only 22.3% of the entire sample size.

4. Cost awareness: (Mean; 0.71, SD = 0.455) - when questioned about the cost of a single blood transfusion which is used to manage the disease; we found that only on 80 respondents were aware out of a total sample of 275. Surprisingly enough, 70.1% of our participants were unaware of the fact and were clueless as to how much a single blood transfusion would generally cost a thalassemic patient in Karachi, Pakistan.

5. Family history of disease: (Mean: 0.52, SD = 0.501) - family history was positive in 132 participants whereas a total of 143 which amounts to more than half of our

sample size mentioned that they've never had an interaction or a close relationship with a thalassemic child before.

Table-3 shows a significant association (p value 0.018) between the "knowledge" which comprised of 5 variables merged altogether (relationship, premarital screening, prenatal screening, termination of pregnancy, costs) and quality of life (QoL) consisting of 7 variables merged together (daily activities, socializing, attitude towards patient, parent's mental health, effect on siblings, family gatherings and blood arrangement).

Table 1: Demographic Characteristics, Spread and Treatment Related Variables

Variables	Frequency (N = 275)	Percent (%)
Age (in years)		
18-22	143	51.4
23-26	95	34.2
27 & above	37	13.3
Mean Age (S. Dev)	24 ± 2.75 years	
Marital Status		
Married	227	81.7
Un-Married	48	17.3
Etiology		
Infectious Disease	34	12.2
Non-Infectious Disease or Metabolic Disease	62	22.3
Hereditary Disease	179	64.4
Transmission		
Blood Transfusion	63	22.7
Parents to offspring	191	68.7
Others	21	7.6
Role of Consanguineous Marriages		
Yes	136	48.9
No, Maybe, Not sure	139	50
Hospital Visits		
Daily	42	15.1
Weekly	149	53.6
1-12 Months	84	30.2
Bottles of blood/ transfusion		
1 bottle	60	21.6
2 bottles	84	30.2
> 3 bottles	18	6.5
Not sure	113	40.6
Genetic Counselling		
Yes	83	29.9
No, Maybe	192	69.1

Table 2: Cross Tabulation of Knowledge of Participants, Etiology and the Role of Consanguineous Marriages in Spread of Thalassemia

Consanguineous Marriages	Yes	No, Maybe, Not sure	p-value
Knowledge of Participants			
Below Average	28	17	0.06
Average	26	40	
Above Average	82	82	
Etiology			
Infectious Disease	12	22	0.14
Non-Infectious Disease or Metabolic Disease	24	38	
Hereditary Disease	100	79	

*= significant p value

Table 3: Cross Tabulation of Bottles of Blood/ Transfusion, Knowledge of Participants and Overall Quality of Life Among Thalassemic Children

Quality of Life	Poor	Satisfactory	Good	p-value
Bottles of blood/ transfusion				
1 bottle	11	28	21	0.006*
2 bottles	28	18	38	
> 3 bottles	6	3	9	
None	26	51	36	
Knowledge of Participants				
Below Average	19	12	40	0.018*
Average	8	29	63	
Above Average	18	25	61	

*= significant p value

DISCUSSION

Beta-thalassemia affects not only the patient but also poses as a challenging task for the parents, therefore this study was aimed to assess the level of knowledge among the participants and the quality of life as perceived by thalassemic child and his/her family.

Only 37 individuals in our study were older than 27 years, with the majority of participants (51.4%) falling into the 18 - 22year age range. This was in line with findings from a study conducted in Malaysia, where 53% of participants were in the 18–20 age bracket.[16] The large proportion of youthful responders can be explained by their greater familiarity with software management and their quicker access to the internet. Married respondents outnumbered single respondents (81.7%) in this study, this is due to the high cultural

preference for early marriage found in South Asian households. Another study conducted in Pakistan, revealed the percentage of married participants to be 71%[17].

While most survey participants were aware of the disease's origin, they were largely ignorant of how thalassemia is transmitted. Most participants (64.4%) in our study agreed that genetics has a role in the disease's spread. This greater figure is the result of awareness initiatives in larger cities and easier access to social media platforms. This is in contrast to another study where a greater proportion of participants (60%) were not aware that thalassemia has a hereditary component[18]. As in another study done in Malaysia, a smaller percentage of participants in our survey believed that thalassemia is a metabolic or non-infectious condition[19].

In our survey, nearly half of the participants agreed that consanguineous marriages are a significant factor in the disease's spread. This is related to the part that culture plays in our day-to-day routine. Individuals typically want to marry within their family. People continue to be unaware of scientific research that strongly highlight the positive role consanguinity plays in the inheritance of thalassemia and hold the belief that human intervention plays no part in the transmission of disease. Government legislation can improve these figures. Similar results were obtained in another investigation that was carried out in India[20]. Our study showed that a relatively large number of participants were not sure about the amount of blood transfused to a thalassemic patient. Although one third of the participants agreed that 2 bottles of blood must be transfused in one sitting to a thalassemic patient. This is in accordance with the fact that they were not related to medical profession, so such precise response could not be expected. This highlights the need to emphasize on directing the attention of general public to such details. This might increase the number of voluntary blood donors in the community.

We also made an effort to assess the participants' thalassemia knowledge. We enquired about the disease's diagnostic procedures. A sizable portion (76%) were unaware regarding the pre-marital and prenatal testing. According to the participants' responses, they didn't know much about the disease's prognosis or course of therapy. Screening is believed to be impacted by a lack of medical awareness and a fear of stigma if one tests positive. Slightly more than half of participants knew that beta-thalassemia major affects daily routine activities of the patient. This is because large number of the participants were educated and knew well about the hindrances caused by this disease. Approximately 52% of participants agreed that thalassemic patient finds it difficult to interact and socialize with other people. This is similar to another study where majority of participants agreed that thalassemic patients might find it difficult to attain marital status. Throughout this investigation, three new findings were noted. First, 53.3% of the general public were found to be aware of how frequently a hospital visit may be required for a blood transfusion. Second, 52.2% of people did not know that arranging blood for a thalassemic child who has received multiple transfusions can be complicated and time-consuming. Finally, 40% of participants were unaware of how many blood bottles are required for each anemic episode.

CONCLUSION

There is a need for holistic management of thalassemia incorporating education, emotional, and physical rehabilitation. Premarital and prenatal screening policies should be introduced in Pakistan.

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Authors' Contributions: FS: led the study design and manuscript drafting. SS and SA: reviewed the literature and refined the content. ZA, HW & AH: contributed to analysis, revisions, and final approval.

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