Original Article

Assessment of level of Awareness regarding Thalassemia Major among Parents of Affected Children

Faizan Fazal[1], Mateen Arshad[2], Haris Mustafa[3], Muhammad Ebad Ur Rehman[4], Usama Tanveer[5], Sidra Hamid[6]

[1,2,3,4,5] Second year MBBS Student, Rawalpindi Medical University, Rawalpindi.
[6] Assistant Professor, Department of Physiology, Rawalpindi Medical University, Rawalpindi.

Author's Contribution
[1,3] *Conception of study*
[3] *Experimentation/Study conduction*
[1,2,3] *Analysis/Interpretation/Discussion*
[1,2,3,4,5] *Manuscript Writing*
[2,4,5] *Critical Review*
[1,2,3,4,5,6] *Facilitation and Material analysis*

Corresponding Author
Mr. Faizan Fazal,
Second year MBBS Student,
Rawalpindi Medical University,
Rawalpindi
Email: Abdalian459@gmail.com

Article Processing
Received: 03/12/2020
Accepted: 16/05/2021

Cite this Article: *Fazal, F., Arshad, M., Mustafa, H., Rehman, M.E.U., Tanveer, U., Hameed, S. Assessment of level of Awareness regarding Thalassemia Major among Parents of Affected Children. Student Supplement of Journal of Rawalpindi Medical College. 12 Dec. 2020; 25(1): 3-7.*
DOI: https://doi.org/10.37939/jrmc.v25i1.1736

Conflict of Interest: *Nil*
Funding Source: *Nil*

Access Online:

Abstract

Background: Thalassemia major is amongst the most common genetic disorders in developing countries like Pakistan. This study aims to assess the level of knowledge among people related to thalassemia and the number of thalassemia cases linked to consanguineous marriages.

Materials and Methods: A cross-sectional study was conducted among parents (n=272) of thalassemia patients registered with Rawalpindi thalassemia welfare society, from June 2019 to September 2019. Parents of thalassemia major patients aged 1 to 16 years were included. Parents who were health care professionals or community workers, and with children who had some other genetic disorders along with thalassemia major were excluded. Non-probability consecutive sampling was used. Data were collected using a self-designed questionnaire that assessed the prevalence of thalassemia in relation to consanguineous marriages and awareness of thalassemia in parents of affected ones. Data were analyzed using Statistical Package for Social Sciences (SPSS), version 22, by applying descriptive statistics.

Results: About 72% of the parents (n=214) had consanguineous marriages. None of them had a thalassemia screening test before their marriage. 98.5% of the parents did not come across any awareness campaign had ever been conducted for thalassemia in their area. 50% of families believed that thalassemia had affected their children's interaction with other children of their age.

Conclusion: This study reveals that parents of thalassemia-affected children are not aware of the high link between thalassemia and cousin marriages and the awareness about premarital testing is almost none.

Keywords: Consanguinity, Beta-thalassemia, Premarital Examination, Taboo, Awareness.

Introduction

Thalassemia is an autosomal recessive, hereditary, chronic hemolytic anemia due to a partial or complete deficiency in the synthesis of α-globin chains (α-thalassemia) or β-globin chains (β- thalassemia) that compose the major adult hemoglobin ($\alpha_2\beta_2$).[1] It is an inherited blood disorder characterized by lower hemoglobin and fewer red blood cells in the body than normal.

Thalassemia major involves a mutation in both genes of Beta-chain synthesis due to which Beta-chain is produced in less amount or is produced with an error. Signs and symptoms include severe anemia, poor growth, and skeletal abnormalities during infancy. Untreated thalassemia major eventually leads to death, usually by heart failure.[2]

Thalassemia is one of the major genetic problems affecting our children since Pakistan is in the thalassemia belt on the world map.[3] Worldwide estimates show that each year, over 50,000 new patients are born with a severe form of thalassemia (beta-thalassemia major and HbE beta-thalassemia) and nearly 80% of these births occur in developing countries. Control of hemoglobinopathies, particularly βthalassemia, in the developing world has been described as a priority by the World Health Organization (WHO).[4]

The number of genetic disorders in South Asian countries, specifically in Pakistan is increasing day by day despite rapid development in the field of medicine and science. The carrier rate in Pakistan ranges between 5-8%, and around 5000 children are diagnosed each year with beta-thalassemia in Pakistan.[10] The reason why these genetic disorders have gone out of hand is the lack of awareness among the public.

Thalassemia major is challenging not only for the affected person but for the family too. The only permanent solution is a bone marrow transplant which the majority of patients cannot afford. In a Nigerian survey involving 196 respondents, 85.2% were of the view that bone marrow transplantation was very expensive for them.[5] Blood transfusions are required at regular intervals, which makes life more challenging for families who are affected by this testing genetic disorder. Without transfusion or transplantation, patients with thalassemia major are expected to die within months of diagnosis.[6] Thalassemia major is largely related to consanguineous marriages. In a study conducted in Bannu, Pakistan, it was revealed that out of 180 subjects, 133(74%) parents of children affected by thalassemia major were cousins.[7]

Despite this alarming figure, the level of awareness in the general public is minimal. In a study conducted in Rahim Yar Khan, Pakistan, sixty percent of parents were unaware of the disease, 25% had little knowledge about the disease and only 15% knew about Thalassemia and its complications.[8]

The primary objective of this research is to assess the level of awareness about the causes and complications of thalassemia major in parents of affected children and the frequency of thalassemia major in cousin marriages. The second objective is to determine the frequency of the premarital test in the Rawalpindi division.

Materials and Methods

Around 3000 tests of Covid-19 were performed in a private lab in Rawalpindi from April to June 2020 in individuals of all age groups. Out of 3000, 1160 In this cross-sectional study, a total of 272 subjects were included, who were all parents of thalassemia major patients admitted to Rawalpindi Thalassemia Welfare Society, Rawalpindi. Subjects were recruited by non-probability consecutive sampling. Data were collected from June 2019 to September 2019. All willing individuals were included in the study except for parents who were health care professionals or community workers, or whose children had other genetic disorders along with thalassemia major. This study was conducted after taking approval from the ethical review board of Rawalpindi Medical University. Before inclusion, the objective of the study was clearly defined and informed consent was taken from every participant. The anonymity of participants was ensured to increase confidentiality.

Data were collected using a self-designed questionnaire developed after an extensive literature search and was approved by the institutional review board. The questionnaire consisted of 25 items pertaining to knowledge of symptoms, causes, and complications of thalassemia, and relation of parents prior to marriage. It also included questions to assess the prevalence of premarital testing. All questions were asked in their native language so that parents could understand them. Statistical Package for Social Sciences (SPSS) version 22 was used to analyze the collected data. Descriptive statistics were applied to analyze data.

Results

Out of 272 parents of thalassemic patients, only 2.2% (n=66) were literate while the majority of them (75.8%) were illiterate. Around 60% of the children suffering from thalassemia major were males and 40 % were females.

None of the parents conducted a premarital test that could rule out the possibility of thalassemia and other genetic disorders in their children. Only 35.3% of parents knew about the premarital test, whereas 64.7% were not aware of it.

About 52% of the participants had married their first cousin. Details regarding the marriage of participants are shown in Figure-1.

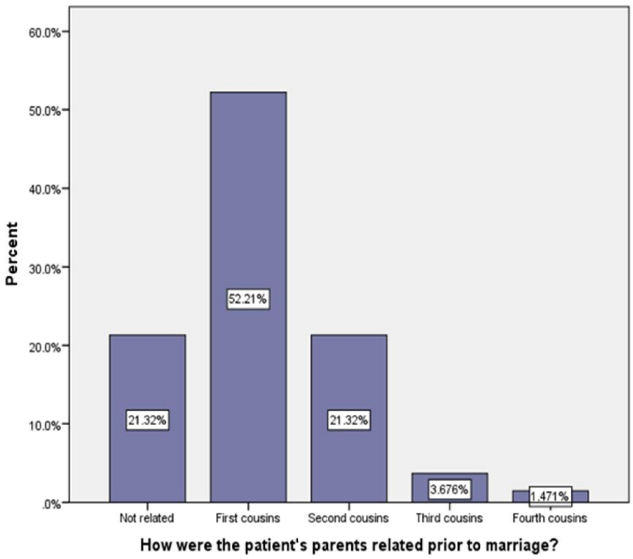

Figure-1 Relationship of patient's parents prior to marriage

According to our results, 84% of parents knew that Thalassemia major is an inherited disorder. Only 16% of parents knew before diagnosis that this disorder is highly linked to cousin marriages. 45% of parents revealed that their thalassemia-affected child faced psychological problems. 52% of patients faced socio-economic problems while encountering this disease. (Table-I)

Table-I Awareness, socio-economic issues of parents and influence of this disorder on child's mental health

Questions (Questions were asked from respondents in the Urdu language)	**Yes**	**No**
Do you know thalassemia major is an inherited disorder?	228 (83.8%)	44 (16.2%)
Does this disorder require repeated and periodic blood transfusions?	210 (78.2%)	62 (22.8%)
Did you know before diagnosis that thalassemia major is largely related to cousin marriages?	44 (16.2%)	228 (83.8%)
Is there any Sibling of the patient who has been diagnosed with thalassemia major?	60 (22.1%)	212 (77.9%)
Do you, as a parent, face socio-economic problems?	144 (52.9%)	128 (47.1%)
Do you know iron overload is a complication in the treatment of thalassemia major?	206 (75.7%)	66 (24.3%)
Was your family history positive for thalassemia major?	80 (30.1%)	190 (69.9%)
Is this disorder affecting his/her normal interaction with other children of the same age?	134 (49.3%)	138 (50.7%)
Does your child face psychological problems like depression?	124 (45.6%)	148 (54.4%)
Has any awareness campaign regarding thalassemia major been conducted where you live?	4 (1.5%)	268 (98.5%)

About 98.5% of the participants recommended that steps should be taken to avoid thalassemia by taking the premarital test before cousin marriage and by running awareness campaigns.

Discussion

Pakistan is among the highest thalassemia-affected countries in the world. Pakistan is said to have approximately 100,000 transfusion-dependent thalassemia patients.[11] This study revealed that the majority of the parents of children affected with thalassemia were married to their cousin and none had premarital thalassemia screening tests. There is a lack of awareness regarding thalassemia in the majority of the participants.

Our study shows that 75% of the parents were illiterate while only 25% were literate. Another study on the association between thalassemia and education conducted in Peshawar, Pakistan revealed that only 1.2% out of 320 patients were matriculate (had studied till 10th standard only).[12] Hence illiteracy is undoubtedly a contributing factor to a decreased level of awareness among the people.

This study shows that out of 272 parents of thalassemia major affected children, 214 (79%) were married to their cousin. This is consistent with a similar study done in district Bannu, Pakistan which stated that out of 180 subjects, 133(74%) parents were cousins, while 47(26%) were unrelated.[13] A study conducted on Gulf countries on the epidemiology of thalassemia also reported cousin marriage as the most important risk factor for thalassemia.[14] This study also revealed that 83% of parents did not know that there are high chances of a thalassemic child being born as a result of cousin marriages, which shows the lack of awareness of thalassemia in our population.

Thalassemia major not only affects the patient but his/her relatives are also involved in some sort of economic stress while countering this disorder. The results of this research have also shown that 53% of parents face socio-economic problems in countering this disorder. In a study from Sri Lanka on analyzing the cost of illness, it was concluded that β-thalassemia major poses a significant economic burden on health services and the families of affected children.[15]

Our study shows that 45% thalassemia-affected children also faces psychological problems. About 49% of parents were of the view that interaction of the affected child with other children of the same age has been affected due to this disorder. This is consistent with the study done on the effect of thalassemia on child mental health in Karachi, Pakistan where it was revealed that 61.7% of patients suffered mild depression.[16]

Another aspect of this study covers the premarital test. A premarital test involves genetic screening of both parents (husband and wife) to check whether any of them is the carrier for the gene of thalassemia major or not. This study also reveals that only 35% of parents knew about the premarital test. And out of 272 parents, no parent underwent premarital testing. In a study on Premarital Screening of Beta Thalassemia done in Iran, it was revealed that a premarital screening program is helpful for the identification and prevention of high-risk marriages. Detecting carrier couples with premarital screening programs is an effective way of controlling thalassemia major.[17] Thus, the reason that has made this disease a prevalent one is the low level of awareness. Family screening can reduce the incidence of thalassemia to satisfying levels. A study was conducted in the UAE to check the effectiveness of premarital screening. Of the 17,826 individuals screened during the studied period, 4.02% (717) were diagnosed as positive for hemoglobinopathies and 0.31 % (28) couples were declared high-risk.[9]

Moreover, 98% of parents in this study agreed that the premarital test should be taken before cousin marriages and a proper awareness campaign on this disease and prevention should be run in residential areas across the country. The only way thalassemia major can be prevented is by ensuring premarital test/screening. The importance of genetic screening in the prevention of thalassemia is proved by a study done in Delhi, India which showed that out of 10983 patients, 7.07% had one or the other hemoglobinopathies and 5.8% had beta-thalassemia trait. Forty-two "at-risk couples" (both husband and wife with thalassemia trait) were identified which could possibly have a thalassemia-affected child in the future.[18] This clearly reveals that genetic screening before marriage/premarital screening can suggest which couples are at high risk and can have a thalassemia-affected child in the future.

Thalassemia is a growing global public health problem with an estimated 900,000 births of clinically significant thalassemia disorders expected to occur in the next 20 years. Proper awareness and genetic counseling can decrease the number of cases to a large extent.[19]

Thalassemia awareness programs, premarital screening, and legislation are the need of the day.[20]

As the premarital thalassemia screening test is one of the effective ways of preventing thalassemia major, this test should be encouraged especially in cases of cousin marriages and in case of family history for

thalassemia major. All the participants involved in this study were from Rawalpindi, multi-centered research needs to be conducted to get a wider perspective about awareness of thalassemia in parents of affected children, especially in rural areas where illiteracy rates are high.

Conclusion

The majority of parents revealed their unawareness regarding the link between thalassemia and cousin marriages. More than 98% of parents had no awareness regarding any campaign on thalassemia in their areas. None of the parents out of 272 conducted premarital screening and only a few heard about it. Thus, our study highlights the dire need for awareness campaigns and premarital screening in high-risk couples which is almost non-existent in our population.

References

1. Fibach E, Rachmilewitz EA. Pathophysiology and treatment of patients with beta-thalassemia - an update. F1000Res. 2017;6:2156.
2. Sharma DC, Arya A, Kishor P, Woike P, Bindal J. Overview on thalassemias: a review article. Medico Res Chronicles. 2017;4(3):325-37.
3. Modell B, Darlison M. Global epidemiology of haemoglobin disorders and derived service indicators. Bull World Health Organ. 2008;86:480-7.
4. Angastiniotis M, Modell B. Global epidemiology of hemoglobin disorders. Ann N Y Acad Sci. 1998;850(1):251-69.
5. Uchechukwu NM, Oluwafemi A, Anthony NO. Cost and Financial Challenges of Accessing Bone Marrow Transplantation: Opinion Survey in a Nigerian Tertiary Institution. Asian Hematol Res J. 2020:18-26.
6. Mohamed SY. Thalassemia Major: Transplantation or transfusion and chelation. Hematol Oncol Stem Cell Ther. 2017;10(4):290-8.
7. Khan MS, Ahmed M, Khan RA, Mushtaq N, Shah MW. Consanguinity ratio in b-thalassemia major patients in District Bannu. J Pak Med Assoc. 2015 Nov;65(11):1161-3.
8. Ghafoor MB. Level of awareness about thalassemia among parents of thalassaemia children. J Rawalpindi Medical College. 2016;20(3):209-11.
9. Salama RA, Saleh AK. Effectiveness of premarital screening program for thalassemia and sickle cell disorders in Ras Al Khaimah, United Arab Emirates. J Genet Med. 2016;13(1):26-30.
10. Asif N, Hassan K. Management of thalassemia in Pakistan. JIIMDC. 2016;5(4):152-3.
11. Zaheer HA, Waheed U, Abdella YE, Konings F. Thalassemia in Pakistan: A forward-looking solution to a serious health issue. Glob J Transfus Med 2020;5:108-10
12. Zaman Q, Salahuddin M. Association between the education and Thalassaemia: a statistical study. Pak J Stat Oper Res. 2006;103-10.
13. Khan MS, Ahmed M, Khan RA, Mushtaq N, Shah MW. Consanguinity ratio in b-thalassemia major patients in District Bannu. J Pak Med Assoc. 2015;65(11):1161-3.
14. Abu-Shaheen A, Heena H, Nofal A, Abdelmoety DA, Almatary A, Alsheef M, AlFayyad I. Epidemiology of Thalassemia in Gulf Cooperation Council Countries: A Systematic Review. Biomed Res Int. 2020;2020:1509501.
15. Vichinsky EP. Changing patterns of thalassemia worldwide. Ann N Y Acad Sci. 2005;1054(1):18-24.
16. Faizan-ul-Haq, Khan MMA, Yaqoob U, Shaikh JR, Salam O et alEffect of Child Thalassemia on the Mental Health of Their Caregivers. Int J Ment Health Psychiatry. 2017;3(3)
17. Hashemizadeh HA, Noori R. Premarital screening of beta thalassemia minor in north-east of Iran. Iran J Ped Hematol Oncol. 2013;3(1):210.
18. Mendiratta SL, Mittal M, Naaz F, Singh S, Anand S. Role of thalassemia screening in prevention and control of thalassemia-a 5 year experience. Int J Reprod Contracept Obstet Gynecol. 2017;5(9):3107-11.
19. Karimzaei T, Masoudi Q, Shahrakipour M, Navidiyan A, Jamalzae AA, Bamri AZ. Knowledge, attitude and practice of carrier thalassemia marriage volunteer in prevention of major thalassemia. Glob J Health Sci. 2015;7(5):364.
20. Ahmed M, Sharif MS, Yaqoob R, Nadeem MS, Haroon Z, Iqbal T. 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 Physio. 2019;15(2):11-5.