ORIGINAL ARTICLE

Awareness and Acceptance of Premarital Carrier Screening of Thalassemia among Adults

SYED HASHIM ALI INAM[1], HAMZA JAMIL[2], NIMRA KLAIR[3], AFSA NAYYAR[4], NAVEEN SHEIKH[5], ANUM ARIF[6]
[1]*House Officer, CMH Lahore*
[2]*HO, Department of Internal Medicine, Military Hospital, Army Medical College, Rawalpindi, Pakistan.*
[3,4,5]*HO, Department of Internal Medicine, Military Hospital, Army Medical College, Rawalpindi, Pakistan*
[6]*Consultant General Surgeon and Fellow Vascular Surgery CMH Lahore,*
Correspondence to Dr. Anum Arif, Email: dranumarif@yahoo.com

ABSTRACT

Aim: To assess the level of awareness and acceptance amongst the population regarding premarital carrier screening of Thalassemia

Methodology: A Cross-sectional descriptive study was carried out From October, 2020 to January, 2021 directed at the general population of Rawalpindi and Islamabad after taking ethical approval. Self-administered questionnaire was used. Random sampling technique was used. Sample size was 181. The questionnaire was designed to evaluate the awareness and acceptance of general population regarding premarital carrier screening of thalassemia. SPSS version-22 was used for data analysis.

Results: Majority of subjects were aged between 20-30 years. 11.6% of subjects out of 181 had never heard about thalassemia.72% who knew about thalassemia had a patient of thalassemia in their family. Common sources of knowledge on subject of thalassemia were their friends (31.3%) and school (26%). After receiving awareness about thalassemia and its screening, majority (80%) of subjects were willing to get themselves screened out of which 93.7% were under the age of 31 and 95.1% were unmarried.

Conclusion: The results of study show that the proportion of population of Rawalpindi and Islamabad screened for thalassemia trait is alarmingly low. Young and unmarried sections of population were highly receptive, and they were willing to get themselves screened after getting knowledge about thalassemia.

Keywords: Acceptance, Premarital screening, Thalassemia,

INTRODUCTION

Pre-marital carrier screening is the act of identifying defective genes that cause genetic diseases with significant morbidity and mortality in the offspring. Among the list of these lethal genetic disorders, thalassemia tops the list[1]. Thalassemia is worldwide prevalent inherited blood disorder in the world characterized by abnormal production of hemoglobin[2]. There are two major types of thalassemia; alpha thalassemia and beta thalassemia[3]. Betathalassemia major is the homozygous form of thalassemia, characterized by complete absence of hemoglobin A and it is managed by repeated blood transfusions and chelation therapy[3].

Prevalence of Beta-thalassemia patients is on the rise because of rising trends of cousin marriages, birth rate, illiteracy rate, marriages at younger age with unawareness and lack of family planning; out of these, consanguinity is the most important factor responsible for this rising trend[4]. Fortunately, with the introduction of modern DNA testing techniques, it seems feasible to tackle these rising trends by carrier screening programs[1]. Thalassemia screening involves Complete Blood Counts (CBC) combined with Single Tube Osmotic Fragility Test (SOFT), and further confirmation by Hemoglobin Electrophoresis; chorionic villous sampling is also applied[5].

A number of countries have started screening programs aiming to decrease the incidence of thalassemia[6]. A research conducted in USA revealed a rising prevalence of thalassemia in USA with approximately 7.5% increase over the last five decades[7]. Newborn screening (NBS) for hemoglobinopathies was started in California to facilitate timely identification of affected individuals, and this program when linked to proper follow up, education and awareness programs proved to reduce the mortality of thalassemia in California[8]. In China, India, Thailand and Malaysia such screening programs have also been started which includes antenatal care (ANC) services or family planning programs that educate and encourage the couples to get screened for thalassemia during early months of pregnancy.[9] Canada and Australia are also providing education about genetic testing and screening in school going children leading to a significant reduction in new thalassemia cases[10].

According to studies conducted in Pakistan, there are approximately 9.8 million carriers of Beta thalassemia among the Pakistani population and approximately 5000 children are diagnosed each year with beta thalassemia in Pakistan[11]. Approximately 6% of the Pakistani population carries the beta thalassemia gene. PND (prenatal diagnosis) program was introduced in Pakistan in the year 1994 and it is available only in big urban centers of Karachi, Lahore and Multan[12]. Unfortunately, in these 25 years, the service has been availed lesser than 3000 times which tells us 80-90% of the carrier couples do not undergo this testing in Pakistan[12]. Recently, Punjab, Sindh and Khyber Pakhtunkhwa provincial assembly have also passed the bill regarding pre-marital screening of thalassemia but practically little work has been done on this issue[13]. Causes for few couples opting for this facility are attributed to lack of awareness, difficult access, religious beliefs and high cost[14].

Received on 28-09-2020
Accepted on 03-01-2021

The aim of this study is to determine awareness and acceptance in general population of Rawalpindi and Islamabad regarding premarital carrier screening for beta thalassemia and their approach towards the preventative strategies available.

METHODOLOGY

This cross-sectional descriptive study was conducted from October 2020 to January 2021 among the general population of Islamabad and Rawalpindi after taking ethical approval from the Institutional Review Board of Army Medical College. Random sampling technique was applied. The study was initiated in October and discontinued in January, so data from only 181 subjects was collected in this duration of 4 months. All those with age greater than 20 years of age and above were included in the study after taking an informed and written consent. The participants were approached in their houses. Individuals with debilitating disease rendering them unable to communicate effectively and those who did not gave consent to fill the questionnaire were excluded.

Data collection was done using a self-designed questionnaire. In addition to the consent form and baseline demographic information, the questions covered the domains of knowledge about thalassemia; personal and family history of thalassemia; knowledge of its etiology, diagnosis, treatment and prognosis; as well as the acceptance of premarital carrier screening. Only those who positively responded to the first question: 'Have you ever heard of thalassemia?' were eligible to answer the subsequent questions. The questionnaire was translated into Urdu. Data was analyzed using SPSS version 22.

RESULTS

181 random subjects from the general population of Islamabad and Rawalpindi were approached. Out of the 181 subjects 90(49.4%) were 20-30 years of age. 64(35.6%) were 31-45 years of age. 25(13.9%) were 46-60 years of age and 2(1.1%) were 61 and above. 145(80%) of the subjects were male and 36(20%) were females. 67/181(37%) were unmarried, 111/181(61%) were married and 3/181(2%) were divorced.88.7% of those approached with the questionnaire had heard of the disease thalassemia, whereas, 11.3% were completely unaware of the term. 21/181(11.6%) people were excluded from answering the rest of the questionnaire because they had never heard of thalassemia. Hence only 160/181(88.4%) were able to answer the rest of the questions on the questionnaire.

Those who had heard about thalassemia were asked for the source they had heard it from. The most common source, 'friends', was given by 50(31.3%) of them, second common being 'school' was answered by 42 participants (26.3%). 28(17.5%) said they had heard from someone in the family. 17(10.6%) knew a patient with thalassemia. Family doctor was the source for 13(8.1%) participants whilst media was the source for 10(6.3%). In order to assess their understanding of the disease, they were asked if they thought blood was deficient in thalassemia. 144(90%) answered yes, 12(7.5%) answered no while 4(2.5%) did not know the answer. In order to assess their knowledge about the etiology of thalassemia, they were asked what kind of disease they thought it was. 138(86.3%) knew it was hereditary. 15(9.4%) thought it was a contagious disease. 6(3.8%) thought it's transmitted sexually while only 1(0.6%) chose allergy as the answer. To assess further their understanding about the hereditary details of thalassemia they were asked if they knew the difference between thalassemia major and minor. 88(55%) said they do not know only 72(45%) said they knew. To gauge deeper their understanding of the hereditary pattern of disease, they were asked if they thought there was a chance of a diseased child if one of the parents had thalassemia minor, 114(71.3%) said yes, 29(18.1%) said no while 17(10.6%) said they did not know. Next, they were asked if they thought there will be increased chances of a diseased child if both parents were thalassemia minor. 128(80%) said yes, 20(12.5%) said no and 12(7.5%) said they did not know. Moving on to assess their idea about the treatment, 91(56.9%) said blood transfusion, 14(8.8%) thought surgery was the treatment option, 10(6.3%) said it could be treated with drugs, 40(25%) said all three treatment options were correct. However, 5(3.1%) said thalassemia is untreatable.

When asked about the health status of a patient having thalassemia disease, 25(15.6%) said yes about the patients showing dangerous clinical symptoms. 113(70.6%) said they don't whereas 22(13.8%) said they did not know. Then they were asked about their idea about the health status of a thalassemia minor person. 56(35%) responded they leave a normal life, 60(37.5%) require blood transfusion. 29(18.1%) thought it could convert into a clinical disease. 12(7.5%) said it could be infectious. 3(1.9%) said they do not know. In order to evaluate their knowledge about the severity of the disease, they were questioned if they knew what happens if thalassemia is left untreated. 145(90.6%) participants said it would worsen, 3(1.9%) said the patient would recover. 8(5%) participants said without treatment there would be no effect on the disease. 4 (2.5%) said they do not know the effects of not treating the patient of thalassemia. They were also assessed for their understanding of the effectiveness of thalassemia treatment. 101(63.1%) of those asked said the patients lead a normal life with treatment. 35(21.9%) said they do not lead a normal life whereas 24(15%) did not know the effect of treatment of the quality of life of the patient. Upon asking subjects what in their opinion was the ideal time for thalassemia screening, 121(75.6%) participants said birth, 31(19.4%) said adolescence whereas 8(5.0%) thought after marriage was the best time. The subjects were asked if they knew thalassemia trait could be tested by a special blood test. 88(55.0%) said yes, 41(25.6%) said no and 30(18.8%) said they did not know.The testing status of subjects was gathered as follows: Only 13(8.1%) said yes and 147(91.9%) said they had never been tested. It was important to know the reasons behind why such a mass majority of people were not screened. When asked, 87(54.4%) did not answer the question. 73(45.6%) said they did not have any signs or symptoms so they never thought of getting themselves screened. Lastly, to evaluate how willing are people about getting themselves tested after they have been given awareness about the disease, 128(80%) said they would.

32(20%) said they still would not. 128(80%) of those surveyed who said they are willing to get screened after getting awareness about thalassemia

DISCUSSION

We concluded in our study that majority of Pakistani population has heard about Beta Thalassemia but the knowledge regarding this disease is low and the proportion of Pakistani population screened for thalassemia trait is alarmingly low. This is a major reason why the proportion of the population shows falling trends of awareness and pre-marital screening for thalassemia. Majority of our population knew that thalassemia is a serious hereditary disease with significant morbidity and mortality and that it runs in the family and is treated by blood transfusions, but they had no idea about the signs and symptoms of thalassemia. Knowledge about health status and quality of life of thalassemia patients was also very low.

A similar study was conducted in United Arab Emirates that showed more than 63% of the sample population had good to excellent knowledge about thalassemia[5]. A study conducted in Thailand also showed higher awareness levels in the study population (79.6%)[15]. Another study conducted on high school students in Southeastern Iran indicated that only 14.7% students had good knowledge towards thalassemia[16]. Population of Middle East had a very good knowledge and attitude towards the thalassemia; these results can be attributed to the fact that most common genetic disorder in Middle East is thalassemia[5]. Our results are consistent with another research conducted in Pakistan that showed that only 54.5% subjects had heard of thalassemia, with a lower mean knowledge score (13.0 ± 4.4 out of 27 questions)[18].

Another aspect of our study was assessing attitude and acceptance of our population towards thalassemia screening. A study conducted in Saudi Arabia indicated 10% reduction in at-risk marriage rates after proper counselling was done[17]. Similar study in Jordan shows 40% reduction in at-risk marriage rates after awareness programs were conducted[17]. We can anticipate similar results in Pakistan from the efforts of Pakistani authorities through powerful awareness campaigns.

In our study, young and unmarried sections of the population were highly receptive, and were willing to get screened after getting knowledge about thalassemia indicating that the limiting factor in widespread thalassemia screening is not the attitude of the population but the level of awareness.

Limitations of study: The limitations in the study were that majority (80%) of the participants were males as mostly males came to the doors of their houses when approached and we were only able to target few areas of both cities.

CONCLUSION

Pakistani population despite having low background knowledge of beta thalassemia has a positive attitude towards premarital screening. This highlights the need of a nation-wide awareness campaign and implementation of screening programs to young adults at an earlier stage so that they can address this issue in the early stages of marriage process. A multi-disciplinary research involving multiple institutions on a much greater scale can expand our recommendations.

Acknowledgement: We would like to thank the medical students from Army Medical College and Rawalpindi Medical College who helped us in data collection.

Conflict of Interest and Financial disclosure: Nil.

REFERENCES

1. Frömmel C. Newborn Screening for Sickle Cell Disease and Other Hemoglobinopathies: A Short Review on Classical Laboratory Methods—Isoelectric Focusing, HPLC, and Capillary Electrophoresis. International Journal of Neonatal Screening. 2018 Dec;4(4):39.
2. Kumar S, Singh D, Garg A. An epidemiological study on the clinico-hematological profile of pediatric patients with congenital hemolytic anemia. International Journal of Contemporary Pediatrics. 2017 Feb 22;4(2):374-7.
3. Rund D. Thalassemia 2016: Modern medicine battles an ancient disease. American journal of hematology. 2016 Jan;91(1):15-21.
4. Asif N, Hassan K. Management of thalassemia in Pakistan. Journal of Islamabad Medical & Dental College. 2016;5(4):152-3.
5. Rabbani SA, Ahmed A, Abdulkader B, Suliman S. Evaluation of Thalassemia awareness among the future healthcare providers of UAE. Nursing. 2017 Feb 1;32:16.
6. Wongprachum K, Sanchaisuriya K, Vidamaly V, Sorpasirth S, Dethvongphanh M, Norcharoen B, Htalongsengchan B, Fucharoen G, Fucharoen S, Schelp FP, Saowakontha S. Pilot screening program for thalassemia in a country with limited resources: a collaboration model between close neighboring countries. The Southeast Asian journal of tropical medicine and public health. 2016 Sep 1;47(5):1040-7.
7. Michlitsch J, Azimi M, Hoppe C, Walters MC, Lubin B, Lorey F, Vichinsky E. Newborn screening for hemoglobinopathies in California. Pediatric blood & cancer. 2009 Apr;52(4):486-90.
8. Fucharoen S, Weatherall DJ. Progress toward the control and management of the thalassemias. Hematology/Oncology Clinics. 2016 Apr 1;30(2):359-71.
9. Crighton G, Wood E, Scarborough R, Ho PJ, Bowden D. Haemoglobin disorders in Australia: where are we now and where will we be in the future?. Internal medicine journal. 2016 Jul;46(7):770-9..
10. Hussain Z, Ansari S. Pre-Marital Screening for Thalassaemia–Legal, Social and Technical Obstacles.
11. Vichinsky EP. Changing patterns of thalassemia worldwide. Annals of the New York Academy of Sciences. 2005 Nov 1;1054(1):18-24.
12. Dehkordi AH, Heydarnejad MS. Enhancement of parents' awareness about ß-thalassemia major disorder through two educational programs.
13. Provinces Asked to Implement Thalassaemia Law [Internet]. 2015 [cited 15/5/2015]. Available : https://www.dawn.com/news/1180896
14. Ansari SH and Shamsi TS. Thalassaemia Prevention Programme. Hematology updates 2010:23-28
15. Xu JZ, Tanongsaksakul W, Suksangpleng T, Ekwattanakit S, Riolueang S, Kaiser BN, Thielman NM, Telen MJ, Viprakasit V. Knowledge, Cultural, and Structural Barriers to Thalassemia Screening in Migrant Populations in Thailand.
16. Miri-Moghaddam E, Motaharitabar E, Erfannia L, Dashipour A, Houshvar M. High school knowledge and attitudes towards thalassemia in southeastern Iran. International journal of hematology-oncology and stem cell research. 2014;8(1):24.
17. Alswaidi FM, O'brien SJ. Premarital screening programmes for haemoglobinopathies, HIV and hepatitis viruses: review and factors affecting their success. Journal of medical screening. 2009 Mar;16(1):22-8.
18. Mirza A, Ghani A, Pal A, Sami A, Hannan S, Ashraf Z, Iqbal S, Malik UZ, Hayat U, Fatmi Z. Thalassemia and premarital screening: potential for implementation of a screening program among young people in Pakistan. Hemoglobin. 2013 Apr 1;37(2):160-70.