DOI: https://doi.org/10.53350/pjmhs2023176253

ORIGINAL ARTICLE

Family Screening of Thalassemic Patients and Effectiveness of Education in Providing Knowledge about Prevention of Thalassemia

RAMEEZ IQBAL[1], SIDRAH[2], ASFAHAN AKHTAR[3], KIRAN MEMON[4], MUHAMMAD RAHIL KHAN[5], MUHAMMAD ANIQUE[6], KIRAN AAMIR[7], AAMIR RAMZAN[8]

[1]*Lecturer, Pathology Department Liaquat university of Medical and health sciences jamshoro Hyderabad*
[2]*Lecturer, Pathology department, Bilawal Medical College for boys , LUMHS Jamshoro Hyderabad.*
[3]*Senior Registrar, Isra university hospital Hyderabad*
[4]*Assistant Professor pathology Department, Indus medical college, Tando Muhammad khan*
[5]*Assistant Professor, Pathology Department, Liaquat university of Medical and health sciences jamshoro Hyderabad*
[6]*Lecturer, Pathology, Department, Bhitai Dental and Medical College Mirpurkhas*
[7]*Associate professor, Pathology Department, Liaquat university of Medical and health sciences jamshoro Hyderabad*
[8]*Lecturer in pathology Department, Liaquat university of Medical and health sciences jamshoro Hyderabad*
Corresponding author: Rameez Iqbal, Email: rameezmemon30@yahoo.com, Cell: 03322056030

ABSTRACT

Objective: To identify the carriers in the families of Beta Thalassemia patients and to determine the effectiveness of education about the prevention of thalassemia**.**

Methods: The Observational - Cross-Sectional study was carried out in 6 months from 01-09-2021 to 31-03-2022 on a sample of 73 thalassemia patients and their families, after taking informed written consent. This study was conducted via non-probability, convenient sampling and was carried out at the Department of Pathology & Diagnostic and Research Laboratory - LUMHS Hyderabad/Jamshoro. The data was analyzed via SPSS 21.0

Results: The mean age of patients was found to be 17.34 + 7.32 years. 52% of participants were males with mean age of 15.23 + 9.67 years and 48% participants were females with mean age of 13.48 + 5.7 years. Most of the patients were having fortnightly blood transfusion i.e. 42.8% followed by 32.9% patients with monthly transfusion. Bi-monthly transfusion were found to be least reported. Consanguineous marriage among parents was found in 87.70% of patients. Hb electrophoresis pattern among siblings of thalassemia patients showed 4.1% occurrence of thalassemia major and 32.8% occurrence of thalassemia minor while 63% patients were found to be having normal electrophoresis pattern. The mean hemoglobin was found to be 09.23 + 3.65 mg/dl among siblings of thalassemia patients with 63.86 + 8.41 mg/dl mean MCV.

Conclusion: The study found out that the around 2/3rd of the families were having positive history of thalassemia with around 1/4th of the patients had history of blood transfusion. The educational session was found to effective in improving the knowledge regarding the thalassemia.

Keywords: Thalassemia, carriers, family history, effectiveness of educational awareness

INTRODUCTION

Thalassemia is one of the most common inherited blood disorder worldwide.[1] It has been estimated that 5.2% of the population worldwide and more than 7 % of pregnant females, carry significant variation in hemoglobin chain. 1.1% of couples are at risk for having children with a hemoglobin disorder, and there are 2.7 per 1000 conceptions are affected with the same disorder worldwide [2]. Pakistan has a population of nearly 200 million and approximately 5% of the people carry the gene for β- thalassemia.[3]

Thalassemia was first illustrated by Cooley and lee in 1925, [4] Thalassemia is not only an important public health problem but also a socioeconomic issue in many countries in the region.[5] "It is a genetic disorder which is passed from parents to their off-spring, [6] the disease originates from genetic mutations in the gene responsible for globin chain biosynthesis. Clinically beta thalassemia is classified according to their severity in to three types, thalassemia major requiring a regular blood transfusion throughout life, thalassemia intermediate characterized by anemia but not of such severity as to require regular blood transfusion, and thalassemia minor which is the symptomless carrier status.[6] It's an inherited disorder lead to decreased and defective production of hemoglobin, a molecule found inside all Red blood cells.[7] The pathophysiology of severe thalassemia involves ineffective erythropoiesis, acute hemolytic and chronic anemia, failure to thrive and iron overloading". [8]

According to the International Classification of Diseases 11 Revision (ICD-11) published by the World Health Organization (WHO) in 2018, "thalassemia is included in the 3A50 subcategory of erythrocytes disorder and blood formation disorder. Some countries across the Mediterranean to South East Asia have a high prevalence of the beta thalassemia, including Italy, Greece, Turkey, Arab, Iran, Iraq, Indonesia etc. [9] As the thalassemia is categorized into three types; major thalassemia, intermedia, and minor thalassemia/carrier, the major and intermedia thalassemia patients need treatment in form of lifetime blood transfusion. They tend to show symptoms such as anemia, delayed growth and development, yellow skin, bloated stomach, and so on. [10] On the other hand, carrier individuals often show little or no symptoms at all (asymptomatic) but only experience mild anemia. [11]

Thalassemia major causes serious health, social and economic problems for patients and their families as well as requiring treatments that place a large enough financial burden on public health budgets. [12] As severe thalassemia affects both physical and mental health, it causes significant distress in patients and their parents; therefore, prevention and control of severe thalassemia are important.[13]

Thalassemia is a disease, the cure is which not widely and easily accessible. Prevention is the only way to reduce its burden. The most effective approach to reduce the burden on the society and reduce the disease incidence through implementation carrier screening program, offering genetic counseling and prenatal diagnosis.[6, 7] Education is one of the thalassemia prevention strategies that have been established upon internationally. Knowledge of how disease is inherited, early symptoms and prevention of thalassemia should be delivered through an attractive and acceptable medium.[14]

Some previous studies shows that siblings identified as β-thalassemia carriers is higher as oppose to carriers in the general population i.e. 62.2% vs 5 to 8% respectively therefore it is practical to focus on siblings of identified thalassemia patients when both resources and budgets are limited".[15]

Aside from the financial impact experienced by patients, families, and the nation, this genetic disease is also associated with mental disorders such as "depression, which is often comorbid with chronic illness and this condition might worsen the individual's general health (Moussavi et al., 2007). [16] The psychological manifestation experienced by teenagers with thalassemia (both major/intermedia and carrier) are depression and anxiety. Depression is very likely to occur in carrier.

Participating in a screening test is a form of health behavior, which is an effort voluntarily made by individuals with little to no symptoms of diseases because they believe their action could prevent them from getting sick or promote their health (Kasl & Cobb, 1966). [17] The health behavior in which a person takes preventive action is called as preventive health behavior (PHB). One of the models that could explain and determine an individual's PHB is called the Health Belief Model (HBM), a cognitive model most commonly used to explain a preventive behavior in medical aspects (Kirscht, 1983). [18] Components which build HBM are the main concepts that can predict someone's intention to perform preventive behavior, to do screening, or to take control over a disease (Champion & Skinner, 2008). [19] They are different than the usual behavior components as they aim to predict the tendency of a specific behavior (Kirscht, 1983). [18] Those components are susceptibility, seriousness, benefits and barriers of the behavior, and cues to action individuals and their physical health might worsen with the onset of depression. [11] Until today, there has not been any medication or treatment that could cure this genetic mutation disease, although a preventive action could be taken by avoiding marriage between carrier individuals and through thalassemia blood-test screening. The prevalence, statistic, severity level, and impact caused by this disease are the reasons why it is important to promote thalassemia screening to society".

MATERIALS AND METHODS

The Observational - Cross-Sectional study was carried out in 6 months from 01-09-2021 to 31-03-2022 on a sample of 73 patients. All male and female patients with a confirmed b-thalassemia diagnosis were considered. Excluded were those with other forms of hemoglobinopathy and those who declined to give written consent.

Each patient had a sample of their blood drawn for analysis. After proper aseptic precautions have been taken, a sample of three milliliters of blood will be obtained using a five-centiliter (cc) disposable syringe and placed in a purple-topped tube containing ethylene diamine tetra acetic acid (EDTA). The Sysmex XN1000 6-part fully-automatic analyzer from Japan will be used to get a comprehensive blood count. The High Performance Liquid Chromatography (HPLC) system will be used to conduct the electrophoresis.

A complete blood count was performed on the EDTA-treated samples using a Sysmex XN1000i Japan 6-part differential automated hematology analyzer.

Microsoft Excel 2016 and SPSS 21.0 were used to analyze the data. No &% symbols were used to denote qualitative information. Quantitative information was written as a mean and standard deviation (X SD) formula. Knowledge adequacy was compared before and after the educational session using a Chi-Square test. Statistical significance will be assumed at a p-value of 0.05.

RESULTS

The mean age of patients was found to be 17.34 + 7.32 years. 52% of participants were males with mean age of 15.23 + 9.67 years and 48% participants were females with mean age of 13.48 + 5.7 years.(Table 1)

Table-1: Gender and age of the cases

Variables	Frequency (73)	Percentage
Mean age (years)	17.34 + 7.32	
Gender		
Male	38	52
Female	35	48

Most of the patients were having fortnightly blood transfusion i.e. 42.8% followed by 32.9% patients with monthly transfusion. Bi-monthly transfusion were found to be least reported.(figure 1)

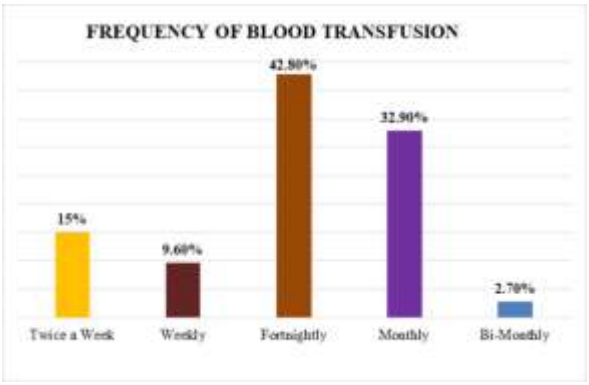

Figure-1: Association of blood transfusion

Consanguineous marriage among parents was found in 87.70% of sample.(figure 2)

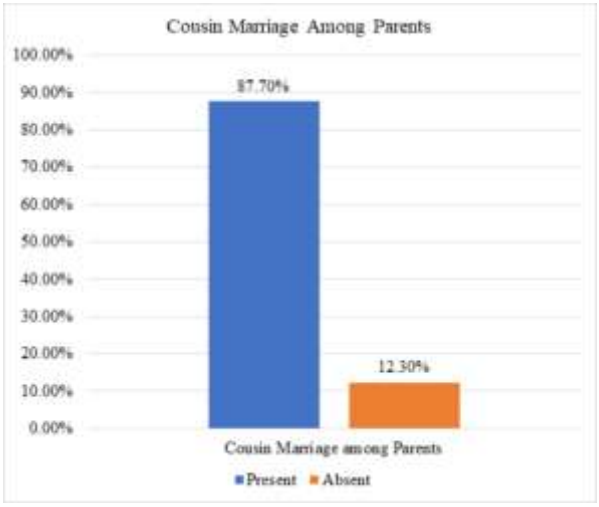

Figure-2: Frequency of cousin marriage

Hb electrophoresis pattern among siblings of thalassemia patients showed 4.1% occurrence of thalassemia major and 32.8% occurrence of thalassemia minor while 63% patients were found to be having normal electrophoresis pattern. (figure 3)

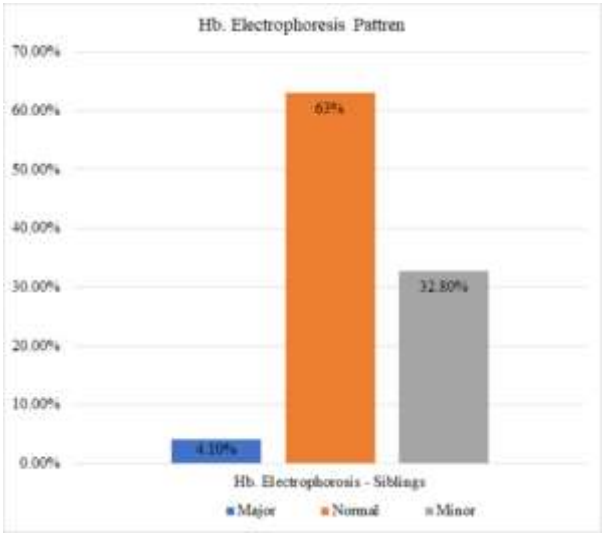

Figure-3: Type of thalassemia (Electrophoresis Pattern)

Table 2: The mean hemoglobin was found to be 09.23 + 3.65 g/dl among siblings of thalassemia patients with 63.86 + 8.41 g/dl mean MCV.(table 2)

Parameter	Mean Values		
Hemoglobin	09.23 + 3.65 g/dl		
MCV	63.86 + 8.41 fl		
Hemoglobin A1	84.5% + 3.2%		
	Major	Normal	Minor
	65% + 2.7%	97.3% + 1.8%	94.8% + 6.3%
Hemoglobin A2	5.8% + 0.4%		
	Major	Normal	Minor
	3.2% + 1.1%	2.7% + 1.6%	5.1% + 0.9%
Hemoglobin F	3.2% + 0.1%		
	Major	Normal	Minor
	32.5% + 2.5%	0.4% + 0.01%	1.2% + 0.8%

DISCUSSION

Thalassemia is an autosomal recessive genetic disease that causes the disturbance of the formation of red blood cells in the patient's body. [19] This condition requires the patient to undergo routine and long-life treatment such as routine blood transfusion and the administration of iron chelation to prevent complications due to the toxic excess of iron in the blood. [20] Preventive measure is essential in determining the possibility of any genetic disorder. Genetic screening in pregnancy is considered very important to be done for any couple with the possession of thalassemia genes. indeed, awareness to genetic screening requires a good understanding of thalassemia. [21]

This study shows that most respondents have poor knowledge. Studies varies in regard to the level of knowledge among teenagers. A study from Iran revealed the same results with less than 20% of students have a good level of understanding about thalassemia. [22] While, other studies from India shows much better results with most respondents have good understanding. [23,24]

Many studies showed variations in regard to the gender in understanding and knowledge related to thalassemia. Research in Iran states that female students who have good knowledge of thalassemia are much more than male students. Study by Basu revealed different results by stating male respondents have better knowledge than women. [24] However, this study does not account any variation on the basis of gender differences.

The general knowledge of respondents about thalassemia is good enough on several topics, namely pathophysiology, symptoms and treatment of thalassemia. Several other topics such as definition, etiology, onset and prevention are still poorly understood by respondents. The topic of aetiology needs to be emphasized on health promotion related to thalassemia because most respondents consider bacteria and unhealthy lifestyles can cause thalassemia. A lack of understanding of the role of genetics in the thalassemia aetiology has an impact on respondents' understanding of the topic of thalassemia prevention. Some of respondents do not know the role of genetic screening as a preventive measure for thalassemia. It is seen that the majority of respondents choose to maintain their diet and have a healthy lifestyle that can prevent the addition of new thalassemia patients.

Respondents also have lack of understanding on the topic of thalassemia onset. Most respondents answered thalassemia suffered since birth, but more than half of respondents assumed that thalassemia could be suffered by someone who was previously normal. Although most respondents understand that blood transfusion is the treatment of thalassemia, quite a number of respondents chose the dialysis answer for that question. Dialysis or hemodialysis is routine treatment for patients with end-stage renal failure and is not associated with thalassemia. [25]

Most respondents said they had never heard information about thalassemia before, while almost one third stated they had heard. The number of respondents who knew of thalassemia earlier in this study was much lower compared to previous studies in several different countries. Research by Ebrahim et al. revealed that 53% of respondents had heard information about thalassemia before. [26] Balcin revealed that more than half of the respondents (57.7%) had heard of thalassemia before, while research in India had a much higher number, as many as 85% of respondents had heard the term disease. [27,28]

The low awareness of the term thalassemia in this study shows that thalassemia is a disease that has not been widely discussed in society, but its prevalence is quite high. Indonesia is one of the thalassemia belt countries which has a relatively high number of thalassemia sufferers. Management of thalassemia which requires lifelong treatment can be a psychological and financial burden on families and countries. Preventive program such as genetic counseling is one step that can be taken by the country to reduce the number of thalassemia cases. [83, 84] People awareness on thalassemia as a genetic disease has massive impact to succession the thalassemia preventive program up to 40% of overall program besides increasing public awareness genetic counseling (33%) and premarital screening (27%). [28]

A person who have a thalassemia gene (carrier thalassemia) are often not known before laboratory tests because they do not experience severe symptoms like thalassemia major patients. Anemia is a symptom that can be found in thalassemia carriers and is commonly known by the public. [28] Knowledge of thalassemia including its severity, causes and symptoms can be the first step to increase public awareness, especially genetic screening students to confirm the thalassemia gene itself. Sources of information selected by respondents to gain knowledge about thalassemia are internet, school, health workers, seminars, relatives/friends and print media. School health programs which consist of regular counseling and seminars that include all majors in high school about thalassemia and other genetic diseases can be a step of thalassemia prevention programs in schools. Seminars in general can be delivered through conventional methods such as lectures or through group discussion methods that involve extension respondents in the process of delivering information. Group discussion is more effective because it can increase students' understanding of new information by up to 50% compared to lecture methods which only produce student understanding of 20%.[25] Thalassemia education as prevention program should be done as soon as possible to teenagers because education is a major factor in increasing knowledge about thalassemia. [24]

CONCLUSION

The study found out that the around 2/3rd of the families were having positive history of thalassemia with around 1/4th of the patients had history of blood transfusion. The educational session was found to effective in improving the knowledge regarding the thalassemia.

REFERENCE

1. Tanveer T, Masud H, Butt ZA."Are people getting quality thalassemia care in twin cities of Pakistan? A comparison with international standards''. Int J Qual Healthcare; 2018; 19:1-7.
2. Wolter K, Medknow. "Antenatal carrier screening for thalassemia and related hemoglobinopathies: Hospital based studies ". J Manag Sci2018; 32(2): 118-121.
3. Ahmed S, Wazir ZJ, et al. "Clinical and Haematological Picture of Multi-Transfused Thalassaemia Major Patients at a Center in Pakistan". J. Islam. Int. Med. Coll 2018; 13(2): 52-56.
4. Ahmed I, Sadiq MA, et al. "impaired glucose tolerance in children with blood transfusion dependant beta thalassemia". Pak Med J 2018;25(9):1402-1405.
5. Kia NS, karami k et al. " Evaluation of an educational intervention based on health belief model on beta thalassemia carrier and final suspect couples". J Educ Health Promot 2018 ; 7:77.
6. Rasid KJ. "Epidemiological Characteristics and Family Relatives among Thalassemic Patients in Sulaimani City, Kurdistan Region, Iraq". Kurd. j. appl. res 2018; 10(2): 62-65.
7. Biswas B, Naskar NN, Dasgupta A ,et al. "Knowledge of the Caregivers of Thalassemic Children Regarding Thalassemia: A Cross-sectional Study in a Tertiary Care Health Facility of Eastern India". Iraqi j. hematol 2018 7(2): 49-53.
8. Rerkwattavorn C, Sirachainan N, Songdej D, et al. " Preventable Severe Thalassemia among Children". Int. j. health sci. res. 2018 ; 1-6.

9. Rathod, D. A., Kaur, A., Patel, V., Patel, K., Kabrawala, R., Patel, V., Patel, M., & Shah, P. (2007). Usefulness of cell counter-based parameters and formulas in detection of ßthalassemia trait in areas of high prevalence. American Journal of Clinical Pathology, 128(4), 585–589.
10. Kementerian Kesehatan Republik Indonesia. (2017). Pedoman pengendalian penyakit thalassemia di fasilitas kesehatan tingkat pertama. Jakarta: Kementerian Kesehatan Republik Indonesia.
11. Keskek, S. Ö., Kirim, S., Turhan, A., & Turhan, F. G. (2013). Depression in subjects with beta-thalassemia minor. Annals of Hematology,92, 1611-1615. doi: 10.1007/s00277-013-1851-9
12. Rakhmilla LE, Susanah S, Rohmawaty E, Effendi SH, "Effectiveness of an Educational Intervention in Providing Knowledge about the Prevention of Thalassemia: An Effort to Reduce New Thalassemia Cases". AJE 2018; 11(2): 60-63
13. Cheng K, Funcharoen S et al. "Effect of health education in severe thalassemia prevention and control in communities in camboelia." Archieves of Public health 2018;76(13):1-8.
14. Rakhmilla LE, Susanah S, Rohmawaty E, Effendi SH. Effectiveness of an educational intervention in providing knowledge about the prevention of thalassemia: an effort to reduce new thalassemia cases. Asian J Epidemiol. 2018;11(2):59-64.
15. Sahiratmadja E, Maskoen AM, Effendy DS, Panigoro R. Exploring the Willingness for Carrier Screening among Extended Family Members of a Thalassaemia Carrier Individual: A Lesson Learned. Journal of Biomedical and Clinical Sciences (JBCS). 2018 Feb 2;2(2):8-10.
16. Moussavi, S., Chatterji, S., Verdes, E.,Tandon, A., Patel, V., & Ustun, B. (2007). Depression, chronic diseases, and decrements in health: Results from the World Health Surveys. Lancet, 370(9590), 851-858. doi:10.1016/S0140-6736(07)61415-9
17. Kasl, S. V., & Cobb, S. (1966). Health behavior, illness behavior, and sick role behavior. I. Health and illness behavior. Archives of Environmental Health, 12(2), 246–266.
18. Kirscht, J. P. (1983). Preventive health behavior: A review of research and issues. Health Psychology, 2(3), 277-300
19. Champion, V. L., & Skinner, C. S. (2008). The health belief model. In K. Glanz, B. K. Rimer, & K. Viswanath (Eds.), Health behavior and health education: Theory, research, and practice (p. 45-65). San Francisco, CA: Jossey-Bass.
20. Marcdante KJ, Kliegman RM. 2015. Nelson Essentials of Pediatrics (7th edition). Philadelphia: Elsevier. pp148-518.]
21. Rund D. Thalassemia: Modern Medicine Battles an Ancient Disease. American Journal of Hematology, 2016;91(1):15-21. doi.org/10.1002/ajh.24231.
22. Dewanto JB, Tansah H, Dewi SP, Napitu H, Panigoro R, Sahiratmadja E. Increased Knowledge of Thalassemia Promotes Early Carrier Status Examination among Medical Students. Universa Medicina, 2016;34(3):220. doi.org/10.18051/UnivMed.2015.v34.220-228.
23. Miri-Moghaddam E, Motaharitabar EM, Erfannia E, Dashipour A, Oushvar M. High School Knowledge and Attitudes towards Thalassemia in Southeastern Iran. International Journal of Hematology-Oncology and Stem Cell Research, 2014;8(1):24-30. www.ncbi.nlm.nih.gov/pmc/articles/PMC3913153/#__ffn_sectitle.
24. Palash D, Srijana M, Kamalika D. Knowledge and Attitude Regarding Premarital Screening of Beta Thalassemia among The Students of Selected College of South-24-Parganas, West Bengal, India. India Journal Research, 2018;7(10):24-26. doi.org/10.36106/paripex.
25. Basu M. A Study on Knowledge, Attitude, and Practice about Thalassemia among General Population in Outpatient Department at A Tertiary Care Hospital of Kolkata. Journal of Preventive Medicine and Holistic Health, 2015;1(1):5-12. www.researchgate.net/publication/282657666
26. Kasper LD, Fauci AS, Longo DL, Braunwald E, Hauser SL, Jameson JL. Harrison's Principles of Internal Medicine (19th edition). 2015. United State: Mc Graw Hill. Pp 598-600.
27. Ebrahim S, Raza AZ, Hussain M, Khan A, Kumari L, Rasheed R, et al. Knowledge and Beliefs regarding Thalassemia in an Urban Population. Cureus, 2019; 11(7). doi.org/10.7759/cureus.5268.
28. Balcin YI, Ergin A, Polat A, Atilgan T, Uzun U, Koyuncu H. Thalassemia Premarital Screening Program: Public View. What has been Done, What Needs to be Done?. International Journal of Hematology and Oncology, 2014;24(4):247-252. doi.org/10.4999/uhod.14569.