RESEARCH ARTICLE

EPIDEMIOLOGICAL FACTORS OF THALASSEMIA IN PATIENTS REPORTING TO THE THALASSEMIA CENTRE IN ISLAMABAD

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ABSTRACT

Objective: *To identify the epidemiological factors of thalassemia in patients reporting to the thalassemia center of Islamabad.*

Design: *Descriptive cross-sectional study.*

Place and Duration: *The study was carried out in the thalassemia center of PIMS, Islamabad, from February to November 2022.*

Materials and Methods: *A descriptive cross-sectional study was carried out on 200 patients reporting to the thalassemia center of PIMS Islamabad, with the help of a self-structured questionnaire. The sampling technique used was non-probability convenience sampling.*

Results: *In this study, 74% of patients were found to be the products of cousin marriage and 77% had a trend of consanguinity in their families. The most common blood group was B+ (36%), followed by O+ (28%). Awareness about risk factors of thalassemia was unknown to 42% of the parents. While 91% of parents had no knowledge about prenatal and premarital screening before the conception of the affected child.*

Conclusion: *Consanguinity and lack of awareness about the disease were the two major risk factors of thalassemia. These risk factors are inescapable due to low socioeconomic position and lack of information about premarital and prenatal screening.*

Keywords: *Consanguinity, Risk Factor, Screening, Thalassemia*

INTRODUCTION

Thalassemia is a set of hereditary hemoglobinopathies caused by abnormalities in the autosomal recessive inheritance of the globin chain of hemoglobin. Unbalance in formation of the globin chain impairs the generation of healthy hemoglobin A and reduces erythropoiesis.[1] Various kinds of this illness exist, the most common among these variants are alpha and beta.[2] In Pakistan, ß- thalassemia is one of the most prevalent hemoglobinopathies with 5.4% carrier frequency. Between 1.5% and 7.5% of people have ß-thalassemia. The Khyber Pakhtunkhwa (KP) province, which is close to the Afghan border, the southern areas of the country, and the region along the coast of the Arabian Sea all have high rates of disease.[3]

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Approximately, 1.5% of the total world population, are thalassemia carriers. In Southeast Asia, rates of thalassemia carriers are comparatively high as well as it is prevalent in most parts of the region. Historically, the prevalence of thalassemia in the Middle East has been high due to the higher thalassemia carrier rate as well as the inclination for consanguineous marriages. However, numerous nations, in the past decade, have introduced and implemented preventive measures thus diminishing the prevalence of thalassemia in the region.[1]

One significant non-biomedical element that causes thalassemia families, to experience severe social, economic, and psychological issues is the lack of information and knowledge about the condition.[4] The two most frequently mentioned biomedical risk factors

for thalassemia are high-risk marriages i.e., couples possessing genetic abnormalities, and consanguineous marriages. According to a study, it is advised that young adults should be targeted and made more aware of thalassemia and its effects to reduce marriages among carriers and the prevalence of such diseases. In Saudi Arabia, consanguineous marriage is a major risk factor for the occurrence of thalassemia. However, prevention is less expensive than therapy in the long run, therefore following actions are potential parts of the preventative strategy. Most important is to identify disease carriers as well as educate those thalassemia carriers regarding the perils of having a child with thalassemia and how to lessen it, which would reduce the number of afflicted childbirths and deaths. Secondly, prenatal diagnosis of couples carrying afflicted children, advising them of the 25% chance of recurrence and encouraging them to minimize family size, which may lower the incidence of births. Along with this, the third important action is the screening of potential carriers across the board. Population screening is beneficial in nations having a relatively higher percentage of consanguineous marriages since it is affordable.[5]

Results of a study showed that cultural risk factors (religious restrictions regarding disease prevention and screening, a strong emphasis on consanguinity), socioeconomic risk factors (lack of social and financial support, sympathetic attitude of doctors and paramedics), and disease-related risk factors (lack of blood and medicine availability, inadequate healthcare system) are among the main risk factors associated with beta-thalassemia major. The study suggests placing a lot of focus on raising parental knowledge of the value of patient screening and counseling. Major beta thalassemia is seen as a susceptible hazard, typically for those from lower socioeconomic backgrounds. Pakistan was ranked 113th out of 120 countries in a 2012 UNESCO study on education, which amplified the poor state of education in our nation. The main barrier to thalassemia awareness and counseling has resulted from this. The inability of the general public to comprehend the technical terms used by health specialists and researchers, language barriers, a lack of access to technologies, and a lack of knowledge of medical information are all barriers to successful communication.[6] This research paper assesses the risk factors associated with thalassemia.

METHODOLOGY

A descriptive cross-sectional study was carried out on patients reporting to the thalassemia center of the Pakistan Institute of Medical Sciences (PIMS), Islamabad from February 2022 to November 2022, after receiving permission from the ethical review board of the institution. The sample size was 200, calculated by Epi Info. The sampling technique was non-probability convenient sampling. Participants of the study were the parents/guardians of patients. Parents of diagnosed thalassemia cases who reported to the thalassemia center PIMS and duly answered the questions were included. Those with interposed language barrier, who refused to participate and incomplete questionnaires formed part of exclusion criteria. The content of questions was verbally explained and oral consent was taken from everyone who participated. The participants were assured of the confidentiality of their responses. A self- structured questionnaire containing 16 questions was used as data collection tool. The questions were asked individually to each participant in such a way that a layman can understand and answer according to his/her knowledge and answers were simultaneously written on the questionnaire. SPSS version 25 was used for data analysis. Mainly descriptive statistics have been reported.

RESULTS

Out of 200 patients, 105 (53%) were males and 95 (47%) were females in age group of 10 (± 4) years with mean hemoglobin levels of 6.3 gm/dl (± 1.3). According to the data collected, majority of the parents were from Punjab (52%) and KP (23%). Fathers of 42% of the patients had private jobs, 33% being self-employed and rest of them having government jobs or running a business. Thalassemia was found to be more prevalent in people of lower socioeconomic status (82%) of the cases. Refer to (Figure 1).

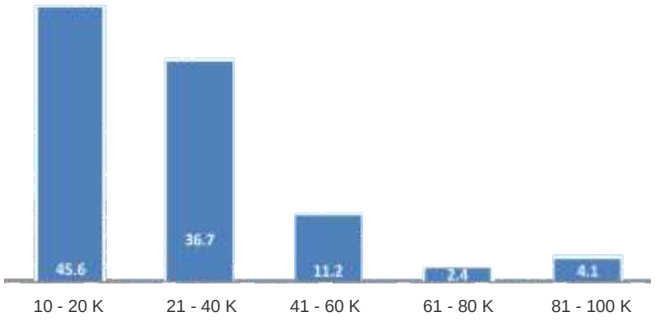

Figure 1: Monthly income of father (PKR)

Consanguinity was proven to be the major risk factor of thalassemia as 148 (74%) of the parents recorded were in cousin marriages out of which (77%) were having trend of cousin marriages in their families (Figure 2). In the current study, family history of thalassemia was found to be 28%. (Table I and II).

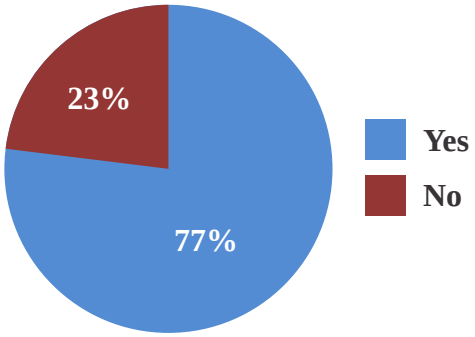

Figure 2: Trend of cousin marriage

Table I: Maternal Family History of Thalassemia

Response	Frequency	Percent
Yes	56	28.0
No	144	72.0
Total	200	100.0

Table II: Paternal Family History of Thalassemia

Response	Frequency	Percent
Yes	54	27.0
No	146	73.0
Total	200	100.0

Thalassemia showed prevalence in particular blood groups, being more common in B+ blood group, 72 out of 200 (36%) and O+ blood group, 57 out of 200 patients (28%). Refer to (Figure 3).

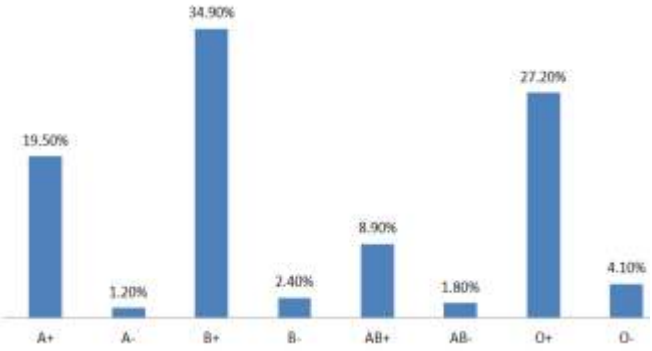

Figure 3: Blood group of the patient

Majority of the parents having low education status had no knowledge about screening of thalassemia prior to conception (Table I and II). Parents, not having awareness about any one of the risk factors of thalassemia, accounted for 42% of the total with 91% of them having no knowledge about premarital and prenatal screening.

Table III: Education of father and awareness about screening of thalassemia before conception

Variable		Did you know about screening of thalassemia before conception?		
		Yes	No	Total (%)
Education of father	Illiterate	4	32	18
	Primary	2	54	28
	Secondary	11	80	45.5
	Bachelors	1	12	6.5
	Masters	0	4	2
Total (%)		9	91	100

Table IV: Education of mother and awareness about screening of thalassemia before conception

Variable		Did you know about screening of thalassemia before conception?		
		Yes	No	Total (%)
Education of mother	Illiterate	7	67	37
	Primary	5	57	31
	Secondary	6	43	24.5
	Bachelors	0	12	6
	Masters	0	4	1.5
Total (%)		9	91	100

DISCUSSION

Globally, thalassemia is a severe public health concern, particularly in South Asian nations where illnesses like thalassemia are not prioritized by the health systems due to their inefficiency and lack of usable resources. This study focuses on investigating the risk factors of thalassemia in patients reporting to the thalassemia center in Islamabad. Consanguineous marriages increase the birth prevalence of autosomal recessive disorders like thalassemia.[7] In our study of 200 participants consanguinity has shown to be a substantial risk factor for thalassemia, with 148 (74%) of the parents being recorded in cousin marriages, and 77% of them having a family trend of cousin marriages. These results are in line with another study where-in 81.7% of patients were the outcome of consanguineous marriages.[8]

The parent's literacy level has a beneficial influence on how they handle the illness or utilize prenatal screening to determine the risk of thalassemia in pregnancy. In this study, 91% of parents did not have knowledge of premarital and prenatal screening and these findings are comparable to another study from university in Ajman, UAE which revealed that 92.7% participants acknowledged the significance of screening tests.[9] Since parents have the role of guardians and care providers therefore, they are the ones to have all the information and knowledge about the prevention, causation,

diagnosis, progression and management of the disease concerned. The most crucial role in this context is played by parents who must possess sufficient knowledge about the causation and progression of thalassemia. According to several surveys, Pakistan's low literacy rate is the biggest barrier to population awareness.[10]

The disease under discussion is very much common in rural areas and in low socioeconomic class which could be due to the fact that people of these areas are not literate as compared to urban areas and hence they do not know much about public health practices and preventive measuresofferedandavailabletourbanizedcommunities. Consistently researches have emphasized the importance of education and pointed out that due to lack of it, people remain uninformed about the manner of causation of diseases. This study found that 82% of parents belonged to low socio-economic class and similar prevalence of thalassemia in rural areas was shown by study conducted in Pakistan which revealed that out of 317 surveyed parents, 196 (69%) were from rural areas.[11]

CONCLUSION

Consanguinity and lack of awareness about the disease were the two main risk factors of thalassemia. Thus, there is a dire need to educate the general public about the disease, its risk factors and the significance of premarital and prenatal screening. This study will help health policymakers to mitigate the risk factors of thalassemia and raise awareness about screening tests available for the disease.

REFERENCES

1. Kattamis A, Fomi GL, Aydinok Y, Viprakasit V. Changing patterns in the epidemiology of P-thalassemia. European Journal of Haematology. 2020;105(6):692–703.
2. Mettananda S, Gibbons RJ. Higgs DR. a-Globin as a molecular target in the treatment of p-thalassemia. Blood. 2015;125(24):3694–701.
3. Khan J, Muhammad D, Ismail M, Khan I. ur Rehman Z, Niaz S. Consanguinity, the driving force behind inheritance of HbS-p thalassemia in Southern Districts of KP. JPMA. 2020; 70(6): 978-83.
4. Canatan D. Corrons JL, Piacentini G, Kara F, Keskinkilnp B. Tezel B, et al. Immigration and screening programs for hemoglobinopathies in Italy, Spain and Turkey. Acta Bio Medica: Atenei Parmensis. 2021 ;92(4).
5. Abu-Shaheen A, Heena H, Nofal A, Abdelmoety DA, Almatary A, Alsheef M, et al. Epidemiology of thalassemia in Gulf Cooperation Council countries: A systematic review. Biomed Res Int. 2020:1509501. Available from: http://dx.doi.org/10.1155/2020/1509501
6. Khan AM, Abbas Z, Ramzan S, Aziz L. Social Epidemiological Analysis of Risk Factors and Psychosocial Burden of Beta Thalassemia Major (BTM) In Pakistan. Elementary Education Online. 2021;20.
7. Bittles AH, Mason WM, Greene J, Rao NA. Reproductive behavior and health in consanguineous marriages. Science. 1991; 252(5007): 789-94.
8. Ishaq F, Hasnain Abid FK. Akhtar A, Malmiood S. Awareness among parents of PP- Thalassemia major patients, regarding prenatal diagnosis and premarital screening. Journal of the College of Physicians and Surgeons Pakistan. 2012; 22(4):218-21.
9. Elsadek AM, Hassan N, Shahwan M, Jairoun AA. Exploring the gap between knowledge and behavior regarding thalassemia among university students: A cross-sectional study in the United Arab Emirates. J Community Health. 2022;47(3):392–9. Available from: http://dx.doi.org/10.1007/s10900-022-01066-8
10. Azhar U. Audit of Beta-Thalassemia Cases At Sheikh Zayed Medical College/Hospital, Rahim Yar Khan. JSZMC. 2015;6(2):811-15.
11. 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 Health Care. 2018;30(3):200-7. Available from: http://dx.doi.org/10.1093/intqhc/mzx198