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Frequency and Awareness of Thalassemia in Families with Cousin Marriages: A Study from Karachi, Pakistan

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Authors' contributions

This work was carried out in collaboration between all authors. Authors MMU, FUH, AS and MMAK developed the concept and designed the study. Authors AA, IT, MI, FA and BN administered the questionnaire, analyzed and interpreted the data. Authors GK, BM, SS, NN and AN drafted the manuscript. Authors FUH and MMU revised the manuscript for intellectual content. All authors read and approved the final manuscript.

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ABSTRACT

Aim of Study: To assess the frequency of thalassemia in families with cousin marriages.
- To assess the level of awareness of Thalassemia in affected patients.
Introduction: Thalassemia is one of the most common inherited diseases in Pakistan. According to few experts approximately 5000 children are diagnosed with this disease every year and according to some sources there are more than 50,000 Thalassemic patients registered in treatment centers all over the country.
Methodology: This cross-sectional study was conducted at OSF, AMTF and Hussaini Blood Bank, Karachi from January to December 2015. A total of 100 sample size was selected. Only

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Thalassemic people having consanguineous marriages were included in the study. Informed consent was taken from both the parents and patients. Structured questionnaire was devised to collect data from patients.

Results: When asked 56% (p=0.00) of patients said they had Thalassemia in their families and 28% (p= 0.268) have had deaths in their families due to Thalassemia. 92%(p=0.00) of parents with cousin marriages had no awareness about Thalassemia before their first Thalassemic child and 63% (p= 0.012) were not aware about the treatment for Thalassemia even after their first Thalassemic child. Regarding screening test for Thalassemia 62% (p= 0.012) were aware of it and 98% (p=0.46) agreed to go through screening.

Conclusion: The study concluded that cousin marriages had high frequency of Thalassemia. Awareness of the patients was inadequate. General public, parents and families of the patients should be educated in this regard.

Keywords: Thalassemia; cousin marriages; awareness; screening test; blood; transfusion; disability; genetics.

1. INTRODUCTION

Thalassemia is one of the most common inherited diseases in Pakistan [1]. The distribution of number of patients shows that the most commonly affected age group is between 5 and 10 years old [2]. Beyond 15 years, the number of cases decreases [2]. Thalassemia is a clinically heterogeneous disorder resulting from different genetic lesions that variably impair globin chain synthesis [3]. The two main types of Thalassemia are alpha and beta: Individuals with alpha Thalassemia do not produce enough alpha chains causing excess beta globulin chains; those with beta thalassemia do not produce beta globulin chain, causing excess alpha chains [4]. The common signs and symptoms of Thalassemic diseases include pale skin, retarded growth and puberty, anemia, enlarged spleen and an increased susceptibility to infections [4]. Almost 70,000 infants are born with beta Thalassemia worldwide each year and 270 million people are carriers of hemoglobinopathies [5]. Beta Thalassemia is most commonly present among populations in all Mediterranean countries, as well as in Southeast Asia, India, Africa, Central America and the Middle East [5]. Analysis of data collected in the present study indicated that almost 70% families have more than one beta Thalassemia child [6]. Beta Thalassemia probably is the most common single gene disorder causing a major genetic health problem in the world [7]. Thalassemia is also classified into homozygous and heterozygous types.

Thalassemia major is one of the most common genetic disorder in Pakistan and over five thousand new patients are added in the pool annually [8]. Heterozygote (-alpha/alpha alpha) and homozygote (-alpha/-alpha) children were 1.5 and 2.5 times more likely to survive malaria than those with normal genotypes [9].

Consanguinity is a deeply rooted social trend and among people currently living in countries where consanguineous marriages are customary, one in every three marriages is between cousins [10]. The rising public awareness on possible preventive measures for congenital disorders has led to an augmentation in the number of couples seeking preconception and premarital counselling on consanguinity [11]. Consanguinity-associated deaths were consistently higher in the neonatal, infant and childhood periods [12]. If two partners who are carriers of Thalassemia get married, based on the probability, their children will have severe Thalassemia (25%), will be healthy (25%) and be carriers of Thalassemia (50%) [13]. Of the beta Thalassemia children born to consanguineous parents, 53.57% were born to the first cousins, which is higher when compared with other degrees of consanguinity [14]. Awareness and knowledge among affected families plays an important role to change the attitude of people towards family extension and reduces the chances of Thalassemia running in family [15].

There are two different treatments of Thalassemia at present: symptomatic treatment and bone marrow transplantation. Symptomatic treatment includes blood transfusion every 2-4 weeks and ongoing medical care, including injections for iron overload due to excessive number of transfusions [16]. The second option of treatment is bone marrow transplantation which in Pakistan is a very costly treatment. As far as a common man is concerned, it costs PKR 9-10 lacs. There is no known cure but prevention is possible, applicable and successfully achieved in some countries [17].

2. METHODOLOGY

2.1 Research Model

It is a cross-sectional study. Parents of patients having other blood disorders like hereditary spherocytosis, sickle cell disease and others were excluded.

2.2 Sample

A total sample size of 100 was recruited in this study.

2.3 Data Collection

A pilot study was conducted to assess the authenticity of questionnaire. Structured questionnaire of 100 sample size was devised to collect the data from patients. It was conducted from January 2015 to December 2015.

2.4 Data Analysis

The data was entered and analyzed using SPSS version 16 with frequencies, percentages, confidence interval and p value of 0.05 was considered being statistically significant. The information was collected by interviewing Thalassemic parents or their close relatives. An informed consent was taken from the parents before the start of interview. The collected information includes diagnosis i.e. Thalassemia, age and method of diagnosis, along with family history of the disease.

3. RESULTS

A survey was conducted to check the frequency of Thalassemia in families who have had cousin marriages, their awareness about the disease in the parents of the patients and point of views about Thalassemia screening and abortion of Thalassemic child. Survey was conducted in Afzal Memorial Thalassemia foundation (50%), Omair Sana foundation (40%) and Hussaini blood bank (10%).

100 questionnaires were filled by the parents of Thalassemic patients. There was a slight preponderance of females which accounted for 52% of the patients and males 48%. The study group includes patients from mean age group 10 years. According to data collected , majority of the patients were from East and Central region, Father of 51% of the patients were laborers, 12% were drivers,21% were involved in other professions and 16% were jobless.

When the parents of the patients were asked about the reason of their marriage in cousins, 82% (P-value 0.00) gave the reason of traditional values, while in 5% of cases the marriage in cousins was by force.

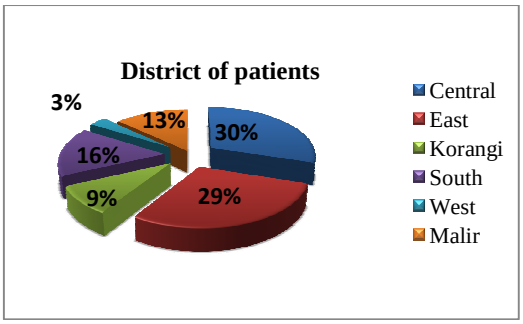

Fig. 1. Most number of patients came from Central and East district

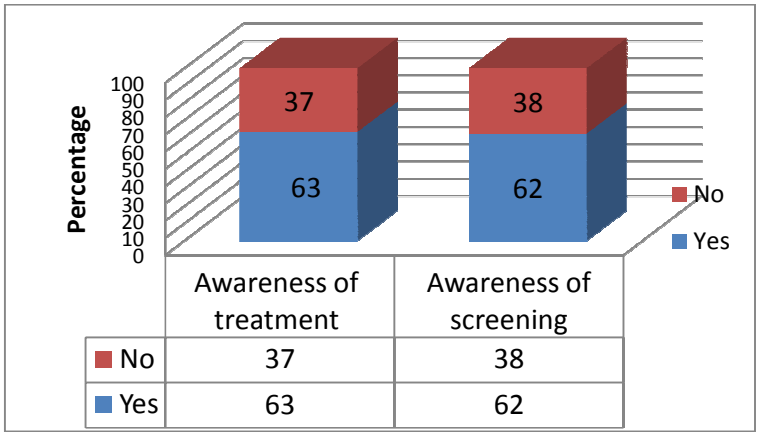

Fig. 2. Majority of the parents were aware of the treatment and screening of thalassemia

Table 1.

S. no.	Questions asked	Percentages %			P-value	Chi-square	Confidence interval 99%	Mean
		Yes	No	Do not know				
1	Prevalence in family	56%	34%	10%	0.000	3.593	0.149 – 0.167	33.3
2	Mortality due to disease	28%	61%	11%	0.000	2.597	0.294 – 0.318	33.3
3	Awareness before Thalassemic child	8%	92%		0.000	0.779	0.510 – 0.530	50
4	Awareness of treatment	63%	37%		0.012	2.239	0.110 – 0.130	50
5	Awareness of screening	62%	38%		0.021	0.624	0.250 – 0.290	50
6	Point of view about screening	98%	2%		0.000	0.177	0.130 – 0.147	50
7	Suggestion about cousin marriage	40%	51%	9%	0.000	3.329	0.306 – 0.330	33.3
8	Point of view about abortion	66%	23%	11	0.000	1.982	0.391 – 0.416	33.3
9	Ignorance of other children	39%	61%		0.036	0.440	0.480 – 0.506	50
10	Knowledge about duration of disease	91%	9%		0.000	0.860	0.204 – 0.219	50

50% (P-value 0.00) of parents were Thalassemia minor, 6% were Thalassemia major, and while 44% did not know about their Thalassemic status.51% and 48% of patients got diagnosed through doctors and medical reports respectively. 56% (P-value0.00) of patients had positive extended family history of Thalassemia in their families while 28% (p= 0.00) had deaths in their families due to Thalassemia. 92% of parents with cousin marriages had no awareness about Thalassemia before birth of their Thalassemic child. 92% (p-value 0.00) of parents first came to know about the disease after having Thalassemic child, while 4% were aware of it before marriage and 4% came to know about it after marriage. 92% (P –value 0.00) of parents got awareness through Doctor/Nurse and 8% got awareness through TV. In 51% of subjects, both, husband and wife had Thalassemia minor, while 44% subjects were unaware of their Thalassemia status.

In our study 63% (P-value 0.012) of parents of the patients were aware about the treatment of Thalassemia. Regarding screening test for Thalassemia 62% (P-value 0.021) of parents were aware of it and 98% (P-value0.00) agreed to go through this before marriage/during pregnancy.

When they were asked if they would have aborted this child if they had known about the Thalassemic child 66% (P-value 0.00) parents said yes, 23% of parents said No and 11% of parents gave no suggestion about it. When the parents were questioned whether other offsprings get neglected because of the diseased child or not, 61% (P-value0.036) of parents denied neglecting of their other children. On the question about duration of disease, 91% (P-value 0.00) were aware that thalassemia is one lifelong disease. 51% (P–value 0.00) of the parents were against cousin marriages now, 40% were still in favor of cousin marriages in their next generation, while 9% gave no suggestion. 50% (P-value 0.00) of patients visit the transfusion center twice a month while 36% visit once a month. In 80% (P-value 0.00) of cases total expenditure per month was found to be between Rs3000 to 5000, which is a huge burden for the poor.

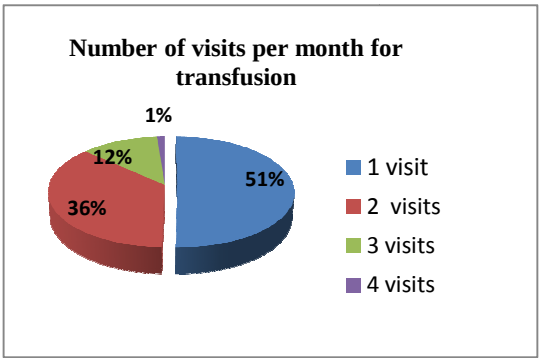

Fig. 3. Majority of the patients visits the transfusion center once a month

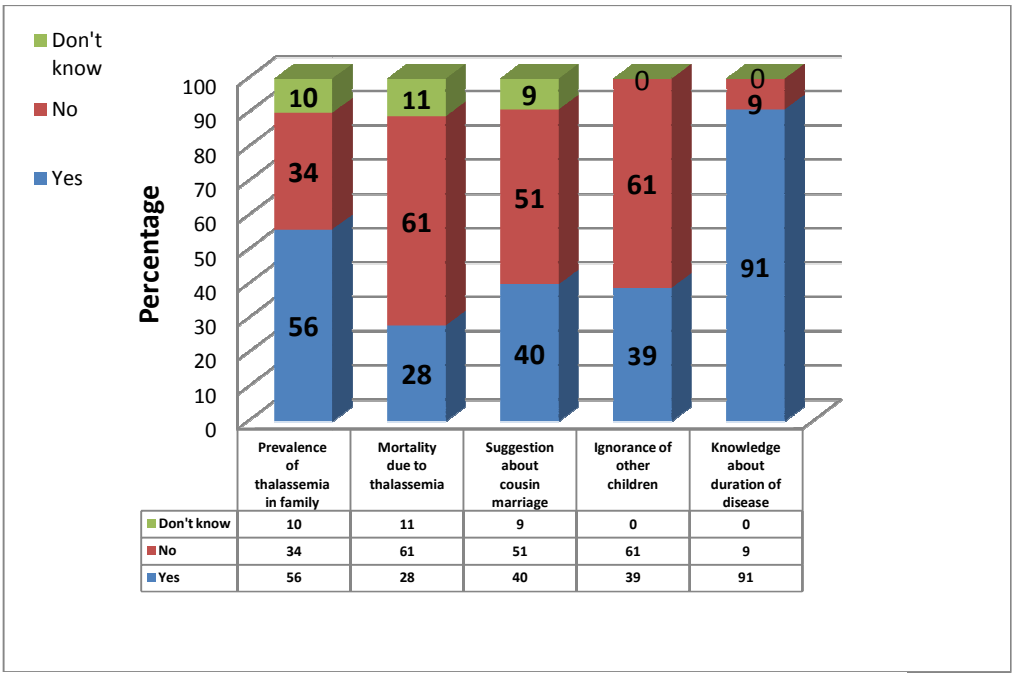

Fig. 4. Thalassemia is present in the majority of the families, most of the families had no case of mortality due to thalassemia, majority of the parents said "No" to the cousin marriages, majority of the parents negate ignorance of other children due to the diseased child. A vast majority of parents were aware about the duration of thalassemia

4. DISCUSSION

According to a research there is a higher risk of inherited disorders like Thalassemia, deafness, blindness, mental disorders, diabetes and muscular disorders in children born to first cousins [18]. Though more common in first-cousin partners, marriages within the same community also carry a higher risk, more than 80 percent of all parents in Pakistan are first cousins, seven percent are related by blood, about six percent belong to the same caste and only about four percent marry outsiders. In many countries, first-cousin marriages are forbidden and children of these marriages stand at 20 percent risk of having genetic problems.

The cross-sectional study was conducted to assess awareness, practices and to devise plans to lessen the occurrence of Thalassemia in off springs of parents with consanguineous marriages. According to our study, out of 100 consanguineous couples, 56% confirmed of cousin marriage to be an extant custom in their family while 34% denied. This greatly advocated the clustering of Thalassemia in individuals of families with cousin marriages [19]. In these customs, it is an obstacle to go against the family traditions. Perhaps that is why 40% of couples supported cousin marriages and would suggest it to the future generation as well.

The mortality is found to be 28% which is high and veils the miserable and low quality of life in the surviving children. Moreover, the striking blow out was from the fact that only 8% of the population had awareness of Thalassemia before the disease occurred in the offspring thus leading to the concept of high prevalence of consanguineous marriages in society. In our cross-sectional study, we noticed that 92% of the parents had no awareness of Thalassemia prior to having a Thalassemic child. Unfortunately, majority of them got acquainted with it after their first Thalassemic child was born. Lack of awareness about this disease, its treatment and screening procedure turned out to be a major source of disappointment [4] In Pakistan, most of the Thalassemic patients come from poor and uneducated families who have a very orthodox ritual of marrying within families [20] which hence breeds genetic errors within the offspring produced [18].

Another significant finding of this study shows the lack of awareness of screening of Thalassemic

trait in an individual before marriage (62% Yes vs 38% No, P=0.021A silver lining was seen when 98% of the couples agreed upon suggesting Thalassemia screening to other couples before marriage.

Majority of parents were aware of the fact that Thalassemia is a lifelong disease (91% Yes vs. 9% No). They came to know about this through the hospital staff and doctors they encountered during their child's blood transfusions and by detailed discussion of their child's condition with them. Also, 63% of parents were well aware of the existence of the treatment of thalassemia. The rest 37% (P=0.012) did not know about the treatment and exact nature of the disease. They required support, expert guidance and directions regarding the fate of their child and quality of life. Most of the parents knew about the treatment because of a previously known Thalassemic child in family/relatives.

When couples were reviewed about their relations with the Thalassemic child and other normal children, 39% gave affirmative response in relation to the ignorance of other children due to the diseased child being their sole priority to look after and due to the time duration for the treatment. While 61% strongly denied this view and practiced treating all their children equally.

A very sensitive issue of abortion, which is considered a taboo in Pakistan, was also raised in our study. A striking number of couples agreed upon aborting if proven to be Thalassemic in utero. 66% had positive views of aborting their Thalassemic child before birth while 23% strongly disapproved and wished to continue their pregnancy as it was against their religious practices and emotional attachments. About 11% of the parents had no idea regarding the abortion of the baby.

Some limitations of our study should be noted. The couples were purposefully selected on the condition of being cousins. Furthermore, the setting in which the interview took place might have made the couples self-conscious as there were other couples sitting nearby. We also faced the trouble of language barrier with few couples which might have hindered the true essence of questions asked. The study could not differentiate on the major minor constituents of Thalassemia separately.

In view of the whole study that we came across, the key factor in prevention of Thalassemia is stressing the need for sensitizing the people against consanguineous marriages [21]. Also Thalassemia screening should be made free and compulsory for every couple, Counseling should be done in all pregnant women and awareness programs should be carried on frequent basis in order to minimize the risk and frequency of Thalassemia in consanguineous marriages [3]. A good initiative is taken by Sindh Assembly that it passed the resolution urging the government to make tests for Thalassemia, HIV and other diseases "mandatory" for couples before marriage.

5. CONCLUSION

The study concludes that cousin marriages had a high frequency of Thalassemia. Awareness of the patients was inadequate but this can be improved by educating general public, parents and families of the patients in this regard. The only thing that we all should do as a part of Pakistani society and citizen of the world is to promote screening before marriage whether it is between cousins or not, to prevent our next generations from suffering from this debilitating disease.

CONSENT

As per international standard or university standard, patient's written consent has been collected and preserved by the authors.

ETHICAL APPROVAL

As per international standard or university standard, written approval of Ethics committee has been collected and preserved by the authors.

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COMPETING INTERESTS

Authors have declared that no competing interests exist.

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QUESTIONNAIRE

Personal Information:

Name:
Age:
Sex:
District:
Contact number (not necessary):
Occupation:
Number of Children:

Questions

1. Why did you bring your child in hospital?

A. For follow up of some illness B. For regular medical checkup
C. For transfusion D. Other

2. Who confirmed Thalassemia in your child?

A. Doctor B. Medical Staff C. Medical Reports D. Medical camp
E. Others

3. Are you suffering from Thalassemia?

A. Yes B. No C. Do not know

4. Does your spouse have Thalassemia?

A. Yes B. No C. Do not know

5. Is anyone in your family suffering from Thalassemia?

A. Yes B. No C. Do not know

6. Did anyone die in your family from Thalassemia?

A. Yes B. No C. Do not know

7. Is cousin marriage/community marriage prevalent or necessary in your family?

A. Yes B. No

8. Why are cousin marriages necessary in your family?

A. Religious reason B. Tradition of cousin marriage in family C. By force D. Other reason

9. Before having child with Thalassemia, were you aware of this disease?

A. Yes B. No C. Don't Know

10. When did you first come to know about Thalassemia?

A. Before marriage B. After marriage C. After birth of your child with Thalassemia

11. How did you come to know about Thalassemia?

A. Television B. Radio C. Newspaper D. Health care professional

12. Did you know before your marriage that Thalassemia is a genetic disease that prevails in family?

A: Yes B: No C: Don't Know

13. Do you think that marrying a cousin or in family can result in having a child with Thalassemia?

A. Yes B. No C. Don't Know

14. Were you aware of Thalassemia screening test before marriage?

A. Yes B. No C. Don't Know

15. Are you aware of treatment of Thalassemia?

A. Yes B. No

16. Are you aware that Thalassemia is a lifelong disease?

A. Yes B. No C. Don't Know

17. Why did you get married in family?

A. Own wish B. Family wishes C. Family tradition

18. Will you continue practice of cousin marriages in your offspring?

A. Yes B. No

19. Will you prefer going for health check-ups during pregnancy?

A. Yes B. No

20. How much do you spend on your child's transfusion needs?

A. 2000-4000 PKR B. 4000-6000 PKR C. 6000-8000 PKR

PKR =Pakistani Rupees

21. How much do you spend on child's nutritional health?

A. 500-1000 PKR B. 1000-1500 PKR C. 1500-2000 PKR

22. Do you feel that your other children are being neglected due to this child?

A. Yes B. No

23. How many times in a month do you take your child to the hospital for follow up?

A. Once B. Twice C. Thrice D. Every week

24. If during pregnancy you come to know that the child is having Thalassemia, will you continue with the pregnancy?

A. Yes B. No

25. Will you advise others to get married in family?

A. Yes B. No

26. Will you advise other people for a Thalassemia screening test before getting married?

A. Yes B. No

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