AWARENESS AMONG THE PARENTS OF CHILDREN WITH THALASSEMIA MAJOR, REGARDING PREMARITAL SCREENING AND PRENATAL DIAGNOSIS

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ABSTRACT

Parental awareness is the best way to prevent thalassemia major disease in children. So the knowledge of premarital screening and prenatal diagnosis helps to decrease the thalassemia disease in Pakistan. The objective of the present study was to assess the awareness among parents of children with thalassemia major regarding premarital screening and prenatal diagnosis. The descriptive study was conducted at two thalassemia centers (i) thalassemia centre, The Children's Hospital & the Institute of Child Health Multan, Pakistan (ii) Fatimid Foundation Multan, Pakistan from 20th March 2019 to 20th June 2019. Total number of 300 parents of thalassemia major patients participated in this study. A structured interview schedule was used as a tool for data collection through convenient sampling. Of the 300 respondents, the majority of the respondents 201(67.0%) were fathers while 99(33.0%) of the respondents were mothers. Greater part of the respondents 177(59.0%) belonged to rural areas. The majority of the respondents 153(51.0%) had primary level of education. The greater part of the respondents' family monthly income was very low between 5100-10000 PKR. Among the total respondents, 131(43.7%) reported that they had 'no knowledge' of premarital screening and prenatal diagnosis 111(37.0%). Majority 207(69.0%) of the respondents did 'strongly agree' that thalassemia disease affected their financial situations. Parents' awareness about premarital screening and prenatal diagnosis was inadequate. There is a need to understand the density of problem and create awareness among thalassemia families and the general public about prevention in order to reduce the burden of disease in Pakistan.

Keywords: Awareness, Parents of Children, Thalassemia Major, Premarital Screening, Prenatal Diagnosis

INTRODUCTION

Thalassemia is the most common inherited blood disorder which is passed from parents to their children. It is an autosomal recessive disorder resulting in reduction of hemoglobin level in the red blood cells, decreased production of hemoglobin and rapid destruction of red blood cells cause severe anemia. About 3% world's population is a carrier of the genes of beta-thalassemia (Michael, Baun, Vichinsky, 2007). It is a major public health problem in Pakistan. Thalassemia carrier status is estimated in Pakistan 5-7 % and more than 5,000 thalassemia major patients are born every year (Ahmed, Saleem, Modell, & Petro, 2002).

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The main factor that leads to high prevalence in Pakistan is consanguinity. About 25,000 thalassemia patients are registered in thalassemia Federation of Pakistan but the actual figure is estimated to be more than 100,000 because the majority of the people are living in rural areas and they are not registered in any thalassemia centers. Thalassemia is a preventable disease found in different countries like Iran, Cyprus, Greece, Italy and Saudi Arabia. These countries established national program of screening and prevention and got significant reduction in the birth of thalassemia affected children. Likewise other studies reported the positive effects of thalassemia education programs that increase parents' knowledge about the disease (Dehkordi & Heydarnejad, 2008; Jetsrisuparb, Wiangnon, 2006). Thalassemia major patients require regular blood transfusions and iron chelation therapy throughout their whole life and thus put financial burden on patients and their families. The management of thalassemia major patients requires regular blood, chelation therapy for iron overload, management of complications including cardiac dysfunction, osteoporosis, endocrine problems, Hepatitis B and C, HIV infection etc. The only cure of thalassemia major is bone marrow transplantation, but it is unaffordable for majority of the patients' families. According to the World Health Organization if the birth rate of any affected infant exceeds 0.1/1000 an effective program for screening must be initiated. Thalassemia is a preventable disease but in Pakistan no national screening program is started. Only few Non Government Organizations (NGOs) are working in their own capacity for thalassemia patients.

After marriage, if a couple is found to be thalassemia carriers, every pregnant female should undergo the test of Chronic Villous Sampling (CVS) at the 10-12 weeks of gestation to determine the status of fetus. As in each pregnancy there are 25% chances that the baby may be suffering from thalassemia major, there are 50% thalassemia minor traits and 25% chances that baby will become normal. If baby is found to be thalassemia major, termination of pregnancy must be offered to the people, as now this is practiced with the permission of religious scholars. In Pakistan due to lack of awareness many thalassemia families are not having knowledge of prenatal diagnosis. In Pakistan prenatal diagnosis was introduced in 1994 but it is still available in big cities like Lahore, Karachi, Rawalpindi, Multan (Ahmed, Saleem, Rashid, Abbas & Malik, 1994; Karimi, Johari & Cohan, 2010).

The main strategies for the prevention of thalassemia include providing of appropriate information of disease, counseling and screening of target families, extended family screening, screening of general population, premarital screening and prenatal diagnosis (Tasleem, Tasleem, Siddiqui, Adil & Rashid, 2007). In Pakistan, public awareness regarding the thalassemia disease is very poor even thalassemia major patients' families know very little about the disease. Naseem et al. reported that the majority of the registered thalassemia major patients' families did not demand prenatal diagnosis (Naseem, Ahmed & Vahidy, 2008). The only effective way for the prevention of thalassemia is mass education through seminars, symposia, electronic and print media, education and training of general medical practitioners, pediatricians, gynecologists, social workers. Furthermore knowledge of the disease may be incorporated in syllabus. Because in Canada and Australia medical education regarding screening and genetic tests is provided at school level children of 14 to 18 years; it is finding promising results in the diagnosis of newly thalassemia cases. Premarital screening facility is available in Pakistan but it is underutilized due to lack of knowledge and high cost. Premarital screening is better as compared to prenatal diagnosis for thalassemia prevention. Unfortunately, in our country awareness of screening and diagnosis of thalassemia is inadequate. In Arab countries like Iran a

significant reduction is found in new thalassemia cases as they have made screening of thalassemia test mandatory before marriage.

In Pakistan, the Punjab Thalassemia Prevention Program was established in 2009. This project was initially started in 04 major districts but now it has been extended in more districts of Punjab and now it is providing awareness to patients, families and general public. Premarital screening and extended family screening are available in many centers but prenatal diagnosis facility (Chronic villus sampling) is limited so the parents have to travel a long distance to avail themselves of this free facility from the government.

In Pakistan a thalassemia major child's overall life expectancy is 10 to 12 years. The management of thalassemia child includes healthy screened blood, iron chelation therapy along with other medicines and psychosocial management these put the enormous burden on families and society. Due to expensive medicines, the majority of thalassemia children in Pakistan are not able to get proper treatment. Thus prevention of thalassemia is important aspect for the management of the disease. Abolghasemi et al (2007) claimed that different countries of the world especially Italy, Greece, Cyprus and Sardinia reduced the birth rates of thalassemia major children through extensive screening program at different levels along with prenatal diagnosis. Iran has reduced the births of thalassemia major patients through an antenatal diagnosis program. Therefore at national level there is an urgent need to start thalassemia screening and control program to reduce the burden of families and society(Abolghasemi et al, 2007). Cao, Rosatelli, Galanello (2007) recommended that screening of thalassemia carriers, prenatal diagnosis and genetic counseling could greatly overcome the births of affected children and improve the diagnosis of affected infants. The present study will be helpful to the health professionals especially federal and provincial health departments, hospitals, thalassemia centers to understand the awareness level of parents of the children with thalassemia regarding premarital screening and prenatal diagnosis which will allow them to take necessary measures in order to make them aware of day to day patient management. The results of the present study will fill the gap by adding into the literature regarding awareness among parents of children with thalassemia, regarding premarital screening and prenatal diagnosis.

METHODS

This descriptive study was conducted at thalassemia centre of The Children's Hospital & the Institute of Child Health Multan, Pakistan and Fatimid Foundation Multan, Pakistan from March 20, 2019 to June 20, 2019. A total number of 300 thalassemia major patients' parents participated in this study through convenience sampling. Structured interview schedule was used as a tool for data collection. Verbal and written consent were taken from the respondents. Before data collection, ethical approval was obtained from the ethical committees of the two thalassemia centers. Only thalassemia major patients' parents were included and the patients of other types of blood disorders like hemophilia, a-plastic anemia, acute lymphoblastic leukemia were excluded. Before actual data collection, a pilot study was conducted to check the validity and reliability. The data were analyzed through the Statistical Package for Social Sciences (SPSS) version 23 and presented in frequency, percentage, Mean and Standard Deviation. The first part was comprised of questions regarding demographic information of the respondents i.e. age, gender, relationship with patient, residential area, education level of the respondents, employment, family monthly income, source of information regarding thalassemia, type of marriage, family structure, awareness of blood screening, premarital screening, prenatal diagnosis, bone marrow

transplantation and in the second part 21 statements were asked to the respondents. The structured interview schedule was discussed with two senior professors having more than 10 year experience working at thalassemia centers of Multan, Pakistan. It was revised to incorporate recommended improvements. A 5-point and 4-point Likert type scales Strongly Agree=5; Agree =4; Undecided=3; Disagree=2; Strongly Disagree=1 and Not knowledgeable=1; Somewhat knowledgeable=2; Knowledgeable=3 and Very Knowledgeable=4 were used to gather responses from the participants. Descriptive statistics was used to describe the data in frequency distribution, percentages, means, and standard deviations.

RESULTS

Table I. Demographic information of the respondents (N=300)

Variables	Frequency (%)	Variables	Frequency (%)
Age		**Occupation of the respondents**	
18-30 Years	135(45%)	Unemployed	20(6.7)
31-40 Years	130(43.3%)	Business	37(12.3)
41-50 Years	25(8.3)	Private	93(31.0)
51-60 Years	7(2.3%)	Government	29(9.7)
➢ 60 Years	03(1.1%)	Laborer	108(36.0)
		Other	13(4.3)
Relationship with the patient		**Monthly income of the respondents**	
Father	201(67.0%)	1000-5000 PKR	42(14.0)
Mother	99(33.0%)	5100-10000 PKR	131(43.7)
Residential area		11000-15000 PKR	62(20.7)
Rural	177(59.0%)	16000-20000 PKR	35(11.7)
Urban	123(41.0)	Above 20000 PKR	30(10.0)
Education level of the respondents			
Primary	153(51.0)		
Middle	35(11.7)		
Matriculation	48(16.0)		
Intermediate	28(9.3)		
Graduation	24(8.0)		
Master	11(3.7)		
Other (Diploma)	1(.3)		

Table I. Of the 300 respondents, 135(45%) respondents were aged between 18-30 years while 130(43.3%) respondents were aged between 31-40 years, 25(8.3%) respondents were aged between 41-50 years, 7(2.3%) respondents were aged between 51-60 years and 3(1.1%) respondents aged were greater than 60 years. The majority of the respondents 201(67.0%) were fathers while 99(33.0%) were mothers. The greater part of the respondents 177(59.0%) belonged to rural areas and 123(41.0%) of the respondents belonged to urban areas. Of the 300 respondents, 153(51.0%) respondents' education level was primary while 35(11.7%) of the respondents' education level was middle, 48(16.0%) respondents' education level was matriculation, 28(9.3%) respondents' education level was intermediate, 24(8.0%) respondents' education level was graduation, 11(3.7%) respondents' education level was masters and only 1(.3%) respondent was diploma holder. So majority of the respondents had primary level of education. As for the occupation of respondents was concerned, 20(6.7%) respondents were unemployed while 37(12.3%) respondents were running their own business, 93(31.0%) respondents were doing private jobs, 29(9.7%) respondents were doing government jobs, 108(36.0%) were laborers and

13(4.3%) respondents were doing other jobs. Of the 300 respondents 42(14.0%) respondents' family monthly income was 1000-5000 PKR, while 131(43.7%) respondents' family monthly income was 5100-10000 PKR, 62(20.7%) respondents' family income was 11000-15000 PKR, 35(11.7%) respondents' family monthly income was 16000-20000 PKR and 30(10.0%) respondents' family income was above 20000 PKR. So the majority of the respondents' family income was very low between 5100-10000 PKR.

Table II Parents' Knowledge about Thalassemia Major Disease (N=300)

Ranks	Statement	N	Not Knowledgeable about = 1	Somewhat Knowledgeable about = 2	Knowledgeable about = 3	Very Knowledgeable about = 4	Mean	SD
1	Do you know thalassemia can be detected by taking blood sample of a patient?	300	82(27.3%)	94(31.3%)	80(26.7%)	44(14.7%)	2.29	1.024
2	Do you have knowledge about iron overload?	300	118(39.3%)	37(12.3%)	88(29.3%)	57(19.0%)	2.28	1.172
3	Do you have knowledge that iron over load can lead complication for thalassemia patients?	300	120(40%)	38(12.7%)	82(27.3%)	60(20.0%)	2.27	1.185
4	Do you have knowledge about prenatal diagnosis?	300	111(37%)	74(24.7%)	74(24.7%)	41(13.7%)	2.15	1.070
5	Have you knowledge about the test for detection of thalassemia carrier?	300	98(32.7%)	101(33.7%)	69(23.0%)	32(10.7%)	2.12	.986
6	Do you know that thalassemia is an inherited blood disorder?	300	132(44.0%)	74(24.7%)	50(16.7%)	44(14.7%)	2.02	1.094
7	Do you know about premarital screening?	300	131(43.7)	78(26.0)	49(16.3%)	42(14.0%)	2.01	1.079
8	Do you know about name of test (Hb Electrophoresis) for the detection of thalassemia	300	156(52.0%)	63(21.0%)	46(15.3%)	35(11.7%)	1.87	1.061
9	Do you have knowledge of bone marrow transplant?	300	149(49.7%)	69(23.0%)	55(18.3%)	27(9.0%)	1.87	1.013
10	Do you have knowledge about leukocyte filter?	300	221(73.7%)	53(17.7%)	18(6.0%)	8(2.7%)	1.38	.719

Scale= Not knowledgeable=1; somewhat knowledgeable=2; Knowledgeable=3; Very knowledgeable=4

Table II. The respondents were asked the various questions regarding knowledge of thalassemia major disease. A question was asked to the respondents if they knew thalassemia could be detected by taking a blood sample of the patients. Out of 300 respondents, 94(31.3%) respondents reported that they had 'somewhat knowledgeable' that blood could be detected through blood sample of the patients (μ=2.29±1.024). A question was asked to the respondents if they had knowledge

about iron overload, 118(39.3%) of the respondents shared that they had 'no knowledge' of iron overload (μ=2.28±1.172). A question was asked to the respondents if they had knowledge that iron over load could lead to complication of thalassemia major child, mostly respondents 120(40.0%) were 'not knowing' that iron overload was a complication for thalassemia major disease (μ=2.27±1.185). A question was asked to the respondents if they had knowledge about prenatal diagnosis, majority of the respondents 111(37.0%) were 'not having knowledge' of prenatal diagnosis (μ=2.15±1.070). Out of 300 respondents 101(33.7%) rated that they had 'somewhat knowledge' about the test for detection of thalassemia carrier (μ=2.12±.986). A question was asked to the respondents if they knew that thalassemia was an inherited blood disorder, majority of the respondents 132(44.0%) reported that they had 'no knowledge' that it was an inherited blood disorder (μ=2.02±1.094). A question was asked to the respondents if they knew about premarital screening, out of 300 respondents 131(43.7%) reported that they had 'no knowledge' about premarital screening (μ=2.01±1.079). A question was asked to the respondents if they knew about the name of test Hb Electrophoresis for the detection of thalassemia major disease, mostly respondents 156(52.0%) did not answer properly that thalassemia major disease could be detected through Hb Electrophoresis (μ=1.87±1.061). A question was asked to the respondents if they had knowledge of bone marrow transplantation, majority of the respondents 149(49.7%) reported that they had 'no knowledge' about bone marrow transplantation (μ=1.87±1.013). A question was asked to the respondents if they had knowledge about leukocyte filter, so majority of the respondents expressed that they had 'no knowledge' of leukocyte filter (μ=1.38±.719). Table III. The respondents were asked the multiple statements regarding the parents' burden of thalassemia major children. A question was asked to the respondents if thalassemia disease affected their financial situation, of the 300 respondents, majority 207(69.0%) of the respondents did 'strongly agree' that thalassemia disease affected their financial situations (μ=4.54±.847). Greater part of the respondents 196(65.3%) did 'Strongly agree' that they wished to acquire more information about thalassemia disease (μ=4.51±.807). Most of the respondents 195(65.0%) did 'strongly agree' to support legislation of mandatory premarital screening before marriage in Pakistan (μ=4.49±.857). A question was asked to the respondents if thalassemia carrier test was available for detection in their area, out of 300 respondents, 136(45.3%) respondents did 'agree' that the test facility was available in few hospitals (μ=3.77±.982). A question was asked to the respondents if they felt hesitation to talk to others about the disease of their children, greater part of the respondents 119(39.7%) did 'strongly agree' that they felt hesitation to talk to others about the disease of their thalassemia children (μ=3.74±1.347). A question was asked to the respondents if thalassemia disease could be treated/ managed by giving blood to the patients, out of 300 respondents, 136(45.3%) respondents did 'Agree' that thalassemia major disease could be treated/ managed by giving blood to the patients (μ=3.41±1.146). A question was asked to the respondents if thalassemia disease affected their family life, out of 300 respondents, majority 169(56.3%) did 'Strongly Agree' that thalassemia disease affected their family life (μ=4.30±1.037). Out of 300 respondents, 110(36.7%) of respondents did 'agree' that their life was successful with thalassemia major child but 93(31.0%) did 'disagree' that their life was not successful because of having thalassemia major child (μ=3.11±1.084). A question was asked to the respondents if thalassemia major was curable/ treatable disease, out of 300 respondents, only 42(14.0%) did 'strongly agree' that thalassemia major disease was curable/ treatable disease while 80(26.7%) respondents did

'disagree' that thalassemia major disease was curable/ treatable (μ=2.70±1.206). Majority of the respondents 134(44.7%) did 'disagree' that thalassemia major disease was treatable by giving medicines likewise 131(43.7%) respondents did 'disagree' that thalassemia disease was transferable from one person to another like infections/ germs (μ=2.47±1.260). In our study majority 297(93.0%) patients were receiving regular medicines and 21(7.0%) were only receiving blood on monthly basis. Majority of the respondents 293(97%) were not ever tested for thalassemia before marriage. Out of 300 respondents, majority 262(87.3%) were not aware of the disease before diagnosis. Only 75(25.0%) respondents indicated that thalassemia centre was available near their home and majority 225(75.0%) reported that thalassemia centre was not available in their areas.

Table III. Parents' burden of Thalassemia major child (N=300)

Ranks	Statement	N	Strongly Disagree=1	Disagree=2	Undecided=3	Agree=4	Strongly Agree=5	Mean	SD
1	Does thalassemia disease affect your financial situation?	300	5(1.7%)	11(3.7%)	7(2.3%)	70(23.3%)	207(69.0%)	4.54	.847
2	Do you wish to have more information about thalassemia?	300	4(1.3%)	6(2.0%)	18(6.0%)	76(25.3%)	196(65.3%)	4.51	.807
3	Do you support legislation of mandatory premarital screen before marriage?	300	6(2.0%)	6(2.0%)	18(6.0%)	75(25.0%)	195(65.0%)	4.49	.856
4	Is thalassemia carrier test available for detection?	300	7(2.3%)	28(9.3%)	60(20.0%)	136(45.3%)	69(23.0%)	3.77	.982
5	Do you feel hesitation to talk to others about the disease of your child?	300	27(9.0%)	42(14.0%)	33(11.0%)	79(26.3%)	119(39.7%)	3.74	1.347
6	Can thalassemia be treated/ managed by giving blood to the patient?	300	23(7.7%)	50(16.7%)	50(16.7%)	136(45.3%)	41(13.7%)	3.41	1.146
7	Does thalassemia disease affect your family life?	300	12(4.0%)	15(5.0%)	12(4.0%)	92(30.7%)	169(56.3%)	4.30	1.037
8	Do you feel that your life is successful?	300	24(8.0%)	69(23.0%)	77(25.7%)	110(36.7%)	20(6.7%)	3.11	1.084
9	Is thalassemia major a curable/treatable disease?	300	33(11.0%)	80(26.7%)	64(21.3%)	81(27.0%)	42(14.0)	3.06	1.240
10	Can thalassemia be treated/managed by giving medicines?	300	38(12.7%)	134(44.7%)	34(11.3%)	67(22.3%)	27(9.0%)	2.70	1.206
11	Is thalassemia transferable (from one person to other like infections/germs)?	300	65(21.7%)	131(43.7%)	37(12.3%)	33(11.0%)	34(11.3%)	2.47	1.260

Scale= Strongly Disagree=1; Disagree=2; Undecided=3; Agree=4; Strongly Agree=5

DISCUSSION

Our study findings are very significant as parents' awareness is very important for the management and prevention of thalassemia major in their children. The level of parents' education was very low in our study because 153(51.0%) respondents' education level was primary. The finding of our study correlates with other study

conducted by (Bashir et al, 2017) where 89.9% couples' education was primary (12). Likewise another study conducted by (Ghafoor et al, 2016) tells that 69% of the parents were illiterate (13). In our study 131(43.7%) respondents' family monthly income was very low 5100-10000 PKR. Similar findings reported by (Maheen et al. 2015) tells that about 51.7% of the respondents' family monthly income was 5000-10000 PKR (14). The findings of our study demonstrated that majority of the respondents 150(57.5%) married first cousins. Similar results found in a study conducted by (Hafeez et al, 2007) that 56.7% of the couples were first cousins (15). Likewise other study conducted by (Cao and Galanello et al., 2002) that cousin marriages have contributed to the increased incidence of this deadly disease (16). Cousin marriages increase the birth of autosomal recessive disorder like thalassemia major (17-18). The study found that main reason for cousin marriages was due to cultural and traditional patterns, so parents prefer to make their children marry in the family without knowing its harmful consequences. In our study 94(31.3%) respondents reported that they were 'somewhat knowledgeable' that blood could be detected through blood sample of patients and 118(39.3%) of respondents indicated that they had 'no knowledge' of iron overload. In this study, the respondents were 'not knowing' that iron overload was a complication for thalassemia major disease. In our study only 7(2.3%) respondents got premarital screening for thalassemia while majority 293(97.7%) respondents had no screening for thalassemia before marriage. A similar finding reported by (Safdar et al, 2017) told that only 1.7% got premarital screening for thalassemia, and 98.2% had never screened for thalassemia before marriage. There is a need to improve the quality of information among thalassemia families and general public by professionals about testing of thalassemia carrier, prenatal diagnosis and termination of pregnancy. In our study 132(44.0%) of respondents reported that they had 'no knowledge' that thalassemia was an inherited blood disorder. In Pakistan for thalassemia prevention and control, premarital screening is more effective instead of prenatal diagnosis as 195(65.0%) respondents in our study rated 'strongly agree' to support legislation of mandatory premarital screening before marriage. Similar results were found in another study conducted by (Ishfaq et al., 2012) that the couples suggested that legislation for mandatory of premarital screening should be made. In our study, respondents 156(52.0%) did not answer properly that thalassemia major disease could be detected through Hb Electrophoresis and mostly respondents 149(49.7%) did not have knowledge of bone marrow transplantation. Due to repeated blood transfusions there is an increased risk of iron overload in thalassemia major patients. So information should be provided to the parents about iron chelation therapy. Our study highlighted that parents had knowledge of iron chelation therapy but very few parents were giving medicines to their children because of affordability and high cost. Thalassemia centers are now providing free of cost medicines to the needy and deserving patient's so the conditions might be improved in future.

CONCLUSION

Parents' awareness about premarital screening and prenatal diagnosis was inadequate. There is a need to understand the density of problem and create awareness among thalassemia families and general public about the prevention in order to reduce burden of the disease in Pakistan.

RECOMMENDATIONS

Thus it is recommended that well efficient National Screening Program should be started to address this national issue, with the emphasis on thalassemia screening in

schools, colleges and universities. Furthermore, there is a need to disseminate awareness of thalassemia prevention in the rural as well as urban areas to overcome the burden of this deadly disease in Pakistan as these promising results have been observed in other countries. Furthermore, a well established primary health care system with well trained staff is available in our country and this could be used to provide good support for prevention of thalassemia disease.

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