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AWARENESS AMONG THE PARENTS OF CHILDREN AFFECTED WITH THALASSEMIA AND IMPACT OF CONSANGUINEOUS MARRIAGES ON THE INCIDENCE OF THALASSEMIA

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Abstract:

Objectives: *This study was conducted to assess the awareness among the caretakers of thalassemia sufferers about the disease, prenatal diagnosis, and premarital screening for carrier detection and to assess the impact of consanguineous marriage in disease transmission in district Sargodha.*

Study Design: *Cross-sectional study.*

Setting: *Sargodha Medical College, Sargodha and Bahawal Victoria Hospital Bahawalpur.*

Period: *April to September 2017.*

Material and Methods: *A sample of 135 thalassemia patients was chosen by random sampling. Caretakers of patients with thalassemia were enrolled for data collection after taking consent. Self-administered questionnaire was given to the respondents and their reflections were observed. Collected data was analyzed using SPSS 16.0.*

Results: *There were 77 male (57%) and 58 female (43%) patients with mean age of 8.16 years. The maximum number of cases was diagnosed at the age of 6 months (27%). About 70 respondents (52%) thought cousin marriages have an impact on incidence of thalassemia. 52 patients (39%) had a positive family history of thalassemia. 91 respondents (67%) told thalassemia as a genetic disease. Only one respondent (0.74%) knew about the pre-marital screening and two respondents knew about the prenatal diagnostic test before the diagnosis of thalassemia in their patients.*

Conclusion: *Insight about thalassemia, prenatal diagnosis, and premarital screening was inadequate among the caretakers even after the diagnosis of the disease. This requires intervention in the form of public health educational programs concentrating on high risk population. Males are affected more than females. The incidence of thalassemia was high in progeny of consanguineous marriages and in castes which strictly marry within their caste.*

Key Words: *Thalassemia, Consanguinity ratio, Awareness of thalassemia.*

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INTRODUCTION:

The thalassemia's are inherited blood disorders and it is the paradigm of monogenic genetic disease and is caused due to mutations in α or β globin chains of the hemoglobin leading to inadequate hemoglobin formation. [1] Abnormal erythroblasts are produced which leads to the anemia. [2] Moreover, ineffective erythropoiesis leads to extramedullary hematopoiesis with bone marrow expansion resulting in skeletal deformities. There is also increased iron absorption by the gastrointestinal tract leading to systemic iron overload or secondary hemochromatosis. [3, 4]

According to an estimate 15% of the world's population are the carriers of β thalassemia and around 50,000 to 60,000 cases are born each year.[5] β thalassemia is prevalent in Mediterranean countries, middle east, southern china, countries along the north African coast and in south America. Highest carrier frequency is reported in Cyprus (14%), Sardinia (10.31%), and Southeast Asia. [6]

In Pakistan thalassemia is the most prevalent genetic blood disorder. Thalassemia carriers have prevalence of 5-7% and in the country 8-10 million populations. According to an estimate, about 100,000 patients suffering from thalassemia major are present in Pakistan and every year this number increases by 5000-9000 [7, 8]. The thalassemia gene is not randomly distributed in Pakistan and is largely restricted to the affected families. According to studies, unawareness about the disease, poverty, and increase number of consanguineous marriages are the prominent factors in increasing the incidence of thalassemia in Pakistan. [9]

The best way to reduce the incidence of thalassemia is by prevention. Throughout the world, the strategies employed for the prevention include prenatal diagnosis, population screening, genetic counseling, termination of pregnancy and parental awareness [10]. Treatment also causes a great financial burden on the state and this can be reduced by preventive strategies. National thalassemia prevention program in Iranian province of Mazandaran demonstrated that an unbearable financial burden can be prevented via such strategies [10,11].

The facilities for prenatal diagnosis and carrier detection are available in Pakistan for over a decade but their use remains limited due to lack of awareness, poor access to services and high procedure costs [12]. In Pakistan the provinces of Punjab, KPK and Sindh have passed laws of making screening mandatory for couples intended to get married. However, due to lack of public awareness and government interests these laws are not properly implemented [13].

This study evaluates the consanguinity ratio and the knowledge of attendants regarding the thalassemia in district Sargodha. A limited number of studies regarding this disease have been conducted in some regions of Pakistan but no such study has been done on designated area. The report aids to fill the gap in literature and contributes to it. This research aims to facilitate the policy makers, philanthropists and health professionals to work conjointly in deterrence of thalassemia.

MATERIALS AND METHODS:

The cross-sectional study was carried out on the patients of Hilal-e-Ahmar Hospital, Sargodha, Safe Thalassemia Center, Sargodha from April 2017 to September 2017, and Thalassemia Center BVH. The approval was obtained from head of institution for conducting the study. A total of 135 attendants of patients suffering from Thalassemia were included in the study. Sampling was done by using convenient sampling technique. A self-administered questionnaire was prepared for data collection and distributed among attendants, after elucidating basis of study and taking verbal consent. The questionnaire was handed over to attendants and requested to return completed questionnaire. Attendants were asked about basic demographic characteristics and variables of interest (Age of the patient at the time of diagnosis, Type of thalassemia, Consanguinity ratio, Family history of thalassemia, Education of respondent, Knowledge about thalassemia, Knowledge about test for prenatal diagnosis, Knowledge about treatment of thalassemia and Knowledge about premarital screening). All the data collected from participants were entered in SPSS version 16.0 and descriptive statistics was used to analyze the data.

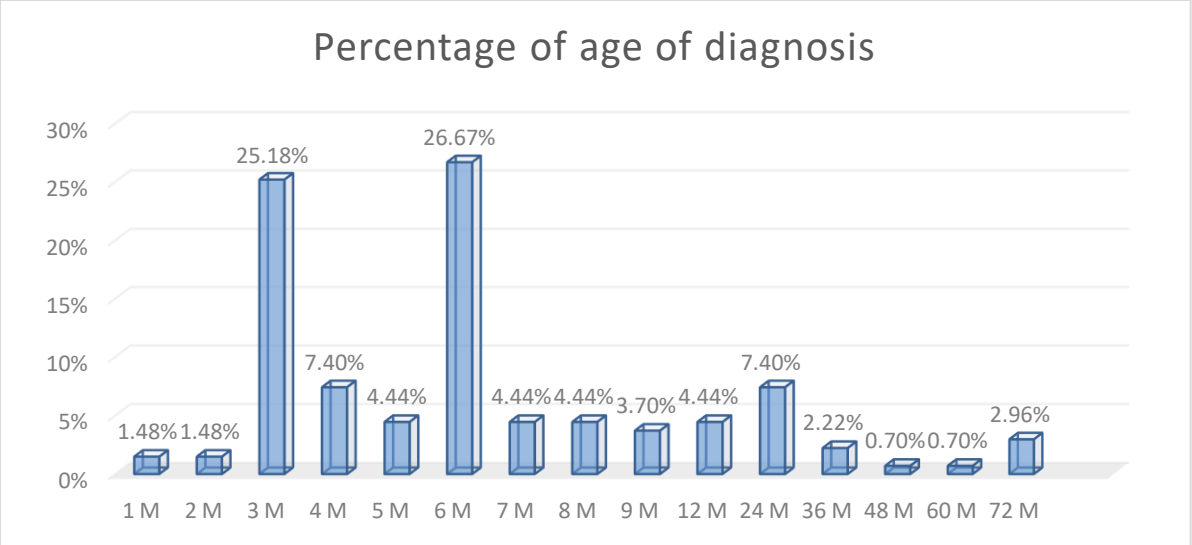

Figure 1: Age wise distribution at which thalassemia was diagnosed in patients

Relationship	Consanguinity Ratio
1st cousin	71.85%
2nd cousin	10.37%
Distant relation	5.19%
No relation	12.59%

Figure 2: Factual consanguinity ratio among the patients of thalassemia included in the study (n=135).

Ninety one respondents (66%) answered beta-thalassemia major as subtype of their patient's disease, 10 respondents (7%) answered beta-thalassemia minor and 34 (25%) respondents were unaware of subtype. Only one respondent knew about subtypes of thalassemia before the diagnosis of this disease in their patient. The rest 134 respondents didn't.

A total of 66 (49%) respondents knew about types of thalassemia after the diagnosis of this disease in their patient, while 69 respondents (51%) didn't have any knowledge in this regard.

A total of 70 respondents 52% thought cousin marriages have impact on prevalence of thalassemia, the remaining 65 respondents (48%) answered that the cousin marriages have no impact. Positive family history of thalassemia was found in 52 (39%) patients and remaining 83 (61%) had negative family history.

Consanguineous marriages were in 111 (82%) and out of this 97 were first cousins (72%), 14 (10%) were second cousins. Out of the remaining 7 (5%) were distant relatives and 17 (13%) had no relation prior to marriage (Figure 2).

Thalassemia is completely curable was the opinion of 38 respondents (28%) while 97 respondents (72%) thought thalassemia had no cure. A total of 81 (60%) respondents answered blood transfusions as the only treatment for thalassemia. Among the respondents, 54 (40%) answered that there is some alternate treatment besides blood transfusions. Out of these, 43% told bone marrow transplant, 33% told medicines, 19% answered iron and 5% answered Hakeem medications, as an alternative treatment to blood transfusions.

Responding to the question about the etiology of the disease, 91 respondents (67%) told thalassemia as a genetic disease, 5 respondents (4%) told it as an infectious disease, 1 respondent thought it was contagious disease and 38 respondents (28%) told other etiology beside these. Only 1 respondent had heard about the pre-marital screening test before diagnosis of their patient while remaining 134 (99%) didn't have any awareness. After the diagnosis of thalassemia in their patients, 105 (78%) of respondents have awareness about the pre-marital screening for thalassemia and only 30 (22%) of respondents had no idea about the screening test. Only 2 respondents had knowledge about the prenatal test before diagnosis of their patient while remaining 133 (98%) didn't have any awareness. After the diagnosis of thalassemia in their patients, 113 (84%) of respondents have awareness about the prenatal test for thalassemia and only 22 (16%) of respondents have no idea about the prenatal test. Summary of the results in given in Figure: 3

Respondent's Awareness/Thinking About disease of their patient	Yes	No	Unsure
About beta-thalassemia major	67%	8%	25%
About types of thalassemia before diagnosis	1%	99%	
About types of thalassemia after diagnosis	49%	51%	
About cousin marriage impact on thalassemia	52%	48%	
Thinking that thalassemia is curable	28%	72%	
Bone marrow transplant is an alternate treatment to blood transfusion	43%	57%	-
Hakeem medicine is an alternate treatment to blood transfusion	5%	95%	-
Iron therapy is an alternate treatment to blood transfusion	19%	81%	-
Medicines are an alternate treatment to blood transfusion	33%	67%	-
Blood transfusion is the only treatment	60%	40%	-
About pre-marital screening before diagnosis	1%	99%	-
About pre-marital screening after diagnosis	78%	22%	-
About prenatal test before the diagnosis	1%	99%	-
About prenatal test after diagnosis.	84%	16%	-
About etiology of disease is genetics	67%	33%	

Figure 3: Summary of Results from Questionnaire

DISCUSSIONS:

It is well established that carrier detection and genetic counseling in high risk population is very effective in preventing thalassemia. It is reported that roughly around 60000 thalassemic babies are born annually worldwide[14]. This estimated number for thalassemia major babies born in Pakistan is over 5000[15]. There is no cure for this disease except bone marrow transplant, which is very expensive and sparse expertise along with very few hospitals available in country like Pakistan. Therefore prevention is the key which is successfully achieved in some countries[16]. We are presenting data regarding awareness among 135 attendants of thalassemia patients from two centers of thalassemia in City of Sargodha and Bahawalpur, Punjab, Pakistan. There were no formal awareness sessions with health care workers at these centers before the collection of data. Findings presented here are of value as awareness of parents and general masses is important for the prevention and good management of thalassemia sufferers[17].

Thalassemia has no sex predilection, in our study number of males (57%) outnumbered females. This study is in line with other studies conducted in Pakistan, Bangladesh and India[18-20]. This gender-ratio difference in thalassemia patients is noteworthy and deserves further investigation considering thalassemia as a single-gene disease transmitted by a recessive mode of inheritance. One possible reason is the fact that the people are more concerned with the health of the male offspring and, hence, are more

likely to seek medical care for them[21]. According to recent census, the population of male (51%) is more than that of females (49%), which could be a reason for more number of male patients. Third reason could be that our study was center based. The less number of females could be due to an attempt on the parent's behalf to hide their daughter's illness for avoiding social stigma and marital problems that can arise from it.

In the current study, just less than half of the patients with thalassemia had a positive family history in contrast to Humaira Zafar et al reported only 5% with family history of thalassemia[22]. This difference could be because our study was conducted in a backward and remote areas of Punjab province where illiteracy dominate as compared to the study done in Islamabad the capital and well developed city of Pakistan. Moreover around 50% of our study participants believed consanguineous marriages has no role in thalassemia spread. This may suggest that despite the respondents being a relative or parent of already existing thalassemia child, their approach, knowledge and behavior for preventative strategy is negative. Similar findings were reported by Maheen et al[23].

In this study we found that majority of Thalassemia children (82%) were born from consanguineous marriages either first or second cousin marriages. Similar results (higher frequency of consanguineous marriage) were reported by different authors in Pakistan[24]. Our study is in contrast with a study in India which had consanguineous ratio 12.7%[25]. The higher rate of consanguineous marriages is due to traditional practice of intra-caste marriages for various social reasons. The castes known for intra-caste marriages have a high incidence of thalassemia,

Jatt (18%), Arain (12%), Rajput (8%), Mughals, Baloch, and Khokhar around (5%).

It is so discouraging that majority of the participants (98%) were not aware about the prenatal diagnosis before the birth of thalassemic child in the family, which is in contrast to similar studies done at Islamabad, Lahore[22]. On the other hand it was encouraging that this figure reduced to 22% after the introduction of a thalassemic child in the family. This indicates that a culturally appropriate linguistically diverse thalassemia prevention programme can change the situation.

In this study 60% of the responded participants said thalassemia could be treated with multiple blood transfusions similar to the results of other studies in this regard[22]. Unfortunately 20% thought iron is the treatment option for these patients which is due to lack of education and understanding as iron chelators are part of treatment in multiply transfused patient rather iron supplement. It has already been proven in previous studies that the incidence of thalassemia major is inversely related to the level of education and knowledge of the parents[26]. Not surprisingly in our study around one fourth of the respondents were illiterate.

CONCLUSIONS:

Consanguineous marriages have high incidence of the disease than the unrelated couples. Illiteracy among caretakers of thalassemia patients is high and knowledge about the disease is insignificant even after the diagnosis of thalassemia in their patient. The incidence of thalassemia is high in those castes which strictly marry within their caste. Present results suggest an urgent need to design a thalassemia education and awareness programme to raise the awareness for extended family screening of the existing patients on priority basis and of the general population as well.

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