

# Awareness Among Parents of $\beta$ -Thalassemia Major Patients, Regarding Prenatal Diagnosis and Premarital Screening

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## ABSTRACT

**Objective:** To assess the knowledge among parents of thalassemia major patients about prenatal diagnosis, premarital screening for carrier detection and impact of consanguineous marriage on disease transmission.

**Study Design:** Descriptive study.

**Place and Duration of Study:** The Thalassemia Centre, Sir Ganga Ram Hospital, Lahore, from July to September 2009.

**Methodology:** One hundred and fifteen parents of  $\beta$ -thalassemia major patients were enrolled in this study. A questionnaire was developed and parents were interviewed to assess their knowledge about preventive measures against thalassemia major. Parents of patients with all other types of blood disorder were excluded from the study.

**Results:** There were 74 male (64.3%) and 41 female (35.7) patients with mean age of  $9.5 \pm 5.1$  years. Eighty-eight patients (76.5%) were accompanied by mothers and the rest by their fathers. Seventy-four parents (32.1%) were illiterate; among the literates only 7 were highly educated (3%). Ninety-four couples (81.7%) had consanguineous marriage. Fifty-two parents (44.6%) knew that thalassemia is an inherited disorder. Thirty-eight (33%) had heard about the test for detecting thalassemia carrier. Premarital screening and prenatal diagnosis was known to 97 (84.3%) and 88 (76.5%) parents respectively. Ninety-nine parents (86.1%) knew about the termination of pregnancy on positive prenatal test but only 69 considered it acceptable religiously (60%). Major source of information to the parents were doctors.

**Conclusion:** Parental knowledge about thalassemia and its preventive measures was inadequate; this requires intervention in the form of public health education programs concentrating on high risk/targeted population.

**Key words:** *Thalassemia major (TM). Chorionic villus sampling (CVS). Prenatal diagnosis. Knowledge.*

## INTRODUCTION

Thalassemia is the world's most common hereditary disease and is a paradigm of monogenic genetic disease. About 3% of the world's population carries gene for beta-thalassemia.<sup>1</sup> In Pakistan, the carrier status is estimated around 5 - 7% and over 5000 thalassemia homozygotes are born every year.<sup>2</sup>

Thalassemia is a clinically heterogeneous disorder resulting from different genetic lesions that variably impair globin chain synthesis. It is a preventable disease demonstrated in countries like Italy, Greece and Cyprus. They were amongst the first to establish successful national programs resulting in significant reduction in the births of affected children. World Health Organization's projections of treatment costs have shown that without prevention programs (to limit affected births) most countries will be unable to offer the optimal treatment to all thalassaemic patients. In Iran, the annual prevention cost is constant but annual treatment costs rise year by

year. National Thalassemia Prevention Program in the Iranian province of Mazandaran demonstrated that an unbearable financial burden can be prevented.<sup>3</sup> Similarly, other studies reported the significant positive effect of such educational programs to increase the knowledge of parents about the disease.<sup>4,5</sup>

The main prevention strategies comprise of providing appropriate information for the public and professionals, screening and counselling of families at risk and screening of general population prior to marriage.<sup>6</sup> Appropriate and extensive screening, accurate detection and counselling of at risk couples along with prenatal diagnosis are promising strategies for the reduction of mortality and morbidity from thalassemia in countries, where it is prevalent.<sup>7</sup> Premarital screening alone has tremendously decreased the birth prevalence of  $\beta$ -thalassemia major.<sup>8</sup>

Prenatal diagnosis and carrier detection is available in Pakistan over a decade but its use remains limited due to lack of public awareness and the cost involved. Naseem *et al.* reported that more than half of the families of the registered thalassaemic patients did not request for prenatal diagnosis.<sup>9</sup> Factors identified for this low utilization of services were lack of awareness, poor access, delay in seeking advice and high cost.<sup>9</sup>

The aim of the current study was to determine the awareness among parents of thalassaemic children

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regarding thalassemia major and their preferences of preventive strategies like prenatal diagnosis and premarital screening.

## METHODOLOGY

This was an observational questionnaire based study conducted from July to September, 2009 at Thalassemia Centre of Sir Ganga Ram Hospital, Lahore. A total of 115 parents (either father or mother) of  $\beta$ -thalassemia major patients were recruited in the study by using non-probability and consecutive sampling technique. A written informed consent was obtained from the parents who were interviewed.

$\beta$ -thalassemia major patients were diagnosed on the basis of history and Hb-electrophoresis with quantification of HbF and HbA2 by high performance liquid chromatography (HPLC).

Parents of the patients with other blood disorders like alpha-thalassemia, thalassemia intermedia, congenital dyserythropoietic anaemia and hereditary spherocytosis etc. were excluded. A semi-structured questionnaire was devised to collect data from study participants. The data was analyzed using Epi-Info version 6, CDC, Atlanta Georgia. Information has been presented in frequency distribution and percentages.

## RESULTS

A total of 115 families of thalassaemic patients were initially enrolled in this study. Among thalassaemic children, 74 were males (64.3%) and 41 were female (35.7%). The mean age of the patients was  $9.5 \pm 5.1$  years, median age of the patients being 8.6 years. A total of 88 patients (76.5%) who visited the Thalassemia Centre were accompanied by mothers and rest by their fathers. Regarding educational status of parents, out of 230 parents (both mothers and fathers) 74 parents (32.1%) were illiterate, 60 had 1-8 years of schooling (26%), 50 had 9-10 years of schooling (21.7%), 25 had 11-12 years of schooling while 14 (6.0%) had schooling of 13-14 years (10.7%) and only 7 had schooling of more than 15 years (3%).

In 115 studied families, 45 families (39.1%) had total earning of below Pakistani Rupees (PKR) 6000, 54 families (47%) were earning between PKR 6000 and 15000 and only 16 were earning more than PKR 15000 per month (13.9%).

The distribution of disease among different caste groups revealed that 77 of parents (33.5%) were *Rajput* followed by 28 *Arain* (12.8%), *Jatt* (11.7%), *Mughal* (9.5%) and *Sheikh* (8.2%). A total of 94 had consanguineous marriages (81.7%); of them 75 were first cousins (65.3%). The remaining 21 had non-consanguineous marriages (18.3%). Sixteen were from the same caste (14%) and only 5 couples (4.3%) had married outside their caste.

**Table I:** Parental knowledge regarding thalassemia carrier screening and prevention strategies (n = 115).

Knowledge assessment	Yes	No
Test available for detecting thalassemia carrier	38 (33%)	77 (67%)
Naming test (Hb. Electrophoresis) for thalassemia carrier	14 (12.2%)	101 (87.8%)
Screening for thalassemia in other children	47 (40.9%)	68 (59.1%)
Premarital screening	97 (84.3%)	18 (15.7%)
Support for legislation of mandatory premarital screening	101 (87.9%)	14 (12.1%)

**Table II:** Parental knowledge regarding prenatal diagnosis of thalassemia (n = 115).

Knowledge assessment	Yes	No
Is there a diagnostic test during pregnancy	88 (76.5%)	27 (23.5%)
Correctly named test-Chorionic Villus Sampling (CVS)	10 (8.7%)	10 (91.3%)
Correct timing of performing test (CVS) during pregnancy	73 (63.5%)	42 (36.5%)
Understanding the need for termination of pregnancy	99 (86.1%)	16 (13.9%)

Among the accompanying parents, 52 knew that thalassemia is an inherited blood disease (44.6%), while 63 parents (55.6%) did not know about its etiology, only 37 parents (32.2%) had knowledge that both parents should be carrier for disease appearance in their children. Table I represents the knowledge of parents of thalassemia major regarding carrier detection, test available for it, premarital screening and its legislation.

Table II shows the parental knowledge about pre-natal diagnosis and understanding the need for termination of pregnancy.

It was noted that 82 parents (71.2%) got information about the disease from doctors, others being paramedics (n=2, 1.8%), electronic media (n=10, 8.7%), print media (n=4, 3.5%), seminars (n=3, 2.6%), internet (n=1, 0.9%) and relatives (n=4, 3.5%).

## DISCUSSION

The acceptability and effectiveness of preventing thalassemia by carrier detection and genetic counselling in high risk population is well established. It is estimated that every year about 60000 thalassaemic babies are born all over the world.<sup>10</sup> There is no known cure but prevention is possible, practicable and successfully achieved in some countries.

In the current study, the numbers of thalassaemic males (64.3%) were more as compared to females (35.7%) in the affected families. Similar, male preponderance was reported by studies from Bangladesh and India.<sup>11,12</sup> On the other hand, a study from Karachi reported 57.5% females affected from thalassemia major.<sup>13</sup>

Level of literacy in the community has a positive impact over parent's behaviour as to how they handle the disease or use prenatal diagnosis to identify the risk of thalassemia in pregnancy. A study conducted in

Rawalpindi, found a significant improvement in the use of prenatal diagnosis of pregnant women with increasing mother's education.<sup>9</sup> On the contrary, in this study, level of literacy among participant mothers was low in comparison.

Majority of the studied thalassemic parents belonged to low income group. This study was carried out in the Government-sponsored Thalassemic Centre where medicines, infusion pumps, syringes, transfusion kits etc. are either provided free of cost or at discounted price and where non-affording parents bring their children for treatment.

Thalassemia major was found to be most common in children belonging to Punjabi *Rajput* caste (33.5%). A study by Hafeez *et al.* also revealed similar result.<sup>14</sup> One obvious reason could be more numbers of *Rajputs* as compared to the other ethnic groups in this region resulting in disproportionate presence of *Rajputs* among study participants.

Consanguineous marriages increase the birth prevalence of autosomal recessive disorders like thalassemia.<sup>15,16</sup> This study found that 81.7% patients were the outcome of consanguineous marriages. These results are comparable with other studies, where frequency of cousin marriages was also found high.<sup>14,17</sup> This high rate of consanguineous marriages is basically due to our local cultural pattern and traditions where people prefer to marry within families without knowing its grave consequences. An Iranian study reported a significant association between  $\beta$ -thalassemia major and first cousin marriages comparing with community at large ( $p < 0.00001$ ).<sup>18</sup>

Knowledge of parents regarding thalassemia as an inherited disorder was comparatively better in this study. In contrast, a study conducted at Civil Hospital, Karachi, documented that only 15% parents were aware of the nature of disease transmission and only 12% knew consanguinity as a risk factor for thalassemia major.<sup>13</sup> One of the reason of this contrast could be that thalassemia Centre, at Ganga Ram Hospital is one of the most established centre in Pakistan where more than 1300 patients are registered and daily attendance in OPD is about 70-80 patients. Moreover, patients and their parents used to attend genetic counselling sessions and seminars more regularly and parent-to-parent interaction is much higher.

There is limited knowledge of accurate frequency and distribution of thalassemia disorder in the developing countries.<sup>19</sup> In Jordan 75% of families have not heard about the disease before having their first affected child.<sup>20</sup> Similarly, insufficient knowledge also have been found in UK based Pakistani families about the risk of carrying a gene that may result in the birth of a thalassemic child.<sup>21</sup> There is a need to improve the quality of information disseminated by professionals

about carrier testing, prenatal diagnosis and termination of pregnancy.

Premarital screening instead of prenatal test (CVS) may be a good strategy in prevention and control of thalassemia in Pakistan as (87.8%) couples in the existing study suggested a need for legislation for mandatory thalassemia screening before marriage. In their view, religious beliefs in society make it difficult to have therapeutic abortion following a positive CVS test even though, facilities for termination of pregnancy is being offered in our hospital soon after diagnosis. Therefore, more emphasis on premarital screening is required to prevent the marriages amongst carriers which may ultimately prevent the birth of thalassemic babies. In this study 88 knew about prenatal diagnosis (76.5%), consistent with another study conducted in Bahawalpur.<sup>22</sup> Among study participants who were aware of prenatal diagnosis only 39.1% mothers had undergone this test during their pregnancies while others did not avail this facility. One possible reason could be high cost of CVS test because most of the participant families belonged to lower socio-income group but another contributory factor may be religious restriction on abortion as the only available option for pregnant woman with affected fetus that prevents them to have prenatal testing. In contrast, a study in North England found that Pakistani women's attitude towards prenatal diagnosis (PND) and termination of pregnancy was influenced by various factors and their religion could not be taken as a proxy for their attitudes either for or against termination of pregnancy.<sup>23</sup>

An important limitation of this study was that subjects were taken only from a single Thalassemia centre whereas views of parents from other centres could not be evaluated in this study.

## CONCLUSION

Parental knowledge about the thalassemia carrier screening and prenatal diagnosis was inadequate. It is the need of time to understand the density of problem and educate general public and create awareness among thalassemic families about the preventive aspects of thalassemia in order to reduce the burden of disease in Pakistan.

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