

## Original Research Article

# Awareness about thalassemia and feasibility of cascade screening in families of thalassemia major patients

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### ABSTRACT

**Background:** The main objective of this study is to assess knowledge and attitude of parents & extended relatives of children diagnosed with Thalassemia major and to evaluate feasibility of screening of extended families for thalassemia trait by correlating their willingness to get screened with actual screening.

**Methods:** This was a cross sectional type of descriptive analytical study, conducted at the thalassemia day care center at a tertiary care teaching hospital in western India among 230 study population using a pre-designed and pre-validated structured questionnaire.

**Results:** We found the overall knowledge score of parents about thalassemia was significantly better than relatives. About 95.83% of parents and only 31.86% of the relatives amongst the study population had adequate knowledge. Majority of study population (84.61% relatives and 100% parents) had positive attitude. The significant contributing factors for knowledge and attitude towards thalassemia were age, marital status, education level, religion and per capita monthly income. 2% relatives agreed to get themselves screened for thalassemia status out of which only 20% got themselves screened.

**Conclusions:** This study has identified key areas which need to be highlighted and emphasized in public awareness campaigns for thalassemia screening in India.

**Keywords:** Attitude, Awareness, Knowledge, Parents, Relatives, Screening, Thalassemia

### INTRODUCTION

Thalassemia is a congenital hemoglobinopathy caused by the absence, or decrease, of one or more of the globin chains in red blood cells, and it represents a serious global public health concern. Over 56,000 affected babies are born annually and at least 5.2% of the global population are carriers.<sup>1</sup> The tropics have the highest incidence, where 7% of pregnant women are carriers.<sup>2,3</sup> The countries mainly affected are those in the Mediterranean, such as Italy, Greece and Cyprus and in Asia countries like China, India and countries in South East Asia including Malaysia. In India it is the most common single-gene disorder.<sup>4</sup> Over thirty million people

are carriers of thalassemia gene in our country.<sup>5</sup> Ten percent of total children with thalassemia in the world are born in India every year.<sup>6</sup> The requirement of regular blood transfusions every month, expensive treatment of iron overload by chelators, lack of adequate thalassemia care centers, inadequate financial resources for various treatment modalities and above all the lack of knowledge regarding thalassemia itself, affect the child and his family economically and socially leading to irregularities in treatment and consequently adverse outcomes for the child.

The effectiveness of preventing thalassemia by carrier detection and genetic counselling in high risk population

is well established. Over the past three decades, regular blood transfusions and iron chelation have dramatically improved the quality of life and transformed thalassemia from a rapidly fatal disease in early childhood to a chronic disease compatible with prolonged life.<sup>7,8</sup> Despite increased life expectancy, complications keep arising. There is an urgent need for making the people aware of this lethal malady. Health education is an important component of the preventive genetic programs. The detection of carriers by premarital screening along with antenatal and inductive or cascade screening seems to be the most fruitful strategy for beta thalassemia. A WHO update on beta thalassemia in India indicated overall carrier frequency of 3-4%, which given the current national population would translate to between 35.6 and 47.5 million carriers of the disorder nationwide.<sup>9</sup> This health burden emphasizes the need for prenatal diagnosis and carrier status - detection to contain the disease and reduce the load of the mutant alleles in the gene pool. Primary prevention, based on a combination of increased awareness of general population, screening of carriers, genetic counselling, and prenatal diagnosis, has led to almost total elimination of thalassemia in Cyprus and, to a considerable extent, in Greece, Italy and Sardinia.<sup>10-12.</sup>

Diseases that are known to run in families and have a high-risk of recurrence become a social stigma when nothing or very little is known about them. In preventing this disease, attitude of population at large and that of families directly involved in care of affected children are of prime importance. Prevention of thalassemia is possible only by sensitizing the problem at the individual, social and state levels. This forms the basis of our study.

## METHODS

This was a descriptive analytic study which was conducted with the aim of assessing the level of knowledge and attitude of parents and extended relatives (2<sup>nd</sup> and 3<sup>rd</sup> degree) of children diagnosed with thalassemia major at a tertiary care center in Mumbai which is a government run major hospital in India. The study period was of 15 months from November 2013 to February 2015. The parents of children diagnosed with thalassemia major who consented were included in the study. Among the relatives, second degree relatives of the index case, who agreed to participate were part of the study. Those did not consent were excluded from the study. Study population was selected via convenience non-probability sampling method. The sample size calculated as per the prevalence of thalassemia in India for an accuracy level of 0.95 with a confidence interval of  $\pm 2.0\%$  was 230. Their awareness was assessed by telephonic interviews through a pre-designed questionnaire.

A 20 item questionnaire was designed to test knowledge and attitude of parents and relatives about various aspects of thalassemia. Questionnaire included 13 Knowledge based questions and the responses categories were “Yes”,

“No”, “Don’t know”, the last being included to discourage guessing. These questions assessed knowledge about various domains of thalassemia such as transmission, symptoms, carrier state, diagnosis, antenatal screening and treatments aspects. Rest 6 questions were based on Attitude and the responses were to be marked on a Likert scale of ‘strongly disagree’ to ‘strongly agree’ continuum, as 1 to 6 respectively. (1-Strongly disagree, 2-Disagree, 3-Somewhat disagree, 4-Somewhat agree, 5-Agree, 6-Strongly agree). The methods used to determine the validity and reliability of the questionnaire were Content and Face Validity and Test-Retest Reliability respectively. Content and face validity of the questionnaire was determined by evaluation by 5 Pediatricians and 10 volunteer subjects and accordingly 5 items were removed from the questionnaire. Inter observer correlation (Cronbach’s alpha) was assessed among two observers after a wash out period of one week and correlation was found to be significant with a correlation coefficient of 0.9. Test-retest reliability was tested by administering questionnaire to patient twice with a wash-out period of 1 week. Test-retest reliability was good with correlation coefficient of 0.9. Cross-cultural adaptation of English version of validated questionnaire to regional language (Marathi and Hindi) was done.

After collection, the data was analyzed by SPSS 21 software using descriptive analytic methods. In this study, the level of significance was considered as  $p < 0.05$ . Quantitative data was represented using mean  $\pm$  standard deviation (SD) and Median and IQR (Interquartile range). Qualitative data was represented in the form of frequency and percentage. The association between qualitative variables was assessed by Chi-Square test.

## RESULTS

A total of 250 subjects (parents and relatives) were approached to participate in the study, amongst them 235 subjects (94%) consented to take part in the study; about 5 subjects (relatives) were excluded from the study as they had not heard about thalassemia and refused to continue the questionnaire. A total of 230 study subjects were therefore included in the analysis, of which 48 were parents and 182 relatives. The socio demographic characteristics of the study population showed of the total 230 study population, 118 (51.3%) were males; mean age was 33.5 years (ranging from 18 to 70 years); 84.78% subjects belonged to urban areas, 79.13% were married; their level of education ranged between illiterate to postgraduate with 4.35% being illiterate, 14.78% received primary education, 36.09% studied till secondary education, 16.96% received higher secondary education, and 27.83% were graduates. Regarding the occupation of the study population 23% were skilled workers, 16.52% were professionals, 10% were self-employed, 33% were homemakers and 11.30% were students.

The knowledge and attitude were assessed by a 20 point questionnaire wherein for each question, a correct response was given a score of one, and an incorrect was scored as zero, total possible score was 0-13, with higher scores indicating better knowledge (Table 1). Study population with scores of 8(60%) and above were considered as having “adequate knowledge” and scores below 8 were interpreted as “inadequate knowledge”. The mean score of knowledge amongst parents was 11.45±0.5 and amongst relatives was 4.29±0.5. We found 95.83% parents and only 31.86% relatives had adequate knowledge about thalassemia. Similarly attitude was assessed using 6 questions, (negative and positive attitude if the correct questions score were less than 50% and more than or equal to 50% correct scores respectively) and it was found 100 % parents had positive towards the disease. While 84.61% relatives compassed positive attitude towards the disease.

**Table 1: Knowledge and attitude score of parents and relatives.**

	Relatives	Parents
Knowledge (13 items)	Number (n)	Number (n)
Adequate (score ≥8 or 60%)	58(31.86%)	46(95.83%)
Inadequate (score ≤7 or 59%)	124(68.13%)	2(4.16%)
Attitude (6 items) (mean score)	Number (n)	Number (n)
Positive (score ≥2.10)	154(84.61%)	48(100%)
Negative (score ≤2.00)	28(15.38%)	0

As per Table 2 parents were relatively well informed about all domains of the disease. Among the relatives only 37.91% knew that it was a hereditary disorder,

**Table 3: Descriptive statistics of attitude of study population about thalassemia.**

Attitude based questions	Parents (n=48)		Relatives (n=182)	
	Agree	Disagree	Agree	Disagree
Individuals who are carrier for thalassemia should not marry each other	87.5%	8.33%	74.7%	7%
Thalassemia carrier couples should not have children	33.3%	64.5%	33.5%	1.8%
Termination of pregnancy with Thalassemia major foetus	100%	0	95%	1%
Screening for thalassemia be made compulsory before marriage.	97.91%	0	96%	4%
Disclosure of thalassemia trait status before marriage affects the prospect of getting married	68.75%	15%	89.5%	4%
Screening oneself or thalassemia carrier status.	48%	0	52%	22.5%

Thalassemia (N=230)

The association between socio demographic characteristics and knowledge and attitude obtained using chi square test. The significant contributing factors (p<0.05) with associated adequate knowledge about thalassemia were middle age (25-45 years), marital status (married), education level (Graduates and above), and

36.26% were aware about antenatal screening of the disease and only 27.47% were informed regarding the carrier state. Most frequently cited sources of information about thalassemia were relatives with thalassemia major children (98.25%).

On assessing the attitude of the study population regarding various aspects of thalassemia (Table 3) majority of parents (87.5%) and relatives (74.7%) believed that thalassemia carriers should not marry each other, 33% of both relatives and parents thought that couples who are thalassemia carrier should not have children, a strong majority of parents thought (100%) and relatives (95%) believed that pregnancy with thalassemia major should be terminated. Again vast majority of parents (97.91%) and relatives (96%) felt that screening for thalassemia be made compulsory before marriage. 68.75% parents and 89.5% relatives believed that disclosure of thalassemia trait status before marriage affects the prospect of getting married.

**Table 2: Distribution of the study population according to knowledge about Thalassemia and sources of information (N=230).**

Various domains of knowledge about thalassemia	Percentage score in parents	Percentage score in relatives
Transmission	91.67%	37.91%
Symptom and nature of disease	89.58%	30.03%
Thalassemia trait	87.5%	27.47%
Diagnosis	95.83%	44.51%
Antenatal screening	95.83%	36.26%
Treatment	79.68%	17.72%

PCMI (class IV and V). Similarly the significant contributing factors towards positive attitude were education (secondary and above), occupation (students and service holders), PCMI (upper socio economic scale) and religion (Hindu religion).

In assessing feasibility of cascade screening in our study population we found only 52% relatives agreed to get themselves screened for thalassemia status (Table 4). Out of which only 19(10.43%) got screened while 41(22.52%) relatives disagreed for getting themselves

screened and none got screened, thus willingness to get screened strongly correlated with actual screening ( $p=0.00006$ ). Additionally among 182 relatives 12 (6.59%) were already screened for thalassemia and only 7 (3.85%) were screened during our study.

**Table 4: Correlation of willingness to get screened and actually got screened for relatives.**

Willingness to get screened	Screened for Thalassemia carrier state	Not screened for carrier state	Total	Chi square p value
Strongly Agree/ Agree	19(10.43%)	76(41.75%)	95	19.42; 0.00006
Strongly disagree/ Disagree	0	41(22.52%)	41	
Indecisive	0	46(25.27%)	46	
Total	19	163	182	

## DISCUSSION

Thalassemia is the commonest monogenic disorder globally with Indian sub-continent sharing one of the highest burdens.<sup>13</sup> In comparison to its prevalence overall awareness about thalassemia is scarce in our population. This is the one of the few studies from India to objectively assess the awareness about thalassemia and the only one to do comparative analysis of knowledge and attitude of parents and extended families of thalassemia major patients.

Our study revealed parents had significantly better knowledge about various domains of thalassemia than the relatives. Previous studies have also highlighted the poor knowledge amongst high risk population in India.<sup>14,15</sup> In contrast to our study, Goyal et al, concluded poor awareness about thalassemia in parents of thalassemia patients in their study.<sup>16</sup> Though overall knowledge about thalassemia was adequate in parents in our study but they were relatively poorly informed about treatment aspect. Only 56% parents were aware about bone marrow transplantation (BMT) as cure of thalassemia. With more and more BMT centers emerging in our country, emphasis should be laid on educating families of such patients about this treatment modality. On contrary the knowledge of extended families about all aspects of thalassemia was poor. Only 14 % of relatives were aware that children of individuals with thalassemia trait can have thalassemia. This implies poor understanding of this high-risk population about carrier state and genetic transmission of this disease, thus hindering the success of any screening program established by government. More health educational programs are required especially focusing the high-risk population in order to make them aware of the transmission and severity of the illness and thus generate insight amongst them to undergo screening for carrier state and prevent transmission of the disease. This idea is supported in the studies by Angastiniotis M et al, Gill PS et al, and Lakhani N, where it has been reported that community health education and outreach

programs have helped in controlling the prevalence of the disease and greatly reduced its health impacts.<sup>17-19</sup> To dispense the information about thalassemia various other modes of communications are very effective in implementing the health education programs. As evident in a study by Saxena A et al, where it has been suggested that effective communication about the disease can be established with audio-visual aids and personal experience sharing.<sup>20</sup>

In our study younger population had better knowledge and more positive attitude towards thalassemia. Thus, student population in colleges should be one of the focus in successful implementation of thalassemia screening programs. Also, our study concluded better education level of parents and relatives was associated with increased knowledge about various aspects of thalassemia. Similarly in a study by Khin Ei H et al, showed parents with low educational level had lesser knowledge about thalassemia thus becoming the main obstacle in preventing the disease. Similarly Paholpak S et al, concluded that with increasing level of education of parents, knowledge about transmission of thalassemia was better.<sup>21,22</sup>

Interestingly it was found in our study despite of having poor knowledge about thalassemia, the extended families majorly had a positive attitude towards the disease. Majority of parents and relatives in our study strongly believed that pregnancy with thalassemia major fetus should be terminated. On the contrary, in a study by Wong PL et al, 68.2% of participants indicated unwillingness to abort the baby if the child has been diagnosed with Thalassemia Major.<sup>23</sup> In our study religious beliefs had significant impact on the attitude towards the disease. Similarly studies conducted by Ahmed S et al, Durosinmi MA et al, and Zahed L et al, showed religious beliefs to be associated with refusal for prenatal diagnosis and termination of affected fetuses among high risk couples.<sup>24-26</sup>

Majority of relatives in our study agreed that screening of thalassemia should be made compulsory before marriage and had shown willingness to get screened though when actually the screening test was offered to them only 3.85% got themselves screened. The main reasons for not taking the screening tests were ignorance, perception of not being at risk, fear of being stigmatized and endangering future prospects of getting married, time and money factor. These views were supported by studies conducted by Chattopadhyay S et al, and Ahmed Set al, where it has been reported that when extended family members were approached to identify carriers in the family tree, the responses were usually unfavorable due to the fear of being stigmatized, in particular, many concerned carrier status may tarnish reputation and affect future marriage prospects.<sup>27,28</sup>

In a study conducted by Zlotogora J it is stated that implementation of mandatory national premarital screening program, and screening young and unmarried women for detection of carriers have dramatically reduced the incidence of infants born with major thalassemia in several countries worldwide.<sup>29</sup> Similarly in studies by Christianson A et al, and Karimi M et al, it is stated that premarital screening to identify carrier couples and subsequently provision of counselling in Iran has resulted in a 70% reduction in the annual birth rate of affected infants and a large amount of medical expenses.<sup>30,31</sup>

Though a large gap remains between attitude and practice regarding screening for thalassemia, inculcating positive attitude towards screening will result in increasing number of relatives (high risk population) getting screened for the disease. Also to minimize the gap between attitude and practice regarding thalassemia screening, addressing of issues such as easy availability of screening tests, appropriate pre and post-test counselling, increasing awareness about thalassemia, subsidizing the cost of the screening test, is required on a large scale.

In our study we found that socio-demographic factors had an important influence on perceptions towards various aspects of thalassemia. This therefore suggests that when thalassemia prevention is offered to the population through carrier screening, premarital or prenatal diagnosis, socio-economic, cultural and religious factors may interfere. Understanding these factors will provide essential insights into successful strategies to reduce births of thalassemia major children with health education playing the most important role.

## CONCLUSION

In our study the high risk population had poor knowledge about thalassemia, though a positive attitude towards the disease suggesting that spreading awareness by various multimedia modalities, strengthening of existing thalassemia units, improving the carrier screening

facilities and implementation of national health programs will lead to acceptance and extensive screening for prevention and control of thalassemia.

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