



Editorial

A National Program on Beta Thalassemia Prevention : Are we ready?

'Prevention is better than cure' holds true for all diseases but is especially true for most genetic disorders where prevention is still the only option. Even for the disorders for which there is a definitive cure, it is often expensive, cumbersome and associated with major risks. Preventive strategies play an important role if the disorder is common in a population. For such disorders nationwide screening and prevention program is cost effective. Beta thalassemia fulfills all these criteria in India for consideration for including in a nationwide screening program. Availability of reliable and sensitive screening tests is one important advantage for beta thalassemia.

Beta thalassemia can manifest with variable severity. Thalassemia major is the most serious form associated with lethality unless optimally treated; but most of the other homozygous cases with the thalassemia intermedia phenotype also have serious problems and disease associated complications and require continued long term treatment. The only curative treatment is bone marrow transplantation available to only lucky ones with HLA matched sib and financial resources. Hypertransfusion therapy and iron chelation, if done on a regular basis and properly, have a good outcome. Availability of oral iron chelators has increased the ease of iron chelation therapy and improved compliance to it. But, given a choice, most families would opt for birth of a child unaffected with thalassemia and this is possible by way of prenatal diagnosis. Most of the families with one child with thalassemia major opt for prenatal diagnosis in the subsequent pregnancies. Prenatal diagnosis for beta thalassemia is available in many parts of India and now it's the time for primary prevention of beta thalassemia.

The carrier frequency for beta thalassemia varies from 3% to 17%. Hence, this is a good candidate for a nationwide prevention program. Mediterranean countries have eradicated thalassemia about two decades ago. Geneticists in India started pilot projects on screening of pregnant women for beta thalassemia carrier state and screening of spouses of

carrier females. Genetic counseling and prenatal diagnosis was offered to the couple if both of them were found to be carriers of beta thalassemia. These projects were sponsored by the Indian Council of Medical Research. These projects in addition to providing experience to the laboratories were effective in many ways by creating awareness amongst obstetricians and common public. There were many other activities for screening population groups and creating awareness amongst laypersons by agencies like ICMR and the Thalassemics Society of India. This has led to increased awareness amongst the general population in major cities and areas with high prevalence of beta thalassemia.

In this issue Dr Rao and his colleagues have shared their experience about the level of awareness about thalassemia in the general population. In spite of some limitations of carrying out such studies, the results are very encouraging. As per the authors, about 40% people from various socioeconomic backgrounds in the study group had heard about 'Thalassemia' and majority of the respondents no longer seemed to view genetic disorders and birth defects as a result of parents' sins. The level of awareness may vary from region to region but in many cities couples are undergoing or being offered beta thalassemia carrier screening during early pregnancy. Early pregnancy or pre-pregnancy period is the best period for carrier screening as the family is more receptive to the issues related to the health of the pregnancy. Screening of college-going students may be easier (our studies as well as those of many others working in the field have shown good take-up of screening tests by college students), but they are less likely to remember the results when necessary and those who test positive for carrier status may also not disclose the fact later to the spouse for fear of stigmatization. Similarly, screening of children and minors for carrier status of any genetic disorder is not ethically justified if the results are not useful medically to the child. In our experience, spreading the message for carrier screening through the family of a child with



Knowledge, Attitude and Practice Study of Beta-Thalassemia in Rural Bengal

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INTRODUCTION

Awareness and education are prerequisites of any screening program for genetic diseases, so that people have informed choice whether to be screened and to weigh benefits and risks of screening.¹ Beta-thalassemia carrier screening programs can be mandatory or voluntary. They can also be based on timing of screening i.e. pre-marital (school children) or pre-pregnancy (couples before or immediately after marriage). In all types of screening programs, international guidelines recommend that genetic screening should be voluntary, informed choice be given to the participants and should invariably be followed by counseling. However a review of international practice on carrier screening for beta-thalassemia found that, there is no universally accepted model to deliver genetic screening programs.²

Efficacy of targeted genetic screening programs largely depends upon correct knowledge and attitude of people towards screening and reflects on the socio-cultural milieu.³ A multi-centric study was conducted by the ICMR (Indian Council of Medical Research) task force on Beta -thalassemia carrier screening program in three metropolitan cities i.e. Delhi, Mumbai, and Kolkata, in which 12,000 school children were screened.⁴ A validation study after 20 years of this carrier screening program among school children in Mumbai found that the education and awareness was ineffective.⁵ Social stigma and negative attitude were largely understood to be the reasons.

However, a report from Mumbai based on a 5-7 year follow up of targeted high risk community screening revealed that carriers had positive attitude and were well aware of all aspects of

disease including prenatal diagnosis and prevention.⁶ Similar positive results were obtained in another study where 99% of 204 married carrier couples (detected before marriage in a prospective extended family cohort) opted for prenatal diagnosis.⁷

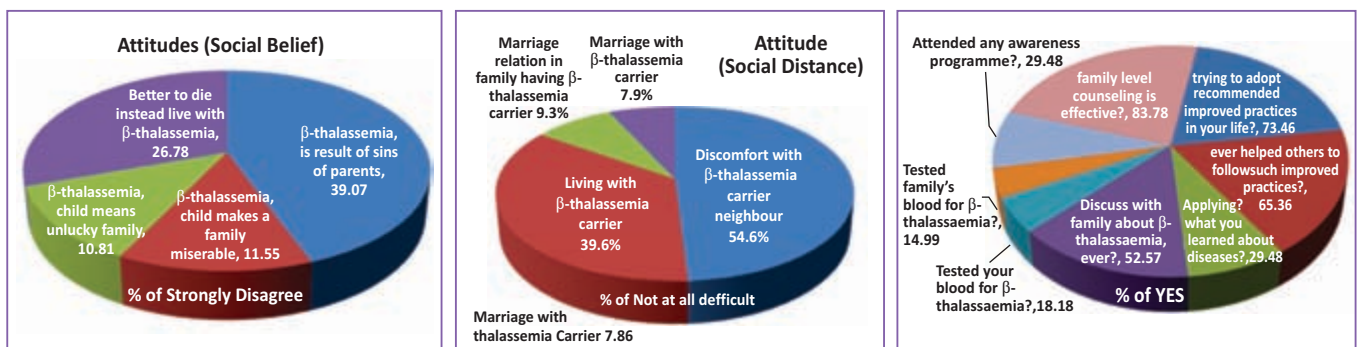
During 2000 -2005, the ICMR task force conducted another beta-thalassemia screening, extending the program to 6 zones in the states of West Bengal, Assam, Gujarat, Maharashtra, Punjab and Karnataka. Two target groups were included for screening at each zone that included 5000 women attending antenatal clinic and 5000 college students, a total of 59892 were covered.⁸ Both programs included education and counseling as inbuilt components of the screening. Assessment of the impact of this carrier screening program is yet to be undertaken.

The results point out two very pertinent facts: beta-thalassemia carrier screening among school children was ineffective, where as targeted high risk community based and extended family screening had a positive effect. In India, traditional family ties still play an important role and ancestry based targeted screening could be the model that works.

In the present study we have undertaken a Knowledge, Attitude and Practice Study (KAPs), before implementing a targeted carrier screening program, in the South 24 paraganas district in West Bengal, considered to be a high risk zone for Beta-thalassaemia.⁹ The program envisages carrier screening coupled with cascade screening of affected families and family counseling. We believe that KAPs study will help to understand social and cultural imperatives, if any, for success or failure of a screening program, so that there will be some perspective/base line data in evaluating the screening program.

Table 1. Knowledge about Beta- thalassemia

Statements	Response (N=407)	
	Yes (%)	No (%)
Do you know the symptoms of beta-thalassemia?	85 (20.88)	322 (79.12)
Is it a blood disorder?	214 (52.58)	193 (47.42)
Is it an inherited disease?	189 (46.44)	218 (53.56)
How many types of thalassemia are there?	50 (12.29)	357 (87.71)
If yes, do you know the names?	15(3.69)	392 (96.31)
Do you know about beta-thalassemia carrier status?	88 (21.62)	319(78.37)
Can a normal person be a beta-thalassemia carrier?	41(10.07)	366 (89.93)
Beta-thalassemia is treatable but not curable	163 (40.05)	244 (59.95)



The attitude of the people in terms of social belief, social distance and application are depicted in pie charts (Figure 1).

MATERIALS AND METHODS

The results of the present study are part of the targeted beta- thalassemia carrier screening program among high risk communities at family level, in South 24 Paraganas district of West Bengal. One thousand seventy three individuals from rural villages, above the age of 13 years were included in this study. The instruments consisted of a structured Beta- thalassemia awareness schedule comprising of Knowledge, Attitude and Practice and a general introductory schedule for the respondent's demographic details. Only 407 (37.93%), who responded positively to the question 'whether you have heard the word beta-thalassemia?' were considered for further evaluation and were administered the KAP schedule. Approval for the study was obtained from the Institutional Ethics Committee of the Anthropological Survey of India.

RESULTS

According to the information provided by the participants, the study population was drawn from a medium to low socioeconomic and educational background. Majority of the respondents replied that cold, cough, diarrhea, fever, gastritis, asthma and arthritis were the general diseases that people suffer in the area and more specifically children suffer from cold, cough, diarrhea, fever, jaundice, pox and acidity. About 54.52% did not know about genetic diseases. However, majority of people (62.16%) said that they had seen/ knew at least one child to whom blood is being given frequently but, only 22.27% knew that it was due to a genetic disease.

Of the 1073 participants, 407 (37.93 %) who had heard the word beta- thalassemia were considered as respondents for further evaluation. The replies to



the knowledge related questions about beta-thalassemia (Table 1) revealed that most of the respondents had no correct knowledge about beta-thalassemia. The response that beta-thalassemia carrier cannot be a normal person was a very important observation, which the education and counseling programs need to consider seriously.

DISCUSSION

The study revealed a positive attitude in the public perception of thalassemia, as majority of the respondents believed that the birth of an affected child was not due to sins committed by the parents. Social distance was not a criterion against beta-thalassemia sufferers but majority of the study participants were unwilling to have marriage relations with the family having a sufferer and to marry a carrier. This may be due to lack of knowledge, since about 90% responded 'No' to the question "Can a normal person be Beta-thalassemia carrier?" That the disease is highly prevalent in the locality was evident, as about 62% of the villagers had seen children to whom blood was given frequently and they were aware of the seriousness of the problem. Majority responded 'not at all difficult' to the question "Feeling discomfort with a neighbor, who is suffering from beta-thalassemia", which showed that 'social stigma' was not an issue.

Feelings of stigmatization or discrimination as undesirable side effects of genetic screening were reported earlier.⁹ However, other studies were emphatic that such feelings were not important.^{10,11,12} In the Indian context, the study by Colah et al (2007) assessing the impact of screening and counseling high school children after 20 years, reflected inadequacy of the onetime carrier screening program.⁵ This was construed as negative impact of carrier screening programs in India, since a similar twenty year outcome of beta-thalassemia carrier screening program among high school children in Montreal was found to be successful.¹³ However, Yagnik et al (1997), in a follow up of 5-6 years in an ancestry based target high risk community beta-thalassemia carrier screening, found the strategy to be highly successful. Similarly, other studies

reported positive impact of beta-thalassemia screening.^{7,14} In India, there are communities with Beta-thalassemia carrier frequency as high as 17%, and ancestry based target carrier screening program with awareness and community participation could be a viable model.¹⁵ The present study, which is a quantitative, descriptive evaluation of the KAP (knowledge, attitude and practice) before screening, among high risk communities in rural West Bengal, found that the perceptions of the people were positive. Proper education and awareness program with local community participation will be effective for generations, since people witness the seriousness of the disease in their day to day life.

Acknowledgements:

The authors acknowledge the Ministry of Culture, Govt of India for support and villagers of the South 24 Paraganas district for participating in the study.

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