## *President's Page*

# Prevention of Thalassemia in India

**Thalassemia syndromes** are a heterogeneous group of single gene disorders, inherited in an autosomal recessive manner, prevalent in certain parts of the world(1). Worldwide, 15 million people have clinically apparent thalassemic disorders. Reportedly, there are about 240 million carriers of b-thalassemia worldwide, and in India alone, the number is approximately 30 million with a mean prevalence of 3.3%(2,3). They are encountered among all ethnic groups and in almost every country around the world.

Every year approximately 100,000 children with Thalassemia Major are born world over, of which 10,000 are born in India. It is estimated that there are about 65,000-67,000 b-thalassemia patients in our country with around 9,000-10,000 cases being added every year(1-7). The carrier rate for b-thalassemia gene varies from 1 to 3% in Southern India to 3% to 15% in Northern India. Certain communities in India, such as Sindhis and Punjabis from Northern India, Bhanushali's, Kutchis, Lohana's from Gujarat, Mahar's, Neobuddhist's, Koli's and Agri's from Maharashtra, & Gowda's and Lingayat's from Karnataka etc. have a higher carrier rate(3,4).

Once a child is diagnosed to have thalassemia homozygous disorders, he/she has to take lifelong treatment. Management includes regular 3 weekly filtered packed red cell transfusions, chelation therapy for iron overload, management of complications of iron overload and transfusions, including osteoporosis, cardiac dysfunction, endocrine problems, Hepatitis B & C, HIV infection, CMV etc. However, this optimal treatment comes at a prohibitive cost. The cost of treatment of an average weight 4-year-old thalassemic child is around Rs. 90,000-100,000 annually in a private set-up. Therefore, not more than 5-10% of thalassemic children born in India receive optimal treatment. Stem cell transplantation as a curative treatment, which costs between 6 and 16 lac rupees is out of reach for majority of children.

Besides bearing the cost of treatment, the psychological stress to both the patient and the parents/family is phenomenal. In fact, it is startling to know from a 15 year old thalassemic child the account of what he has undergone so far. He has received around 250 units of packed red cells and 4000 injections of desferioxamine. He has had a needle in his body for over 40,000 hours of his life. His family has already spent Rs. 16,20,000 for chelation alone. If this child lives for 50 years, then he would require 2000 units of packed red cells, 15,000 desferrioxamine injections, which translates into 1.5 lac hours of a needle in his body and Rs 90 lacs for chelation alone (personal communication). This is besides the cost incurred by the hospital where he receives his regular treatment including packed red cell transfusions and other medical care.

The birth of a thalassemic child thus places considerable strain not only on affected child and family but on society at large. Therefore there is emphasis for shift from treatment to prevention of birth of such children in future. This can be achieved by:

- Population education
- Mass screening of high risk communities for thalassemia minor
- Genetic counseling of those who test positive for thalassemia minor
- Prenatal Diagnosis

The question that arises in the mind is whether prevention at a national level is cost-effective. The answer to this is Yes, it is cost-effective and we should strive to prevent the birth of a thalassemic child. In a study done in Iran, it was concluded that the ratio of cost of treatment to prevention of thalassemia is 16:1.6 (5). A similar result was published by Ostrowsky *et al*. (6) from Quebec, Canada. They found that the total cost per case prevented was less than the cost of a single year of treatment for an individual with the disease. Further, they found that with current rates of marriage and endogamy, the costs that would accrue by having to

treat individuals with thalassemia were more than twice those incurred by the delivery of the prevention program. A World Health Organization committee, in a study, stated that in Cyprus, the annual cost of running the screening and prenatal diagnosis program was found to be about equal to the cost of treatment of all existing patients for five years. In Sardinia, the committee estimated that a 90 per cent reduction in the incidence of thalassemia disease would recoup the cost of setting up a prevention program in three years from the start of the services; thereafter the total cost of treatment would fall steadily over the next five years to about one-fifth of the projected cost of treatment(7).

Prevention of thalassemia, therefore is practical, feasible and the answer to the agony of so many children, families and nations. The methods would include creating awareness amongst high risk communities about the prevalence and the difficulties in management of this condition. Screening young people amongst all high risk communities before marriage is the right way to go. If screening is performed in childhood, it is often forgotten around the time they get married. Hemoglobin electrophoresis is the confirmatory test to diagnose thalassemia minor or carrier status. All at-risk couples need to be counseled about the prenatal diagnosis to confirm the thalassemic status of the fetus. If the fetus is not affected, the pregnancy should be continued. If the fetus is affected, the choice of terminating the pregnancy is offered.

A concerted effort is needed to have a National Thalassemia Prevention Program in place. This needs involvement of all government health agencies, scientific research bodies, institutions caring for thalassemia, parents' societies, dedicated and committed social workers along with the medical fraternity to be able to successfully eradicate thalassemia from the country. With this in mind Indian Academy of Pediatrics has envisaged Thalassemia Prevention Program under IAP Vision 2007. A national consultative meeting is planned this year to formulate the guidelines for an ideal thalassemia prevention program which will be followed by a series of advocacy and sensitization meetings in high risk states. If countries like Cyprus, Sardinia and Greece could achieve, there is no reason why we cannot achieve the same.

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