Review Article

Thalassemia Major: The Future Prospective

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Abstract: Thalassemia is one of the most common inherited diseases worldwide with a varied clinical spectrum with Thalassemia Major being the most devastating form requiring long term treatment. It prevails in significant numbers, especially in Indian subcontinent, with the carriers and diseased children increasing in alarming proportions each year. Thalassemia brings immense health implications, financial burden and psychosocial trauma both for the family and the child. It is the felt need of the hour that more and more emphasis should be placed on prevention of Thalassemia by Mass Education and Awareness programs, Carrier screening programs, Genetic Counselling, Antenatal Diagnosis and Permissible Abortions. Formulation of a National Health policy in this regard and its strict implementation is strongly recommended.

Keywords: Thalassemia, Autosomal Recessive disease, psychosocial, dysmorphic, Cascade

Introduction

Thalassemia, one of the most challenging hematologic disorder, is an Autosomal Recessive disease characterised by reduction of or absence of globin chain synthesis in haemoglobin with a varied clinical spectrum.[1] One of the most prevalent single gene disorder, thalassemia, has huge impact on health along with financial, psychosocial and emotional impact on its patients and their families. The cure of this ailment is costly and limited by both scanty availability of compatible bone marrow donors and limited number of medical centres offering the treatment. The best path thus to free the humanity from the clutches of this ruthless disease is by formulating and implementing a strong health plan aiming at its prevention rather than struggling with its cure.

Disease Burden

Thalassemia is an emerging global health problem with its predominant prevalence in the population of Middle East, Transcaucasus, Central Asia, the Indian subcontinent and Far East. Worldwide, there are approximately 15 million people who are estimated to suffer from thalassemic disorder. Reportedly, there are about 240 million carriers of beta thalassemia worldwide i.e about 1.5% of world population. The number of thalassemia carriers in India alone is 30 million approximately. The carrier rate of beta thalassemia is average of 3.2% in India. This means that on an average 1 in every 25 Indians is a carrier of thalassemia.[2,3]

In India, every year 12000 infants are added in the prevailing belt of thalassemia major. So by so, one child is born every hour who suffers from this genetic disorder.

Psychosocial Burden in Thalasemics

Thalassemia, at present is one of the most challenging hematologic disorders. It challenges the individual at physical, emotional and cognitive levels and disrupts the quality of life. Added financial burden over the individual and his or her family is also considerable due to regular need of blood transfusions and chelation therapy.

The psychosocial burden of the disease is perceived even more in adolescence when the teenager is confronted with various difficulties like identity formation, developing intimate relationships and entering the working world. Accepting and integrating thalassemia into one's identity becomes a difficult task at this stage.[4]

Thalassemic children have an adverse impact on their education in terms of frequent leaves from school and hampered academic performance due to disease and related problems.[5]

These children have negative self concept and have physical weakness leading to limited

indulgence in outdoor play activity and poor sports performance.[5] Psychologically, the kids have a fear of being ridiculed and have lack of trust on peers and depend on immense support from family members. Anxiety, poor psychiatric adaptation with dysmorphic moods and fear of pain and death is also common among thalassemic adolescents especially those with physical abnormalities.[6,7]

Along with the thalassemic kid who is struggling with the disease, also is of equal consideration, the family, especially involving the parents who undergo a similar degree, if not more, of physical, emotional, financial and psychological trauma fighting with thalassemia along with their kid.

The biggest irony is that such a large proportion of population is living in the clutches of thalassemia, fighting daily with the stress of the disease in all abovementioned aspects and all this is happening for a disease that is totally preventable.

Programmes for Prevention of Thalassemia

History has witnessed pilot population programs directed to prevent beta thalassemia major by carrier screening, prenatal diagnosis and public awareness and education. The need of the hour is to consider formulating a strong National program, as there is none in India, aiming at the prevention of thalassemia.

Education

Lack of awareness amongst not only in the population at risk of disease but also policymakers and health professionals is a major obstacle in the control of thalassemia. Periodic meetings can be held among physicians especially paediatricians and obstetricians, family planning workers, nurses, social workers to sensitize towards the need and methods of thalassemia prevention.

Population education makes use of mass media, posters and informational booklets that could be left at various key sites such as family planning clinics, marriage registries and counselling rooms.[8] It is important to emphasize that the carrier state can be easily recognized at the thalassemia centres and is not associated with stigmatization.

The families where both the partners are carriers should also be taught regarding the various reproductive options including prenatal diagnosis.

Formal education on thalassemia can be introduced in educational curriculum at the end of secondary school to create awareness at the right age.

Carrier Detection

A carrier detection screening procedure should be chosen on the basis of its ability to not miss even a single couple at risk. MCV, MCH and HbA 2 estimation together form a strong carrier detection program.[8] HbA 2 done by Hb Electropheresis also enables to identify HbF and enables to detect various other Hb Variants.

The various carrier detection screening programs can be Mass screening, Cascade screening, adolescent and Antenatal (or Prenatal) screening program.

Mass Screening in a country like India with enormous population numbers can be economically a non feasible or costly affair. More feasible options would include Cascade screening (screening of the relatives of the patient already suffering from thalassemia), Adolescent screening or screening at the time of marriage. At least this option can be exercised in high prevalence communities. In all parts of country, prenatal screening using HbA 2 estimation should be made mandatory and if the foetus is found to be suffering from thalassemia, termination of the pregnancy should be opined.

Genetic Counseling

Genetic counselling is aimed at providing information to couples or unmarried individuals so as to enable them to take a decision regarding reproduction. The aim is to make people understand the simple genetics of this Autosomal recessive disease. If one of the couple is carrier and one is normal, there is 50% chance of an offspring being carrier and 50% that the offspring would be absolutely normal. Similarly, if both the parents are carriers, there is 50% chance of an offspring being carrier, 25% chance that the offspring would be a thalassemic major and 25% that the offspring would be normal. Clearly, if the marriage between two carriers could be prevented, the probability of birth of a thalassemic major is nil. If a person is found to be a carrier as detected by increased HBA2 levels, the

discussion should include various options including birth control, mate selection, adoption, foetal testing including prenatal diagnosis or preimplantation diagnosis, artificial insemination by normal donors along with the details of natural history of the disease.

Antenatal diagnosis

Antenatal diagnosis was in the past done by Foetal Blood sampling by fetoscopy or placentocentesis. Now-a-days, thalassemia is detected directly by the analysis of amplified DNA from foetal trophoblast or amniotic fluid cells. Foetal DNA is obtained transabdominally from Chorionic Villous sampling. The advantage of this technique is that it can be done in first trimester and is associated with a low risk (< 1 %) of foetal mortality. PCR based technique of prenatal diagnosis is another reliable advancement. Further foetal anaemia in case of Hb Barts Hydrops foetalis can be diagnosed in mid trimester gestation by quantification of middle cerebral artery peak systolic velocity. Now-a-days several attempts have been made to clinch this diagnosis with non invasive techniques like analysis of foetal cells in maternal circulation.

Abortions

Any aberrant foetus should be offered termination as per National policy as the health implications, psychosocial trauma and financial burden on the family in case of continuation of this pregnancy are huge.

Thus, the need to shift the focus from the treatment to prevention of thalassemia is a major need of hour. The policymakers should take up sincere responsibility to formulate and implement the prevention of thalassemia as a national program.

Conclusion:

As paediatrician, we daily come across the physical, psychological, emotional and financial trauma being faced not only by the thalassemic patients but also their whole family. So many resources are being spent on the treatment of a condition which can be prevented. So management of thalassemia should be started even before the disease starts to exist. It being a genetic disorder clusters in families, so we can form local protocols for diagnosis & prevention of thalassemia. We have eradicated many preventable diseases, so why not thalassemia↑ So let's work for it, goal for a thalassemic free society & make the world a better place for the entire human race.

Conflict of Interest: None

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