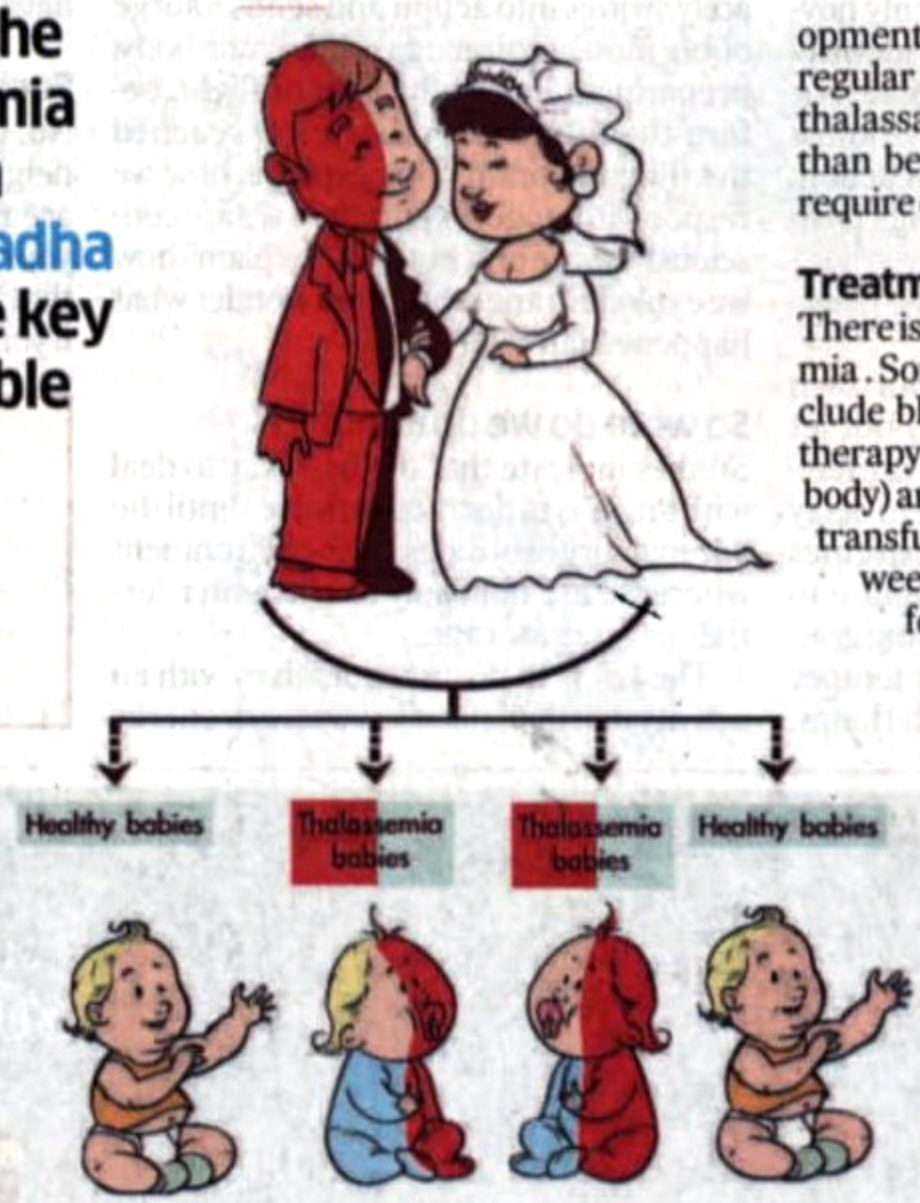
Battling thalassaemia

BLOOD BASICS With the number of thalassaemia cases on the rise each year, Dr Sheetal Sharadha says awareness is the key to prevent the incurable disorder



India has one of the largest numbers of people suffering from thalassaemia. There are over 40 million carriers and over 100,000 thalassaemia majors undergo blood transfusion every month. Keeping this in mind, it is important to create awareness about the disease.

Thalassaemia is an inherited blood disorder in which the body makes a decreased quantity of haemoglobin. In India, thalassaemia is one of the causes for haemolytic anaemia amongst infants. This form of anaemia is associated with an excessive number of red blood cells being destroyed by the body (due to defective haemoglobin). Marriages with close members of the family or with members amongst the local communities result in at least 3-17% of thalassaemia cases in India.

The types

Normal adult haemoglobin consists of two alfa chains and two beta chains. Alpha thalassaemia occurs when there is a mutation in the genes encoding the alfa chain. More than 100 different genetic mutations have been associated with alpha thalassemia. Similarly, beta thalassaemia occurs when there are mutations in the gene which encodes for the beta chain. More than 400 different mutations have been identified in beta thalassaemia. These mutations lead to different types of haemoglobin variants. The most common haemoglobin (Hb) variants worldwide in descending order of prevalence are HbS, HbE, HbC, and HbD.

Inheritance & severity

Thalassaemia major is inherited in an autosomal recessive manner. Being an autosomal recessive condition means that two copies of the defective gene are required for the disorder to occur in a severe form. If only one copy of the gene is defective, it is known as thalassaemia minor.

Silent carriers of alpha thalassaemia and persons with alpha or beta thalassaemia trait are asymptomatic and require no treatment. However, as carriers of one defective copy of the gene, they have a higher risk of passing on this abnormal gene copy to their children.

Thalassaemia major occurs when a person inherits a defective copy of the same gene from both the parents, resulting in a severe form of the disease. Individuals affected by thalassaemia major, present grave symptoms like severe anaemia, jaundice, enlargement of the heart, liver and spleen, delay in growth and development. Affected children will require regular lifelong blood transfusions. Beta thalassaemia intermedia is less severe than beta-thalassaemia major and may require episodic blood transfusions.

Treatment

There is no permanent cure for thalassaemia . Some of the available treatments include blood transfusions, iron chelation therapy (removal of excess iron from the body) and bone marrow transplant. Blood transfusions at intervals of two to three weeks are prescribed to patients affected with thalassaemia major. However, with consistent blood transfusions, the iron content of the body increases and there is a likelihood of the excess iron causing problems to vital organs like the heart and the liver. Under such conditions, experts suggest iron chelation therapy to remove the excess iron from the body.

Bone marrow transplants can be curative for some children with beta-thalassaemia major. It is an expensive therapy and involves matching a compatible bone marrow donor with the patient.

Prevention

Couples planning to get married should undergo carrier testing along with genetic counselling. This is critical in case of thalassaemia, as carriers are at a greater risk of having a child with thalassaemia major especially if they are married to another person who is also a carrier. Prenatal diagnosis can be offered to such couples.

In the current Indian context, the use of advanced genetic testing for carrier screening of the parents combined with genetic counselling is the best method to reduce the occurrence of thalassaemia. Genetic analysis can help the couple to make informed reproductive decisions for their future. Organising special camps for parents and encouraging parents to help spread awareness about the disease.

(The author is clinical geneticist, MedGenome)