ORIGINALLY PUBLISHED IN "THE SANGAI EXPRESS" ON 12TH JUNE 2013

Thalassemia Trait and Testing – Salam Ghanapriya

There was this shocking news published on the front-page of The Telegraph in its 29th May 2013's edition under the headline "Father mulls mercy killing for kids – With 3 children suffering from thalassemia, man considers ultimate step". We had read the news with dismay at the way Abdul Rahim, a resident of Them Mawbah of Meghalaya, who has been struggling to meet the expenses of his three ailing children took the shattering decision to knock on the doors of the judiciary and plead mercy killing for his children. Rahim has to spend at least Rs. 45,000/- a month for regular blood transfusions for his children being treated at a Guwahati hospital. "I can no longer see my children suffering. I have nothing left but to seek permission to take away the lives of my children before I commit suicide", Rahim told the journalists gathered that afternoon at the Shillong Press Club.

WHAT IS THALASSEMIA?

Thalassemia is an inherited blood disorder in which the body's ability to produce haemoglobin is compromised, causing anaemia that can range from mild to life-threatening. Haemoglobin helps carry oxygen through the blood to all parts of the body. While severe forms of thalassemia require life-long follow-up care and regular blood transfusions, some other forms are more manageable and require little or no treatment. At the mildest end, thalassemia "minor" also called thalassemia trait, isn't a disease at all. However, children with thalassemia "major", as was in the case of Rahim's children, regular treatment and blood transfusions are required to stay healthy. Untreated, thalassemia major can be fatal.

INCIDENCE OF THALASSEMIA

Thalassemia is a very common genetic disorder. Worldwide, several million people have thalassemia disorders and about 300 million people carry the genetic trait for thalassemia. There are roughly 60 million people in India who carry the thalassemia trait. Since the trait doesn't impact them directly, most of them are unaware of this. However, if two people carrying the thalassemia trait get married, there is a 25% chance that their children will have the dangerous thalassemia major disorder. Approximately, over 10,000 thalassemia major children are born in India every year.

RISK FACTORS

Thalassemia mostly occur in persons of Southeast Asian, Chinese, Mediterranean, or of African American origin. As mentioned before, if both husband and wife are diagnosed as thalassemia minor, there is a heightened risk of a thalassemia major child being born in the family.

Parent status	Children
Both parent normal	All normal
One parent thalassemia minor/ other parent normal	• No thalassemia major • 50% chance of thalassemia minors • 50% chance of normal children
Both parents thalassemia minors	• 25% chance of thalassemia majors • 50% chance of thalassemia minors • 25% chance of normal children

CAN YOU PREVENT THE BIRTH OF A THALASSEMIA MAJOR CHILD IN YOUR FAMILY?

It can be prevented only if you are aware that you are a thalassemia minor.

- If you are not a thalassemia minor, no precaution is required.
- If you are a thalassemia minor and unmarried, you should be aware of all the risks associated in marrying another thalassemia minor.
- If you are thalassemia minor and married, get your spouse tested for thalassemia minor condition. If your spouse is not thalassemia minor, no precaution is required.
- If you and your spouse are thalassemia minors, consult your doctor for genetic counselling before conception.
- If both husband and wife are thalassemia minors and wife is already pregnant, then prenatal diagnosis should certainly be done.

THALASSEMIA TRAIT TESTING

Finding out if one has the genetic trait for thalassemia begins by determining the size of one's red blood cells. Individuals with thalassemia trait have red blood cells that are slightly smaller than normal. Testing for the thalassemia trait usually happens in two parts:

Indicative Tests:

- Complete Blood Count Test (or Complete Haemogram) is a snapshot of the cells and fluids in the bloodstream. This is the same simple blood test most employees go through during pre-employment check-ups and during routine testing of most illnesses. In it, the Mean Corpuscular Volume (MCV) reading determines the size of one's red blood cells. If the MCV reading indicates that one may have thalassemia trait, one should undergo additional tests for confirmation.
- Blood Smear (also called peripheral smear and manual differential). With thalassemia, the red blood cells are often microcytic (low MCV).
- Iron Studies. These may include iron, ferritin, unsaturated iron binding capacity (UIBC), total iron binding capacity (TIBC), and percent saturation of transferrin. These tests help determine whether an iron deficiency is causing and/or exacerbating a person's anaemia and to monitor the degree of iron overload in an individual with thalassemia.

Confirmatory Tests:

- Hb HPLC or Hb Electrophoresis for HbA_2 is a reliable way of determining whether or not a person has thalassemia trait (for beta thalassemia minor).
- DNA analysis is used to investigate deletions and mutations in the alpha and beta globin producing genes. More definitive "molecular" testing is performed to determine the presence or absence of thalassemia trait. As these genetic tests are developed further, they will be used more widely to test for thalassemia trait.

***

(The writer is Junior Biochemist, BABINA Diagnostics, Imphal)