Qure
Center for Complete Care
Blood Diseases. Cancer. Stem Cell Transplantation

What Is Thalassemia?

Thalassemia is an inherited blood disease that affects the red blood cells. Red blood cells circulate in the blood and contain hemoglobin, which carries oxygen to all parts of the body. The normal hemoglobin protein is made up of two alpha and two beta globin protein chains, and when the beta globin gene is affected the resulting lack of normal hemoglobin leads to destruction of red blood cells and anemia (beta thalassemia).

If both beta globin genes are severely affected then patients need monthly transfusions (Thalassemia major), whereas if the defect is milder they can make some normal hemoglobin and need only intermittent transfusions (Thalassemia intermedia). Patients who have only one of their pair of beta globin genes affected are carriers but lead normal healthy lives (Thalassemia trait).

How common is Thalassemia?

The word thalassemia comes from "thalassa" = "great sea" because it was first described in Italians, Greeks and other people who live around the Mediterranean. We now know it is found in many other countries, and each year 100,000 children with thalassemia are born around the world, including 10,000 born in India. It is estimated that there are 60-70,000 thalassemia patients in our country.

In some communities in India, such as Sindhis and Punjabis from Northern India, Bhanushalis, Kutchis, Lohanas from Gujarat, Mahars, Neobuddhists, Kolis and Agris from Maharashtra and Gowdas and Lingayats from Karnataka 5-15% of the population are beta thalassemia carriers (having at least one abnormal beta globin gene). Although carriers have healthy lives, if they marry they are at risk of having a child with thalassemia major.

What are the signs of thalassemia?

Babies with thalassemia major are normally healthy at birth, but develop symptoms after the first few months of life which include:

- Pale skin
- Acting cranky or upset
- Not growing as much as expected
- Swelling of the belly
- The skin or white part of the eyes turning yellow
- The bones of the face or skull being wider than normal

Many of the symptoms happen as the body frantically tries to make red cells, including in parts of the body that don't normally make red blood cells such as the liver and spleen, despite which there is increasing anemia.

How is thalassemia diagnosed?

A few simple tests will usually confirm the diagnosis. Sometimes both parents need to be checked before the diagnosis is certain.

- Complete blood count: this is a simple blood test which will usually show severe anemia with small red blood cells which vary in size, and many immature red blood cells. This may be strongly suggestive of the diagnosis.

- Hemoglobin electrophoresis: This is a special blood test that will show an absence of normal hemoglobin that is characteristic of thalassemia.
- Genetic testing: Numerous mutations in the beta globin gene can cause thalassemia but this test is important if you are planning to screen future pregnancies.

How is Thalassemia Major treated?

People with thalassemia major need lifelong monthly red cell transfusions. Although blood transfusions help treat thalassemia, they can also cause problems. That's because blood has iron in it and when people get a lot of blood transfusions, their body gets too much iron which can damage the heart and liver. People getting blood transfusions for their thalassemia need treatment to get rid of the extra iron that builds up in their bodies, and take daily pills to get rid of this extra iron which is called "iron chelation."

Rarely patients who need unusually large amounts of red cells will need to have their spleen removed by surgery which increases the risk of infection or blood clots in the body.

Is there a permanent cure for Thalassemia?

Severe thalassemia can be cured with a procedure called a "bone marrow transplant" (BMT) which involves replacing the cells that cannot make enough red blood cells with healthy cells from another person (the donor), typically a brother or sister. BMT is only offered to patients who have thalassemia major, since it can cause many side effects including death.

How can patients with Thalassemia avoid complications?

- See your doctor for regular follow-ups, and follow all instructions about tests and treatment, especially regular red cell transfusions and iron chelation.
- Avoid taking vitamins or supplements with iron in them.
- Take a vitamin called folic acid daily

Iron load from routine blood transfusions can impact almost any organ in the body but especially the heart and liver. Imaging tests such as T2*MRI and echocardiography can check for iron overload and heart function. In addition bone abnormalities can occur and calcium and Vitamin D supplements may be prescribed. Growth and hormone function can be affected and special blood tests may be done to check on these.

What if you want to get pregnant?

It is important to stay in good physical condition with regular transfusions and adequate chelation. Depending on your partner it is possible that your future children could have thalassemia, discuss with your doctor to find out how likely this is. If you do get pregnant, you can choose to test your unborn baby for thalassemia if needed.

What can I do if I have more questions?

If you have more questions, you can discuss these with your (or your child's) hematologist at QURE Hematology Clinic.