



World Thalassemia Day-08 May 2015

What is Thalassemia?

Thalassemia is a blood disorder passed down through families (inherited) in which the body makes an abnormal form of haemoglobin, the protein in red blood cells that carries oxygen. The disorder results in excessive destruction of red blood cells, which leads to anaemia. Anaemia is a disorder in which your body doesn't have enough normal, healthy red blood cells (RBC).

This disease is inherited, meaning that at least one of your parents must be a carrier of the disease. It is caused by either a genetic mutation, or a deletion of certain key genes.

The two main forms of thalassemia are alpha thalassemia and beta thalassemia. In alpha thalassemia, at least one of the alpha globin genes has a mutation or abnormality. In beta thalassemia, the beta globin genes are the ones affected.

Each of these two forms of thalassemia has several distinct types. The exact form you have will affect the severity of your symptoms and your prognosis.

Causes and Risk Factors for Thalassemia

Thalassemia occurs when there is an abnormality or mutation in one of the genes involved in haemoglobin production. This genetic defect is inherited from your parents.

If only one of your parents is a carrier for thalassemia, you may develop a form of the disease called "thalassemia minor." If this occurs, you will probably not have symptoms, but you will be a carrier of the disease. Some people with thalassemia minor do develop minor symptoms.

If both of your parents are carriers of thalassemia, you have a 25 percent chance of inheriting a more serious form of the disease.

Who carries Thalassemia?

Thalassemia is most common in people from Southeast and Central Asia, the Mediterranean, the Middle East, India, and North Africa.

What Are the Symptoms of Thalassemia?

The symptoms of thalassemia depend on which type you have.

Mild thalassemia usually doesn't cause any symptoms.

Moderate or severe disease may cause symptoms of anaemia. For example, you may feel weak, tire out more easily, and feel short of breath. Other symptoms also can occur depending on how severe your disease is and what problems it causes.

Children with severe thalassemia may grow slowly (failure to thrive), have skull bones that are not shaped normally, and have problems with feeding, frequent fevers, and diarrhoea.

What is Thalassemia Minor?

People with a thalassemia mutation only in one gene are known as carriers or are said to have thalassemia minor. Thalassemia minor results in no anaemia or very slight anaemia. People who are carriers do not require blood transfusion or iron therapy, unless proven to be iron deficient.

What is Thalassemia Major?

Children born with thalassemia major usually develop the symptoms of severe anaemia within the first year of life. Lacking the ability to produce normal adult haemoglobin, children with thalassemia major:

- are chronically fatigued
- fail to thrive, and
- do not grow normally

Prolonged anaemia will cause bone deformities and eventually will lead to death within the first decade of life. The only treatment to combat severe anaemia is regular blood transfusions.

There are two main types: alpha and beta. Beta thalassemia is the most common.

Beta thalassemia

Beta thalassemia comes in two serious types: thalassemia major (also called Cooley's anaemia) and thalassemia intermedia. The symptoms of thalassemia major (Cooley's anaemia) generally appear before a child's second birthday. The severe anaemia related to this condition can be life-threatening. Other symptoms include:

- fussiness
- paleness
- frequent infections
- poor appetite
- failure to thrive
- jaundice (yellowing of the skin and whites of the eyes)
- enlarged organs

This form of thalassemia is usually so severe that it requires regular blood transfusions.

Thalassemia intermedia is a less severe form of beta thalassemia. While people with this condition still have anaemia, they do not need blood transfusions.

Alpha thalassemia

This type occurs when one or more of the four alpha-globin genes that make haemoglobin are missing or damaged.

- **If one gene is missing or damaged:** Your red blood cells might be smaller than normal. You will have no symptoms and you will not need treatment. But you are a **silent carrier**. This means you don't have the disease but can pass the defective gene to your child.
- **If two genes are missing or damaged:** You will have very mild anaemia that will typically not need treatment. This is called **alpha thalassemia minor** or **alpha thalassemia trait**.
- **If three genes are missing:** You will have mild to moderately severe anaemia. This is sometimes called **haemoglobin H disease**. If it is severe, you may need blood transfusions.

Haemoglobin H disease can cause bone issues. The cheeks, forehead, and jaw may all overgrow. In addition to the bone issues and the anaemia associated with any kind of thalassemia, haemoglobin H disease can cause these other symptoms:

- Jaundice
- Extremely enlarged spleen
- Malnourishment
- **If all four genes are missing:** This is called **alpha thalassemia major** or hydrops fetalis. The fetus will be stillborn, or the child will die soon after birth.

Hydrops fetalis is an extremely severe form of thalassemia. It occurs before birth, and most individuals with this condition are either stillborn or die shortly after being born.

How is thalassemia diagnosed?

Your doctor will do an exam and ask about your health history. Tests you may need include:

- A complete blood count (CBC).
- A gene test to see if you have the genes that cause thalassemia.
- An iron level test.

- A blood test that measures the amounts of different types of haemoglobin, to help find out which type of thalassemia you have.
- Abnormally shaped red blood cells are a symptom of thalassemia. So another test may be performed called haemoglobin electrophoresis, this test separates out the different molecules in the red blood cells, allowing the abnormal type to be identified.

Depending on the type and severity of the thalassemia, a physical examination might also help in the diagnosis. For example, a severely enlarged spleen might suggest to your doctor that you have haemoglobin H disease.

How is it treated?

Treatment depends on how severe your condition is.

Most large medical centres have treatment centres for blood disorders. They are an excellent resource to help you and your family get the best care.

- Mild thalassemia, the most common form, does not need treatment.
- Moderate thalassemia may be treated with blood transfusions and folic acid supplements. Folic acid is a vitamin that your body needs to produce red blood cells.
- Severe thalassemia may be treated with:
 - Blood transfusions.
 - Bone marrow transplant
 - Medications and supplements
 - Possible surgery to remove the spleen and /or Gallbladder.
- You may be instructed not to take vitamins or supplements containing iron and don't take extra vitamin C, which can increase how much iron you absorb from food. This is especially true if you require blood transfusions. People who receive blood transfusions receive extra iron that the body can't easily get rid of and iron can accumulate in tissues, which can be potentially fatal, this can damage your heart and other organs.
- If you are receiving a blood transfusion, you may also need chelation therapy, which generally involves receiving an injection of a chemical that binds with iron (and other heavy metals). This helps remove extra iron from your body.
- Get a flu vaccine each year. Also talk to your family physician about getting a pneumococcal vaccine. These vaccines may protect you from severe infections, which can make anaemia worse and cause severe illness in people who have thalassemia.

What Is the Long-Term Outlook for Thalassemia?

The prognosis for thalassemia depends on the type of the disease involved. Hydrops fetalis, for example, is usually fatal either before or shortly after birth.

On the other hand, people who have mild or minor forms of thalassemia can typically lead normal lives.

Other forms of thalassemia can fall anywhere in between. In severe cases, heart failure may occur in the 20s.

Your doctor can give you more information about your personal prognosis, as well as how your treatments can help improve your life or increase your expected lifespan.

So please.....



Of Thalassemia Child.

Dr. Karuna Milind, Sr. Medical Officer.

