

Thalassemia

What is Thalassemia?

Thalassemia is a group of genetic blood disorders that affects the body's ability to produce a protein in the red blood cells called hemoglobin. Hemoglobin carries oxygen and nutrients throughout the body. When there is not enough oxygen, our organs and tissues will not function properly. People with thalassemia have less hemoglobin than normal, thus suffer from mild to severe anemia.

Who is at risk?

Because thalassemia is a genetic disorder, it can only be inherited from parents who have the defective genes. Genes determine our characteristics including the color of our eyes, our skin tone, and even some genetic diseases. A child who inherits one normal gene from one parent and a thalassemia gene from another is a carrier of the disease. A carrier has no symptoms of thalassemia and lives a healthy life. When two carriers become parents, for each child they bear, the chances of the child developing thalassemia are:

- one in four chance that he/she will receive a thalassemia gene from each parent. In such case, the child will develop thalassemia.
- one in four chance that their child will receive normal genes from both parents.
- one in two chance that he/she inherit one normal gene from one parent and a thalassemia gene from another, becoming a carrier of the disorder.

People from the following origins tend to have a higher risk of developing thalassemia:

Asia, Mediterranean, Greece,
Italy, Middle East, Africa

What are the different types of thalassemia?

Hemoglobin, the oxygen-carrying protein in red blood cells, is made up of two chains, an alpha chain and a beta chain. These two chains are made from specific genes we inherit from our

parents. When these specific genes are not working properly, hemoglobin production is affected. There are two major types of thalassemia.

Alpha Thalassemia

A child inherits four genes, two from each parent, that control the production of hemoglobin alpha chain. Alpha thalassemia occurs when one or more of these genes fail to work properly. The severity of alpha thalassemia depends on the number of defective genes:

1. **Silent Carrier**
With one defective gene, the body still makes hemoglobin. Therefore, the person will not feel any symptoms and can lead a normal and healthy life.
2. **Alpha Thalassemia Minor**
The loss of two normal genes causes the red blood cells to be smaller than usual. Except for possible mild anemia, patients remain in good health.
3. **Hemoglobin H Disease**
Hemoglobin made from only one gene does not carry oxygen properly. Patients with hemoglobin H disease can suffer from severe anemia.
4. **Alpha Thalassemia Major**
With all four genes failing to produce the alpha chain, the body has a significant loss of hemoglobin which results in a severe form of anemia.

Beta Thalassemia

Unlike the alpha chain, the production of hemoglobin beta chain is determined by two genes, one from each parent. The severity of beta thalassemia depends on whether one or both of the genes fail to work properly.

Beta Thalassemia Minor occurs when one beta gene is defective. People with beta thalassemia minor have smaller red blood cells, but no major health problems.

Beta Thalassemia Major (also known as Cooley's anemia) is the most severe form of thalassemia in which both beta genes fail. The body makes little or no beta chain which results in severe anemia.

What tests can be done to diagnose thalassemia?

Simple blood tests can determine the amount of hemoglobin and the size of red blood cells. It can also measure the amount of iron to rule out iron-deficiency anemia.

Prenatal screenings such as chorionic villus sampling (CVS), in which a small piece of placenta is removed for sampling, and amniocentesis, a procedure to test amniotic fluid, can detect thalassemia trait.

What are the symptoms of thalassemia?

People with mild cases of thalassemia usually do not present any symptoms. In more severe cases of thalassemia, symptoms may include: weakness, pale skin or jaundice, dark urine, fatigue, lightheadedness, rapid heartbeat, abnormal facial bones and poor growth, protruding abdomen with enlarged spleen or liver.

How is thalassemia treated?

In mild cases of thalassemia, treatment is not necessary, since patients do not have any symptoms. People with moderate to severe thalassemia may require blood transfusion. The blood they receive consists of new and healthy red blood cells, which allow oxygen and nutrients to enter body cells.

One of the side effects of blood transfusion is an excess load of iron in the body. Extra iron can lead to damage in body parts such as the heart, lungs, and liver. In order to prevent the

damage, patients with regular blood transfusion also undergo iron chelation therapy. The treatment involves administering a medication under the skin to get rid of excess iron in the body.

The cause of anemia in thalassemia patients is different from those who suffer from iron-deficiency anemia. Therefore, eating iron-rich foods or taking iron supplements will not treat thalassemia. On the contrary, as described above, those who go through blood transfusion as a treatment for severe thalassemia can have excess iron level that is harmful to the body. As a result, thalassemia patients are advised to avoid iron-rich foods, such as spinach, beef, pork, lamb, liver, and dried beans.

Thalassemia is a genetic disease that can only be inherited from parents who have the disease or are carriers. Parents' genes are passed on to their children randomly.

Therefore, there is nothing a parent can do to prevent passing on genetic disorders to their children. If you and your partner are suspected of having the thalassemia trait, genetic counselors can provide extensive medical information to help you make plans for future families.

For more information about thalassemia, contact the following agency:

Northern California Comprehensive
Thalassemia Center
www.thalassemia.com
(510) 428-3885 x4398