WHAT IS THALASSEMIA?

Thalassemia is an inherited blood disorder, which means it is passed from parent to child through genes. People with thalassemia are not able to make enough of a protein called hemoglobin. Hemoglobin is an important part of red blood cells that helps them carry oxygen to other parts of the body. Without hemoglobin, your body's red blood cells cannot function well and don't live as long as they should. With fewer red blood cells in the bloodstream, the rest of the body's cells do not get enough oxygen, which results in a condition called anemia.

There are two main types of thalassemia: alpha and beta. Symptoms of thalassemia vary, depending on the type. Some people may have very mild symptoms or none at all. This is called "minor" thalassemia. Someone with the more severe form called "major" thalassemia may have serious symptoms and need regular blood transfusions.

What are the symptoms of thalassemia?

The symptoms of thalassemia are caused by anemia and can include:

- Fatigue
- Dizziness and fainting
- Shortness of breath
- Fast heartbeat
- Headaches
- Leg cramps
- Difficulty concentrating
- Pale skin

How do I know if I have thalassemia?

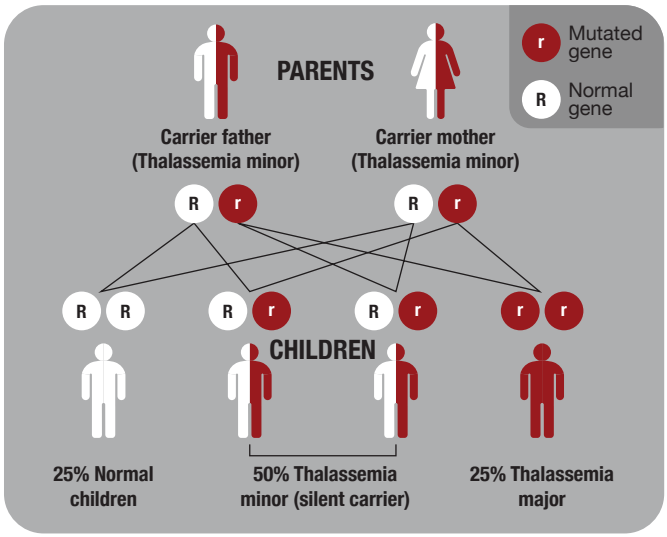
Thalassemia is screened for at birth, which is when people with more severe forms are typically diagnosed. They are also more likely to exhibit symptoms early in life. However, people with milder thalassemia often get diagnosed later, when they begin having symptoms of anemia or when their doctor finds evidence of anemia from blood tests.

If your doctor suspects you have thalassemia, they may order the following blood or genetic tests:

- **Complete blood count (CBC)** to measure the types of cells in your blood and how much hemoglobin you have.
- **Special hemoglobin tests** to measure the type of hemoglobin in your blood.
- **Genetic testing** to find out what type of thalassemia you have.

What causes thalassemia?

Thalassemia is caused by mutated, or altered, genes that you inherit from your parents. In alpha thalassemia, the mutations are in the gene that makes the alpha part of the hemoglobin protein. The number of mutated genes you inherit affects the severity of your symptoms. If you inherit the gene from only one parent, you are a "silent carrier," meaning you have no signs of illness but can pass the gene to a child.



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If you inherit two mutated genes, you have alpha thalassemia trait and may have either mild anemia or no symptoms. If you inherit three or more genes from both parents, you may have hemoglobin H disease, a type of thalassemia that causes moderate to serious symptoms.

In beta thalassemia, you can inherit one or two altered genes. If you inherit one altered gene you have beta thalassemia minor, which causes mild symptoms. With two, you will have either beta thalassemia intermedia with moderate symptoms or beta thalassemia major with more serious ones.

Because thalassemia is inherited, the disease tends to run in families. People from certain parts of the world are more likely to have thalassemia than others. The genes that cause thalassemia are more common in people from the Middle East, the Mediterranean, Asia, and Africa.

How is thalassemia treated?

Treatment for thalassemia depends on how serious the symptoms are. Someone with the trait or with only mild symptoms may not need treatment. **Treatments for more serious thalassemia can include:**

- Blood transfusions to provide your body with more red blood cells.
- Iron chelation therapy to remove excess iron from your body.
- Blood and bone marrow transplant or gene therapy to help your body make working red blood cells.
- Folic acid to help red blood cells develop and also to treat anemia.

Other treatments may involve medications that reduce the symptoms or a splenectomy – surgery to remove your spleen, which can become enlarged and make anemia worse.

Not all treatments are right for all patients, so talk to your healthcare provider if your treatment plan doesn't seem to be working for you.

What do I need to know about pregnancy and thalassemia?

If you have thalassemia and are planning to become pregnant, meet with a genetic counselor who can explain your risk of passing the condition to your children and your options for prenatal diagnosis and testing.

Having thalassemia might make it more difficult to become pregnant. It could also increase the chances of health risks during pregnancy. Talk with your healthcare provider to learn how you can safely manage your condition and lower your risk of pregnancy complications.

How do I live well while managing my thalassemia?

It is important to be proactive about your health, manage your disorder, and adopt a healthy lifestyle. Work with your healthcare provider and follow their treatment plan. Get regular checkups and stay up to date on recommended vaccines, especially if you have had a splenectomy. If you have been prescribed blood transfusions or chelation therapy, it is important that you stick to that schedule to prevent anemia or organ damage from iron overload.

Work on building good habits:

- Eat a healthy diet
- Exercise regularly
- Stop smoking
- Get enough quality sleep

Learn more about thalassemia at nhlbi.nih.gov/health/thalassemia

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