Alpha Thalassemia

Thalassemias are a group of blood disorders that affect the way the body makes hemoglobin. Hemoglobin is a protein found in red blood cells that carries oxygen throughout the body. It's made up of alpha globin and beta globin.

The body contains more red blood cells than any other type of cell, and each has a life span of about 4 months. Each day, the body produces new red blood cells to replace those that die or are lost from the body.

With thalassemia, the red blood cells are destroyed at a faster rate, leading to anemia, a condition that can cause fatigue and other complications.

Thalassemias are inherited conditions — they're carried in the genes and passed on from parents to children. People who are carriers of a thalassemia gene show no thalassemia symptoms and might not know they're carriers. If both parents are carriers, they can pass the disease to their kids. Thalassemias are not contagious.

While there are many different types of thalassemias, the main two are:

- **Alpha thalassemia:** when the body has a problem producing alpha globin
- **Beta thalassemia:** when the body has a problem producing beta globin

When the gene that controls the production of either of these proteins is missing or mutated, it results in that type of thalassemia.

**About Alpha Thalassemia**

Normally, each person has four genes for alpha globin. Alpha thalassemia happens when one or more of the genes that control the making of alpha globins is absent or defective. It can cause anemia ranging from mild to severe and is most commonly found in people of African, Middle Eastern, Chinese, Southeast Asian, and, occasionally, Mediterranean descent.

Some children with alpha thalassemia have no symptoms and require no treatment. Others with more severe cases need regular blood transfusions to treat anemia and other symptoms.
A child can only get alpha thalassemia by inheriting it from his or her parents. Genes are "building blocks" that play an important role in determining physical traits and many other things about us.

Alpha globin is made by a gene on chromosome 16. So, if any gene that tells chromosome 16 to produce alpha globin is missing or mutated, less alpha globin is made. This affects hemoglobin and decreases the ability of red blood cells to transport oxygen around the body.

**Types of Alpha Thalassemia**

Alpha globin is made by four genes and one or more can be mutated or missing, so there are four kinds of alpha thalassemia:

- **One missing or abnormal gene** makes a child a silent alpha thalassemia carrier. Silent alpha thalassemia carriers have no signs or symptoms of the disease, but are able to pass thalassemia on to their children.

- **Two missing or mutated genes** is a condition called alpha thalassemia minor or having alpha thalassemia trait. Children with this condition may have red blood cells that are smaller than normal (microcytosis) and sometimes very slight anemia.
  - **People with alpha thalassemia minor usually don't have any symptoms at all, but can pass thalassemia on to their children.** The two abnormal genes can be on the same chromosome (called the cis position) or one on each chromosome (called the trans-position). If two genes on the same chromosome are affected, the person can pass along a two-gene defect to his or her child. This situation is much more common in people of Asian descent.

- **Three missing or mutated genes** is called hemoglobin H disease. Signs and symptoms will be moderate to severe.

- **Four missing or mutated genes** is a condition known as alpha thalassemia major or *hydrops fetalis*. **This almost always leads to a fetus dying before delivery or a newborn baby dying shortly after birth.** However if this disease is suspected because of a history in the family, it can be diagnosed prenatally. Sometimes, if treatment is initiated before the baby is even born, the baby can survive.

**Complications**

In addition to anemia and *hydrops fetalis*, severe cases of alpha thalassemia and hemoglobin H disease can lead to serious complications, especially if untreated. Complications of alpha thalassemia include:
• **Excess iron.** When children have alpha thalassemia, they can end up with too much iron in their bodies, either from the disease itself or from getting repeated blood transfusions. Excess iron can cause damage to the heart, liver, and endocrine system.

• **Bone deformities and broken bones.** Alpha thalassemia can cause bone marrow to expand, making bones wider, thinner, and more brittle. This makes bones more likely to break and can lead to abnormal bone structure, particularly in the bones of the face and skull.

• **Enlarged spleen.** The spleen helps fight off infections and filters out unwanted materials, such as dead or damaged blood cells, from the body. Alpha thalassemia can cause red blood cells to die off at a faster rate, making the spleen work harder, which makes it grow larger. A large spleen can make anemia worse and may need to be removed if it gets too big.

• **Infections.** Children with alpha thalassemia have an increased risk of infection, especially if they've had their spleens removed.

• **Slower growth rates.** The anemia resulting from alpha thalassemia can cause children to grow more slowly and also can lead to delayed puberty.

**Symptoms**

The signs and symptoms of alpha thalassemia vary depending on the type that a child has and how severe it is. **Children with the alpha thalassemia trait and those who are silent carriers have no symptoms at all.**

Some of the more common symptoms of alpha thalassemia include:

- Fatigue, weakness, or shortness of breath
- A pale appearance or a yellow color to the skin (jaundice)
- Irritability
- Deformities of the facial bones
- Slow growth
- A swollen abdomen
- Dark urine

**Diagnosis**

In most cases, alpha thalassemia is diagnosed before a child's second birthday or through newborn screening, a blood test given when the child is first born. Children with alpha thalassemia major may have a swollen abdomen or symptoms of anemia or failure to thrive.

If the doctor suspects alpha thalassemia, he or she will take a blood sample for testing. Blood tests can reveal red blood cells that are pale, varied in shape and
size, or smaller than normal. They also can detect low red blood cell counts and cells with an uneven distribution of hemoglobin, which causes them to look like a bull’s-eye when seen through a microscope.

Blood tests also can measure the amount of iron in the blood, evaluate hemoglobin, and test a child's DNA for abnormal hemoglobin genes.

If both parents are carriers of the alpha thalassemia disorder, doctors can conduct tests on a fetus before birth. This is done through either:

- **Chorionic vilius sampling**, which takes place about 11 weeks into pregnancy and involves removing a tiny piece of the placenta for testing.
- **Amniocentesis**, which is usually done about 16 weeks into the pregnancy and involves removing a sample of the fluid that surrounds the fetus.

**Treatment**

The amount of treatment that alpha thalassemia requires depends on how severe the symptoms are. **For those with alpha thalassemia trait or silent carriers with only mild anemia from time to time, no medical treatment is necessary.**

However, the blood counts in the alpha thalassemia trait look a lot like the blood counts in iron deficiency anemia, which is a very common disorder. It’s important for doctors to know when children have alpha thalassemia trait so that they do not treat them with iron if it’s not needed.

Doctors also might recommend a folic acid supplement for kids with hemoglobin H disease to help the body make new red blood cells. In addition, these kids may require an occasional blood transfusion, particularly after surgery. Less commonly, children with severe cases of hemoglobin H disease may require regular blood transfusions their entire lives to keep them healthy. During blood transfusions, they're given blood from donors with matching blood types. Over time, this can cause a build-up of iron in the body, so kids who receive frequent blood transfusions may have to take medications to remove excess iron from their bodies.

Currently, the only cure for thalassemia is a procedure called a bone marrow transplant (also called a stem cell transplant). Bone marrow, which is found inside bones, produces blood cells. In a bone marrow transplant, a person is first given high doses of radiation or drugs to destroy the defective bone marrow. The bone marrow is then replaced with cells from a compatible donor, usually a healthy sibling or other relative. Bone marrow transplants carry many risks, so they usually are done only in the most severe cases of thalassemia.
Because living with alpha thalassemia can be challenging, people who are carriers of alpha thalassemia trait may want to seek genetic counseling if they're considering having children.

If your child has alpha thalassemia, support groups are available to help your family cope with the obstacles presented by the disease.

For more information, call 775-982-3892.