Alpha thalassemia is an inherited blood condition. It is most often found in people whose ancestors come from Asian countries, including China, the Philippines, Malaysia, Thailand, Cambodia, Laos, Vietnam, Burma, India, and Sri Lanka, but it can be found in people with ancestry from other parts of the world. To understand this condition, it helps to know more about how your blood is made.

**Hemoglobin**

Your blood contains millions of red blood cells. Each of your red blood cells has hemoglobin, which gives blood its red color and carries oxygen throughout your body. Hemoglobin is made by combining a “heme” portion (iron) and a “globin” portion (protein). The iron comes from the food you eat and your body makes the globins.

There are different kinds of hemoglobin that the body can make. The most common kind of hemoglobin in an adult is hemoglobin A. For hemoglobin A, your body puts two “alpha” globin chains together with two “beta” globin chains. If you don’t make enough alpha chains it is called alpha thalassemia.

The instructions for making globin chains are part of the genetic information you inherit from your parents. Genetic instructions are called genes. You inherit your genes in pairs, with one copy of each gene coming from each parent. One particular gene, the alpha globin gene, is responsible for telling the body how to make alpha globin chains. Unlike most genes, there are actually two pairs of alpha globin genes, for a total of four copies of the gene. For most people all four copies of the gene can make alpha globin. Alpha thalassemia happens when one or more of these genes are not working.

**Alpha thalassemia**

There are different forms of alpha thalassemia, based on how many alpha globin genes are working. All forms of alpha thalassemia can be passed to offspring.

**Silent carrier**

A person with three working copies of the alpha globin gene is a silent carrier. This is not a disease and does not affect your health. This form of alpha thalassemia is usually not detected by routine laboratory tests.

**Alpha thalassemia trait**

A person with two working copies of the alpha globin gene has alpha thalassemia trait. Like the silent carrier, alpha thalassemia trait is not a disease and does not affect your health. Alpha thalassemia trait causes the size of the red blood cells to be smaller than usual. Sometimes, this is mistaken for low iron levels (iron-deficiency anemia). However, taking an iron supplement does not change the size of the red blood cells.

**Hemoglobin H disease**

A person with only one working copy of the alpha globin gene has hemoglobin H disease. In hemoglobin H disease, the red blood cells are broken down by the body faster than usual and there are fewer red blood cells in the body. This causes a lifelong mild to moderate form of anemia. Despite having anemia, most people with this condition have no serious health problems. However, medical problems can range from none, to an occasional need for blood transfusions, to severe chronic anemia. A person with hemoglobin H disease should receive regular medical care. In rare cases, the spleen
can become enlarged or gallstones can form. There are also certain medications and other substances (like moth balls and fava beans) that can cause severe anemia in someone with hemoglobin H disease. Your doctor can tell you what to avoid.

**Alpha thalassemia disease (alpha thalassemia major)**

Alpha thalassemia disease happens when there are no working copies of the alpha globin gene. This causes a very severe disease that affects the developing baby before birth. The baby cannot make any normal hemoglobin. This leads to stillbirth or death shortly after birth. In some cases, treatment for the baby has been done during pregnancy.

When the baby has alpha thalassemia major, there are also risks for the pregnant woman. A woman carrying a baby with alpha thalassemia disease has a higher chance for developing pre-eclampsia or "toxemia" (protein in the urine, high blood pressure, swollen ankles and feet). This can become a very serious medical problem and requires close medical supervision.

**Inheritance and prenatal diagnosis.**

If you have any form of alpha thalassemia, it is possible to pass it to your children. The form of alpha thalassemia your children could inherit depends on the form of alpha thalassemia in both parents. Blood tests can find out what exactly what form of alpha thalassemia you have. Then, your genetic counselor can tell you the chance that a child of yours could inherit the severe form of alpha thalassemia.

When only one parent has alpha thalassemia trait and the other does not, there is no chance of having a baby with the most severe form of alpha thalassemia. However, each child has a 50% (or 1 in 2) chance to inherit the trait from the parent. This can be helpful for your baby’s pediatrician to know, so your baby is not treated with iron needlessly.

Hemoglobin H can happen when one parent has alpha thalassemia trait and the other parent is a silent carrier. In this situation, there is a 25% chance for hemoglobin H disease, but the pregnancy is not at risk for the severe form of alpha thalassemia disease.

The most severe form of alpha thalassemia, alpha thalassemia disease (alpha thalassemia major) can only happen when both parents have alpha thalassemia trait. When both parents have alpha thalassemia trait, there usually is a 25% (or 1 in 4) chance in each pregnancy for the baby to have alpha thalassemia disease and a 75% (or 3 in 4) chance that the baby will not have this disease.

It is possible to test a developing baby for alpha thalassemia disease as early as 10 weeks in pregnancy. If testing shows the baby has alpha thalassemia disease, parents can choose whether or not to continue the pregnancy.
**Alpha thalassemia**

Common Inheritance Patterns

**KEY TO SYMBOLS**

- Four working alpha globin genes
- Three working alpha globin genes
- Two working alpha globin genes
- One working alpha globin gene
- No working alpha globin genes

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**One parent with alpha thalassemia trait**

- Trait
- Trait

Each pregnancy has a 50% chance to have **trait**. NOT at risk for disease

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**One parent with alpha thalassemia trait and One parent a silent carrier**

- Trait
- Silent carrier
- Hemoglobin H disease

Each pregnancy has a 25% chance to have **hemoglobin H disease**

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**Both parents with alpha thalassemia trait**

- Trait
- Trait
- Alpha thalassemia disease

Each pregnancy has a 25% chance to have **alpha thalassemia disease**

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*Genetics.kp.org*

This information is not intended to diagnose health problems or to take the place of medical advice or care you receive from your physician or other health care professional.

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