

## Alpha Thalassemia Trait

Alpha thalassemia is common in people of African, Southern Chinese, Southeast Asian, Middle Eastern and Mediterranean descent.

- Alpha thalassemia affects the amount of hemoglobin in the red blood cells.
- Adult hemoglobin (hemoglobin A) is made of alpha and beta globins.
- Normally, people have four (4) genes for alpha globin with two (2) genes on each chromosome ( $\alpha\alpha/\alpha\alpha$ ).
- People with alpha thalassemia trait only have two (2) genes for alpha globin, so they make slightly lower amounts of hemoglobin.
- All red blood cells contain hemoglobin, which carries oxygen from the lungs to all parts of the body.

### What is Alpha Thalassemia?

Alpha thalassemia genes can mix in different ways; this can range from the loss of one alpha thalassemia gene to the loss of four alpha thalassemia genes. Based on the number of alpha thalassemia gene(s) a person is missing, the different outcomes are listed below:

- Missing one alpha thalassemia gene (silent carriers): ( $\alpha\alpha/\alpha-$ ), a silent carrier does not have any symptoms.
- Missing two alpha thalassemia genes (alpha thalassemia trait): ( $\alpha\alpha/--$ ) or ( $\alpha-/α-$ ), this normally does not cause health problems, but can cause low blood levels (anemia) and small red blood cells.
- Missing three alpha thalassemia genes (Hemoglobin H disease): ( $\alpha-/--$ ), this disease causes health problems and requires treatment by a physician.
- Missing four genes (Hydrops fetalis): ( $--/--$ ), this is a life threatening disease.

### What is Bart's Hemoglobin?

In a new baby, the presence of Bart's Hemoglobin indicates alpha thalassemia. Alpha thalassemia can cause low blood levels (mild anemia), this can be confused with iron deficiency. It **should not** be treated with iron supplements, unless iron deficiency is present in addition to Alpha thalassemia. Iron deficiency should be verified through iron studies in people with alpha thalassemia. Notify your physician if your child has Bart's Hemoglobin in the new born screen.

1. A small amount of Bart's Hemoglobin indicates that your child is a silent carrier ( $\alpha\alpha/\alpha-$ ) or has alpha thalassemia trait ( $\alpha-/α-$ ) or ( $\alpha\alpha/--$ ).
- If a small amount of Bart's Hemoglobin is present at birth, it will disappear shortly after birth and may not be detectable with standard blood tests.
- The only way to determine a silent carrier, other than by DNA testing, is through the presence of Bart's Hemoglobin at birth.
2. If there is a large amount of Bart's Hemoglobin at birth, it may indicate the loss of 3 or more alpha thalassemia genes. In this case, the Bart's Hemoglobin will probably not disappear from the blood. This may indicate the presence of Hemoglobin H disease.

### What is Alpha Thalassemia trait?

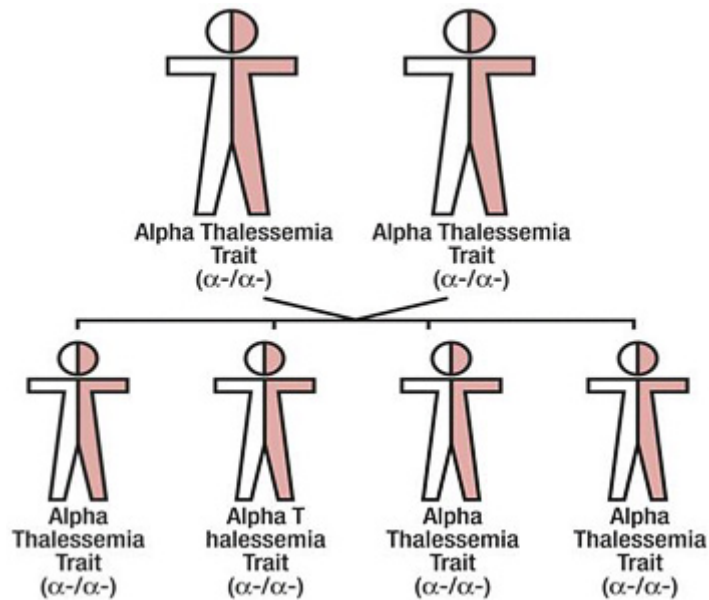
There are two different types of alpha thalassemia trait.

1. The first type of alpha thalassemia trait has one alpha gene missing on each chromosome ( $\alpha-/α-$ ). This is called the **trans** form of alpha thalassemia trait.
- The **trans** form of alpha thalassemia trait ( $\alpha-/α-$ ) is common in African-Americans (20-30%) and in people of African descent. It is rare for African-Americans to have the **cis** form of alpha thalassemia, but it can happen.
2. The second type of alpha thalassemia trait has two missing alpha genes on the same chromosome ( $\alpha\alpha/--$ ). This is called the **cis** form of alpha thalassemia trait.
3. Both types of alpha thalassemia trait are common in people of Southeast Asian, Southern Chinese, Mediterranean and Middle Eastern descent, however, the **cis** type of trait is more common.

People with alpha thalassemia trait do not develop Hemoglobin H cell disease or Hydrops Fetalis later in life.

### Why is it important to know if I have Alpha Thalassemia trait?

- Alpha thalassemia trait is inherited from one's parents, like hair or eye color
- If both parents have the **trans** form of alpha thalassemia trait ( $\alpha-/α-$ ), all of their children will have alpha thalassemia trait.
- Alpha thalassemia trait normally does not cause any health problems.
  - People with alpha thalassemia trait can have small red blood cells and a low red blood count (mild anemia).
  - Alpha thalassemia **should not** be treated with iron. This treatment will not help people with alpha thalassemia trait. Alpha thalassemia can be treated with iron, only if iron deficiency is present in addition to Alpha thalassemia and is confirmed by your doctor

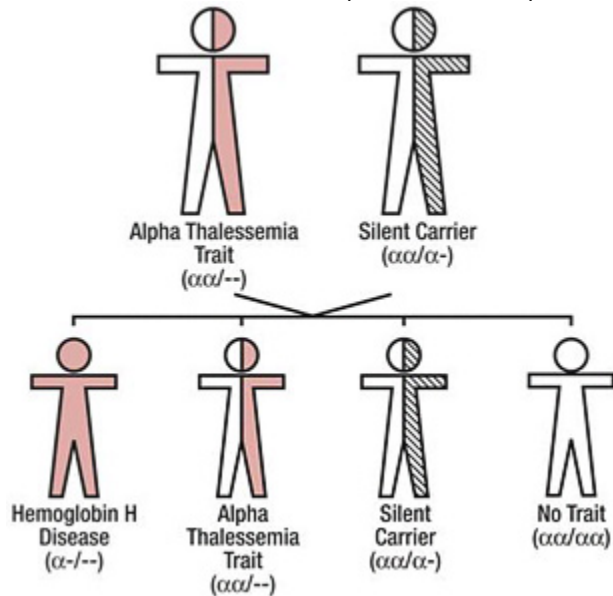


**What if one parent has Alpha Thalassemia trait and the other parent is a silent carrier?**

If one parent has the *cis* form of alpha thalassemia trait ( $\alpha\alpha/--$ ), and the other parent is a silent carrier ( $\alpha\alpha/\alpha^-$ ), there is a 25 percent (1 in 4) chance **with each pregnancy** of having a child with Hemoglobin H disease.

- People with Hemoglobin H disease can have an enlarged spleen, low red blood cell counts, gallstones, developmental delays, and other health problems with Hemoglobin H disease.
- Hemoglobin H disease is rare in people of African descent.

Hemoglobin H disease is lifelong illness that can result in serious health problems and requires treatment by a physician



**What if both parents have the *cis* form of Alpha Thalassemia trait?**

- If both parents have the *cis* form of alpha thalassemia trait ( $\alpha\alpha/--$ ), there is a 25 percent (1 in 4) chance **with each pregnancy** of having a child with Hydrops Fetalis ( $--/--$ ).
- Hydrops fetalis is a very serious health condition and usually causes death before or shortly after birth.
- Infants born with Hydrops Fetalis normally do not survive because they are unable make enough hemoglobin.

