St. Jude Children's Research Hospital

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Alpha Thalassemia Trait

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This document is not intended to replace counseling by a trained health care professional or genetic counselor. Our aim is to promote active participation in your care and treatment by providing information and education. Questions about individual health concerns or specific treatment options should be discussed with your doctor. For general information on sickle cell disease and other blood disorders, please visit our Web site at *www.stjude.org/sicklecell*.

Alpha thalassemia trait

All red blood cells contain hemoglobin (HEE muh glow bin), which carries oxygen from your lungs to all parts of your body. Alpha thalassemia (thal uh SEE mee uh) trait is a condition that affects the amount of hemoglobin in the red blood cells.

- Adult hemoglobin (hemoglobin A) is made of alpha and beta globins.
- Normally, people have 4 genes for alpha globin with 2 genes on each chromosome (αα/αα).

People with alpha thalassemia trait only have 2 genes for alpha globin, so their bodies make slightly less hemoglobin than normal. This trait was passed on from their parents, like hair color or eye color.

A trait is different from a disease

Alpha thalassemia trait is not a disease. Normally, a trait will not make you sick.

Parents who have alpha thalassemia trait can pass it on to their children. Also, their children might be at risk for hemoglobin H disease or hydrops fetalis.

In this booklet we will tell you about:

Alpha thalassemia trait;

Hemoglobin H disease;

Hydrops fetalis; and

Bart's hemoglobin.

Who can have alpha thalassemia trait?

Alpha thalassemia is common in people whose ancestors came from Africa, Southern China, Southeast Asia, the Middle East, and the Mediterranean region. It is possible for a person of any nationality to have alpha thalassemia trait.

What is alpha thalassemia trait?

Alpha thalassemia is a condition caused by having fewer alpha globin genes than normal.

Normally, people have 4 genes for alpha globin. People with alpha thalassemia can be missing one (1), 2, 3, or 4 alpha globin genes.

1. People missing one (1) alpha globin gene (αα/α-) are called silent carriers of alpha thalassemia. This means they can pass on the condition of having one (1) gene missing. However, being without a gene does not affect their health or the way they feel.
    - A silent carrier does not have any symptoms.
    - If you are a silent carrier, you **do not** have hemoglobin H disease and cannot develop it later in life.
2. People with 2 missing alpha globin genes (αα/-- or α-/α-) have alpha thalassemia trait.
    - This normally does not cause health problems but can cause low red blood cell levels (anemia) and small red blood cells.
    - If you have alpha thalassemia trait, you **do not** have hemoglobin H disease and cannot develop it later in life.
3. People with 3 missing genes have hemoglobin H disease (α-/--).
    - This disease causes health problems. People with this disease need a doctor's treatment.

4. People with 4 missing genes have hydrops fetalis (--/--).
    - This is a life-threatening condition.

Two types of alpha thalassemia trait

There are 2 types of alpha thalassemia trait.

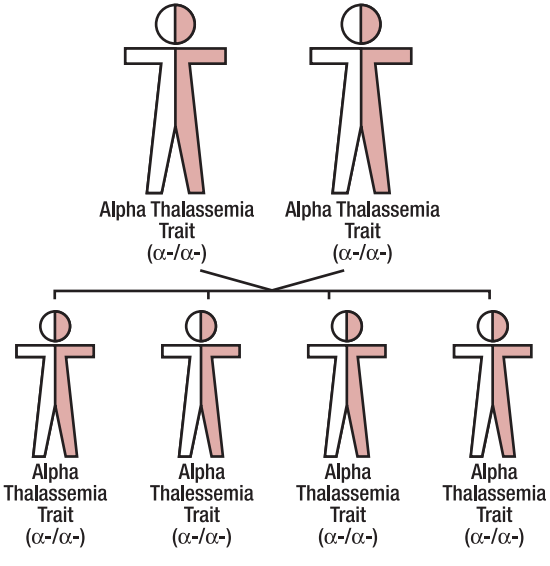
1. People with the first type of alpha thalassemia trait have one (1) alpha globin gene missing on each chromosome (α-/α-). This is called the ***trans*** form of alpha thalassemia trait.
    - The *trans* form of alpha thalassemia trait (α-/α-) is common in African-Americans (20–30 percent) and people of African descent.
2. People with the second type of alpha thalassemia trait have 2 missing alpha globin genes on the same chromosome (αα/--). This is called the ***cis*** form of alpha thalassemia trait.

The ***trans*** and ***cis*** types of alpha thalassemia trait are found most often in people whose ancestors come from Southeast Asia, Southern China, the Mediterranean, and the Middle East.

People with alpha thalassemia trait *do not* develop hemoglobin H disease or hydrops fetalis later in life.

Why do I need to know if I have alpha thalassemia trait?

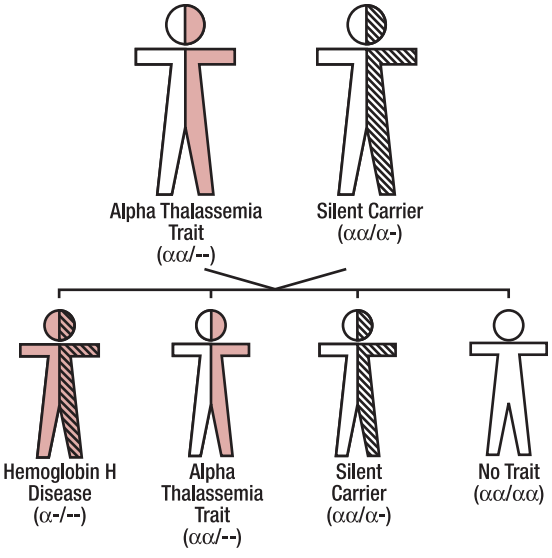
- You can pass on alpha thalassemia trait to your children, like you would hair color or eye color.
- If both parents have the ***trans*** form of alpha thalassemia trait (α-/α-), all of their children will have alpha thalassemia trait.
- Alpha thalassemia trait normally does not cause any health problems.
  - People with alpha thalassemia trait can have small red blood cells and a low red blood cell count (mild anemia).
  - Alpha thalassemia *should not* be treated with iron because it will not help. Alpha thalassemia can be treated with iron only if the person has low iron levels in addition to alpha thalassemia. Your doctor will tell you if this happens.



What is hemoglobin H disease?

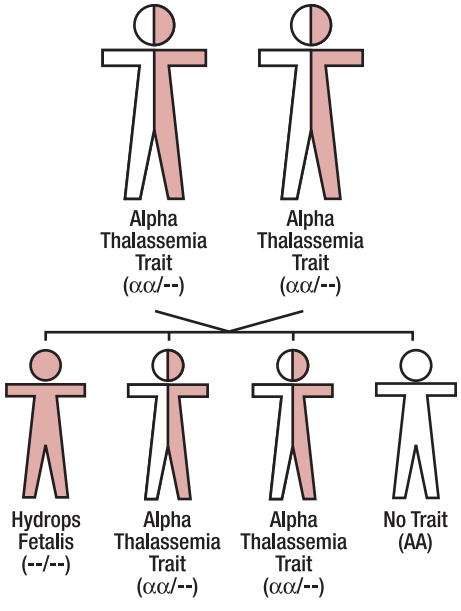
If one (1) parent has the cis form of alpha thalassemia trait (αα/--), and the other parent is a silent carrier (αα/α-), there is a 25 percent (1 in 4) chance **with each pregnancy** of having a child with hemoglobin H disease.

- People with hemoglobin H disease can have an enlarged spleen, low red blood cell counts, and gallstones. They also may have other health problems.
- Hemoglobin H disease is a lifelong illness that can cause health problems. It requires a doctor's treatment.



What is hydrops fetalis?

- If both parents have the cis form of alpha thalassemia trait (αα/--), there is a 25 percent (1 in 4) chance **with each pregnancy** of having a child with hydrops fetalis (--/--).
- Hydrops fetalis is a serious health condition that usually causes death before or shortly after birth.
- Babies born with hydrops fetalis normally do not live, because they cannot make enough hemoglobin.



What is Bart's hemoglobin on the newborn screening test?

A new baby with Bart's hemoglobin on the newborn screening means that alpha gene deletions are present, and the baby might have alpha thalassemia, alpha thalassemia trait, or hydrops fetalis.

Alpha thalassemia can cause low red blood cell levels (mild anemia) and should not be confused with not having enough iron in the blood.

Tell the doctor if your child's newborn screening test showed Bart's hemoglobin.

- If a small amount of Bart's hemoglobin is present at birth, it will usually disappear shortly after birth. This means your child has a one (1) or 2 alpha gene deletion and has alpha thalassemia trait or is a silent carrier. Normally, a second newborn screening test will not detect this condition.
- If a large amount of Bart's hemoglobin and Hemoglobin H genes are present on the newborn screening test, it usually means the baby has hemoglobin H disease (a 3 gene deletion).

- Hydrops fetalis is a condition that results from a 4 gene deletion. Usually, the fetus will not survive unless it has a blood transfusion while in the womb and continues blood transfusions after birth until a permanent treatment, such as bone marrow transplant is offered.

Recap: Alpha thalassemia facts

- People with alpha thalassemia usually do not have any health problems caused by the trait.
- People with alpha thalassemia trait can never develop hemoglobin H disease or hydrops fetalis.
- Bart's hemoglobin on the newborn screening test indicates alpha thalassemia.
- Parents who have alpha thalassemia trait can pass it on to their children. Also, their children might be at risk for hemoglobin H disease or hydrops fetalis.
  - Hemoglobin H is a blood disease that requires medical treatment.
  - Hydrops fetalis is usually fatal before or shortly after birth.

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