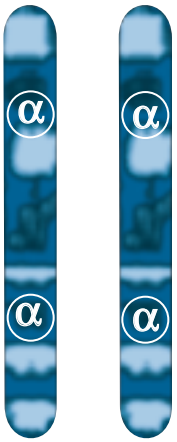
Alpha Thalassemia

Physicians often mistake alpha thalassemia trait for iron deficiency anemia and incorrectly prescribe iron supplements that have no effect on the anemia.



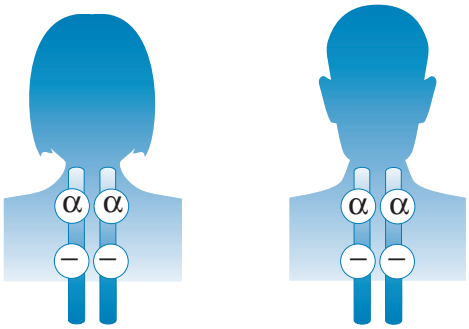
Normal alpha globin genes found on chromosome 16

People who do not produce enough alpha globin protein chains have alpha thalassemia. It is commonly found in Africa, the Middle East, India, Southeast Asia, southern China, and occasionally the Mediterranean region.

Alpha globin is made by four genes, two on each strand of chromosome 16. Individuals who have one or two abnormal alpha globin genes have alpha thalassemia trait.

An individual with one abnormal alpha globin gene is said to be a **silent carrier** of **alpha thalassemia**. This condition, in which one of the four alpha globin genes is missing or defective, generally causes no health problems because the lack of alpha globin protein is so small that there is no anemia.

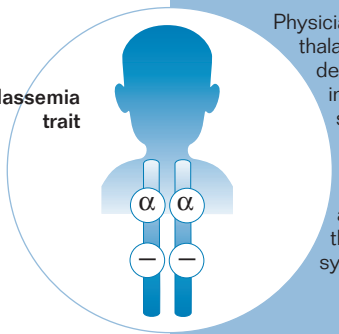
It is called "silent carrier" because of how difficult it is to detect. Silent carrier state is "diagnosed" by deduction when an apparently normal individual has a child with hemoglobin H disease or alpha thalassemia minor. It can also be diagnosed by special DNA testing.



If **both parents** carry **alpha thalassemia trait** in which the two abnormal genes of each parent are on opposite chromosomes, their children will inherit **alpha thalassemia trait.**

An individual with two abnormal alpha globin genes is said to have **alpha thalassemia trait**. The two abnormal genes can be on the same chromosome or on each chromosome in the pair.

Alpha Thalassemia Trait.
In this condition, in which two of the four alpha globin genes are missing or defective, the lack of alpha globin protein is somewhat greater. Both abnormal alpha globin genes may be on the same chromosome (cis position) or one may be on each chromosome in the pair (trans position).



Physicians often mistake alpha thalassemia trait for iron deficiency anemia and incorrectly prescribe iron supplements that have no effect on the anemia. Patients with this condition have smaller red blood cells and a mild anemia, although they do not experience symptoms.

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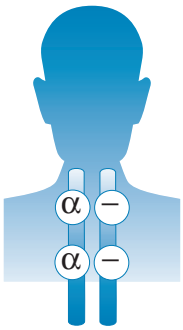
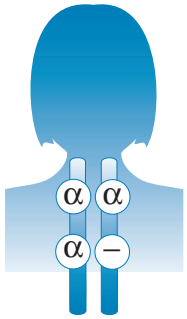
The alpha thalassemia traits combine in different ways to produce blood disorders that range from mild to severe in their effect on the human body.

if...



one parent has **alpha thalassemia trait** with two abnormal alpha globin genes on the same chromosome



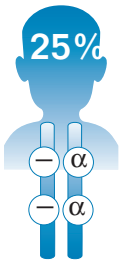
and **the other parent** has the **silent carrier state** with a single abnormal alpha globin gene on one chromosome

..then

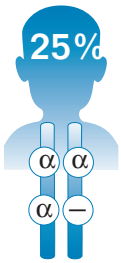
there is a **25%** chance with **each** pregnancy that their child will be born with **hemoglobin H disease** in which three of the four alpha globin genes are abnormal.

In this condition, the lack of alpha protein is great enough to cause moderate to severe anemia and may cause serious health problems such as an enlarged spleen, bone deformities and fatigue.

normal hemoglobin

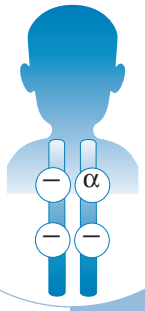


alpha thal trait

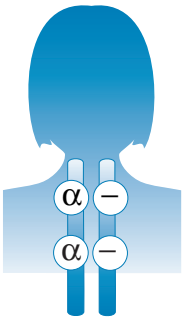


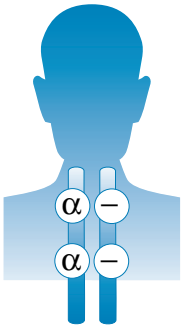
alpha thal silent carrier

25%
hemoglobin H disease



if...



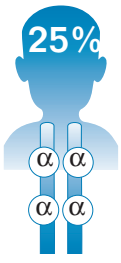


both parents carry **alpha thalassemia trait** with two abnormal alpha globin genes on the same chromosome

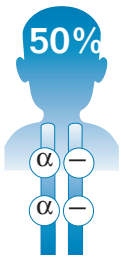
..then

there is a **25%** chance with **each** pregnancy that their child will inherit **hydrops fetalis** or **alpha thalassemia major**, in which all four alpha globin genes are abnormal.

Most individuals with this condition die before or shortly after birth. In some extremely rare cases, in utero blood transfusions have allowed the birth of children with hydrops fetalis who then require lifelong blood transfusions and extensive ongoing medical care.

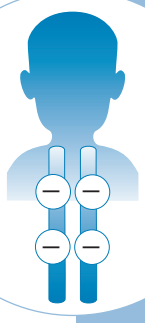


normal hemoglobin



alpha thal trait (cis Type)

25%
hydrops fetalis (alpha thal major)



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