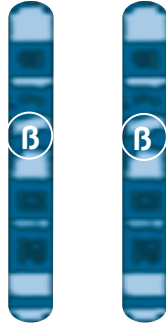


# Beta Thalassemia

Beta thalassemia is found in people of Mediterranean, Middle Eastern, African, South Asian (Indian, Pakistani, etc.), Southeast Asian and Chinese descent.

1



Normal beta globin genes found on chromosomes 11



A person with beta thalassemia trait has one abnormal beta globin gene

To understand how thalassemia affects the human body, you must first understand a little about how blood is made.

Blood carries oxygen from your lungs to other parts of your body using a protein called hemoglobin found in red blood cells. Hemoglobin is made of two different kinds of protein chains, called alpha and beta globins.

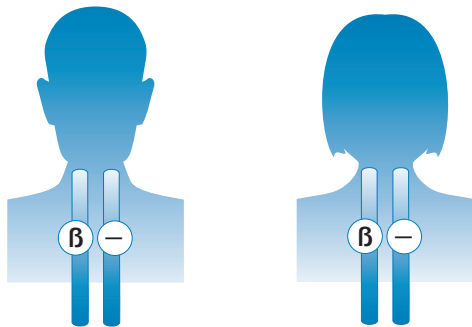
Beta globin is made by two genes, one on each chromosome 11. Individuals who have one abnormal beta globin gene have beta thalassemia trait (also known as beta thalassemia minor).

## BETA THALASSEMIA TRAIT/MINOR

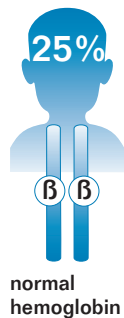
In beta thalassemia trait, one of the two beta globin genes is abnormal but the lack of beta globin is not great enough to cause problems in the normal functioning of the hemoglobin. A person with this condition simply carries the genetic trait for beta thalassemia and will usually experience no health problems other than a mild anemia.

Physicians often mistake the small red blood cells of the person with beta thalassemia minor as a sign of iron-deficiency anemia and incorrectly prescribe iron supplements that have no effect on the anemia.

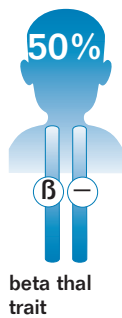
if...



both parents carry the beta thalassemia trait,

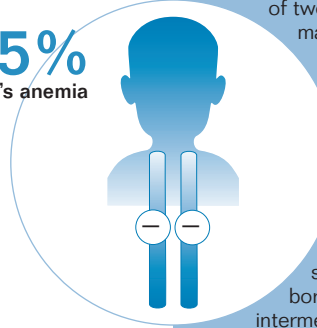


normal hemoglobin



beta thal trait

25%  
Cooley's anemia



..then

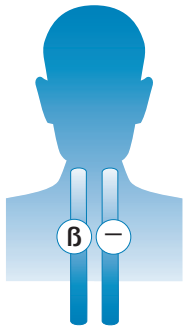
there is a **25%** chance with **each** pregnancy that their child will inherit two abnormal beta globin genes. In its most severe form, this may cause **beta thalassemia major** or **Cooley's anemia**, a blood disorder in which the lack of beta globin causes a life-threatening anemia that requires regular blood transfusions and extensive ongoing medical care. Lifelong transfusions lead to iron overload which must be treated with chelation therapy to prevent early death from organ failure.

In a somewhat milder form, the inheritance of two abnormal beta globin genes may cause **beta thalassemia intermedia**, in which the lack of beta globin in the hemoglobin causes a moderately severe anemia and significant health problems including bone deformities and enlargement of the spleen.

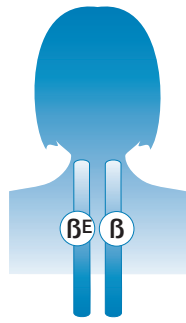
Due to the wide range in severity of this condition, the borderline between thalassemia intermedia and thalassemia major can be confusing. When a patient is dependent on blood transfusions, he is likely to be classified as thalassemia major.

The beta thalassemia trait can also combine with "variant" hemoglobins to produce other related blood disorders.

if...



one parent carries the beta thalassemia trait



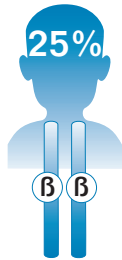
and the other parent carries the hemoglobin E trait

..then

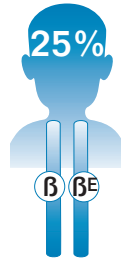
there is a **25%** chance with **each** pregnancy that their child will be born with **E beta thalassemia**, a moderately severe anemia that has similar symptoms to beta thalassemia intermedia but on occasion may be as severe as thalassemia major.

Hemoglobin E trait is one of the most common abnormal hemoglobins. It is usually found in people of Southeast Asian ancestry, such as Cambodians, Vietnamese and Thai.

**25%**  
E beta thalassemia



normal hemoglobin



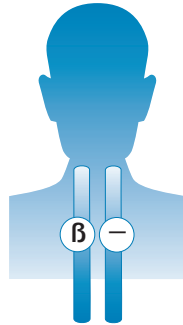
hemoglobin E trait



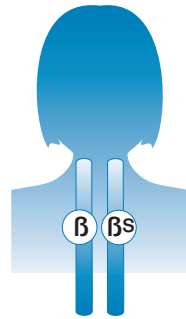
beta thalassemia trait

2

if...



one parent carries the beta thalassemia trait



and the other parent carries the hemoglobin S trait (the abnormal hemoglobin found in people with sickle cell disease)

..then

there is a **25%** chance with **each** pregnancy that their child will be born with **sickle beta thalassemia**. The severity of this condition varies according to the amount of normal beta globin produced by the beta globin gene. When no beta globin is produced by the beta globin gene, the condition is almost identical to sickle cell disease. When some beta globin is produced by the beta globin gene, the condition is less severe.

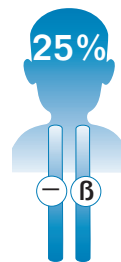
**25%**  
sickle-beta thalassemia



normal hemoglobin



sickle cell trait



beta thal trait

Hemoglobin S trait is commonly found in people of African or Mediterranean ancestry, such as Africans, Italians, Greeks, Turks, and in people from the Caribbean.