Beta Thalassemia Trait and Beta Thalassemia Disease

Beta thalassemia trait is an inherited blood disorder. It is more often found in people with Italian, Greek, Asian, or African heritage, but it can be found in people with ancestry from any part 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 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 beta chains it is called beta 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 beta globin gene, is responsible for telling the body how to make beta globin chains. In beta thalassemia, the gene for making beta globin chains doesn’t work normally.

Beta Thalassemia Trait
When you have one working copy of the beta globin gene and one non-working copy you have beta thalassemia trait. Your body makes less of the beta chains, but is still able to make plenty of hemoglobin A. This is not a disease and does not affect your health. This trait causes 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.

A person with beta thalassemia trait will never develop beta thalassemia disease. The importance of identifying beta thalassemia trait is that it helps find couples whose children may be born with beta thalassemia disease.

Beta Thalassemia Disease (Beta thalassemia major)
Beta thalassemia disease is a lifelong condition that can include serious health problems, but it affects each person differently. The most common form of the disease requires regular blood transfusions and extensive medical care due to severe anemia. Starting treatment early in life helps decrease the symptoms of the disease, but repeated transfusions can cause medical problems from iron build-up in the body.

Beta thalassemia disease has been cured using bone marrow transplantation, but the procedure has serious risks and requires a suitable donor.
Other forms of hemoglobin disease

Most of the time beta thalassemia disease happens when both parents have beta thalassemia trait. But sometimes one parent carries a different blood trait, such as hemoglobin E or hemoglobin S (sickle cell). When one of these other hemoglobins combines with beta thalassemia trait, the result is a different form of hemoglobin disease. Other diseases that can be related to beta thalassemia include:

- Hemoglobin E-beta thalassemia disease
- Sickle-beta thalassemia disease

Some forms of hemoglobin disease have more medical problems than others.

Inheritance and prenatal diagnosis.

If you have beta thalassemia trait, it is possible to pass it to your children. However, the chance for a hemoglobin disease depends on the kind of blood trait in both parents. Blood tests can find out exactly what trait you have, if any. Then, your genetic counselor can tell you the chance that a child of yours could inherit beta thalassemia disease or another form of hemoglobin disease.

When only one parent has beta thalassemia trait and the other does not have a blood trait, there is essentially no chance of having a baby with any form of beta thalassemia disease. However, each child has a 50% (or 1 in 2) chance to inherit beta thalassemia trait from the parent.

Beta thalassemia disease can only happen when both parents have beta thalassemia trait. When both parents have beta thalassemia trait, there is a 25% (or 1 in 4) chance in each pregnancy for the baby to have beta thalassemia disease. There is a 75% (or 3 in 4) chance that the baby will not have this disease.

When one parent has beta thalassemia trait and the other parent has a related blood trait (hemoglobin E or hemoglobin S), there is a 25% (or 1 in 4) chance in each pregnancy for the baby to have a hemoglobin disease. There is a 75% (or 3 in 4) chance that the baby will not have this disease.

It is possible to test the developing baby for beta thalassemia disease as early as 10 weeks in pregnancy. If testing shows the baby has beta thalassemia disease, parents can choose whether or not to continue the pregnancy. Early detection can also allow the family to prepare for the birth of a baby who may need specialty care in infancy.

In California, all babies are routinely tested for beta thalassemia disease through the Newborn Screening Program. Beta thalassemia trait is not usually detectable in the newborn period.
Beta Thalassemia
Common Inheritance Patterns

KEY TO SYMBOLS

<table>
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<tr>
<th>Symbol</th>
<th>Description</th>
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| AA     | Hemoglobin A  
- Two working beta globin genes |
| AB     | Beta thalassemia trait  
- One working beta globin gene  
- One non-working beta globin gene |
| BB     | Beta thalassemia disease  
- Two non-working beta globin genes |
| AE     | Hemoglobin E trait  
- One working beta globin gene  
- One hemoglobin E gene |
| BE     | E-Beta thalassemia disease  
- One non-working beta globin gene  
- One hemoglobin E gene |

One parent with beta thalassemia trait

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

Both parents with beta thalassemia trait

Each pregnancy has a 25% chance to have beta thalassemia disease

One parent with beta thalassemia trait and One parent with hemoglobin E trait

Each pregnancy has a 25% chance to have E-beta thalassemia disease

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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