

# BETA THALASSEMIA TRAIT

## Positive Carrier Screening Result

**You have recently learned that you have beta thalassemia trait. Here is important information about being a trait carrier.**

Hemoglobin is found in red blood cells and moves oxygen through the body. Hemoglobin A (Hb A) is the most common type of hemoglobin. Everyone has two copies of genes that make Hb A. A person with beta thalassemia trait has one copy of a gene that makes a usual amount of Hb A and one copy that makes less or no Hb A. When there is not enough Hb A it can cause an inherited blood condition called **beta thalassemia disease**.

**Beta thalassemia trait does not affect your health.** You have enough Hb A and will not develop beta thalassemia disease. Most beta thalassemia trait carriers (Hb A/ $\beta$ ) have small red blood cells and mild anemia, but no treatment is needed. There is a 1 in 2 (50%) chance of passing this trait to any of your children.

**If you are pregnant, the next step is to test your partner.** Both parents must carry a hemoglobin trait to be at-risk for a baby with a blood condition. Your partner can be tested at any Kaiser Permanente laboratory at no cost. A genetic counselor will let you know how to arrange testing.

### Why test your partner?

- You want to know the chance that your baby could have a blood condition.
- You want to be prepared if your baby is found to be at-risk for a blood condition.
- You might consider testing the baby for a blood condition during pregnancy.

### Why would you not test your partner?

- You do not want to learn about a risk for a blood condition during pregnancy.
- The result would not change anything you do during pregnancy.

### What happens if my partner is a trait carrier?

When both parents are trait carriers, there is a 1 in 4 (25%) chance for a baby to have a blood condition. There is also a 3 in 4 (75%) chance that a baby will not have a blood condition. If you are pregnant, you will be offered prenatal testing to find out if your baby has a blood condition. A procedure, such as amniocentesis or CVS, is able to diagnose blood conditions during pregnancy. Prenatal testing could help with pregnancy care and delivery plans. Some parents may use the results to consider whether to continue or end the pregnancy. Testing could also be done after birth through the newborn screening program.

### MORE ABOUT: THE BETA THALASSEMIA GENE (*HBB*)

The beta-globin protein is made by a gene called *HBB*. In order to make Hb A, the body combines equal parts of two proteins: beta-globin and alpha-globin. Alpha-globin is made by a different gene called *HBA*. Changes (variants) in the *HBB* gene can affect the amount of Hb A in the body. *HBB* variants are labeled by how much beta-globin is made.

- $\beta^0$  (beta-zero): no beta-globin is made.
- $\beta^+$  (beta-plus): a reduced amount of beta-globin is made.

Since everyone has two copies of *HBB*, a person can have a mix of variants. For example, a person could inherit  $\beta^0$  from both parents ( $\beta^0/\beta^0$ ). This causes a more severe type of beta thalassemia disease because no beta-globin is made. A person who inherits  $\beta^0$  from one parent and  $\beta^+$  from the other parent ( $\beta^0/\beta^+$ ) still makes some Hb A. Special testing is usually needed to determine the *HBB* variants.

## Are there different types of beta thalassemia disease?

There are two main types of beta thalassemia disease based on when symptoms start and what treatment is needed. Sometimes we are not able to know which type a baby has before birth.

- **Transfusion-dependent (beta thalassemia major)** - Symptoms start between 6 months and 2 years of age. Regular blood transfusions are needed to survive. Medicine is needed to remove extra iron from the body. People with this condition usually inherit a  $\beta^0$  variant from both parents ( $\beta^0/\beta^0$ ).
- **Non-transfusion dependent (beta thalassemia intermedia)** - Symptoms usually start after 2 years of age to early adult. Blood transfusions may be needed occasionally, but are not required for survival. There are many beta thalassemia conditions that are non-transfusion dependent. These can be caused by different genetic variants. For example, this can happen when a person inherits a  $\beta^0$  variant from one parent and a  $\beta^+$  variant from the other parent ( $\beta^0/\beta^+$ ).

### Other blood conditions related to beta thalassemia disease:

The severity of these conditions can vary depending on the type of beta thalassemia variant ( $\beta^0$  or  $\beta^+$ ).

**Hb E-beta thalassemia disease** - People with this condition inherit a beta thalassemia variant from one parent and Hb E from the other parent. Some people have mild anemia and no serious medical problems. Some people have severe anemia and require regular blood transfusion.

**Hb S-beta thalassemia disease** - People with this condition inherit a beta thalassemia variant from one parent and Hb S from the other parent. This is a form of sickle cell disease. The red blood cells can collapse into a sickle shape, instead of the usual round shape. Sickled cells can get stuck in blood vessels, causing pain and damage to any area where this occurs. Symptoms are different for each person and may include severe anemia, repeat infections, pain, and fatigue.

**Hb C-beta thalassemia disease** - People with this condition inherit a beta thalassemia variant from one parent and Hb C trait from the other parent. This usually causes mild to moderate anemia. A person may need occasional visits with a doctor and might develop gall stones or an enlarged spleen as they get older. However, most people do not need regular transfusions.

**Hb D-beta thalassemia disease** - People with this condition inherit a beta thalassemia variant from one parent and Hb D from the other parent. This usually causes mild to moderate anemia. A person may need occasional visits with a doctor, but does not usually need medical treatment.

**Alpha thalassemia disease** - Since alpha globin is also needed to make Hb A, changes in the *HBA* gene can affect the symptoms of beta thalassemia. Extra copies of the *HBA* gene in a person with beta-thalassemia trait can cause severe anemia which could require transfusions. Testing the *HBA* gene is included in partner testing.

## Testing Your Partner for Hemoglobin Traits

Anyone can carry a hemoglobin trait, even if there is no history of blood conditions in the family. You usually don't know if you're a carrier without special testing. Carrier screening detects most hemoglobin trait carriers. The chance for your partner to be a trait carrier depends on their ancestry or ethnic background.

- **Beta thalassemia trait (Hb A $\beta$ )** is more common in people with Asian Indian, Mediterranean, Middle Eastern, Chinese, and Southeast Asian ancestry.
- **Hb E trait (Hb AE)** is more common in people with Southeast Asian or Asian Indian ancestry.
- **Hb S trait (sickle cell trait or Hb AS)** is more common in people with African and Hispanic/Latino ancestry.
- **Hb C trait (Hb AC)** is more common in people with West African, Mediterranean, and Middle Eastern ancestry.
- **Hb D trait (Hb AD)** is more common in people with Asian Indian, Mediterranean, Caribbean, and Middle Eastern ancestry.
- **Alpha thalassemia trait ( $\alpha\alpha/--$ )** is more common in people with Chinese, Southeast Asian, Mediterranean, Middle Eastern, Asian Indian, and African ancestry.

People with these ancestries are more likely to be trait carriers, but it is possible for any person to carry a hemoglobin trait. The chance could be higher if there is any history of blood conditions in the family. Let your provider know if someone in the family has a blood condition or a hemoglobin trait.

## Newborn Screening for Inherited Blood Conditions



In California, all babies are screened for inherited blood conditions in the first few days after birth. Newborn screening helps identify most types of beta thalassemia disease. If your baby has one of these conditions, a screening coordinator will contact you with the results. Newborn screening does not identify babies with beta thalassemia trait.

### Limits of newborn screening:

- Some forms of beta-thalassemia disease are not detected.
- Some babies with other inherited blood conditions may not be identified (this is rare).