

ALPHA THALASSEMIA TRAIT

Positive Carrier Screening Result

You have recently learned that you have alpha thalassemia trait or another type of alpha thalassemia.

Everyone has genes that make hemoglobin. Hemoglobin is found in red blood cells and moves oxygen through the body. A person with alpha thalassemia trait makes less than the usual amount of hemoglobin.

The gene responsible for alpha thalassemia is called the alpha-globin gene. Most people have 4 copies of this gene (2 from each parent). However, some people have less than 4 copies. This can affect the type and amount of hemoglobin made.

Alpha thalassemia trait does not affect your health.

A person with trait has 2 working copies of the alpha-globin gene instead of 4 copies. This is not a disease because the body still makes enough hemoglobin. Usually there are no signs of being a trait carrier, except small red blood cells. A person with this trait may also have mild anemia, but this does not require any treatment. There is a 1 in 2 (50%) chance of passing this trait to any of your children.

Alpha thalassemia major (ATM or “hydrops fetalis”) happens when a developing baby has no working alpha genes. **Both** parents must carry alpha thalassemia trait to be at-risk for a baby with ATM. This condition causes fetal death if untreated. ATM may also cause serious pregnancy problems and could lead to maternal death.

If you are pregnant, the next step is to test your partner.

If your partner has a normal blood test result, the chance that a baby will have ATM is very small. Your partner can be tested at any Kaiser Permanente laboratory at no cost. If you are not pregnant, your partner can arrange testing through their own health care provider. A genetic counselor will let you know how to arrange testing.

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 an alpha thalassemia disease. There is also a 3 in 4 (75%) chance that a baby will not have an alpha thalassemia disease. If you are pregnant, you will be offered prenatal testing to find out if your baby could have an alpha thalassemia disease. A procedure, such as amniocentesis or CVS, is able to diagnose alpha thalassemia diseases during pregnancy. Prenatal testing is very helpful in determining pregnancy care and delivery plans. Some parents may use the results to consider whether to continue or end the pregnancy.

Common types of alpha thalassemia:

Silent carrier– Silent carriers have 3 working alpha-globin genes. They have normal or slightly small red blood cells. No treatment is needed. Routine blood tests do not detect silent carriers. Silent carriers can only be confirmed by genetic testing.

Hb Constant Spring (Hb CS) trait - One alpha-globin gene makes a different type of hemoglobin (Hb CS) which does not work as well as regular hemoglobin. Carriers may have slight red blood cell changes or none at all. No treatment is needed. Routine blood tests do not detect Hb CS. This trait can only be confirmed by genetic testing.

Alpha thalassemia trait – People with this trait have 2 working alpha-globin genes. Routine blood tests show small red blood cells and may also show mild anemia. No treatment is needed. Genetic testing is needed to confirm alpha thalassemia trait.

Triplicated alpha genes- People with this trait have more than 4 alpha-globin genes. Extra alpha genes are not found by routine screening. Genetic testing is needed to confirm the presence of extra alpha genes. No treatment is needed. Extra copies of alpha globin genes in a person with beta-thalassemia trait can cause severe anemia, which could require transfusions.

Homozygous Hb Constant Spring People with this trait have 2 normal working alpha-globin genes and two alpha genes producing Hb Constant Spring. This causes mild anemia and may lead to an enlarged spleen or liver. Treatment may be needed. A fetus with this condition should be monitored for signs of severe anemia.

Hb H disease - People with this condition have 1 working alpha-globin gene. This causes mild to moderate anemia. Treatment may be needed and certain medications avoided. Genetic testing is needed to confirm Hb H disease.

Hb H-Constant Spring Disease - People with this condition have 1 working alpha-globin gene. This condition happens when a person has both alpha thalassemia trait and Hb CS. Moderate to severe anemia is present. Treatment may be needed and certain medications avoided. A fetus with this condition should be monitored for signs of severe anemia.

Alpha Thalassemia Major (ATM) - Fetuses with ATM have no working alpha-globin genes. Affected fetuses develop problems during pregnancy that can usually be seen by ultrasound. Genetic testing can confirm this condition. If left untreated, ATM leads to fetal or newborn death, and may cause severe maternal problems or death.

Carrier screening detects most trait carriers.

Anyone can carry a hemoglobin trait, even if there is no history of blood conditions in the family. The chance for your partner to be a trait carrier depends on their ancestry or ethnic background.

Testing Your Partner for Hemoglobin Traits

Why test your partner?

- You want to know the chance your baby could have an **alpha thalassemia disease**.
- You want to be prepared if your baby is at-risk for **alpha thalassemia major**. This may include maternal and fetal intervention.
- You want to be prepared if your baby is at-risk for an **alpha thalassemia disease** which involves severe fetal anemia. This may include the need for fetal intervention.
- You might consider testing the baby for alpha thalassemia during pregnancy.

Why would you not test your partner?

- You do not want to learn about a risk for alpha thalassemia major during pregnancy.
- The result would not change anything you do during pregnancy. Pregnancy monitoring and obstetrical intervention may still be needed.

Newborn screening can look for inherited blood conditions.



All babies are screened for inherited blood conditions in the first few days after birth. ATM is usually diagnosed during pregnancy. Newborn screening can identify Hb H disease and Hb H-CS disease. If your baby has either of these conditions, a screening coordinator will contact you with the results. Newborn screening does not identify babies with alpha thalassemia trait, Hb-CS trait, or silent carriers.

Limits of newborn screening:

- Some babies with other inherited blood conditions may not be identified (this is rare).

Alpha thalassemia trait 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.

More about Alpha Thalassemia Diseases

Alpha thalassemia major (ATM) happens when a baby inherits alpha thalassemia trait from both parents. The baby does not make any normal hemoglobin and usually dies before birth due to heart failure and fluid build-up (hydrops). A pregnant person can also develop serious health problems that need obstetrical intervention to prevent maternal and fetal death. Fetal treatment can be done to help babies survive to birth; however, if they survive, they will need lifelong blood transfusions and medical care.

Hb H disease (Hb H disease) People who have this condition inherited alpha thalassemia trait from one parent and a single alpha gene deletion from the other parent. This condition has variable severity, with anemia that ranges from mild to moderate anemia. Common symptoms may include gallstones and a large spleen. Symptoms are treated with occasional blood transfusions. Oxidative medications should be avoided.

Hb H-Constant Spring disease (Hb H-CS disease) People who have this condition inherited alpha thalassemia trait from one parent and Hb Constant Spring (CS) from the other parent. Medical problems include moderate to severe anemia, growth problems, gallstones, and a large spleen. Medical problems are most often treated by blood transfusions. Oxidative medications should be avoided. Fetuses with this condition should be monitored for the risk of hydrops.