KAISER PERMANENTE®

INFORMATION SHEET

SICKLE CELL TRAIT

Positive Carrier Screening Result

You have recently learned that you have sickle cell trait. Here is important information about being a trait carrier.

Everyone has two copies of a gene that makes hemoglobin. Hemoglobin is found in red blood cells and moves oxygen through the body. A person with sickle cell trait (Hb AS) has one copy of the gene that makes the usual hemoglobin (Hb A), and one copy that makes hemoglobin S (Hb S). Hb S can cause sickle cell disease when combined with a hemoglobin other than Hb A.

Sickle cell trait does not usually affect your health. Trait carriers will not develop sickle cell disease and usually have no signs of being a carrier. Serious health problems in trait carriers are rare, but can happen under extreme conditions, such as severe dehydration or high-intensity physical activity. 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 sickle cell disease. If your partner has a normal blood test result (Hb AA), the chance that a baby will have sickle cell disease is very small. 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?

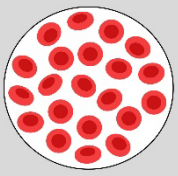
- You want to know the chance that your baby could have sickle cell disease.
- You want to be prepared if your baby is found to be at-risk for sickle cell disease.
- You might consider testing the baby for sickle cell disease during pregnancy.

Why would you not test your partner?

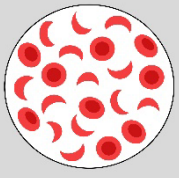
- You do not want to learn about a risk for sickle cell disease during pregnancy.
- The result would not change anything you do during pregnancy.

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 sickle cell disease. There is also a 3 in 4 (75%) chance that a baby will not have sickle cell disease. If you are pregnant, you will be offered prenatal testing to find out if your baby has sickle cell disease. A procedure, such as amniocentesis or CVS, is able to diagnose sickle cell disease 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. You could also wait until after birth to test your baby through the newborn screening program.



Normal blood



Sickle cell anemia

What is sickle cell disease?

Sickle cell disease refers to a group of inherited blood conditions that have high levels of Hb S inside the red blood cells. This causes the usually round red blood cells to collapse into a sickle shape. Sickled red blood cells get stuck in blood vessels causing pain and damage to any area where this occurs. This can happen in any part of the body, including bones, spleen, liver, heart, lungs, kidney, brain, and muscles. Sickled cells also break down more quickly, which leads to anemia.

Signs of sickle cell disease can start in the first year of life. Symptoms are different for each person, but often include repeat infections, pain, and fatigue.

Research is being done to find ways to keep people with sickle cell disease as healthy as possible. Early diagnosis and treatment can help. Some treatments can be started in infancy. Treatments may include medications, frequent doctor visits, blood transfusions, and overnight hospital stays.

Types of sickle cell disease:

Hb SS disease – People with this condition inherit Hb S from both parents. This is usually the most severe form of sickle cell disease.

Hb SC disease- People with this condition inherit Hb S from one parent and Hb C from the other parent. Severity can vary.

Hb S-Beta Thalassemia disease- People with this condition inherit Hb S from one parent and a beta thalassemia variant from the other parent. Severity can vary depending on the type of beta thalassemia variant. Further blood tests are needed to determine the type of beta thalassemia variant.

Hb SD disease, Hb SE disease, or Hb S-HPFH disease - People with this condition inherit Hb S from one parent and a different hemoglobin (Hb D, Hb E, or HPFH) from the other parent. This type of sickle cell disease is less common. Severity can vary.

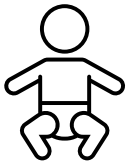
Testing Your Partner for Hemoglobin Traits

Anyone can carry a hemoglobin trait, even if there is no history of sickle cell disease or other blood conditions in the family. The chance for your partner to be a trait carrier depends on their ancestry or ethnic background. Carrier screening detects most hemoglobin trait carriers.

- **Sickle cell trait** (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.
- **Beta thalassemia trait** (Hb Aβ) is more common in people with Chinese, Southeast Asian, Mediterranean, Middle Eastern, and Asian Indian 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 sickle cell trait, sickle cell disease, or another blood condition or hemoglobin trait.

Newborn Screening for Sickle Cell Disease



In California, all babies are screened for sickle cell disease in the first few days after birth. Newborn screening helps identify sickle cell disease as early as possible. If your baby has sickle cell disease, a screening coordinator will contact you with the results. Newborn screening will also identify babies with sickle cell trait.

Limits of newborn screening:

- Some babies with sickle cell disease may not be identified (this is rare).