Sickle cell disease is an inherited blood disorder. It is most often found in people with African heritage, but it can also be found in people with ancestry from other parts 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. Sickle cell happens when there is a difference in how the beta globin chains are made. 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. Sickle cell disease happens when both copies of the beta globin genes are not working in the usual way.

**Hemoglobin S**

In sickle cell disease, the body makes hemoglobin S instead of the most common kind of hemoglobin, hemoglobin A. Although hemoglobin S is able to carry oxygen around in the blood, a slight chemical difference makes it more likely to collapse into a sickle (banana-like) shape instead of the usual round (donut-like) shape. This makes the red blood cells more rigid and sticky.

**Sickle Cell Trait**

A person with one normal copy of the beta globin gene and one copy making hemoglobin S has sickle cell trait. A person with sickle cell trait makes a small amount of hemoglobin S, but also makes plenty of hemoglobin A. About 10% of the African-American population has sickle cell trait.

A person with sickle cell trait will never develop sickle cell disease and usually has no medical problems from the trait. Very rarely, if you have sickle cell trait, your blood cells can sickle (change shape) when your body is not getting enough oxygen, such as during travel to high altitudes or when doing very strenuous exercise.

The importance of identifying sickle cell trait is that it helps find couples whose children may be born with sickle cell disease.

**Sickle Cell Disease**

Sickle cell disease is a lifelong condition that can include serious health problems, but it affects each person differently. When the blood cells become sickle-shaped, they can get stuck in the blood vessels and create blockages. This leads to pain in the area of the blockage and may cause damage to that area. Bones are very often affected, but it can happen in any part of the body, like the spleen, liver, heart, lungs, kidney, brain, and muscles. Sickled cells also get broken down more quickly by the body, which causes chronic anemia and fatigue.
Sickle Cell Trait and Sickle Cell Disease

A person with sickle cell disease needs regular medical visits to get routine care and watch for problems related to sickle cell disease. Medical problems can start very early, so, knowing that a baby has sickle cell disease allows early medical treatment.

Sickle cell disease has been cured using bone marrow transplantation, but the procedure has serious risks and requires a suitable donor.

**Types of sickle cell disease**

Most of the time sickle cell disease happens when both of the beta globin genes are making hemoglobin S, instead of hemoglobin A. This is called hemoglobin SS disease or sickle cell anemia. But there are changes in the beta globin gene that lead to other differences in hemoglobin, such as hemoglobin C, hemoglobin D, hemoglobin E, and beta thalassemia. When one of these other hemoglobin traits combines with hemoglobin S, the result is a form of sickle cell disease. The less common types of sickle cell disease are hemoglobin SC disease, hemoglobin SD or SE disease, and hemoglobin S-beta thalassemia disease. Some types of sickle cell disease have more medical problems than others.

**Inheritance and prenatal diagnosis**

If you have sickle cell trait, it is possible to pass it to your children. However, the chance for sickle cell 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 any form of sickle cell disease.

When only one parent has sickle cell trait and the other has normal hemoglobin, there is **no chance** of having a baby with any form sickle cell disease. However, each child has a 50% (or 1 in 2) chance to inherit sickle cell trait from the parent.

Sickle cell disease can only happen when both parents have sickle cell trait (or a related blood trait). When both parents have sickle cell trait, there is a 25% (or 1 in 4) chance in each pregnancy for the baby to have sickle cell disease and a 75% (or 3 in 4) chance that the baby will not have this disease.

**Testing for sickle cell disease**

It is possible to test the developing baby for sickle cell disease as early as the tenth week of pregnancy. If testing shows the baby has sickle cell 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 sickle cell disease through the Newborn Screening Program. This testing can also identify babies with sickle cell trait.
KEY TO SYMBOLS

**AA**  Hemoglobin A  
- Two working beta globin genes

**AS**  Sickle cell trait  
- One working beta globin gene  
- One sickle cell gene

**AB**  Other hemoglobin trait  
- One working beta globin gene  
- One non-working beta globin gene (beta thalassemia, hemoglobin C, hemoglobin D, etc)

**SS**  Sickle cell Anemia  
- Two sickle cell genes

**SB**  Other sickle cell disease (SC disease, sickle-beta thalassemia, etc)  
- One sickle cell gene  
- One non-working beta globin gene (beta thalassemia, hemoglobin C, hemoglobin D, etc)

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**One parent with sickle cell trait**

**Both parents with sickle cell trait**

**Each pregnancy has a 25% chance to have sickle cell anemia**

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

**One parent with sickle cell trait and One parent with another hemoglobin trait**

**Each pregnancy has a 25% chance to have sickle cell disease**