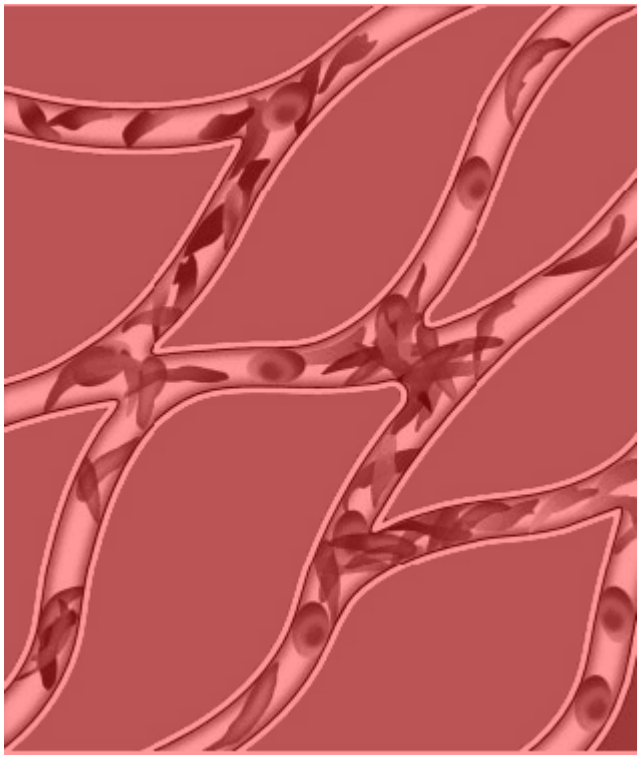
What Is Sickle Cell Anemia?

Sickle cell anemia is a serious disease in which the body makes abnormally shaped red blood cells. Normal red blood cells are smooth and round like a doughnut without a hole. They move easily through blood vessels to carry oxygen to all parts of the body. In sickle cell anemia, the body produces red blood cells that are shaped like a sickle (or crescent). These "sickle cells" are hard and sticky and they don't move easily through blood vessels. They tend to get stuck and block the flow of blood to the limbs and organs. This can cause pain, organ damage, and a low blood count (anemia).

Sickle cell anemia is an inherited (genetic) disorder. People who have sickle cell anemia are born with it. It is a lifelong disease.



The sickle-shaped red blood cells tend to get stuck in blood vessels, blocking the flow of blood.

Sickle cell anemia affects millions of people. Effective treatments exist for the symptoms and complications of the disease, but in most cases there is no cure. (Some researchers believe that bone marrow transplantation may offer a cure in a small percentage of cases.) Over the past 30 years, doctors have learned a great deal about the disease. They know what causes it, how it affects your body, and how to treat many of the complications. Today, with good health care, many people with the disease:

- Are in reasonably good health much of the time
- Live fairly normal lives
- Live 40 to 50 years or more

Anemia

Anemia is the term for having a shortage of red blood cells in your blood. In sickle cell anemia, this shortage of red blood cells occurs because sickle cells do not last very long. Red blood cells are produced in the spongy marrow inside the large bones of the body. The bone marrow constantly makes new red blood cells to replace old ones. Normal red blood cells last about 120 days in the bloodstream and then die. Sickle cells die much faster, usually after only about 10 to 20 days. The bone marrow can't make new red blood cells fast enough to replace the dying ones, so anemia (low level of red blood cells) results.

Sickle Cell *Trait* Versus *Anemia*

The condition called *sickle cell trait* is different from *sickle cell anemia*. A person with sickle cell trait does not have the disease but carries the gene that causes the disease. People with sickle cell trait can pass the gene on when they have children. For more information on sickle cell trait, see the section on What Causes Sickle Cell Anemia.

What Causes Sickle Cell Anemia?

Sickle Cell Anemia

Sickle cell anemia is a genetic disease. People with sickle cell anemia inherit two copies of the sickle cell gene, one from each parent.

The sickle cell gene tells the body to make a variant (different than normal) form of a protein called hemoglobin (HEE-muh-glow-bin). Hemoglobin is the protein inside red blood cells that carries oxygen to all parts of the body and gives blood its red color. The variant hemoglobin is what causes the red blood cells to become hard and curved like a sickle. It takes two copies of the sickle cell gene for the body to make the variant hemoglobin.

Sickle Cell Trait

Children who inherit only one copy of the sickle cell gene (from one parent) will not have sickle cell *anemia*. They will have sickle cell *trait*.

People with sickle cell trait:

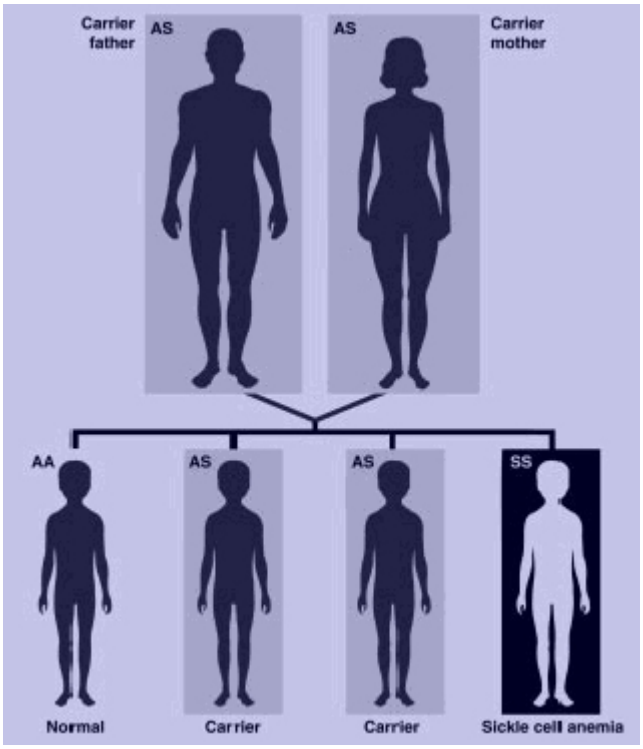
- Generally have no symptoms
- Live normal lives
- Can pass the sickle cell gene on to their children

When two people with sickle cell trait have a baby, there is a:

- One in four chance (25 percent) the baby will inherit two sickle cell genes and have the disease.
- One in four chance (25 percent) the baby will inherit two normal genes and not have the disease or trait.

- Two in four chance (50 percent) the baby will inherit one normal gene and one sickle cell gene. The baby will not have the disease but will have sickle cell trait like the parents.

Inheriting Sickle Cell Genes



The presence of two sickle cell genes (SS) is needed for sickle cell anemia. If each parent carries one sickle hemoglobin gene (S) and one normal hemoglobin gene (A), with each pregnancy, there is a 25 percent chance of the child's inheriting two SS genes and having sickle cell anemia; a 25 percent chance of inheriting two AA genes and not having the disease or being a carrier; and a 50 percent chance of being an unaffected carrier (AS) like the parents.

How Is Sickle Cell Anemia Treated?

Effective treatments exist for the symptoms and complications of sickle cell anemia, but in most cases there is no cure. Some researchers believe that bone marrow transplantation may offer a cure in a small percentage of cases. Researchers are working on developing new treatments for sickle cell anemia, including gene therapy and more safe and effective bone marrow transplants.

People with sickle cell anemia need regular medical care. Some doctors and clinics specialize in treating people with the disease.

The goals of treating sickle cell anemia are to relieve pain, prevent infections, and control complications if they occur. The treatments include:

- Medicines
- Blood transfusions
- Specific treatment for complications

Treating Pain

Painful crises are the leading cause of emergency room visits and hospitalizations of people with sickle cell anemia. The usual treatments for acute pain crises are pain-killing medicines and fluids, either by mouth or through a vein, to prevent dehydration. The pain-killing medicines most often used are:

- Acetaminophen
- Nonsteroidal anti-inflammatory drugs (NSAIDs)
- Opioids, such as morphine, oxycodone, hydrocodone, and others

The treatment of patients with mild-to-moderate pain usually begins with NSAIDs or acetaminophen. If pain continues, an opioid may be added. Moderate-to-severe pain is treated with opioids. The opioid may be used alone or together with NSAIDs or acetaminophen.

In adult patients with severe sickle cell anemia, doctors may give a medicine called hydroxyurea to reduce the frequency of painful crises. This medicine is used only to prevent these crises, not to treat them when they occur. Given daily, it can reduce the frequency of painful crises and of acute chest syndrome. Patients taking the medicine may also need fewer blood transfusions. Patients taking hydroxyurea must be monitored carefully because the medicine can cause serious side effects, including an increased risk of dangerous infections. Because of the risks of the medicine, it is usually only used in adults with severe complications of sickle cell anemia. It is not approved for use in children.

Preventing Infections

Infection is a major complication of sickle cell anemia. In fact, pneumonia is the leading cause of death in children with the disease. Other infections common in people with the disease include meningitis, influenza, and hepatitis.

To prevent infections in babies and young children, treatments include:

- Daily doses of penicillin. Treatment may begin as early as 2 months of age and continue until the child is at least 5 years old.
- Vaccinations for pneumonia, meningitis, influenza, and hepatitis.

Children with sickle cell anemia should have a flu shot each year. If a child with sickle cell anemia shows early signs of an infection, such as fever, it is very important to get treatment right away.

Adults with sickle cell anemia should also have a flu shot every year. In addition, they should be vaccinated for pneumonia.

Preventing Eye Damage

Children may develop damage to the blood vessels in the back of their eyes. Parents should ask the child's doctor about regular checkups with an eye doctor who specializes in diseases of the retina. The retina is a thin layer of tissue inside the back of the eye.

Blood Transfusions

Blood transfusions are used to treat worsening anemia and sickle cell complications. A sudden worsening of anemia resulting from infection or enlargement of the spleen is a common reason for a transfusion. Some, but not all, patients need transfusions to prevent life-threatening events such as stroke or pneumonia.

Treating Complications

Acute chest syndrome is a frequent cause of death in children and adults with sickle cell anemia. Treatment usually requires hospitalization and may include oxygen therapy, transfusions, antibiotics, and pain medicine.

Hand-foot syndrome is an early complication seen in sickle cell anemia. The syndrome may start at less than 1 year of age. Treatment includes the use of pain medicine and fluids.

Leg ulcers may be treated with cleansing solutions and medicated creams or ointments. Skin grafts may be needed if the condition continues. Leg ulcers can be painful, and patients may be given strong pain medicine. Bed rest and keeping the leg raised to reduce swelling are useful, although not always possible.

Regular Health Care for Children

Children with sickle cell anemia should get regular health care, just like children without the disease. They need to have their growth checked and to get the usual shots that all children receive.

Before age 2, children with sickle cell anemia should see the doctor every 2 or 3 months. After age 2, children should see the doctor at least every 6 months. These visits are good chances for parents to talk with their child's health care provider and ask questions about the child's care.

Penicillin is generally given to all children with sickle cell anemia until age 5. Many patients are prescribed a vitamin called folic acid (folate) to help prevent some of the complications of sickle cell anemia.

New Treatments

Today, research on sickle cell anemia is looking at new medicines, bone marrow transplants, and gene therapy. The hope is that these studies can provide new treatments and find a possible cure for sickle cell anemia. Researchers are also looking for a way to predict the severity of the disease.

Bone marrow transplant can be a very effective treatment for sickle cell anemia, but, because of its risks, only some patients can or should have this procedure. The bone marrow transplant procedure carries the risk of serious complications and even death. It is usually reserved for younger patients with severe disease, but the decisions are made on a case-by-case basis. It requires a donation of bone marrow from a closely matched donor, usually a close family member, who does not have the disease.