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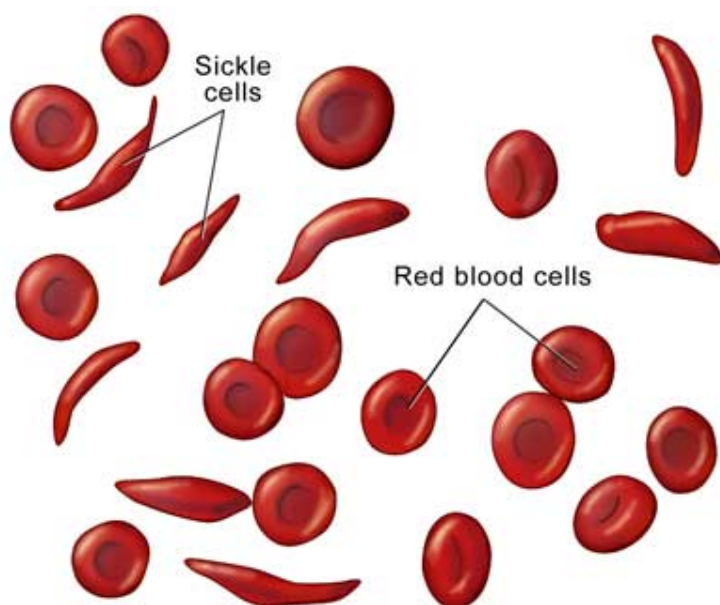
Sickle Cell Anemia

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Introduction

Sickle cell anemia is an inherited form of anemia — a condition in which there aren't enough healthy red blood cells to carry oxygen throughout your body.

Under normal circumstances, your red blood cells are flexible and round, and they move easily through your blood vessels to carry oxygen to all parts of your body. In people with sickle cell anemia, the red blood cells become rigid and sticky and are shaped like sickles or crescent moons.



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Normal red blood cells are round. In sickle cell anemia, some red blood cells become deformed, so they look like a sickle used to cut wheat. These unusually shaped cells give the disease its name.

These irregular-shaped blood cells die prematurely, resulting in a chronic shortage of red blood cells. Plus, they can get stuck when traveling through small blood vessels, which can slow or block blood flow and oxygen to certain parts of the body. This produces pain and can lead to the serious complications of sickle cell anemia.

There's no cure for most people with sickle cell anemia. However, treatments can relieve pain and prevent further problems associated with sickle cell anemia.

Signs and symptoms

People with sickle cell trait have one gene for the disease. They don't develop the disease and usually have no signs and symptoms. Approximately one in 12 black Americans has sickle cell trait.

People with sickle cell anemia have two genes for the disease — one from each parent. They usually show some signs and symptoms after 4 months of age. Some people with sickle cell anemia have few symptoms. For others, the disease is more severe and they may require repeated hospitalizations.

Signs and symptoms of sickle cell anemia include:

- **Anemia.** Sickle cells are fragile. They break apart easily and die, leaving you chronically short on red blood cells to carry oxygen to your tissues — a condition known as anemia. Without enough red blood cells in circulation, your body can't get the oxygen it needs to feel energized. That's why anemia causes fatigue.
- **Episodes of pain.** Periodic episodes of pain, called crises, are a major symptom of sickle cell anemia. Pain develops when sickle-shaped red blood cells block blood flow through tiny blood vessels to your chest, abdomen and joints. Pain can also occur in your bones. The pain may vary in intensity and can last for a few hours to a few weeks. Some people experience only a few episodes of pain. Others experience a dozen or more crises a year. If a crisis is severe enough, you may need hospitalization so that painkillers can be injected into your veins (intravenously).
- **Hand-foot syndrome.** Swollen hands and feet are often the first signs of sickle cell anemia in babies. The swelling is caused by sickle-shaped red blood cells blocking blood flow out of the hands and feet.
- **Jaundice.** Jaundice is a yellowing of the skin and eyes that occurs because of liver damage or dysfunction. Occasionally, people who have sickle cell anemia have some degree of jaundice because the liver, which filters harmful substances from the blood, is overwhelmed by the rapid breakdown of red blood cells. In people with dark skin, jaundice is visible mostly as yellowing of the whites of their eyes.
- **Frequent infections.** Sickle cells can damage your spleen, an organ that fights infection. This may make you more vulnerable to infections. Doctors commonly give infants and children with sickle cell anemia antibiotics to prevent potentially life-threatening infections, such as pneumonia.
- **Stunted growth.** Red blood cells provide your body with the oxygen and nutrients you need for growth. A shortage of healthy red blood cells can slow growth in infants and children and delay puberty in teenagers.
- **Vision problems.** Some people with sickle cell anemia experience vision problems. Tiny blood vessels that supply your eyes may become plugged with sickle cells. This can damage the retina — the portion of each eye that processes visual images.

Causes

Sickle cell anemia is caused by a mistake in the gene that tells your body to make hemoglobin — the red, iron-rich compound that gives blood its red color. Hemoglobin is a component of every red blood cell in your body. It allows red blood cells to carry oxygen from your lungs to all parts of your body, and to carry carbon dioxide waste from other parts of your body to your lungs so that it can be exhaled.

Under normal circumstances, your body makes healthy hemoglobin known as hemoglobin A. People with sickle cell anemia make hemoglobin S — the S stands for sickle.

The sickle cell gene is passed from generation to generation in a pattern of inheritance called autosomal recessive inheritance. This means that both the mother and the father must pass on the defective form of the gene for a child to be affected. Most often, sickle cell disease is passed down the family tree by parents who have sickle cell trait.

People with sickle cell trait have one normal hemoglobin gene and one defective form of the gene. So their bodies make both normal hemoglobin and sickle cell hemoglobin. Their blood may contain some sickle cells, but they usually don't experience symptoms unless they're in an area with low oxygen — such as at high altitudes on an airplane or on a mountain. However, they are carriers of the disease, which means they can pass the defective gene on to their children.

Two carriers have a 25 percent chance of having an unaffected child with normal hemoglobin, a 50 percent chance of having a child who also is a carrier, and a 25 percent chance of having a child with sickle cell anemia. These chances are the same in each pregnancy.

Evolution of a defective gene

Researchers believe the defective hemoglobin gene that causes sickle cell anemia evolved many years ago, among people living in parts of Africa, the Mediterranean, the Middle East and India. At that time, malaria epidemics killed many people in those regions.

But some people in those regions had a genetic mutation that caused some of their red blood cells to change shape — a condition now known as sickle cell trait. The sickle cells actually interfered with the growth of the parasite that causes malaria. So people with sickle cell trait often survived malaria outbreaks.

Over time, these survivors migrated and continued on with their lives. In some cases, two people with the sickle cell trait had children. And some of their children inherited two copies of the mutated gene, which results in sickle cell anemia. Today, millions of people all over the world have sickle cell anemia.

How defective hemoglobin causes anemia

Red blood cells with normal hemoglobin are smooth and round and glide through blood vessels. Red blood cells with defective hemoglobin may become hard, sticky and shaped like a sickle used to cut wheat. These crescent-shaped cells can get stuck in small blood vessels, blocking blood flow and causing episodes of pain and damage to organs.

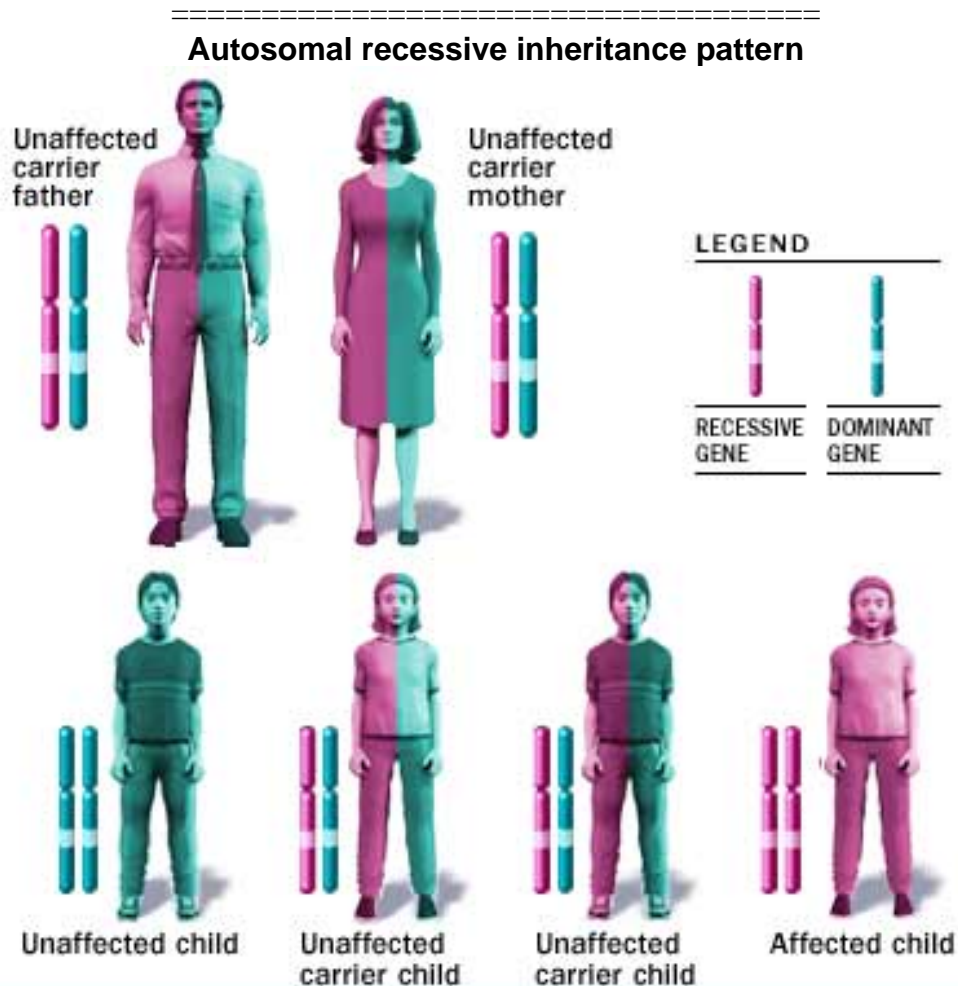
Your bone marrow — the red, spongy material found within the cavities of many of your large bones — regularly produces red blood cells. Bone marrow also produces white blood cells to fight infections and platelets to help blood clot. These two types of blood cells aren't directly involved in sickle cell anemia.

Once red blood cells leave your bone marrow, they normally live for about 120 days before they die and need to be replaced. However, sickle cells die after only 10 to 20 days. So, it's difficult for your body to produce enough replacements. The result is a chronic shortage of red blood cells, known as anemia.

Risk factors

The risk of inheriting sickle cell anemia really comes down to genetics. For a baby to be born with sickle cell anemia, both parents have to carry the sickle cell gene.

The gene is particularly common among people with African, Spanish, Mediterranean, Middle Eastern and Indian ancestry. In the United States, it most commonly affects blacks and Hispanics.



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To have an autosomal recessive disorder, you inherit two mutated genes, one from each parent. These disorders are usually passed on by two carriers. Their health is rarely affected, but they have one mutated gene (recessive gene) and one normal gene (dominant gene) for the condition. Two carriers have a 25 percent chance of having an unaffected child with two normal genes (left), a 50 percent chance of having an unaffected child who also is a carrier (middle), and a 25 percent chance of having an affected child with two recessive genes (right).

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Screening and diagnosis

A blood test can check for hemoglobin S — the defective form of hemoglobin that underlies sickle cell anemia. In most states in the United States, this blood test is part of routine newborn screening done at the hospital. But older children and adults can be tested too.

In adults, a blood sample is drawn from a vein in the arm. In young children and babies, the blood sample is usually collected from a finger or heel. The sample is then sent to a laboratory, where a technician can screen for hemoglobin S.

If the screening test is negative, there is no sickle cell gene present. If the screening test is positive, the technician will conduct further tests to determine whether one or two sickle cell genes are present. People who have one gene — sickle cell trait — have a fairly small percentage of hemoglobin S. People with two genes — sickle cell disease — have a much larger percentage of the defective hemoglobin.

Additional steps

To confirm any diagnosis, a sample of blood is examined under a microscope to check for large numbers of sickle cells — a marker of the disease. If you or your child has the disease, a blood test to check for anemia — a low red blood cell count — will be done.

If you or your child has sickle cell anemia, you'll likely be referred to a doctor who specializes in blood-related diseases (hematologist). And your doctor may suggest additional tests to check for possible complications of the disease. If you or your child carries the sickle cell gene, you may be referred to a genetic counselor — an expert in genetic diseases.

It's possible to detect sickle cell anemia in an unborn baby by sampling some of the fluid surrounding the baby in the mother's womb (amniotic fluid). A test can determine whether an unborn baby has sickle cell anemia or carries the sickle cell gene (sickle cell trait).

Complications

Sickle cell anemia can lead to a host of complications, including:

- **Stroke.** A stroke can occur if sickle cells block blood flow to an area of your brain. Stroke is one of the most serious complications of the disease. Signs of stroke include seizures, weakness or numbness of your arms and legs, sudden speech difficulties, and loss of consciousness. If your baby or child has any of these signs and symptoms, seek medical treatment immediately. A stroke can be fatal.
- **Acute chest syndrome.** This life-threatening complication of sickle cell anemia causes chest pain, fever and difficulty breathing. Acute chest syndrome is similar to pneumonia, but is caused by a lung infection or trapped sickle cells in the blood vessels of your lungs. It requires emergency medical treatment with antibiotics, blood transfusions and drugs that open up airways in your lungs. Recurrent attacks can damage your lungs.
- **Organ damage.** Sickle cells can block blood flow through blood vessels, immediately depriving an organ of blood and oxygen. In sickle cell anemia, blood is also chronically

low on oxygen. Chronic deprivation of oxygen-rich blood can damage nerves and organs in your body, including your kidneys, liver and spleen. Organ damage can be fatal.

- **Blindness.** Tiny blood vessels that supply your eyes can get plugged with sickle cells. Over time, this can damage the retina — the portion of each eye that processes visual images — and lead to blindness.
- **Other complications.** Sickle cell anemia can cause open sores, called **ulcers**, on your legs. Sickle cells can block blood vessels that nourish your skin, causing skin cells to die. Once skin is damaged, sores can develop.

Gallstones also are a possible complication. The breakdown of red blood cells produces a substance called bilirubin. Bilirubin is responsible for yellowing of the skin and eyes (jaundice) in people with sickle cell anemia. A high level of bilirubin in your body can also lead to gallstones.

Men with sickle cell anemia may experience painful erections, a condition called **priapism**. Sickle cells can prevent blood flow out of an erect penis. Over time, priapism can damage the penis and lead to **impotence** in men with sickle cell anemia.

Treatment

Bone marrow transplant offers the only potential cure for sickle cell anemia. But very few people have a suitable donor for transplant.

As a result, treatment for sickle cell anemia is usually aimed at avoiding crises, relieving symptoms and preventing complications. If you have sickle cell anemia, you'll need to make regular visits to your doctor to check your red blood cell count and monitor your health. You may also require treatment from specialists at a hospital or sickle cell anemia clinic. Treatments may include medications to reduce pain and prevent complications, blood transfusions and supplemental oxygen, as well as bone marrow transplant.

Medications

Medications used to treat sickle cell anemia include:

- **Antibiotics.** Children with sickle cell anemia usually begin taking the antibiotic penicillin when they're about 2 months of age and continue until they're 5 years old. Doing so helps prevent infections, such as pneumonia, which can be life-threatening to an infant or child with sickle cell anemia. Antibiotics may also help adults with sickle cell anemia fight certain infections.
- **Pain-relieving medications.** To relieve pain during a sickle crisis, your doctor may advise over-the-counter pain relievers and application of heat to the affected area. You may also need stronger prescription painkillers.
- **Hydroxyurea (Droxia, Hydrea).** This prescription drug, normally used to treat cancer, may be helpful for adults with severe disease. When taken daily, it reduces the frequency of painful crises and may reduce the need for blood transfusions. It seems to work by stimulating production of fetal hemoglobin — a type of hemoglobin found in newborns that helps prevent the formation of sickle cells. There is some concern about the

possibility that long-term use of this drug may cause tumors or leukemia in certain people. Your doctor can help you determine if this drug may be beneficial for you.

Blood transfusions

In a red blood cell transfusion, red blood cells are removed from a supply of donated blood. These donated cells are then given intravenously to a person with sickle cell anemia.

Blood transfusions increase the number of normal red blood cells in circulation, helping to relieve anemia. In children with sickle cell anemia at high risk of stroke, regular blood transfusions can decrease their risk of stroke.

Blood transfusions carry some risk. Blood contains iron. Regular blood transfusions cause an excess amount of iron to build up in your body. Because excess iron can damage your heart, liver and other organs, people who undergo regular transfusions must often receive treatment to reduce iron levels. In 2005, the Food and Drug Administration approved deferasirox (Exjade), the first oral medication that can reduce excess iron levels, for use in people older than the age of 2.

Supplemental oxygen

Breathing supplemental oxygen through a breathing mask adds oxygen to your blood and helps you breathe easier. It may be helpful if you have acute chest syndrome or a sickle cell crisis.

Bone marrow transplant

This procedure allows people with sickle cell anemia to replace their bone marrow — and its sickle-shaped red blood cells — with healthy bone marrow from a donor who doesn't have the disease. It can be a cure, but the procedure is risky, and it's difficult to find suitable donors. Researchers are still studying bone marrow transplants for people with sickle cell anemia. Currently, the procedure is recommended only for people who have significant symptoms and problems from sickle cell anemia.

In a bone marrow transplant, your bone marrow is first destroyed using chemotherapy or radiation. A matched donor's healthy bone marrow is removed using a minor surgical technique. You then receive a transfusion of that healthy marrow, and the healthy marrow may start producing normal blood cells. Sometimes, however, the transplant doesn't work or the recipient's body rejects the new marrow.

The procedure requires a lengthy hospital stay. After the transplant, you'll need drugs to help prevent rejection of the donated marrow.

Treating complications

Doctors treat most complications of sickle cell anemia as they occur. Treatment may include antibiotics, blood transfusions, pain-relieving medicines, other medications and possibly surgery, such as to correct vision problems or to remove a damaged spleen.

Experimental treatments

Scientists continue to gain new insights into the symptoms and causes of sickle cell anemia. Some possible new treatments researchers are studying include:

- **Gene therapy.** Because sickle cell anemia is caused by a defective gene, researchers are exploring whether correcting this gene and inserting it into the bone marrow of people with sickle cell anemia will result in the production of normal hemoglobin. Scientists are also exploring the possibility of turning off the defective gene while reactivating another gene responsible for the production of fetal hemoglobin — a type of hemoglobin found in newborns that prevents sickle cells from forming.
- **Butyric acid.** Some studies show that this commonly used food additive may increase the amount of fetal hemoglobin in the blood.
- **Clotrimazole.** Normally used to treat fungal infections, this over-the-counter medication helps prevent a loss of water from red blood cells, which may reduce the number of sickle cells that form.
- **Nitric oxide.** People with sickle cell anemia have low levels of nitric oxide, a gas that helps keep blood vessels open and reduces the stickiness of red blood cells. Treatment with nitric oxide may prevent sickle cells from forming.

Prevention

If you carry the sickle cell trait, you may wish to see a genetic counselor before trying to conceive a child. A genetic counselor can help you understand your risk of having a child with sickle cell anemia. He or she can also explain possible treatments, preventive measures and reproductive options.

There is an in vitro fertilization procedure that improves the chances for parents who both carry the sickle cell gene to have a child with normal hemoglobin. This procedure is known as preimplantation genetic diagnosis. First, eggs are taken from the mother. Then, sperm is taken from the father. In a laboratory, the eggs are fertilized with the sperm. The fertilized eggs are then tested for the presence of the sickle cell gene. Fertilized eggs free of the sickle cell gene can be implanted into the mother for normal development. However, this procedure is expensive and not always successful.

Self-care

Taking steps to stay healthy is critical for anyone with sickle cell anemia. Eating well, getting adequate rest and protecting yourself from infections are good ways to maintain your health and prevent crises.

Infants and children with sickle cell disease need to receive regular childhood vaccinations. Children and adults with sickle cell anemia also should have a yearly flu shot and be immunized against pneumonia.

If you or your child has sickle cell anemia, follow these suggestions to help stay healthy:

- **Take folic acid supplements daily, and eat a balanced diet.** Bone marrow needs folic acid and other vitamins to make new red blood cells.
- **Drink plenty of water.** Staying hydrated helps keep your blood diluted, which reduces the chance that sickle cells will form.

- **Avoid temperature extremes.** Exposure to extreme heat or cold can trigger the formation of sickle cells.
- **Avoid stress.** A sickle crisis can occur as a result of stress.
- **Exercise regularly, but don't overdo it.** Talk with your doctor about how much exercise is right for you.
- **Fly on commercial airplanes with pressurized passenger cabins.** Unpressurized aircraft cabins don't provide enough oxygen. Low oxygen levels can trigger a sickle crisis.
- **Avoid high-altitude areas.** Traveling to a high-altitude area may also trigger a crisis because of lower oxygen levels.

Coping skills

If you or someone in your family has sickle cell anemia, you may need help handling the stresses of coping with this lifelong disease. Your doctor can talk with you about your concerns. Sickle cell centers and clinics also can provide information and counseling. Many areas have sickle cell support groups for families affected by the disease.

It's especially important to find ways to control — and cope with — pain. Different techniques work for different people, but it might be worth trying heating pads, hot baths, massages or physical therapy. Prayer, family and friends also can be a source of support.

If you have a child with sickle cell anemia, the best way to help is to learn as much as you can about the disease and to make sure your child gets the best health care possible. A child with sickle cell disease has special needs and requires regular medical care to stay as healthy as possible. Your doctor can explain how often to bring your child for medical care and what you can do if he or she becomes ill.

You may also want to let teachers and caregivers know about your child's illness. Help them understand what kinds of exercises and situations can be harmful to your child, and teach them to recognize signs of infection. Having other people help care for your child is good for your child — and good for you.

[By Mayo Clinic Staff](#)

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