Health Basics

Sickle Cell Disease

What is sickle cell disease?
Sickle cell disease (also called sickle cell anemia) is an inherited condition in which a mutated gene causes production of abnormal hemoglobin, a protein in red blood cells that carries oxygen through the body.

Healthy red blood cells are round and flexible, but people with sickle cell disease have rigid cells shaped like crescent moons. This irregular shape causes the cells to clump together and get stuck in small blood vessels, impeding the flow of blood and reducing delivery of oxygen. What's more these abnormal cells have a shorter lifespan, which can leave patients with inadequate red blood cells, or anemia.

For most people, there has been no cure for sickle cell disease, though the condition can be managed with medications. Some patients benefit from regular blood transfusions. Stem cell transplants and new gene therapies approved in December 2023 can potentially cure the condition.

According to the Centers for Disease Control and Prevention, sickle cell affects approximately 100,000 Americans. In the United States, most people with sickle cell are Black. It occurs among about 1 out of every 365 African-American births and about 1 in 13 Black babies is born with sickle cell trait.

What are the symptoms of sickle cell disease?
Signs of sickle cell disease usually appear after an infant is 4 months old. Symptoms may include anemia (a low red blood cell count and inadequate hemoglobin), periodic episodes of pain (called crises), swollen hands and feet (called hand-foot syndrome), frequent infections, delayed growth and vision problems.

What causes sickle cell disease?
Sickle cell disease is caused by a gene mutation that instructs the body to make abnormal hemoglobin, which causes red blood cells to become rigid and misshapen.

The sickle cell gene is passed from generation to generation, but for a child to develop the disease, both biological parents must pass on the defective gene. If only one parent passes the sickle cell gene to the child, that child will be a carrier of the disease without developing the condition, but they can pass it on to their own children. The gene is more common in families from

Africa, India, the Mediterranean, parts of the Middle East and South and Central Americas.

What are the complications of sickle cell disease?

Sickle cell disease can lead to the following medical problems:

Stroke occurs when blood flow to the brain is blocked; strokes may characterized by weakness, paralysis or numbness (often on one half of the face or body); seizures, sudden speech difficulties or vision problems and loss of consciousness
Acute chest syndrome, a life-threatening condition that causes chest pain, fever and difficulty breathing
Pulmonary hypertension, high blood pressure in the lungs that causes difficulty breathing
Organ damage, caused by abnormal sickle cells blocking blood flow, which stops blood and oxygen from reaching an organ
Blindness, caused by damage to the retina, the part of the eye that processes visual images
Leg ulcers, open sores on the legs
Gallstones, accumulation of material in the gallbladder when the hemoglobin from damaged red blood cells forms bilirubin. This chemical can then combine with calcium and fat to form pebble-like gallstones.
Priapism, painful, long-lasting erections, which can occur when sickle cells block the blood vessels in the penis. This can damage the penis and eventually lead to impotence.

How is sickle cell disease diagnosed?

Genetic screening: This is done when a baby is born. Once the condition is diagnosed, parents are usually referred to a doctor who specializes in blood disorders (hematologist).
Blood testing: This screening checks for the presence of hemoglobin S, the abnormal form of hemoglobin that characterizes sickle-cell disease. In the United States, this test is part of routine newborn screening done at the hospital. But older children and adults can be tested, too. If the blood test is negative, the sickle-cell gene is not present. If the test is positive, doctors order further tests to determine whether one or two sickle-cell genes are present. People with one abnormal gene are diagnosed as having the sickle-cell trait, while those with two such genes are diagnosed with sickle-cell disease.
Amniotic fluid sampling: This test allows doctors to diagnose sickle cell disease in an unborn baby. The test involves taking a sample of the fluid surrounding the fetus in the womb to look for the sickle cell gene. Doctors recommend this test if a prospective parent has sickle cell disease or sickle-cell trait.

How is sickle cell disease treated?

Gene therapy: Two cell-based gene therapies for sickle cell disease were approved in December 2023. Casgevy (exagamglogene autotemcel) uses CRISPR-based therapy—it is, in fact, the first approved CRISPR therapy—while Lyfgenia (lovotibeglogene autotemcel) uses a viral vector to modify blood-producing stem cells. These gene therapies involve a procedure similar to a stem cell transplant but using the patient's own cells.
Stem cell transplant (bone marrow transplant): A stem cell transplant from a matched donor is a potential cure for sickle cell disease, but finding a compatible donor is difficult, and the procedure has serious risks.

- Blood transfusions: These procedures help increase the number of normal red blood cells in circulation and decrease the risk of stroke in children with sickle cell disease. But frequent blood transfusions can cause excess iron to build up in the body, and people who receive regular transfusions may need treatment to reduce iron levels to avoid damage to the heart, liver and other organs.
- Disease-modifying therapy: Hydroxyurea can increase hemoglobin levels, which lessens the frequency of sickle cell crises and helps prevent strokes. Newer disease-modifying therapies include Endari (L-glutamine), Adakveo (crizanlizumab) and Oxbryta (voxelotor).
- Symptomatic therapy: Various medications and procedures, including pain relievers, antibiotics and breathing exercises (incentive spirometry), may be used to lessen sickle cell symptoms and reduce complications.
- Lifestyle and behavior modifications: A healthy diet, regular exercise, adequate sleep and reducing stress can all help people with sickle cell disease avoid complications.

Last Reviewed: December 7, 2023

© 2026 Smart + Strong All Rights Reserved.

http://beta.docker.realhealthmag.com/basics/health-basics/sicklecell-anemia