

Sickle Cell Anemia

Definition

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

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.

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.

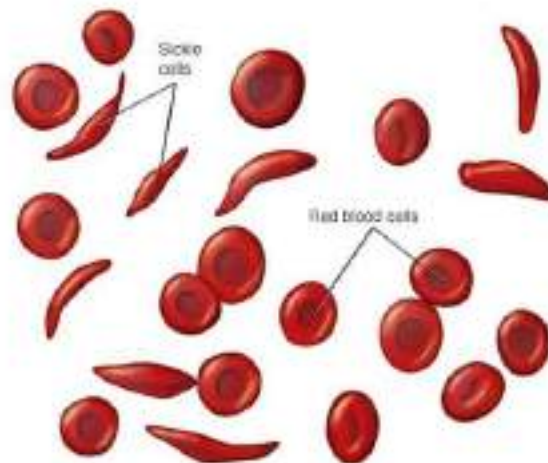
People with sickle cell anaemia have two genes for the disease — one from each parent. They usually show some signs and symptoms after 4 months of age. Some people with sickle cell anemia have few symptoms. For others, the disease is more severe and they may require repeated hospitalizations.

Signs and symptoms of sickle cell anaemia include:

- **Anaemia.** 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 anaemia. 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-toot syndrome.** Swollen hands and feet are often the first signs of sickle cell anaemia 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 anaemia 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.
- **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 anaemia 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.

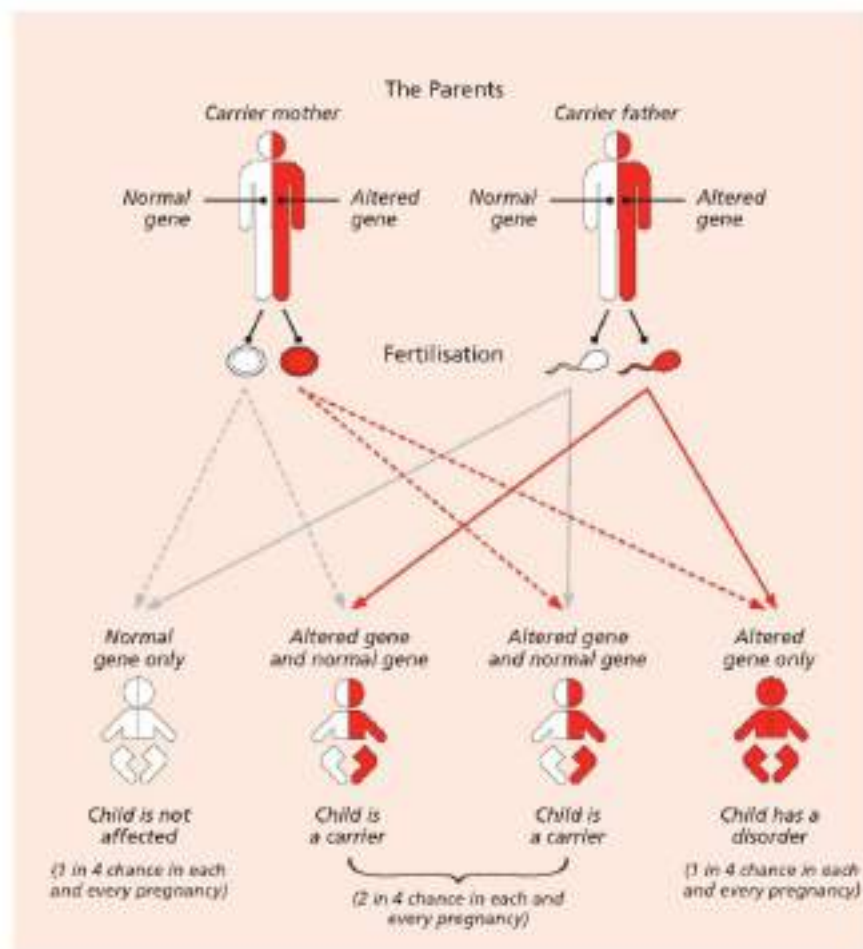


Normal red bloods are round. In sickle cell anaemia some red blood cells become deformed, so they look like a sickle used to cut wheat. These unusually shaped cells give the disease its name.

Causes

Sickle cell anaemia 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 anaemia make hemoglobin S the S stands for sickle.



To have an autosomal recessive disorder, you inherit two mutated gene, 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, a 50 percent chance of having an unaffected child who also is carrier and a 25 percent chance of having an affected child with two recessive genes.

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.