Sickle cell anemia

Sickle cell anemia is a disease passed down through families in which red blood cells form an abnormal sickle or crescent shape. Red blood cells carry oxygen to the body and are normally shaped like a disc.

Causes, incidence, and risk factors

Sickle cell anemia is caused by an abnormal type of hemoglobin called hemoglobin S. Hemoglobin is a protein inside red blood cells that carries oxygen.

- Hemoglobin S changes the shape of red blood cells. The red blood cells become shaped like crescents or sickles.
- The fragile, sickle-shaped cells deliver less oxygen to the body's tissues.
- They can also get stuck more easily in small blood vessels, as well as break into pieces that can interrupt healthy blood flow. These problems decrease the amount of oxygen flowing to body tissues even more.

Sickle cell anemia is inherited from both parents. If you inherit the sickle cell gene from only one parent, you will have sickle cell trait. People with sickle cell trait do not have the symptoms of sickle cell anemia.
Sickle cell disease is much more common in people of African and Mediterranean descent. It is also seen in people from South and Central America, the Caribbean, and the Middle East.

Symptoms

Symptoms usually do not occur until after age 4 months.
Almost all patients with sickle cell anemia have painful episodes (called crises), which can last from hours to days. These crises can cause pain in the bones of the back, the long bones, and the chest.
Some patients have one episode every few years. Others have many episodes per year. The crises can be severe enough to require a hospital stay.
When the anemia becomes more severe, symptoms may include:

- Fatigue
- Paleness
- Rapid heart rate
- Shortness of breath
- Yellowing of the eyes and skin (jaundice)

Younger children with sickle cell anemia have attacks of abdominal pain.
The following symptoms may occur because small blood vessels may become blocked by the abnormal cells:

- Painful and prolonged erection (priapism)
- Poor eyesight or blindness
- Problems thinking or confusion caused by small storkes
- Ulcers on the lower legs (in adolescents and adults)

Over time, the spleen no longer works. As a result, people with sickle cell anemia may have symptoms of infections such as:

- Bone infection (osteomyelitis)
- Gallbladder infection (cholecystitis)

- Lung infection (pneumonia)
- Urinary tract infection

Other symptoms include:

- Delayed growth and puberty
- Painful joints caused by arthritis

Signs and tests

Tests commonly performed to diagnose and monitor patients with sickle cell anemia include:

- Bilirubin
- Blood oxygen
- Complete blood count (CBC)
- Hemoglobin electrophoresis
- Serum creatinine
- Serum potassium
- Sickle cell test

Treatment

The goal of treatment is to manage and control symptoms, and to limit the number of crises. Patients with sickle cell disease need ongoing treatment, even when they are not having a painful crisis. It is best to receive care from health care providers and clinics that take care of many patients with sickle cell anemia.

Folic acid supplements should be taken. Folic acid is needed to make new red blood cells.

Treatment for a sickle cell crisis includes:

- Blood transfusions (may also be given regularly to prevent stroke)
- Pain medicines
- Plenty of fluids

Other treatments for sickle cell anemia may include:

- Hydroxyurea (Hydrea), a medicine that may help reduce the number of pain episodes (including chest pain and difficulty breathing) in some people
- Antibiotics to prevent bacterial infections, which are common in children with sickle cell disease

Treatments that may be needed to manage complications of sickle cell anemia include:

- Dialysis or kidney transplant for kidney disease
- Counseling for psychological complications
- Gallbladder removal in people with gallstone disease
- Hip replacement for avascular necrosis of the hip
- Surgery for eye problems
- Treatment for overuse or abuse of narcotic pain medicines
- Wound care for leg ulcers

Bone marrow or stem cell transplants can cure sickle cell anemia. However, they are currently not an option for most patients. Sickle cell anemia patients are often unable to find well-matched stem cell donors.

People with sickle cell disease must reduce their risk of infections. This includes receiving certain vaccinations, including:

- Haemophilus influenzae vaccine (Hib)
- Pneumococcal conjugate vaccine (PCV)

- Pneumococcal polysaccharide vaccine (PPV)

Support Groups

Joining a support group where members share common experiences can relieve the stress related to caring for someone with a chronic disease. See: Sickle cell anemia - support group

Expectations (prognosis)

In the past, sickle cell patients often died between ages 20 and 40. Thanks to a better understanding and management of the disease, today patients can live into their 50s or beyond.

Causes of death include organ failure and infection.

Calling your health care provider

Call your health care provider if you have:

- Any symptoms of infection (fever, body aches, headache, fatigue)
- Pain crises
- Painful and long-term erection (in men)