CONDITION

Sickle cell disease

Sickle cell disease is an inherited blood condition that causes anemia, pain episodes, and possible organ damage.

WHAT IT IS
Inherited condition affecting red blood cells

BODY SYSTEM
Blood, circulation, immune system, and organs

MAIN SYMPTOMS
Pain episodes, anemia, fatigue, frequent infections

WHAT CAUSES IT
Inherited changes in the beta-globin gene

HOW IT IS DIAGNOSED
Newborn screening and hemoglobin testing; sometimes genetic testing

MAIN TREATMENT
Medicines, pain management, transfusions, and specialist care

CAN IT BE CURED?
Sometimes - stem cell or gene therapy can cure

IS IT AN EMERGENCY?
Sometimes - breathing difficulty, stroke signs, or fever

LONG-TERM OUTLOOK
Lifelong condition; modern care improves health, but complications remain

Overview

Sickle cell disease is a group of inherited blood conditions. It affects hemoglobin, the protein in red blood cells that carries oxygen around the body.

Healthy red blood cells are flexible and usually round. In sickle cell disease, an altered form of hemoglobin can make some cells stiff, sticky, and curved like a crescent or sickle. These cells can break down earlier than normal, causing anemia. Anemia means the blood has fewer healthy red blood cells available to carry oxygen.

Sickled cells can also block small blood vessels. This reduces blood flow and may cause sudden pain, known as a pain crisis or vaso-occlusive episode. Repeated blockages and ongoing anemia can gradually affect organs such as the lungs, brain, kidneys, eyes, bones, and spleen.

There are several types. Sickle cell anemia, usually called HbSS, is one of the most severe. Other types include HbSC and sickle beta thalassemia. Symptoms and complications can differ even among people with the same type.

Sickle cell trait is not the same as sickle cell disease. A person with the trait carries one altered gene and usually does not have the ongoing symptoms of the condition. They can still pass the gene to their children.

Symptoms

Symptoms often begin during infancy, after levels of fetal hemoglobin naturally fall. Fetal hemoglobin is the type of hemoglobin present before birth and during the first months of life.

Symptoms may include:

Pain episodes may last for hours or days. Their frequency and severity are different for each person. Some have few episodes, while others have frequent pain or develop chronic pain.

Certain conditions can contribute to sickling, including dehydration, low oxygen, illness, temperature extremes, or physical stress. An episode does not always have an obvious trigger.

Other symptoms may point to a complication. These include chest pain, breathing difficulty, severe headache, weakness on one side, vision changes, a painful erection lasting longer than expected, or sudden enlargement of the abdomen in a child.

- Episodes of pain in the chest, back, abdomen, arms, legs, or joints
- Tiredness, weakness, shortness of breath, or pale skin from anemia
- Yellowing of the eyes or skin, called jaundice
- Swollen and painful hands or feet, especially in babies
- Frequent or serious infections
- Slower growth or delayed puberty
- Ongoing pain between sudden pain episodes

Causes and risk factors

Sickle cell disease is caused by changes in the HBB gene, which provides instructions for making part of hemoglobin. It is inherited in an autosomal recessive pattern. This means a child usually develops the condition after receiving an altered gene from each biological parent.

When both parents have sickle cell trait, each pregnancy has a possibility of producing a child

with sickle cell disease, a child with the trait, or a child with neither. The outcome of one pregnancy does not affect the next.

The condition is more common in people whose families come from parts of Africa, the Mediterranean, the Middle East, India, the Caribbean, and Central or South America. However, it can occur in any ethnic group.

Your genes determine whether you have sickle cell disease. Lifestyle, diet, stress, or something a parent did during pregnancy does not cause it.

The type of sickle cell disease influences the chance of complications, but it does not predict one exact course. Access to preventive care, treatment, and regular monitoring also affects long-term health.

Diagnosis

In many countries, sickle cell disease is found through newborn blood-spot screening. Early diagnosis allows preventive treatment and education to begin before symptoms or serious infections develop.

Diagnosis is confirmed with blood tests that identify the types of hemoglobin present. These may be called hemoglobin electrophoresis, high-performance liquid chromatography, or capillary electrophoresis. A complete blood count can show anemia, but it cannot confirm sickle cell disease by itself.

Genetic testing may be used when hemoglobin test results are unclear, to identify the exact type, or when testing relatives. Testing may also distinguish sickle cell disease from sickle cell trait.

If sickle cell disease is suspected later in life, the same hemoglobin tests can be used. Recent blood transfusions may affect the results, so the testing team needs to know about them.

Carrier screening can identify sickle cell trait before or during pregnancy. Prenatal testing can determine whether a fetus has inherited the condition, but this is a personal choice that requires clear information about the test and its risks.

Treatment and management

Care is usually coordinated through a specialist sickle cell service. Treatment aims to reduce pain episodes, prevent infections and organ damage, and treat complications early. The plan depends on age, sickle cell type, symptoms, previous complications, and personal

circumstances.

Hydroxyurea is a disease-modifying medicine used for many people with sickle cell disease. It increases fetal hemoglobin, which is less likely to sickle. It can reduce pain episodes, acute chest syndrome, hospital treatment, and the need for some transfusions. Regular blood tests are needed because it can affect blood cell counts. Pregnancy and family-planning considerations should be discussed with the prescribing team.

Other disease-modifying medicines may be available in some countries or for certain age groups. Their benefits, risks, and availability differ.

Pain treatment may include fluids, warmth, rest, and pain medicines used according to an agreed care plan. Severe pain may require hospital treatment. Persistent pain can need a broader approach involving physical activity, sleep support, psychological care, and specialist pain services.

Blood transfusions provide healthy red blood cells. They may be used for severe anemia, acute chest syndrome, stroke, surgery, or other complications. Some people receive regular transfusions to lower stroke risk. Repeated transfusions can cause iron buildup or immune reactions against donated blood, so monitoring is needed.

Preventive care may include recommended vaccinations and, for young children, antibiotics like amoxicillin or doxycycline to lower the risk of serious bacterial infection. Regular checks may look for stroke risk, kidney problems, eye disease, lung complications, and iron overload.

A stem cell transplant can cure some people by replacing blood-forming cells with healthy donor cells. It carries important risks, including serious infection, infertility, and graft-versus-host disease, in which donor immune cells attack the recipient's tissues.

Gene therapies can also be potentially curative for some people with severe disease. They involve collecting and altering a person's own blood-forming cells, followed by intensive chemotherapy. Access is limited, and long-term follow-up is essential.

Complications

Sickle cell disease can affect almost any organ. Complications may occur suddenly or develop over years.

Acute chest syndrome happens when sickling, infection, or another problem reduces oxygen exchange in the lungs. It may cause chest pain, fever, cough, or breathing difficulty and needs urgent hospital treatment.

Blocked blood flow to the brain can cause a stroke. Children may have no warning symptoms

before a first stroke, which is why some receive regular ultrasound screening of blood flow in the brain.

Damage to the spleen increases the risk of serious infection. In young children, blood may suddenly collect in the spleen, causing rapid anemia and abdominal swelling. This is called splenic sequestration.

Other possible complications include kidney damage, gallstones, leg ulcers, bone or joint damage, eye disease, hearing problems, heart strain, and pulmonary hypertension, which is high pressure in the blood vessels of the lungs.

Priapism is a painful erection that continues without sexual stimulation. A prolonged episode can damage tissue and requires urgent treatment.

Pregnancy with sickle cell disease has higher risks for both the pregnant person and baby. Specialist care before and during pregnancy can help manage these risks.

Prevention

Sickle cell disease cannot be prevented in someone who has inherited the gene changes.

Carrier screening and genetic counselling can help people understand the chance of passing sickle cell disease or trait to a child. These services explain reproductive options without directing personal decisions.

Complications cannot always be prevented, but regular care can lower some risks. Helpful measures include keeping vaccinations current, attending recommended screening, taking prescribed preventive medicines, and following an individual pain and fever plan.

Drinking enough fluid, avoiding extreme temperatures, and increasing physical activity gradually may reduce avoidable stress on the body. These steps do not guarantee that pain episodes will stop, and having an episode does not mean you caused it.

Living with it and what to expect

Sickle cell disease is lifelong. Its course is not fully predictable. Symptoms may be mild for long periods and then become more active, or they may require frequent care from childhood.

During childhood, infection prevention and stroke screening are major priorities. Teenagers gradually take more responsibility for medicines, appointments, hydration, and knowing when to seek help. A planned move from children's to adult services can reduce gaps in care.

In adulthood, some people develop more persistent pain, fatigue, or organ problems. Others

continue to have long periods with few symptoms. Regular reviews remain important even when you feel well because kidney, eye, or lung changes may begin without clear symptoms.

Sickle cell disease can affect school, employment, exercise, travel, sleep, relationships, and mental health. Many people study, work, have families, and take part in activities they value, although adjustments may be needed during pain episodes or periods of fatigue.

A written care plan can explain your usual pain treatment, baseline blood results, transfusion history, and emergency warning signs. Carrying this information may help when you receive care from a team that does not know you.

Long-term health has improved with newborn screening, preventive care, disease-modifying treatment, and better management of complications. Even so, sickle cell disease remains a serious condition that can shorten life. Ongoing specialist care matters throughout adulthood.

When to get medical help

Arrange routine follow-up if pain is becoming more frequent, fatigue is worsening, medicines are causing problems, or symptoms are interfering with sleep, school, work, or daily activities. New vision changes, swelling, reduced urination, or ongoing shortness of breath also need prompt assessment.

Follow your sickle cell care plan for fever. Fever can be the first sign of a serious infection, especially in children, and usually requires urgent medical assessment.

Seek urgent or emergency care for:

Emergency symptoms should not wait for a routine appointment.

- Chest pain, breathing difficulty, rapid breathing, or blue or grey lips
- Stroke signs, such as facial drooping, one-sided weakness, confusion, trouble speaking, or a sudden severe headache
- Severe pain that is not controlled by the agreed home plan
- A painful erection that continues or keeps returning
- Fainting, unusual sleepiness, or difficulty waking
- Sudden abdominal swelling, marked paleness, or weakness, especially in a child
- Fever with severe illness, breathing problems, neck stiffness, or reduced alertness

Common questions

Is sickle cell disease contagious?

No. It is inherited through genes and cannot spread through contact, coughing, sex, blood exposure, or sharing food.

Is sickle cell trait the same as sickle cell disease?

No. People with sickle cell trait usually have one altered HBB gene and one usual gene. They generally do not have the ongoing anemia and blood-vessel blockages seen in sickle cell disease. Rare health problems can occur under extreme conditions.

Can sickle cell disease be cured?

Sometimes. A stem cell transplant or certain gene therapies can be curative for selected people. These treatments involve intensive care and significant risks, and they are not suitable or available for everyone.

What triggers a sickle cell pain crisis?

Possible triggers include dehydration, infection, low oxygen, extreme temperatures, overexertion, or major stress on the body. Many episodes have no clear trigger.

Can someone with sickle cell disease have children?

Yes. Sickle cell disease may affect fertility, pregnancy risks, and the chance of passing an altered gene to a child. Pre-pregnancy care, carrier testing for a partner, and genetic counselling can provide useful information.

Why does sickle cell disease cause so many different problems?

Red blood cells travel throughout the body. When they break down early or block small blood vessels, they can reduce oxygen delivery to many different tissues and organs.

Sources

- Centers for Disease Control and Prevention: About Sickle Cell Disease
- National Heart, Lung, and Blood Institute: Sickle Cell Disease
- American Society of Hematology: Sickle Cell Disease Clinical Practice Guidelines
- GeneReviews: Sickle Cell Disease
- US Food and Drug Administration: FDA Approves First Gene Therapies to Treat Patients with Sickle Cell Disease