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Overview of SCD

What is sickle cell disease (SCD)?

SCD is a group of inherited disorders that affect red blood cells. In the United States, most people with SCD are of Black or African ancestry, but the disorder is also common in people with Hispanic backgrounds.[1] SCD should be thought of as a chronic, progressive, debilitating disease that causes a lifetime of physical, emotional, and mental challenges—both for those who have it and for their families.

During the 1970s, in the United States, life expectancy for someone with SCD was less than 20 years of age. Due to advances, today, people with SCD often live well into their 40s.[2]

How does SCD affect the body?[1]

Hemoglobin is a protein in red blood cells that carries oxygen throughout the body. Red blood cells with normal hemoglobin are disc-shaped and flexible, so they can travel through the arteries easily and deliver oxygen to all tissues in the body. Abnormal hemoglobin in red blood cells, called ***hemoglobin S***, causes SCD. Hemoglobin S causes red blood cells to sickle, or change shape. Sickle cells are rigid and cannot change shape easily and, because of their inflexibility, they break apart. Simultaneously, blood cells become sticky and form clusters in the blood vessel. This can cause blockages that slow or stop the flow of blood and oxygen to nearby tissues.

Pain crises can happen when lack of oxygen to tissues causes sudden, severe attacks that may require treatment in a hospital.

Normal red blood cells and sickle red blood cells[1,3]

Healthy individual

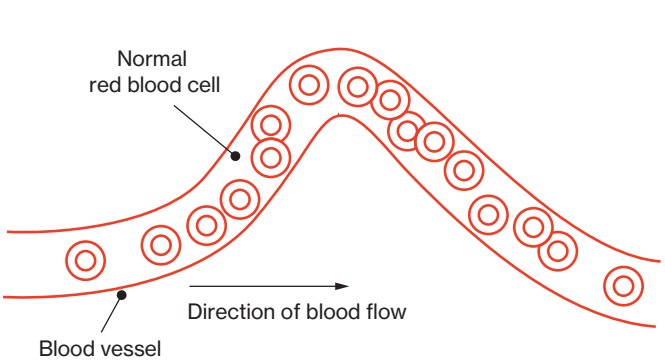


Figure A shows normal red blood cells flowing freely in a blood vessel.

Individual with sickle cell anemia

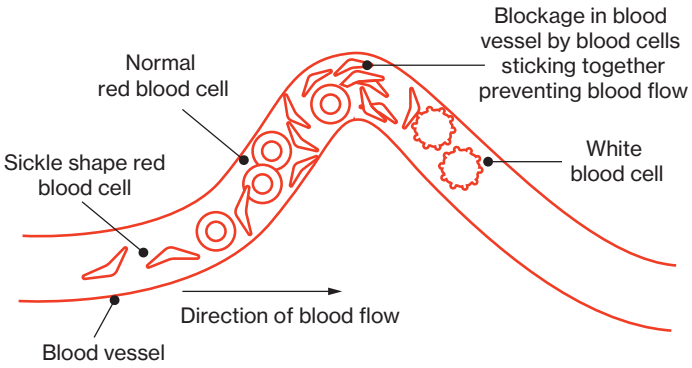
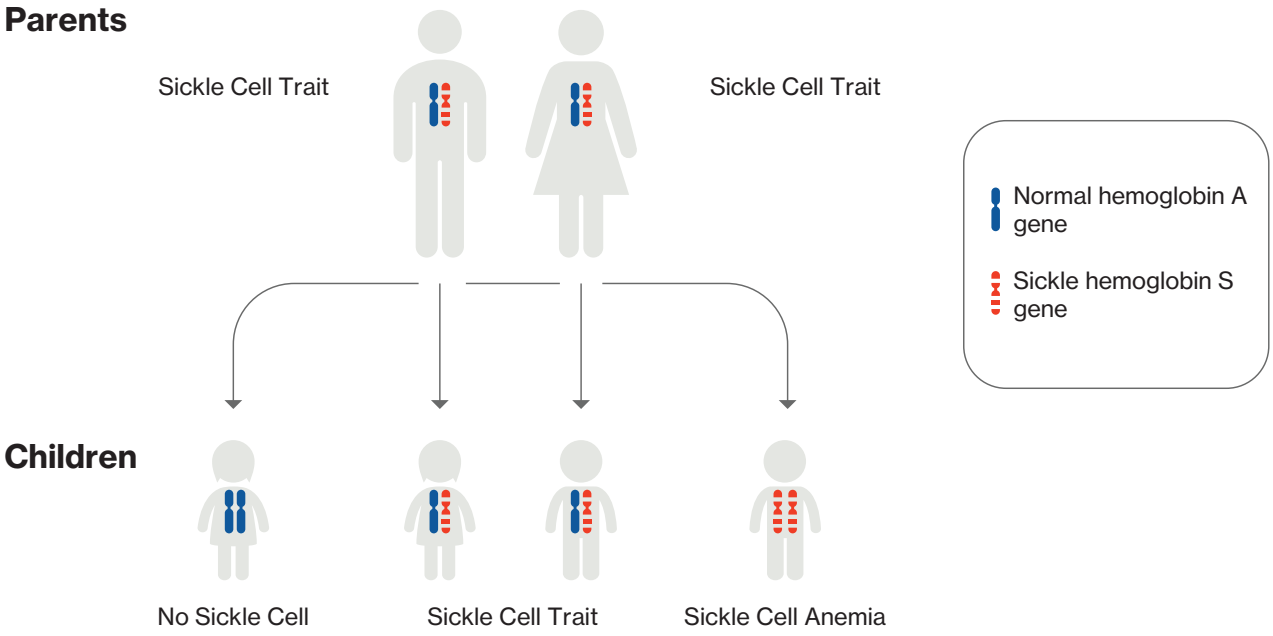


Figure B shows blood cells sticking together and blocking blood flow in a blood vessel.

How is SCD inherited?[1]

If a child inherits hemoglobin S from both parents, the child will be born with SCD. If the hemoglobin S gene is passed to a child from only 1 parent, the child will have ***sickle cell trait***. When both parents have sickle cell trait, each of their children has a 25% chance of being born with SCD.



People who have sickle cell trait rarely have complications like those seen in people who have SCD, but they do carry the defective hemoglobin S gene and can pass it on if they have a child.

How prevalent is SCD?[4]

The majority of children born with SCD are of Black or African ancestry. In the United States, it is estimated that:

1 of every 365 African-American babies is born with SCD. About 100,000 people have SCD, and ~3,000,000 people have sickle cell trait.

References: 1. National Heart, Lung, and Blood Institute. Sickle cell disease. https://www.nhlbi.nih.gov/print/4933. Accessed April 9, 2021. **2.** Adams-Graves P, Bronte-Jordan L. Recent treatment guidelines for managing adult patients with sickle cell disease: challenges in access to care, social issues, and adherence. *Expert Rev Hematol*. 2016;9(6):541-552. **3.** Conran N, Franco-Penteado CF, Costa FF. Newer aspects of the pathophysiology of sickle cell disease vaso-occlusion. *Hemoglobin*. 2009;33(1):1-16. **4.** American Society of Hematology. Understanding the impact of sickle cell disease. http://www.scdcoalition.org/pdfs/ASH_Infographic.pdf?_ga=2.202673880.830594348.1617381148-715304286.1617381148. Accessed April 9, 2021.

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Diagnosing SCD

A diagnosis of sickle cell disease (SCD) can be very difficult for people to receive[1]

When people find out that their child has SCD and that they themselves have the sickle cell trait, it can cause a wide range of normal, emotional feelings such as:

- Shock, if parents didn't know they have sickle cell trait; disbelief, especially if the diagnosis is unexpected
- Upset, angry, or guilty that they have unknowingly given SCD to their child
- Helpless because neither they nor anyone else can take away SCD from their child
- Confused, anxious, depressed, and/or scared because they don't know how SCD will affect their child and the rest of the family

Screening for SCD[2]

Newborn screening
As part of a required screening program, all babies born in the United States are tested for SCD at birth.

Prenatal screening
Parents may choose to have prenatal screening for SCD. These tests can be performed as early as 8 to 10 weeks into pregnancy, by testing tissues from the placenta or amniotic fluid.

Genetic testing
A blood test can be performed to learn if someone has SCD.

Early signs, symptoms, and warning signs of complications

Normally, if someone has SCD, it is detectable at birth and parents are notified immediately. However, most newborns don't show signs or symptoms of SCD until ~5 months of age or later.[2] This is because ***baby (or fetal) hemoglobin*** protects red blood cells from sickling. At around 4 to 5 months of age, a baby's fetal hemoglobin is replaced by abnormal hemoglobin (hemoglobin S) that causes red blood cells to begin to sickle.[3]

Some typical early signs and symptoms of SCD include[2]:

- Jaundice (yellowing of the skin) or icterus (yellow in the whites of the eyes) happens when a large number of red blood cells are destroyed in the spleen and other parts of the body (called ***hemolysis***)[4]
- Fatigue or fussiness due to anemia
- Swelling in the hands and feet caused by sickle cells inhibiting blood flow in and out of the hands and feet (hand-foot syndrome)[2,5]

Early problems will vary among individuals and can change over time.

References: 1. Oni L, Dick M, Walters J, Rees D. *A Parent's Guide to Managing Sickle Cell Disease*. 3rd ed. National Health Service. Brent Sickle Cell & Thalassaemia Centre, London; 2012. **2.** National Heart, Lung, and Blood Institute. Sickle cell disease. https://www.nhlbi.nih.gov/print/4933. Accessed April 9, 2021. **3.** Centers for Disease Control and Prevention. Complications and treatments of sickle cell disease. https://www.cdc.gov/ncbddd/sicklecell/treatments.html. Accessed April 9, 2021. **4.** National Heart, Lung, and Blood Institute. Hemolytic anemia. https://www.nhlbi.nih.gov/health-topics/hemolytic-anemia. Accessed April 9, 2021. **5.** Ballas SK, Kesen MR, Goldberg MF, et al. Beyond the definitions of the phenotypic complications of sickle cell disease: an update on management. *ScientificWorldJournal*. 2012;2012:949535.

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Complications of SCD

These are the common complications that people with sickle cell disease (SCD) might experience:

Pain crisis, also known as vaso-occlusive crisis (VOC), can happen suddenly anywhere in the body when sickle cells block blood flow and inhibit oxygen delivery, causing pain.[1]

- Pain may be stabbing, sharp, intense, or throbbing; pain can be excruciating
- Pain commonly strikes in the abdomen, lower back, chest, arms, and legs
- Can be brought on by dehydration, stress, high altitudes, or changes in temperature

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Acute chest syndrome is lung damage and improper oxygen exchange that can occur when sickle cells block blood vessels in the lungs.[1]

- Symptoms are: chest pain, coughing, difficulty breathing, and fever
- Often starts a few days after a pain crisis begins
- Immediate treatment in a hospital is needed

Anemia is a common complication of SCD. In SCD, red blood cells die early, resulting in a lack of healthy red blood cells to carry oxygen throughout the body. This causes multiple symptoms, including[2]:

- Fatigue and irritability
- Dizziness and lightheadedness
- Fast heart rate
- Difficulty breathing
- Pale skin color or jaundice
- Slow growth and delayed puberty

People with SCD usually have mild to moderate anemia. However, severe anemia can occur in newborns or children with SCD and can be life-threatening. Severe anemia may be caused by[1]:

- **Aplastic crisis**—usually brought on by a parvovirus B19 infection that causes bone marrow to stop making new red blood cells for a while, causing severe anemia
- **Splenic sequestration crisis**—the spleen suddenly enlarges because red blood cells get trapped there, resulting in fewer blood cells circulating through the body

Parents may notice that a baby or child with severe anemia is sluggish and doesn't want to eat.

Brain injuries from clinical or silent strokes
Strokes can be symptomatic (clinical) or asymptomatic (silent stroke).[1]

- Silent stroke is a common complication of SCD in both adults and children
- Up to 35% of children with SCD have silent strokes[3]
- Silent strokes can result in learning problems, difficulty making decisions, or holding down a job

Joint complications – Sickling in hip bones and (less commonly) other joints can cause severe joint damage due to oxygen deficiency.[1]

Priapism – A long-lasting, painful erection can happen when blood flow out of an erect penis gets blocked by sickle cells.[1]

- If priapism lasts longer than 4 hours, men or boys should seek emergency medical treatment
- Over time, priapism can permanently damage the penis and lead to impotence

Visual problems, including blindness – SCD can injure blood vessels in the eyes. The most common site of injury is the retina. Retinal damage or detachment can occur, and may lead to vision loss.[1]

Spleen damage – When sickle cells get jammed up in the spleen's blood vessels, the spleen can enlarge rapidly. It can cause anemia, pain, and reduced ability to fight infection.[1]

Leg ulcers – Sickle cell ulcers are sores that usually begin to occur after the age of 10. They may start small but grow larger. Rates of healing vary among individuals.[1]

Gallstones – A common problem with SCD. When red blood cells die, hemoglobin breaks down into bilirubin, which can form stones in the gallbladder.[1]

Pregnancy complications – During pregnancy, women with SCD are at increased risk of[1]:

- High blood pressure and blood clots
- Miscarriage, premature birth, and low-birth-weight babies

References: 1. National Heart, Lung, and Blood Institute. Sickle cell disease. https://www.nhlbi.nih.gov/print/4933. Accessed April 9, 2021. **2.** Centers for Disease Control and Prevention. Complications and treatments of sickle cell disease. https://www.cdc.gov/ncbddd/sicklecell/treatments.html. Accessed April 9, 2021. **3.** Dowling MM, Quinn CT, Rogers ZR, Buchanan GR. Acute silent cerebral infarction in children with sickle cell anemia. *Pediatr Blood Cancer.* 2010; 54(3):461–464.

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Treatment for SCD

At this time, blood and bone marrow transplants are the only cure for sickle cell disease (SCD). However, treatments are available to help patients manage complications of the disease.

Medicines – Medicines for SCD can help patients manage complications of the disease. Some approved treatment options include different medicines that help treat symptoms of sickle cell disease.

Blood transfusions – Patient's physician may recommend transfusion to treat and prevent certain SCD complications. Types of transfusions include:

- Acute transfusion may be used to treat severe anemia, an acute stroke, acute chest crises, and multiorgan failure or, presurgically, to prevent complications
- Red blood cell transfusions increase the number of normal red blood cells that are more flexible, resulting in better oxygen delivery to tissues in the body
- Ongoing transfusions are often given to people with SCD who have had an acute stroke, to lower the risk of additional strokes. Transfusions may be given to some children to reduce the chance of having a first stroke

Blood and bone marrow transplant – Transplant aims to replace the stem cells in the bone marrow of someone with SCD with healthy stem cells from a matching donor.

Talk with your patient to determine the right treatment option for them.

Reference: National Heart, Lung, and Blood Institute. Sickle cell disease. https://www.nhlbi.nih.gov/print/4933. Accessed April 9, 2021.

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Treatment adherence in patients with SCD

Adherence is an important factor in the efficacy of medications, the reduction of SCD complications, and decreased health care costs.[1]

Care managers can assist with adherence

Nonadherence is problematic within the SCD patient population, especially among adolescents and young adults.[2] Care managers can offer guidance and support to help patients overcome barriers to adherence.

Understand barriers and suggest ways to address them[1]

BARRIERS	POTENTIAL SOLUTIONS
Lack of access to clinics for treatments, screenings, communication with doctors	Connect patient with free local transportation services, if available
Failure to fill or refill prescriptions due to:	
• Patient or family beliefs about the value and necessity of taking medicine	Probe for beliefs about the severity of SCD and the use of medications
• Being forgetful or too busy	Talk with pharmacist to set up refill alerts; ask provider for a 90-day supply of medicine
• Fear of side effects	Ensure patient understands common side effects, importance of recognizing potential complications, and when to contact their doctor
• Insurance problems	Connect patient with an assistance program or local source of support; suggest talking with provider and contacting insurance carrier to learn about coverage
Balks at receiving needles	
• Fear of and pain caused by needles	If the needle hurts in one place, ask if it can be injected in a different place

BARRIERS	POTENTIAL SOLUTIONS
Skipped doses of medication either on purpose or due to:	
• Lack of understanding that, over time, missed doses may lead to complications	Help people understand the importance of taking medicine exactly as directed
• Stress caused by competing demands	Direct people to social support for themselves or for their child with SCD; consider talking with a counselor or joining a support group
• Forgetfulness	Offer tips for adherence: • Technology can help them remember — Use smartphones, laptops, or tablets to receive daily text message medication and adherence reminders — Set alarms on phone for times of medicine doses — Use smartphone calendar to enter dates for infusions, screenings, appointments, medicine refills — Use an online treatment log to keep track of medicine doses — Go online to get educated about SCD and its treatment; find an online community of people living with SCD • Try using a weekly pill box • Link taking medicine to something they do every day at the same time, like brushing their teeth
Experiencing side effects	Stay in close touch with the provider because he or she can help manage any side effects they might be having; don't stop taking medicine without talking with their provider first
Cost of medication	• Connect patient with an assistance program or local source of support • Suggest talking with provider and contacting insurance carrier to learn about coverage

References: 1. Walsh KE, Cutrona SL, Kavanagh PL, et al. Medication adherence among pediatric patients with sickle cell disease: a systematic review. *Pediatrics*. 2014;134(6):1175-1183. **2.** Badawy SM, Thompson AA, Liem RI. Technology access and smartphone app preferences for medication adherence in adolescents and young adults with sickle cell disease. *Pediatr Blood Cancer*. 2016:63(5):848-852.

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Helping patients manage life with SCD

There are things you can do to help both patients and caregivers manage life with sickle cell disease (SCD), such as:

- Encourage patients and caregivers to learn all they can about SCD
- Help patients, parents, and caregivers to recognize their feelings
- Highlight the importance of adhering to treatment, receiving follow-up care and recommended screenings, and communicating often and openly with the health care team
- Discuss the importance of a healthy lifestyle

Understanding feelings[1]

Living with SCD may bring up a wide range of feelings that may often change. Parents may feel disbelief, anger, and guilt when they first learn their child has SCD. People who have SCD or care for a family member who has it sometimes feel resentful, overwhelmed, and worried about the future. All of these feelings are common and natural, and they may come and go depending on the circumstances. It's important for patients and parents to acknowledge feelings and not feel guilty about having them.

Know the key steps to living with SCD

Care managers should ensure that patients, parents, and caregivers understand steps that can be taken to help patients live with SCD.

- Stay hydrated
- Control environment temperature when possible
- Try to avoid exposure to high altitude – flying, mountain climbing
- Skip strenuous exercise, like heavy training or mountain climbing, that can lower oxygen levels in the body

Explain key elements of adopting a healthy lifestyle while living with SCD[2]:

- Be physically active but avoid strenuous exercise; pacing yourself is important to avoid SCD fatigue
- Get enough sleep
- Hydrate frequently throughout the day
- Limit alcohol
- Follow a personalized heart-healthy eating plan
- Quit smoking

Highlight ways people can care for their mental and emotional health[2]:

- Receive support from others to help lower stress and anxiety, to express feelings and be heard
- Seek help from a mental health expert. He or she may recommend talk therapy, possible medications for depression or anxiety, and alternative therapies

Pediatric to adult transition is a critical step for teens with SCD

Life with SCD is never easy, but it can be especially stressful when teens have to transition from pediatric to adult care. Parents should work with their teen's provider to formulate a plan for this transition. Teens commonly have more hospital admissions and medical problems during this transition period for the following reasons[2]:

- As teens with SCD age, organ damage and disabilities may increase
- The shift from pediatric to adult care often occurs simultaneously with changes in teens' physical, emotional, social, and academic lives. Greater independence and self-management may be difficult for teens to cope with
- Treatment plan adherence may suffer
- There may be fewer medical and social resources in a given region for adults than there are for children with SCD

References: 1. Oni L, Dick M, Walters J, Rees D. A parent's guide to managing sickle cell disease. National Health Service. Brent Sickle Cell & Thalassaemia Centre, London. 3rd Edition; 2012:1-120. **2.** National Heart, Lung, and Blood Institute. Sickle cell disease. https://www.nhlbi.nih.gov/print/4933. Accessed April 9, 2021.

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SCD support and assistance

There are many support and assistance resources for adults with sickle cell disease (SCD), parents of a child with SCD, and family caregivers. Here are some of them:

EDUCATION, SERVICES, AND SUPPORT

Sickle Cell Disease Association of America (SCDAA)

www.sicklecelldisease.org

This is a national organization with over 50 local chapters. Come to this site to find free educational materials about living and coping with SCD. SCDAA also offers:

- Screening tests recommended for people living with SCD
- Counseling and social services
- Transportation
- Summer camp
- A sickle cell chat room

American Sickle Cell Anemia Association

www.ascaa.org

This national organization provides many services, including testing, counseling, and support services to people and families who are at risk of or already have SCD.

LEARN MORE ABOUT SCD

National Heart, Lung, and Blood Institute (NHLBI)

www.nhlbi.nih.gov/health-topics/sickle-cell-disease

NHLBI is part of the National Institutes of Health (NIH). Here you will find much information about SCD, research, and ways to manage SCD.

Centers for Disease Control and Prevention

www.cdc.gov/ncbddd/sicklecell

Learn more about SCD, who has it, and how SCD health problems can be prevented.

KidsHealth

www.kidshealth.org/en/kids/sickle-cell.html

This website has separate sections for kids, teens, and parents to learn about SCD in simple, easy-to-understand language. You can also learn about all kinds of other diseases and health problems, from cancer to warts. The website includes articles, slideshows, and videos.

ONLINE COMMUNITIES

Sick Cells

www.sickcells.org

This is an online community for people with SCD. Its purpose is to make sure the voices of people with SCD are heard.

Generation S: Sickle Cell Awareness

www.joingens.com

This online community helps people with SCD learn about SCD and find support. Visitors to the website can read and share stories about living with SCD.

SCD RESEARCH

American Society of Hematology (ASH)

https://www.hematology.org/advocacy/sickle-cell-disease-initiative

The national organization for hematologists (blood experts). Each year, at their annual meeting, experts present the newest research results about SCD. Visit their website to learn all about it.

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