Sickle Cell Disease

Carol J. Adams DNP MSN RN
Clinical Program Manager

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What is Sickle Cell Disease?

Sickle Cell Disease (SCD) is an inherited blood disorder caused by a genetic gene (HBB gene), which causes red blood cells (RBC) to become rigid, sticky, and crescent shaped.

The origin of Sickle Cell was a gene mutation that originated as a protective trait against malaria.

- One study suggests it dates back approximately 7,300 years ago to a single ancestor in Africa.

SCD affects millions of people worldwide and is especially common in people whose ancestors come from:

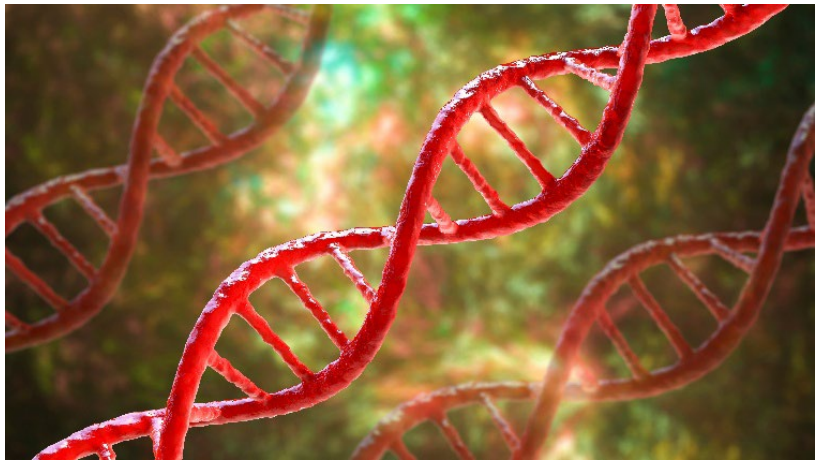
- **Africa:** Particularly West and Central Africa (Senegal, Cameroon, Benin, Nigeria).
- **Middle East:** Saudi Arabia, Lebanon**.**
- **Mediterranean:** Greece, Turkey, Italy.
- **Asia:** India.
- **Americas:** Descendants of African slaves and populations in the Caribbean, Central, and South America. (CDC, 2025)

What is Sickle Cell Disease?

The exact number of people living with SCD in the United States is unknown.

- The estimated life expectancy of those with SCD is over 20 years shorter than the average expected.

- The quality-adjusted life expectancy is more than 30 years shorter. (CDC, 2024)



What causes Sickle Cell Disease?

SCD is inherited by being passed from parent to child through their genes. Genes come in pairs—you get one of each pair from each parent:

- A child who inherits two faulty hemoglobin genes (one from each parent) will have SCD.
- A child who gets a sickle cell gene from one parent and a normal hemoglobin gene from another parent has Sickle Cell Trait (HbAS).

- If both parents are carriers (meaning both parents have Sickle Cell Trait):
  - There is a 25% chance their child will have SCD.
  - There is a 50% chance their child will have Sickle Cell Trait.
  - There is a 25% chance their child will have either SCD or Sickle Cell Trait (CDC, 2025).

What causes Sickle Cell Disease?

SCD is a group of conditions where the red blood cells are not shaped as they should be. Healthy red blood cells are round, flexible and disc shaped. With Sickle Cell Disease, they are shaped like sickles or crescent moons.

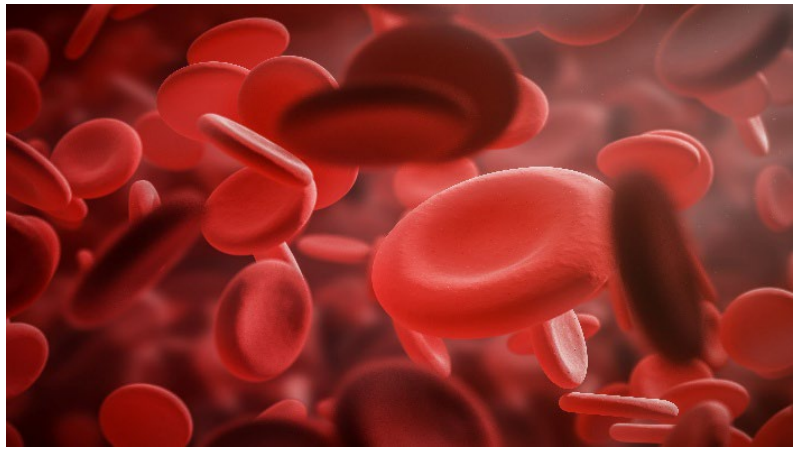
These sickle shaped cells can cause problems. They are stiff, sticky, and block small blood vessels when they get stuck together. This stops blood from moving as it should, which can lead to pain and organ damage.

They break down faster than normal red blood cells, causing too few red blood cells, a condition called anemia. (Nemours, 2026)

What causes Sickle Cell Disease?

SCD causes red blood cells to:

- Change and become more fragile.
- Become shaped like crescents or sickles.
- Deliver less oxygen to the body's tissues.



In small blood vessels, they become stuck and can break into pieces.

- This interrupts healthy blood flow and further decreases the oxygen being delivered to the body's tissues. (MedlinePlus, 2024)

What causes Sickle Cell Disease?

There are different types of SCD (March of Dimes, 2026) (CDC, 2025):

- Sickle Cell Anemia (Hbss) - usually the most severe form of SCD.
- Hemoglobin SC Disease (HbSC) - usually a milder form of SCD.
- Hemoglobin S-beta Thalassemia (HbS beta Thalassemia) - another milder form of SCD.
- Sickle Cell Trait (HbAS) – typically, you don’t have any signs of SCD.

Sickle Cell Disease in Adults

- SCD is a lifelong disorder that can lead to chronic anemia, severe pain crisis, and organ damage.
- SCD is often diagnosed at birth, but adults can be diagnosed through blood tests that identify abnormal hemoglobin. (Mayo Clinic, 2026)
- Women can be tested for SCD during pregnancy. (March of Dimes, 2026)

Common symptoms include:

- Chronic pain
- Fatigue
- Severe pain crises (Vaso-occlusive crises) that occur when sickled cells block blood flow. (Mayo Clinic, 2026)

Sickle Cell Disease in Adults

Complications may include:

- Anemia
- Acute and chronic pain
- Acute chest syndrome (life threatening emergency)
- Kidney, liver, and heart disease
- Pneumonia
- Priapism (in males, persistent and painful erection of the penis)
- Pulmonary hypertension (high blood pressure in the arteries of the lungs)
- Stroke
- Vision problems/Sickle Cell Retinopathy
- Increased susceptibility to infections (Mayo Clinic, 2026) (CDC, 2024)

Sickle Cell Disease in Children

- A child can get SCD when they receive a sickle cell gene from both parents (two sickle cell genes).
- A child who inherits only one sickle cell gene from one parent has Sickle Cell Trait.
    - Most individuals with Sickle Cell Trait do not experience symptoms of SCD.
- Babies can be tested for SCD as part of newborn screening tests. (March of Dimes, 2026)

Sickle Cell Disease in Children

In children, symptoms usually do not occur until after the age of 4 -6 months.

Symptoms may include:

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

Younger children with SCD can have attacks of abdominal pain.

Other signs and symptoms include:

- Delayed growth and puberty
- Painful joints caused by arthritis
- Heart or liver failure due to too much iron (from blood transfusions)

(MedlinePlus, 2024) (Mayo Clinic, 2026)

Symptoms of Sickle Cell Crisis

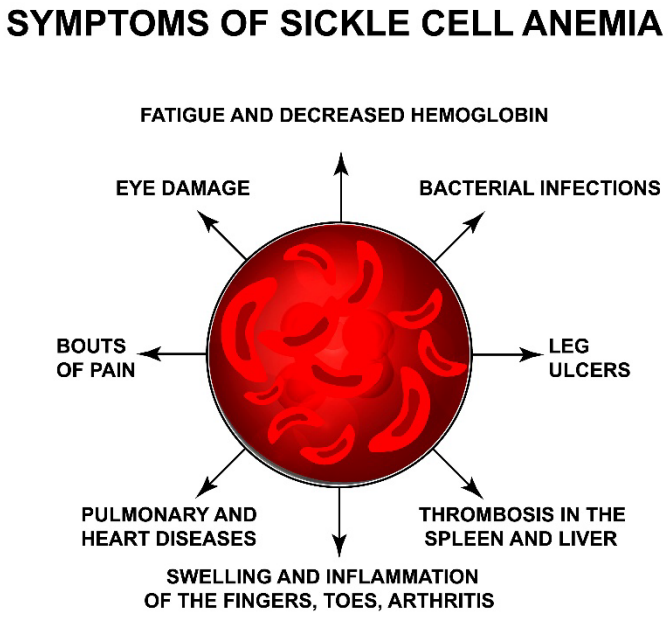
Most people with SCD have painful episodes called ‘crisis’.

- Crises can cause pain in the lower back, legs, joints and chest.
- Crises can last from hours to several days.
- Crises, if severe, may require a hospital stay.

As Sickle Cell becomes more severe, symptoms may include:

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

(MedlinePlus, 2024)



Symptoms of Sickle Cell Crisis

Other signs and symptoms include:

- Delayed growth and puberty
- Painful joints caused by arthritis
- Heart and liver failure from too much iron from blood transfusions

If the spleen is affected by SCD, infections can occur including:

- Bone infections (osteomyelitis)
- Gallbladder infections (cholecystitis)
- Lung infections (pneumonia)
- Urinary tract infections (UTI)

(MedlinePlus, 2024)

When to call the Doctor

Get emergency medical care right away if you or your child has any of these symptoms:

- Fever of 101°F (38°C) or higher
- Pain that isn't getting better with medicine
- Chest pain
- Severe headaches or dizziness
- Severe stomach pain or swelling
- Shortness of breath or trouble breathing
- Extreme tiredness
- Skin that's yellow or very pale
- A penile erection that is not going away or is painful
- Sudden change in vision
- Seizures
- Weakness or trouble moving part of the body
- Slurred speech
- Loss of consciousness (passing out)
- Numbness or tingling (Nemours, 2026)

Treatment for Sickle Cell Disease

Management of SCD is focused on avoiding pain episodes, relieving symptoms, and preventing complications through medicines and blood transfusions.

Medications to treat SCD may include:

- **Hydroxyurea (Droxia, Hydrea) -** Reduces the frequency of pain crises and the need for blood transfusions.
  - May increase the risk of infections.
- **Endari (L-glutamine) oral powder -** Reduces the frequency of pain crises.
- **Adakveo (Crizanlizumab) -** Injection to help reduce the frequency of pain crises in adults and children over the age of 16.
  - Common side effects may include nausea, joint pain, back pain, and fever.
- **Gabapentinoids (Gabapentin, pregabalin) -** May be used to manage chronic or neuropathic pain in some individuals with SCD.
- **Duloxetine and Amitriptyline -** Reduces neuropathic and chronic pain.
- **Pain-relieving medications** (Ibuprofen, Acetaminophen, Opioid medications)

(Mayo Clinic, 2026) (NIH, 2026) (CDC, 2020)

Treatment for Sickle Cell Disease

Other treatments for SCD may include:

- Blood transfusions of red blood cells
- Preventing infections with:
  - Penicillin
  - Vaccines against pneumonia, meningitis, hepatitis B, and flu shots

Procedures which may cure SCD:

Stem cell transplant (bone marrow transplant) - Replacing bone marrow affected by Sickle Cell Anemia with bone marrow from a donor.

Stem cell gene addition therapy - Removal of a person's own stem cell and a gene to produce typical hemoglobin is injected. The stem cells are then given back to the person through transplant.

Gene editing therapy - Stem cells are removed from the body, and the sickle gene is changed (edited), to help restore the cells' ability to make healthy red blood cells. The treated stem cells are then returned to the body through blood transfusions.

- In December 2023, the FDA approved two gene therapies, Casgevy and Lyfgenia, for eligible patients aged 12 years and older. (Mayo Clinic, 2026) (NIH, 2026) (CDC, 2020) (FDA, 2023)

Additional Therapies

Additional Therapies for SCD may include:

- **Cognitive Behavioral Therapy (CBT**) - focusing on thoughts, feelings, and behaviors that are interconnected.
- **Guided Audiovisual Relaxation** - uses techniques of spoken words, music, and visualization to create a relaxed state.
- **Psychological Counseling** - involves a licensed professional to help overcome feelings of distress, improve functioning and quality of life.
- **Physical and Occupational Therapy**
- **Yoga**
- **Massage**
- **Art and Music Therapy**

Key Resources for Sickle Cell Disease

Centers for Disease Control and Prevention (CDC): "Living Well with SCD" toolkit.

- Weblink: https://www.cdc.gov/sickle-cell/sickle-cell-trait/sickle-cell-trait-toolkit.html

Sickle Cell Disease Association of America (SCDAA):

- Phone: 800-421-8453
- Weblink: https://sicklecelldisease.org

Sick Cells:

- Weblink: https://sickcells.org/

American Sickle Cell Anemia Association (ASCAA):

- Phone: (216) 229-8600
- Email: irabragg@ascaa.org
- Weblink: https://ascaa.org/

PA Health & Wellness Resources

Learn more about PA Health & Wellness's Disease Management programs:

- Visit the PA Health & Wellness website:
  https://www.pahealthwellness.com/
- Call Participant Services: **1-844-626-6813 (TTY:711)**
- Email: **phwcasemanagement@pahealthwellness.com**
- QR CODE:

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Questions ?

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