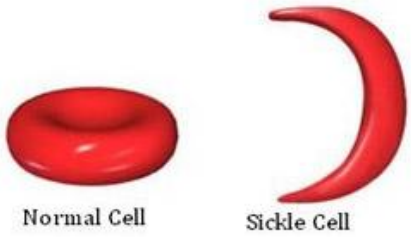
About Sickle Cell Disease

https://youtu.be/HP11Mvx1Vtg

○ What is SCD?

WHAT IS SICKLE CELL DISEASE?

Sickle cell disease (SCD) is an inherited blood disorder that affects the red blood cells. Healthy red blood cells are round and move through small blood vessels carrying oxygen to all parts of the body. In SCD, the red blood cells become hard and sticky and look like a C-shaped farm tool called a "sickle". While normal red blood cells can live up to 120 days, sickle blood cells only live for about 10 to 20 days. This causes a constant shortage of red blood cells. The sickle cells can get stuck in small blood vessels and block the flow of blood and oxygen to organs in the body. These blockages cause repeated episodes of several pain, organ damage, serious infections, or even stroke.



SCD is the most common genetic disease in the United States. SCD affects approximately 100,000 Americans. SCD occurs in 1 out of every 365 Black or African American births. SCD occurs among about 1 out of every 16,300 Hispanic American births. Worldwide, approximately 300,000 infants are born with sickle cell anemia annually. About 3000 people are living with sickle cell disease in the state of Mississippi.

○ Types of SCD

TYPES OF SICKLE CELL DISEASE

The most common types of SCD are:

- **Hemoglobin SS (HbSS)-** Inheritance of one sickle cell gene (hgb S) from each parent; known as sickle cell anemia.
- **Hemoglobin SC (HbSC)-** Inheritance of sickle cell gene (hgb S) from one parent and an abnormal hemoglobin (hgb C) from the other parent.
- **Hemoglobin beta thalassemia-** Inheritance of a sickle cell gene (hbg S) from one parent and abnormal beta globin gene from the other parent. The 2 types of thalassemia are "0" and "+".
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WHAT IS HEMOGLOBIN?

Hemoglobin (Hb or Hgb) is a protein inside the red blood cell that carries oxygen throughout the body. With SCD, the hemoglobin forms into still rods within the red blood cells which changes the shape of the red blood cells. Low hemoglobin or anemia is a condition in which a person lack enough healthy red blood cells to carry adequate oxygen inside the body. This can make you feel tired and weak.

○ Cause of SCD

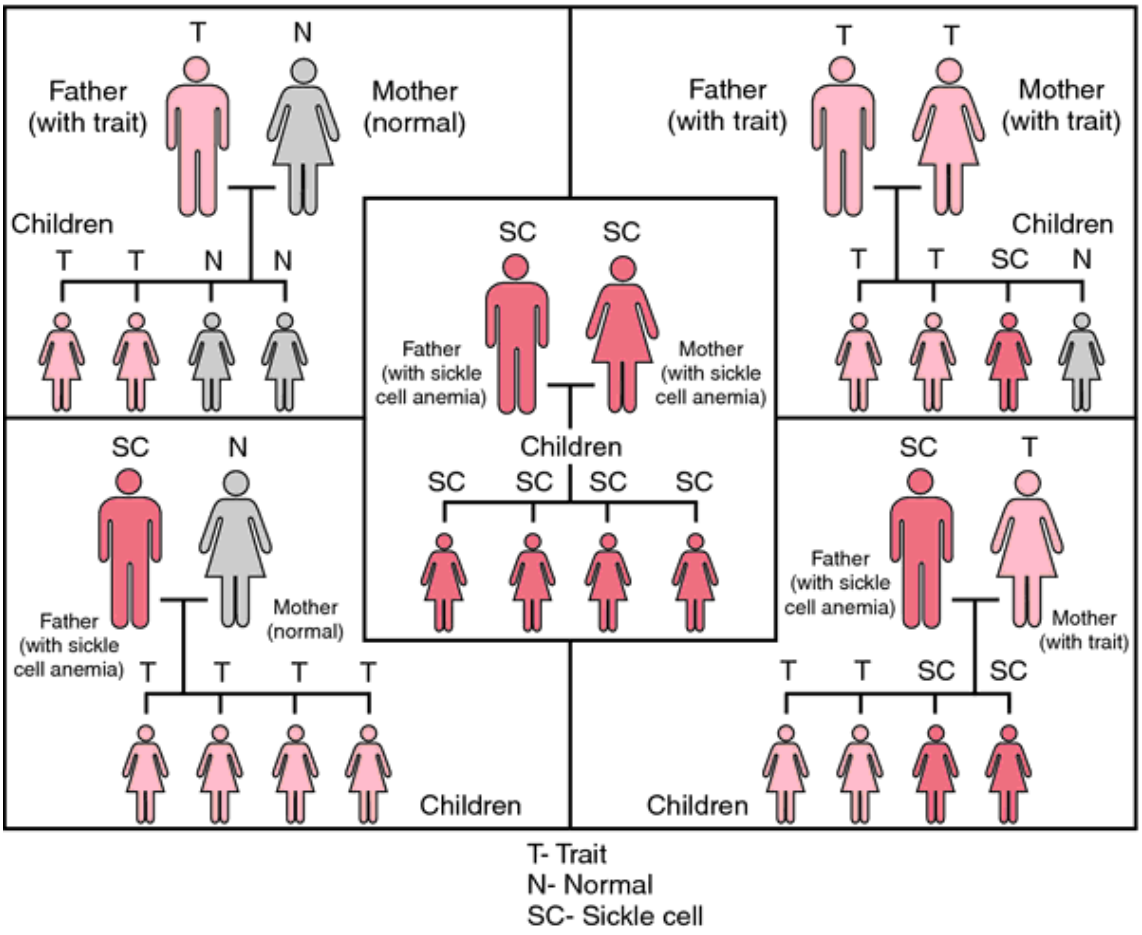
WHAT CAUSES SICKLE CELL DISEASE?

SCD is inherited in the same way that people get the color of their eyes, skin, and hair. A person with sickle cell is born with it. They inherit a sickle hemoglobin from each parent. ***People cannot catch SCD from being around a person with SCD.***

It is important to identify carriers of an abnormal hemoglobin so they will be aware of their risk of having children with sickle cell disease.

If both parents are carriers of SCT, there is a 1 in 4 chance (25%) the child will have SCD, 1 in 4 chance (25%) the child will not have SCT or SCD and 1 in 2 chance (50%) the child will have SCT.

If only one parent has SCT and the other parent has a normal gene, the child will have 1 in 2 chance (50%) chance of having SCT or 1 in 2 chance (50%) that they will not have SCT (normal gene).



○ Diagnosis

HOW IS SICKLE CELL DIAGNOSED?

SCD is diagnosed with a simple blood test. All babies are screened for SCD as part of their newborn screening. Prenatal testing can be done to find out if a baby will have SCD, SCT, or neither one.

○ Complication and Treatment

Please note: the recommended treatments listed below are recommendation based on research. Please consult with your doctor before taking any of these treatments.

WHAT ARE SOME COMMON HEALTH PROBLEMS OF SICKLE CELL DISEASE?

People with SCD show signs of the disease as early as five months. Symptoms and complications of SCD are different for each person and can range from mild to severe.

- **Infections**- People with SCD, especially infants and children often experience harmful infections such as flu, meningitis, and hepatitis.
- **Stroke**- Sickle cells can clog blood flow to the brain and cause a stroke. A stroke can result in lifelong disabilities and learning problems.
- **Acute Chest Syndrome (ACS)**- blockage of the flow of blood to the lungs can cause ACS. ACS is like pneumonia and cause chest pain, fever, and difficulty breathing. ACS is life threatening and should be treated in the hospital.
- **Pain/Pain Crisis-** When sickle cells get stuck and do not move easily through small blood vessel, they clog the blood flow which cause pain that can start suddenly, be mild to severe, and can last for any length of time.
- **Hand-Foot Syndrome**- Swelling in the hands and feet, often along with a fever. This is caused by the sickle cells getting stuck in the blood vessels and blocking the blood from flowing freely through the hands and feet.
- **Eye Disease**- SCD can affect the blood vessels in the eye and can lead to long term damage.
- **Leg Ulcers**- Poor circulation due to blockage of blood vessels by sickle cell can lead to sores that ae difficult to heal. These often occur around the ankles.
- **Priapism-** is an unwanted and sometimes prolonged painful erection in men. This happens when blood flow out of the erect penis is blocked by sickled cells. Priapism can cause permanent damage and lead to impotence. Priapism that lasts more than 4 hours is a medical emergency.

Other problems caused by SCD are jaundice, enlarged spleen, gallstones, heart problems such as coronary heart disease and pulmonary hypertension, joint problems, and organ damage.

Complication during pregnancy can raise the risk of high blood pressure and blood clots in people who have SCD. Although a woman with SCD can have a healthy baby, SCD increases the risk of miscarriage, premature birth, and low birth weight babies.

HOW IS SICKLE CELL DISEASE TREATED/MANAGED?

- **Antibiotics to try to prevent infections.**

To prevent infections, doctors may prescribe a daily dose of penicillin. **Penicillin** taken twice a day helps lower children's chance of having a serious infection in the bloodstream. Newborns need to take liquid penicillin. Older children can take tablets.

Many healthcare providers will stop prescribing penicillin after a child has reached the age of 5. Some providers prefer to continue to prescribe this antibiotic throughout a person's life, particularly if the person has hemoglobin SS or hemoglobin Sβ0 thalassemia, since people who have sickle cell disease are still at risk. All people who have had surgical removal of the spleen (called a splenectomy*)* or a past infection with pneumococcus should keep taking penicillin throughout their lives.

- **Pain relievers for acute or chronic pain**

Pain crises, also known as a sickle cell crisis, are unpredictable and extremely painful, lasting anywhere from a few hours to a few weeks, and are the most common reason for hospitalization. There can be two types of pain: acute pain which happens quickly and usually goes away when after the underlying cause is treated or has been resolved. Chronic pain is the second pain type. Chronic pain lasts longer than six months despite medication and treatment. Frequency, duration, and intensity of pain crises can vary with age. In addition to causing extreme pain, these crises can affect any part of the body and can happen in more than one spot at a time.

To manage the pain, non-steroidal anti-inflammatory drugs (NSAIDs) can either be purchased over the counter or prescribed by a doctor. Moderate-to-severe chronic pain usually is treated with opioids. Long-term opioid therapy (LTOT), or long-term use of opioids, such as oxycodone, hydrocodone, and morphine, can be prescribed by providers to treat moderate to severe pain but can also have serious risks and side effects. **Have detailed discussions with your provider about whether LTOT is right for you.** Many opioids are taken in pill form, some are administered through a vein, by injection or through an IV.

- **Four FDA-approved medications**

Four drugs have been approved by the FDA for the treatment of sickle cell disease. Before taking any medication, please speak with your document.

- **Hydroxyurea (Droxia, Hydrea, Siklos).** Daily hydroxyurea reduces the frequency of painful crises and might reduce the need for blood transfusions and hospitalizations. But it can increase the risk of infections. Don't take the drug if you're pregnant.
- **L-glutamine oral powder (Endari).** ENDARI is a medication to reduce the acute complications of sickle cell disease in adults and children 5 years and older. ENDARI is pharmaceutical grade l-glutamine oral powder that has been shown to reduce the

acute complications of sickle cell disease in adults and children 5 years and older when taken as directed. The FDA recently approved this drug for treatment of sickle cell anemia. It helps in reducing the frequency of pain crises. See https://www.endarirx.com

- **Crizanlizumab (Adakveo).** Adakveo is given by injection and can help reduce the frequency of pain crises in adults and children older than 16. Side effects can include nausea, joint pain, back pain, and fever. See https://www.us.adakveo.com/sickle-cell-disease

- **Voxelotor (Oxbryta).** Oxbryta is used to treat sickle cell disease in adults and children older than 12. Taken orally, this drug can lower the risk of anemia and improve blood flow throughout the body. Side effects can include headache, nausea, diarrhea, fatigue, rash, and fever. See https://www.oxbryta.com/about-oxbryta

- **Vaccines (especially flu)**
  Childhood vaccinations are important for preventing disease in all children. They're even more important for children with sickle cell anemia because their infections can be severe. Your child's doctor should ensure that your child receives all the recommended childhood vaccinations, as well as vaccines against pneumonia, meningitis, hepatitis B and an annual flu shot. Vaccines are also important for adults with sickle cell anemia.

  During the COVID 19 pandemic, people with sickle cell anemia should take extra precautions, such as staying isolated at home as much as possible and for those who are eligible, getting vaccinated.

- **Blood Transfusions**
  If you have sickle cell disease (SCD), you may need one or more blood transfusions (healthy blood from a donor put into your body) during your lifetime. During a blood transfusion, your blood and the donated blood must have matching antigens, or special proteins on the surface of each red blood cell.

Other forms of treatment can be massages, using heating pads, yoga classes, etc. Staying hydrated, eating a healthy diet, exercise and getting plenty of rest may also help with pain crises. Before any of these treatments and/or pain medications, please make sure you discuss with your doctor. It is very important that you have a team of doctors for your medical care and keep all scheduled doctor's appointments.

Cure

Stem cell or bone marrow transplants are the only cure for sickle cell disease. Most transplants are currently performed in children who have had complications such as strokes, acute chest crises, and recurring pain crises. The transplant requires a matched donor. These transplants are riskier in adults with sickle cell disease. There is NO universal cure for sickle cell disease.

What is Sickle Cell Trait

WHAT IS SICKLE CELL TRAIT?

Sickle Cell Trait (SCT) occurs when a person inherits one sickle cell gene from one parent and a normal gene from the other parent. Approximately one in ten African Americans carries sickle cell trait. People who are carriers generally do not have any major medical problems. If you have SCT, you cannot develop SCD. Certain conditions could be harmful to people with SCT such as increased pressure in the atmosphere, low oxygen levels in the air, dehydration, and high altitude. SCT is diagnosed with a simple blood test. One in 12 blacks or African Americans in the United States have SCT.

Sickle Cell Trait in Athletes

Some athletes experience heat strokes and muscle breakdowns when participating in intense workouts and competitive sports. To prevent these complications, athletes should drink plenty of water, set their own pace, rest often, keep body temperate cool, and seek medical care when feeling ill.