New Diagnosis Handbook for Pediatric Patients

Sickle Cell Disease

Information and Resources

Updated 2024

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Pediatric Specialists of Virginia

Children's National INOVA

Letter from Pediatric Specialists of Virginia (PSV) Sickle Cell Clinic

Sickle Cell Disease is a serious condition, but with today's medical knowledge and technology, qualify of life for children with sickle cell disease can be great. Now that your child is diagnosed with sickle cell disease, it is important to maintain consistent care with Pediatric Specialists of Virginia's (PSV) Sickle Cell Clinic, as well as with your child's pediatrician. Parents, families, and patients themselves play the mosts important role in the well-being of children with sickle cell disease. PSV's Sickle Cell Clinic is here to provide family-centered care and help coordinate multidisciplinary specialty care for our patients.

The Sickle Cell Team at PSV is composed of physicians, nurse practitioners, physician assistants, registered nurses, nurse coordinators, social workers, and psychologists.
Our goal is to provide comprehensive care to pediatric patients with sickle cell disease.

Elizabeth Yang, MD, PhD: Hematologist
Robin Dulman, MD: Hematologist
Angela Lewis, FNP: Nurse Practitioner
Noravy Briere, CPNP: Nurse Practitioner
Nupur Gupta, PsyD: Psychologist
Quinn Mashuda, RN: Nurse coordinator
Kendle Valcourt, MSW, Social Worker

This booklet provides helpful information regarding the care of your child. We look forward to getting to know you and your child as we work together to provide the best sickle cell care possible.

Sincerely,

The Sickle Cell Team
Pediatric Specialists of Virginia
Center for Cancer and Blood Diseases of Northern Virginia (CCBD-NV) Inova Schar Cancer Institute
8081 Innovation Park Drive Building B, Suite 765 Fairfax,
VA 22031
571.472.1717
https://psvcare.org/northern-virginia-comprehensive-pediatric-sickle-cell-program

When to call your doctor

- Fever 100.4 F (38 C) or higher for younger than 6 months
- Fever 101 F (38.5 C) or higher for 6 months or older
- Difficulty breathing or chest pain
- Pain not relieved with home care
- Unable to hydrate
- Abdominal swelling
- Severe headache
- Sudden weakness or loss of feeling or movement
- Seizure
- Painful erection of penis lasting more than 30 minutes

When to go to the Emergency Room

If you feel the child is experiencing an emergency that cannot wait for a return phone call, please do not hesitate to go to the Emergency Room. Be sure to call the on-call physician on

If symptoms occur during normal operating hours for the Sickle Cell Clinic at Pediatric Specialists of Virginia, **please call us first at 571.472.1717**. The nurse will either get you a **clinic appointmen**t ASAP or arrange for an **Emergency Room** visit.

AFTER HOURS: 571.472.1717 will be routed to the Inova Fairfax Hospital operator 703.776.4000 who will page the **on-call pediatric hematologist**. Someone is available to speak with you 24 hours a day.

Emergency Room	Inova Children's Hospital
Emergency Phone Number	703-776-4000 (Inova Fairfax operator)
Address	Inova Fairfax Hospital
	3300 Gallows Road, Falls Church, VA 22042

People you will meet in the clinic

The **front desk staff** will check your child into clinic and make appointments. The **medical assistants** will take vital signs, height, and weight. **Phlebotomists** will draw blood for lab tests. Some results will be available at the time you see the doctor.

Doctors specializing in sickle cell disease are hematologists (blood disorders specialist) who direct the medical care of your child. Sickle cell doctors will work with families to determine the best therapy for the patient. They educate the patient and family about sickle cell disease, and help each individual patient and family manage the patient's sickle cell health. PSV's sickle cell doctors are well-versed in the latest medical advances so that patients and families can benefit from new knowledge and discovery.

Sickle Cell Nurse Coordinator and Educator helps patients and families navigate the complex healthcare system. The Nurse Coordinator and Educator provides medical education, health advice, support for patients and families, and connects patients and families to needed resources, such as other specialists. The Nurse Coordinator and Educator also prepares teenagers for transitioning to adult medical care so that by the early 20s, your young adult will be equipped with the skills and knowledge to be transferred to adult sickle cell care, with parental support in the background.

Nurse Practitioners and Physician Assistants see patients for routine checkups and sick visits. They provide education and continuity of care. They help families understand the importance of health maintenance and assist parents along the way as the child grows.

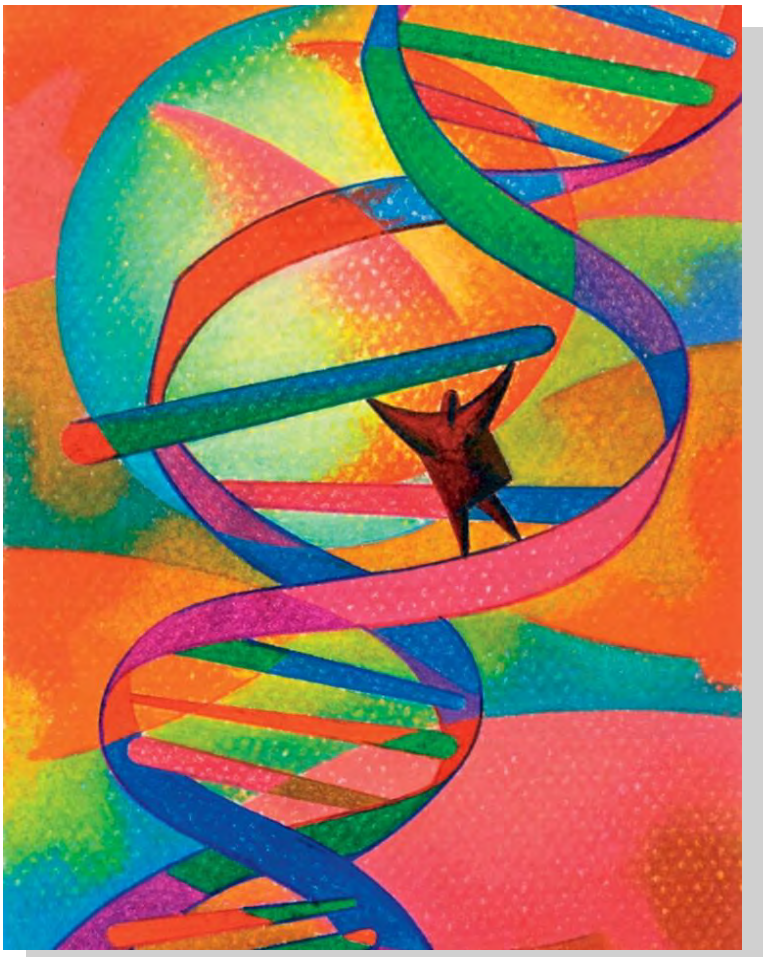
Nurses in the clinic administer treatment and medication when your child is ill and give medical advice on the phone.

Social Worker assesses the needs of patients and families, provides access to social services and community resources, assists with insurance issues, and offers support and counseling throughout the course of the child's care.

Psychologist provides support and counseling for patients to help cope with issues related to their disease or other contributing factors.

About Sickle Cell Disease

- **What is sickle cell disease?**
- **The sickling problem**
- **How is sickle cell disease diagnosed?**
- **Inheritance of sickle cell trait and disease**
- **Common types of sickle cell disease**
- **How children can be affected by sickle cell disease**
- **Other medical complications to monitor as the child grows**

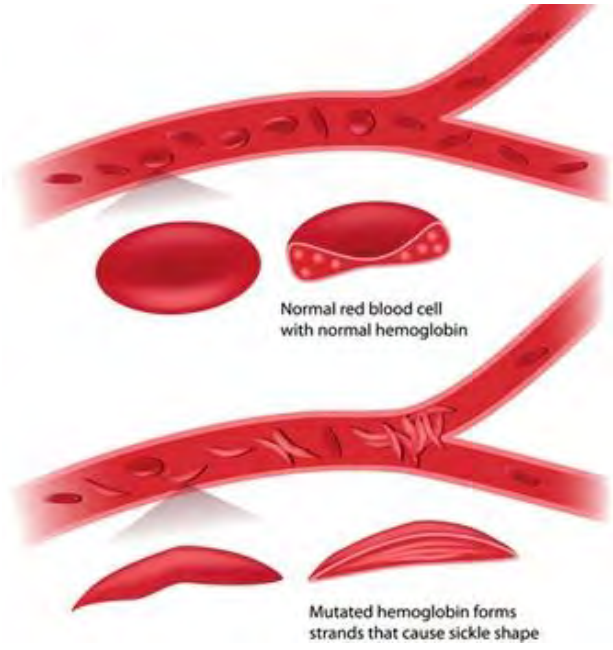


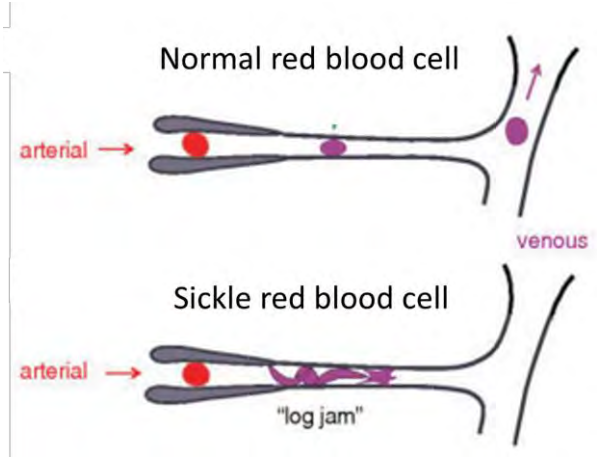
What is sickle cell disease?

Sickle cell disease is a group of inherited red blood cell disorders. Healthy red blood cells are round, flexible, and move easily through blood vessels to deliver oxygen to all parts of the body. Oxygen is carried by hemoglobin, the main component inside red blood cells. In sickle cell disease, mutation in hemoglobin causes the blood cells become hard, sticky, and resemble a C-shaped farm tool called the sickle. Sickle cells break down easily, lasting only a fraction of the lifespan of a normal red blood cell, resulting in a constant shortage of red blood cells, called anemia. Because the C-shaped cells are rigid, when sickle cells travel through small vessels, they get stuck and clog the blood flow, keeping blood and oxygen from reaching parts of the body. This can cause pain in the arms, legs, chest, or back. This also causes chronic damage to all organs of the body, including the brain. Sickle cell disease is a serious condition that can be detrimental to daily life and long-term health. Fortunately, with modern medical advancements, many complications can be prevented, and children with sickle cell disease can thrive!

Sickle cell anemia refers to the most genetically severe forms of sickle cell disease.

Sickle cell disease encompasses of disorders involving sickle hemoglobin

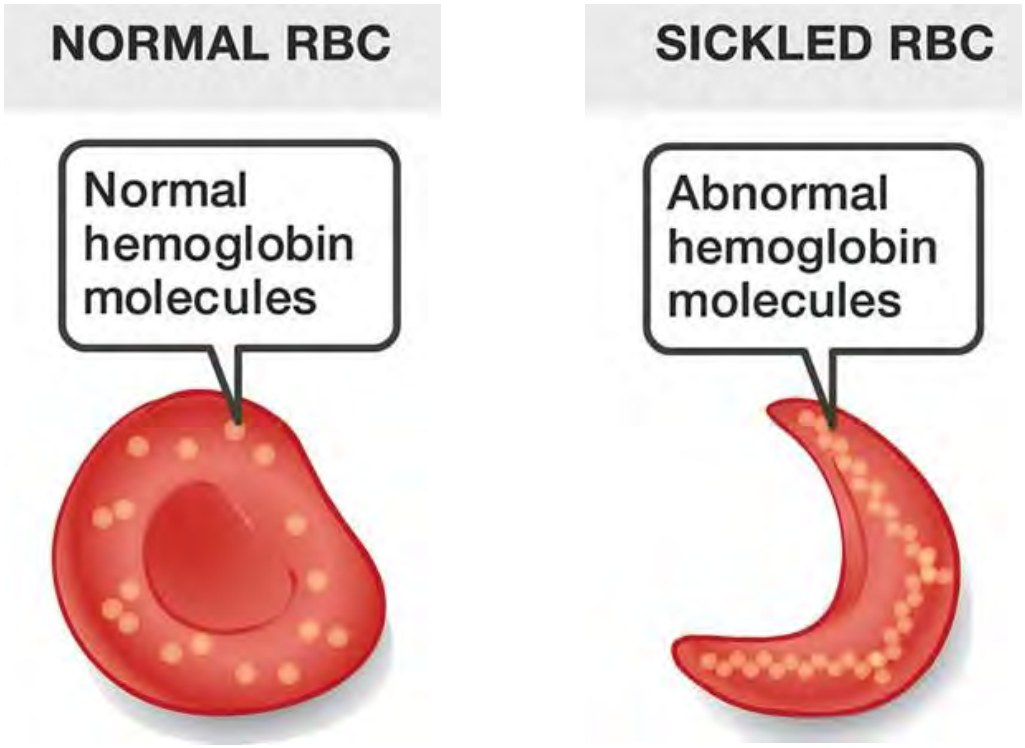




Sickle shaped red blood cells —> block blood flow —> less oxygen —> damage tissue

The Sickling Problem

Hemoglobin molecules carry oxygen (O2) in the blood stream and unload the oxygen to the tissues. The normal hemoglobin is called Hemoglobin A. The abnormal form of hemoglobin in sickle cell disease is called Hemoglobin S. When hemoglobin S molecules are not carrying oxygen, they stack to form chains, and many chains come together to form rods. The rods kink the red cell into the sickle shape. This rigid, non-deformable sickle shape clogs blood flow. **The stacking together of hemoglobin S, called sickle hemoglobin polymerization, is the root of the problems in sickle cell disease.**



Hemoglobin S forms rods

Vaso-occlusion (clogging of blood vessels)

Decreased oxygen delivery

Pain, organ damage

How is sickle cell disease diagnosed?

Early diagnosis of sickle cell anemia is very important. Children who have the disease need prompt and proper treatment.

In the United States, all states mandate testing for sickle cell disease as part of newborn screening.

The baby's blood sample from a heel stick is used for all routine newborn screening tests, including sickle cell disease testing. The test can show whether a newborn infant has sickle cell disease or sickle cell

Newborn screen results are sent to the baby's primary care doctor. This is the doctor you name on the paperwork filled out at the hospital before giving birth. It is important to provide correct contact information to the hospital. This allows your baby's doctor to get the test result as quickly as possible.

If the test shows sickle cell disease, a second blood test is done to confirm the diagnosis. The second test should be done as soon as possible and within the first month of birth. If the second test confirms sickle cell disease, the primary care doctor will send you to a hematologist. This is a doctor who specializes in blood disorders. In Northern Virginia, babies with abnormal hemoglobin test results on the newborn screen are referred to Pediatric Specialists of Virginia.

It is also possible to diagnose sickle cell anemia before birth. This is done using a sample of amniotic fluid or a small piece of the placenta. Amniotic fluid is the fluid in the sac surrounding the baby. The placenta is the organ that attaches the umbilical cord to the mother's womb.

Inheritance of sickle cell trait and disease

Who is at risk for sickle cell disease?
Sickle cell disease affects millions of people in the world. It is most common in people who have families from Africa, South or Central America, Caribbean islands, Mediterranean countries (such as Turkey, Greece, and Italy), India, and Saudi Arabia.

In the United States, sickle cell disease affects about 100,000 people. The disease occurs in about 1 out of every 365 African American births. Sickle cell disease also affects Hispanic Americans and occurs in 1 out of every 16,300 births. Children of parents with sickle cell trait or another abnormal hemoglobin are at risk of sickle cell disease.

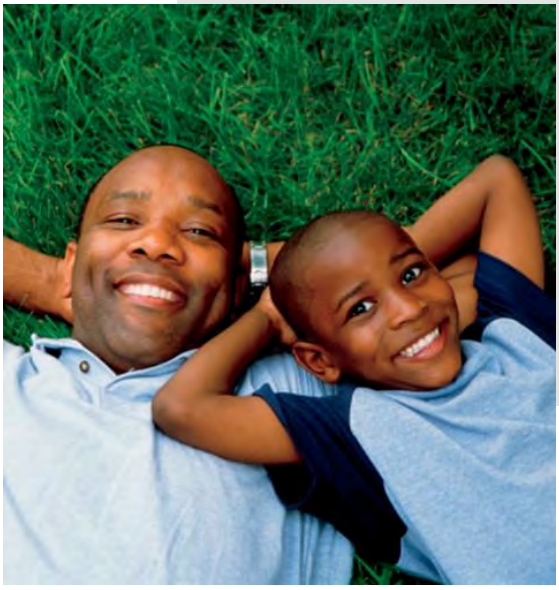
What is sickle cell trait?
Sickle cell trait is different from sickle cell disease. A person with sickle cell trait does not have the disease but has one copy of the abnormal gene that can cause disease. Persons with sickle cell trait can pass the abnormal gene to their children. It takes two abnormal genes to cause sickle cell disease. About 2 million Americans carry this trait. Sickle cell trait occurs in about 1 in 13 African American babies born in the US. About 1/100 Latinos carry sickle cell trait. You can be Caucasian and have sickle cell trait, the prevalence is 0.02%.

Normal hemoglobin is called type A. Sickle hemoglobin is called S. A person with sickle cell trait carries one sickle hemoglobin producing gene (S) inherited from one parent and one normal hemoglobin gene (A) from the other parent. This person has hemoglobin AS and has sickle cell trait. Persons with sickle cell trait can pass the sickle cell gene on to their children.

Sickle cell trait is **NOT** a disease.
Sickle cell anemia **is** a disease.

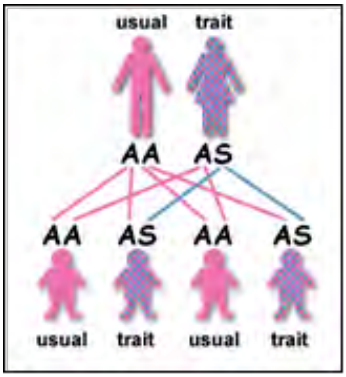
Persons with sickle cell trait generally have no symptoms in daily life, but can have symptoms during extreme exercise in high altitude/low oxygen situations, such as professional athletes doing conditioning at high altitudes, or tourists traveling to high mountainous areas.

How is sickle cell disease inherited?



Everyone has two copies of the hemoglobin gene in every cell in their body, except eggs and sperm, which only carry one copy of each gene. Each person inherits one gene from their mother and one gene from their father. For a person with sickle cell trait, each egg or sperm has either the A gene or the S gene. The genes the baby gets depends on the genes carried by the egg and sperm that joined together.

Sickle cell disease is called a recessive condition, meaning you must have two abnormal hemoglobin genes to have the disorder. Sickle hemoglobin is often shortened to S or Hgb S. If you have only one copy of the sickle hemoglobin, along with one copy of the usual hemoglobin, or Hgb A, you have sickle cell trait, Hgb AS. This is means you "carry" the gene and can pass it on to your children. If your partner also has sickle cell trait or sickle cell disease, then your children can inherit one copy of Hgb S from each parent and end up with two copies of sickle hemoglobin gene, Hgb SS, and have sickle cell disease. **Genes from both parents contribute to the child's sickle cell disease.**



In these diagrams, the usual hemoglobin gene is shown in pink and the sickle hemoglobin gene is shown in blue.

In the diagrams on these pages, the lines coming into each baby show that one gene has come from the mother and one gene has come from the father. In these diagrams, inheritance of the usual hemoglobin gene A is shown by a pink line and inheritance of the sickle hemoglobin gene S is shown by a blue line. **In all the diagrams, you will get the same possibilities if the genes in the mother and father are swapped.**

	A	S
A	AA	AS
A	AA	AS

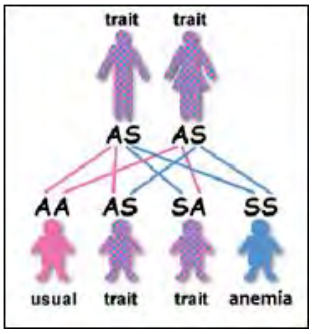
Another way of showing the inheritance of sickle genes is by Punnett Square. One parent's genes are on top and one parent's genes are on in the left column. One gene on top comes together with one gene from the left column to give the baby's genes.

Unaffected and affected individuals

If one parent has sickle cell trait (Hgb AS) and the other does not carry the sickle hemoglobin gene at all (Hgb AA), then none of the children will have sickle cell disease. There is a one in two chance (50%) that any given child will inherit one copy of the Hgb S gene and therefore have the sickle cell trait, Hgb AS. It is equally likely that any given child will inherit two Hgb A genes and be completely unaffected, Hgb AA.

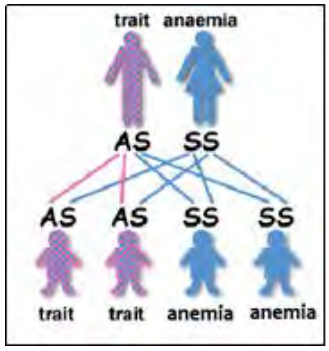
Both parents have sickle cell trait

If both parents have sickle cell trait (Hgb AS) there is a one in four chance (25%) that any given child could be born with sickle cell disease. There is also a one in four chance (25%) that any given child could be completely unaffected. There is a one in two chance (50%) that any given child will get the sickle cell trait.



To get sickle cell disease, a child must inherit the sickle hemoglobin gene S from one parent and a sickle gene S or another abnormal hemoglobin gene from the other parent.

	A	S
A	AA	AS
S	AS	SS



	A	S
S	AS	SS
S	AS	SS

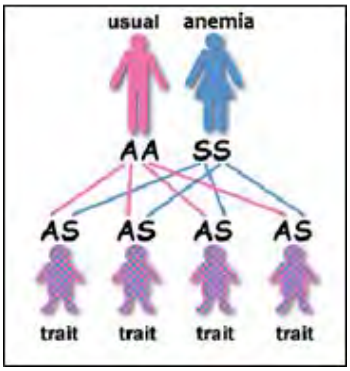
One parent has sickle cell trait and one parent has sickle cell disease

If one parent has sickle cell trait (Hgb AS) and the other has sickle cell disease (Hgb SS), there is a one in two chance (50%) that any given child will inherit sickle cell trait and a one in two chance (50%) that any given child will inherit sickle cell disease. No child will be completely unaffected.

One parent without sickle cell and one parent with sickle cell disease

If one parent has sickle cell disease (Hgb SS) and the other is completely unaffected (Hgb AA), then all the children will have sickle cell trait. None will have sickle cell anemia. The parent who has sickle cell disease (Hgb SS) can only pass one sickle hemoglobin gene to each child.

	A	A
S	AS	AS
S	AS	AS



One parent with sickle cell trait and one parent with another abnormal hemoglobin

	A	S
A	AA	AS
X	AX	SX

Some forms of sickle cell disease result from one parent having sickle cell trait and the other parent carrying another abnormal hemoglobin, such as hemoglobin C or beta thalassemia. In this Punnett square, the abnormal hemoglobin is represented by "X." If one parent has sickle cell trait, then, before having children, the other parent needs to have his/her blood tested for any abnormal hemoglobin, not just hemoglobin S.

To summarize, when two people with sickle cell trait have a baby, there is:

- One in four chance **(1/4, or 25%**) the baby will inherit two sickle cell genes and have the **disease**.
- One in four chance (**1/4, or 25%)** the baby will inherit two normal genes and **not have the disease or trai**t.
- Two in four chance (**1/2, or 50%**) the baby will inherit one normal gene and one sickle cell gene. The baby will not have the disease, but will have sickle cell trait, like the parents.

Understanding chance

To help you to think about chance, it can be useful to consider examples that we can all understand. When a woman has a baby, there is a one in two (50%) chance that the baby will be a girl and a one in two (50%) chance that the baby will be a boy. Although over the whole population there are about equal numbers of men and women, within any family there may be all girls, all boys, or a mixture of both. It is important to understand that the same chances apply to each pregnancy.

The one in two chance applies to each pregnancy. The1/2 chance tells you that nature will choose one out of two different possibilities. If the chance is one in four, there are four different possibilities, and the outcome will be one of these. This is very important in understanding what sort of children you will have if you carry the sickle hemoglobin gene. Just as there are families with all boys and families with all girls, when both parents have sickle cell trait, or one parent has sickle cell trait, and another parent has another abnormal hemoglobin, all of the children can have sickle cell disease, or all of the children can be unaffected, or there can be any combination of children with sickle cell trait, sickle cell disease, or totally unaffected status. Having had a child with sickle cell disease does not ensure the next child will not. Similarly, having had a child without sickle cell disease does not mean the next child will also not have disease. Each pregnancy is a new round of chances.

How can I find out if I carry sickle cell trait?

A simple blood test called **"hemoglobin electrophoresis"** can be done by your doctor or local lab. It requires that blood be taken from a vein. This is not a routine test and must be requested. The test will tell if you are a trait carrier or if you have any abnormal hemoglobin. The test will not only detect Hgb S, but other abnormal genes that can cause sickle cell disease like Hgb C or beta thalassemia.

Pregnancy planning

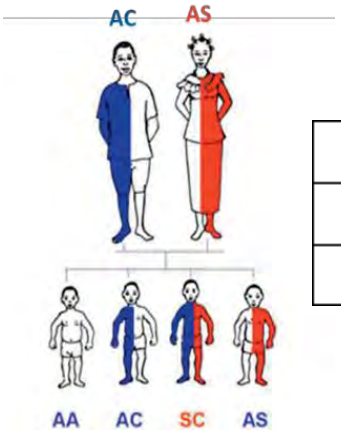
If you are planning to have a baby, you and your partner can have genetic testing at medical centers. A genetic counselor can refer a couple for DNA testing and a doctor can order hemoglobin electrophoresis. Either a genetic counselor or a hematologist can correctly interpret the results and discuss possible risks to your children.

Common types of sickle cell disease

Hgb SS: People who have this form of sickle cell disease inherit two sickle cell genes ("S"), one from each parent. This is the most common form of sickle cell disease and the most severe. The standard of care is to treat all children with Hgb SS with the medication **Hydroxyurea** starting in infancy to prevent sickle cell complications. Children treated with Hydroxyurea have good quality

	A	S
A	AA	AS
S	AS	SS

Hgb SC: People who have this form of sickle cell disease inherit one sickle cell S gene and one gene for an abnormal type of hemoglobin called "C." Hgb SC is usually a milder form of sickle cell disease.



	A	S
A	AA	AS
C	AC	SC

Hgb S beta thalassemia: People who have this form of sickle cell disease inherit one sickle cell gene and one gene for beta thalassemia, another type of anemia. There are two types of beta thalassemia, "beta 0 (zero)" and "beta + (plus)." Those with Hgb S beta 0 thalassemia have no Hgb A and have a severe form of the disease similar to Hgb SS disease. Sickle beta 0 thalassemia is treated the same as Hgb SS. People with Hgb S beta + thalassemia have some Hgb A and have a milder form of the disease.

	A	S
A	AA	AS
beta thal	A/beta thal	S/beta thal

Hgb SD, Hgb SE, Hgb SC^{Harlem}, Hgb SO^{Arab}: People who have these forms of sickle cell disease inherit one sickle cell gene and one gene of an abnormal type of hemoglobin. The severity of these rarer types of the disease varies. The disease symptoms and complications of Hgb SD, Hgb SC^{Harlem}, and Hgb SO^{Arab} are similar to those of a person with "SS," while Hgb SE is usually milder, similar to Hgb S beta+ thalassemia.

	A	S
A	AA	AS
X	AX	SX

Sickle cell disease, type Hgb SC

Sickle cell disease type Hgb SC is the second most common type of sickle cell disease. It occurs when a hemoglobin S gene is inherited from one parent and a hemoglobin C gene is inherited from the other parent. Hgb SC is generally a milder form of the disease. There is only one hemoglobin S gene, and while the hemoglobin C by itself is harmless, when hemoglobin C is combined with hemoglobin S, it can participate in hemoglobin polymerization to cause sickling. **Hemoglobin SC is sickle cell disease.**

A patient with Hgb SC can have all of the same symptoms as the more severe forms of SCD, including pain, but usually much less frequently or not at all. However, every patient is different. Some patients are minimally affected by the disease while others have more serious complications. It is important to continue regular follow up appointments with a hematologist, no matter how mild the symptoms. The same precautions should be taken to prevent symptoms, including staying well hydrated, avoiding extreme temperatures, avoiding low oxygen situations, and adhering to proper infection prevention.

Eye problems can occur in all types of sickle cell disease but is more frequent in Hgb SC. Sickle cell retinopathy and retinal detachment can cause blindness; this is a medical emergency. It is important to have the eyes checked regularly by an ophthalmologist (not an optometrist who checks the vision). The ophthalmologist can visualize the retina and treat any complication. It is recommended to have a retinal exam once a year starting at age 10.

Splenic sequestration is another common symptom for Hgb SC. This occurs when sickle cells become trapped in the spleen and the spleen is enlarged. Severe splenic sequestration can be life threatening, but not if detected early and properly monitored or treated.

If a patient with Hgb SC has frequent **pain episodes** or **acute chest syndrome**, treatment with **Hydroxyurea** is very effective. Hydroxyurea does not appear to prevent eye problems or splenic sequestration, but it does prevent pain and acute chest syndrome. Hydroxyurea leads to a much improved quality of life with decreased hospitalizations and fewer missed school days.

How children can be affected by sickle cell disease

Possible complications in children with sickle cell disease :

Anemia (A low number of red blood cells or low hemoglobin): A normal blood cell has a lifespan of 3 to 4 months. A sickle cell has a life span of 1 to 2 weeks. Because of their sickled shape and rigidity, they are destroyed easily. The body constantly makes new red blood cells but not as fast as they are destroyed. Because of this, a person with sickle cells has fewer red blood cells and lower hemoglobin, which is called anemia. People with anemia may tire easily. Anemia means that there are not enough healthy red blood cells to carry oxygen throughout the body.

Typical hemoglobin level for **healthy non-sickle cell patient = about 12–14**
Typical hemoglobin level for **untreated Hgb SS or Hgb S beta O = about 6–8**
Typical hemoglobin level for **Hgb SC or Hgb S beta+ = about 10–11**

Symptoms of Anemia:

Tiredness
Irritability
Dizziness or lightheadedness
Fast heart rate
Difficulty breathing
Pale skin color
Slow growth

*When red blood cells die, the hemoglobin is broken down into something called bilirubin, which is yellow. The excess bilirubin can cause the whites of the eyes and skin to look yellow; this is called **jaundice**.

Hand-and foot syndrome or dactylitis: Babies with sickle cell disease may have pain and swelling in their hands or feet. This is usually the first noticeable symptom of sickle cell disease in babies.

Aplastic crisis: Children with sickle cell disease may stop making red blood cells for a short time and hemoglobin may fall very low. Signs include paleness, less activity than normal, fast breathing, and fast heartbeat. Aplastic crisis can occur when the child is sick with infection. A child with these signs must be seen quickly by a doctor.

Pain crisis: This is the most common symptom of sickle cell disease**.** Pain mostly in the arms, hands, legs, feet, or back happens when sickle cells plug blood vessels and block the flow of blood. This is called vaso-occlusion. Pain due to vaso-occlusion is a painful episode, or crisis, also called vaso-occlusive crisis, or VOC.

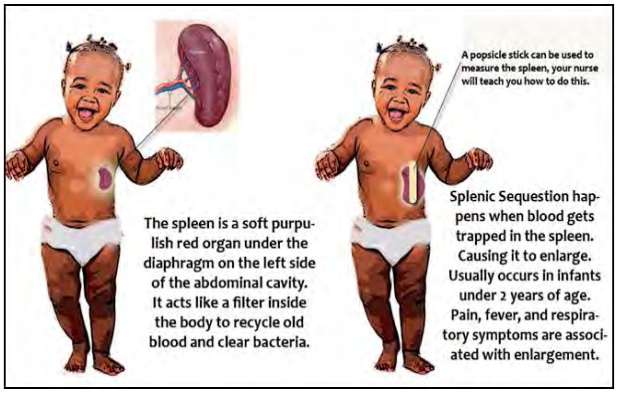
How to prevent pain crisis:

- Keep the child properly hydrated.
- Try not to let the child get too hot or cold (avoid temps <40F or >90F) for prolonged time periods.
- Avoid places of high altitude (flying for many hours, high mountain climbing, or being in cities with traveling to places with very high altitude) especially if the child is sick, even with the common cold.
- Avoid places or situations with low oxygen (extreme exercise or prolonged physical exertion).
- **Give your child Hydroxyurea.**

Splenic sequestration crisis: The spleen is the organ that filters blood. In children with sickle cell disease, the spleen can enlarge rapidly from trapped red blood cells, and the amount of red blood cells in the circulation can become very low. This condition is called "splenic sequestration crisis" and can be life-threatening. Babies and young children are at greatest risk for splenic sequestration, both in Hgb SS and Hgb SC disease. Complications can develop as young as 2 months of age, but usually occur between ages of 6 months to 5 years. Parents of children with sickle cell disease should learn how to feel and measure the size of their child's spleen.

Symptoms of splenic sequestration:

Sudden weakness
Pale lips
Fast breathing
Abdominal pain on left side of abdomen
Fast heart beata
Refusal to walk



Acute chest syndrome: This can be a life-threatening condition in sickle cell anemia. It is similar to pneumonia. The condition is caused by a combination of infection and sickle cells trapped in the lungs. This leads to decreased oxygen delivery to the bloodstream. Acute chest syndrome is a major problem caused by sickle cell disease and can happen at all ages, including toddler age. People who have this condition usually have chest pain and fever. They also often have abnormal chest X ray and need supplemental oxygen. Sometimes transfusions are necessary to stop acute chest syndrome from getting worse. Over time, lung damage from repeated acute chest syndrome may lead to chronic lung disease and pulmonary arterial hypertension. **Hydroxyurea is highly effective in preventing acute chest syndrome.**

Bacterial infections: The child with sickle cell anemia is at increased risk for serious infections of certain types of bacteria, such as sepsis (a blood stream infection), meningitis, and pneumonia. The risk of infection is increased because the spleen does not function normally.

How to pevent infections:

- **Vaccinations:** Keep updated with all scheduled shots at your pediatrician's office.
- **Additional pneumococcal vaccines, previously Pneumovax 23** at 2 years and then 5 years later, now being replaced by **Prevnar 20 (PCV 20)**.
- **Meningococcal vaccine (ACWY)** to prevent meningitis is given at age 9 months and 12 months (for Menveo) or age 2 (for Menactra) and then every 5 years.
- **Meningococcal** B Vaccine is given at age 10 (2 dose series), then every 3 years.
- **Flu vaccine:** Every year after 6 months of age (all family members should also be vaccinated annually).
- **COVID vaccination and booster** to begin at age 6 months.
- **Penicillin** (twice daily) for children starting at birth–5 years old or when vaccinations are complete, to prevent bacterial infection while the child gains immunity through vaccination.

Stroke: Stroke prevention is an important part of sickle cell treatment. Stroke happens when arteries in the brain are narrowed and/or blocked by sickled red blood cells. For children with genetically severe forms of sickle cell disease, including Hgb SS and Hgb S beta 0, a screening test called a Transcranial Doppler Ultrasound, or TCD, is done aannually starting at age 2. If this test is abnormal, your child is at risk for a stroke. In the past, these children required regular blood transfusions to prevent stroke. However, now we know that regular use of Hydroxyurea may be just as good in preventing strokes in these patients. Stroke can lead to lifelong disabilities and learning problems. Hydroxyurea prevents abnormal

Transcranial Doppler

Photo Credit: http://www.ihtc.org/patient/blood-disorders/sickle-cell-disease/stroke-intervention/ and https://en.wikipedia.org/wiki/

A child with any of these signs must be seen by a doctor immediately:

Seizure
Difficulty with speech
Weakness of the arms and legs
Loss of consciousness

Strokes are much less frequent in milder forms of sickle cell disease, therefore, children with Hgb SC or Hgb S/beta+ thalassemia do not need routine TCDs.

Other medical complications to be aware of as the child grows:

Eye Problems: Sickle cells can also clog the small blood vessels that deliver oxygen-rich blood to your eyes. The retina is a thin layer of tissue at the back of your eyes that takes the images you see and sends them to your brain. Decreased blood flow to the retina can cause bleeding, abnormal blood vessel formation called proliferative **sickle retinopathy**, and detachment of the retina leading to blindness. Starting at age 10, patients with any type of sickle cell disease should receive annual monitoring retina exams, so they can receive treatment early if abnormalities are detected. Retina monitoring is performed by an eye specialist called an ophthalmologist or a retina specialist (not an optometrist who mainly manages need for glasses or contacts) who conducts a thorough examination of your child's eyes and perform laser surgery if abnormalities are detected.

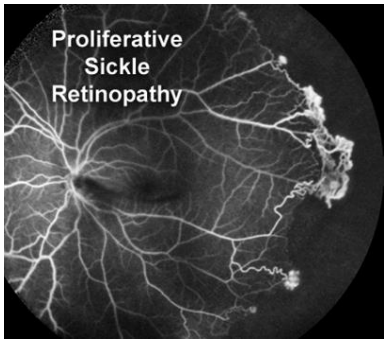


Photo Credit:
http://retinagallery.com/displayimage.php

Kidney problems: Impairment of blood flow in small blood vessels in the kidney caused by sickling of red blood cells can cause protein in the urine, blood in the urine, and tissue death in parts of the kidney, which is called papillary necrosis. Annual urine screening begins at age 5. If protein is detected in the urine, it may be a sign of kidney damage and referral to a nephrologist, or kidney specialist may be necessary. The kidneys of patients with sickle cell disease are not able to reabsorb water as well, and the urine is more dilute. People with sickle cell disease also produce more urine. This is another reason why hydration is important in sickle cell disease.

Gallstones: When red blood cells are broken, they release the hemoglobin that is normally inside the red blood cells into the blood stream. The body breaks down hemoglobin into a molecule called bilirubin. Too much bilirubin in the body can cause stones to form in the gallbladder. Gallstones may cause pain that lasts for 30 minutes or more in the upper right side of the abdomen, under the right shoulder, or between the shoulder blades. The pain may happen after eating fatty meals. People who have gallstones may have nausea, vomiting, fever, sweating, chills, clay-colored stools, or jaundice. Treatment is surgical removal of the gallbladder, usually done laparoscopically.

Silent cerebral infarcts (SCI) or silent strokes: These are abnormal findings on brain MRIs that are believed to be due to small blood vessels not delivering adequate oxygen to brain tissue. Unlike the usual overt strokes, no symptoms are associated with silent strokes (thus the name), and one cannot tell if a child has silent strokes without doing a brain MRI. Before children were treated with Hydroxyurea, up to 30% of children with sickle cell disease had silent infarcts by school age. Hydroxyurea may reduce the occurrence of silent infarcts, but this has not been proven.

Priapism: Males who have sickle cell anemia may have painful and unwanted erections. This condition is called priapism (PRI-a-pizm). It happens because the sickle cells block blood flow out of an erect penis. Repeated priapism can damage the penis. **You must notify your doctor immediately if this happens.**

A**vascular necrosis (AVN)**: Due to lack of sufficient oxygen, the tissues of some bones in the body can die, causing pain and limitation of range of motion. Common places of AVN are the hip joint (femoral head), shoulder joint (humoral head), and knees, as well as other less common sites. AVN is not common in the age of hydroxyurea.

Leg ulcers: Decreased blood flow to areas on the body with thin skin and not much fat under the skin, such as lower legs and ankles, can cause chronic ulcers that are difficult to heal. Hydroxyurea does not help leg ulcers. Leg ulcers are uncommon in children.

Delayed growth and puberty: Children who have sickle cell anemia who are not treated often grow more slowly and reach puberty later than other children. Starting Hydroxyurea at an early age may help growth and development.

Cognitive Problems: People with untreated sickle cell disease can develop cognitive (thinking) problems that may be hard to notice early in life. Some children may require cognitive testing and an individualized education plan (IEP) to help them be successful in school. We know Hydroxyurea helps with blood flow in the brain. Children treated with Hydroxyurea early in life tend not to have school problems and can achieve like other children, but it remains to be seen how much Hydroxyurea helps cognitive function in children.

Treatment

The following pages contain information about FDA-approved treatments and NIH guidelines.

- Hydroxyurea
- Other Medications: Voxelotor (Oxbryta™)
- Transfusions
- Immunizations and preventive antibiotics
- Hydration
- Pediatric Sickle Cell Clinic Health Maintenance Schedule

Can sickle cell disease be cured?

- Bone Marrow Transplant
- Gene Therapy and Gene Editing

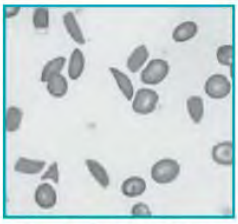
Hydroxyurea

Hydroxyurea is a medication that improves the symptoms of sickle cell disease. It is not a cure for sickle cell disease, but can help prevent many of the complications of the disease, including:

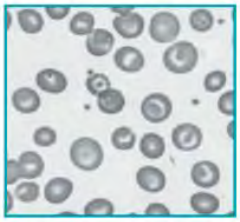
- Acute chest syndrome
- Pain crises
- Trips to the hospital
- Frequency of blood transfusions

Sickle cell patients who take Hydroxyurea live longer. It is safe and beneficial to start Hydroxyurea in infancy.

Hydroxyurea must be taken as directed to help reduce some of the complications. It works well if taken every day. It takes some time for the drug to take full effect. Hydroxyurea is taken orally in capsule form. The capsule is opened into food for young children who can't swallow the capsule.



Blood cells before taking hydroxyurea



Blood cells after taking hydroxyurea

From St. Jude's Hydrozureas Guide for Parents

Hydroxyurea is prescribed for anybody with the severe forms of sickle cell disease, including Hgb SS, Hgb S beta 0, Hgb SD, Hgb SC^{Harlem}, Hgb SO^{Arab}, etc.

Hydroxyurea is also given to people with milder forms for sickle cell disease, such as Hgb SC or Hgb S beta+, if they have significant symptoms, such as pain or acute chest syndrome.

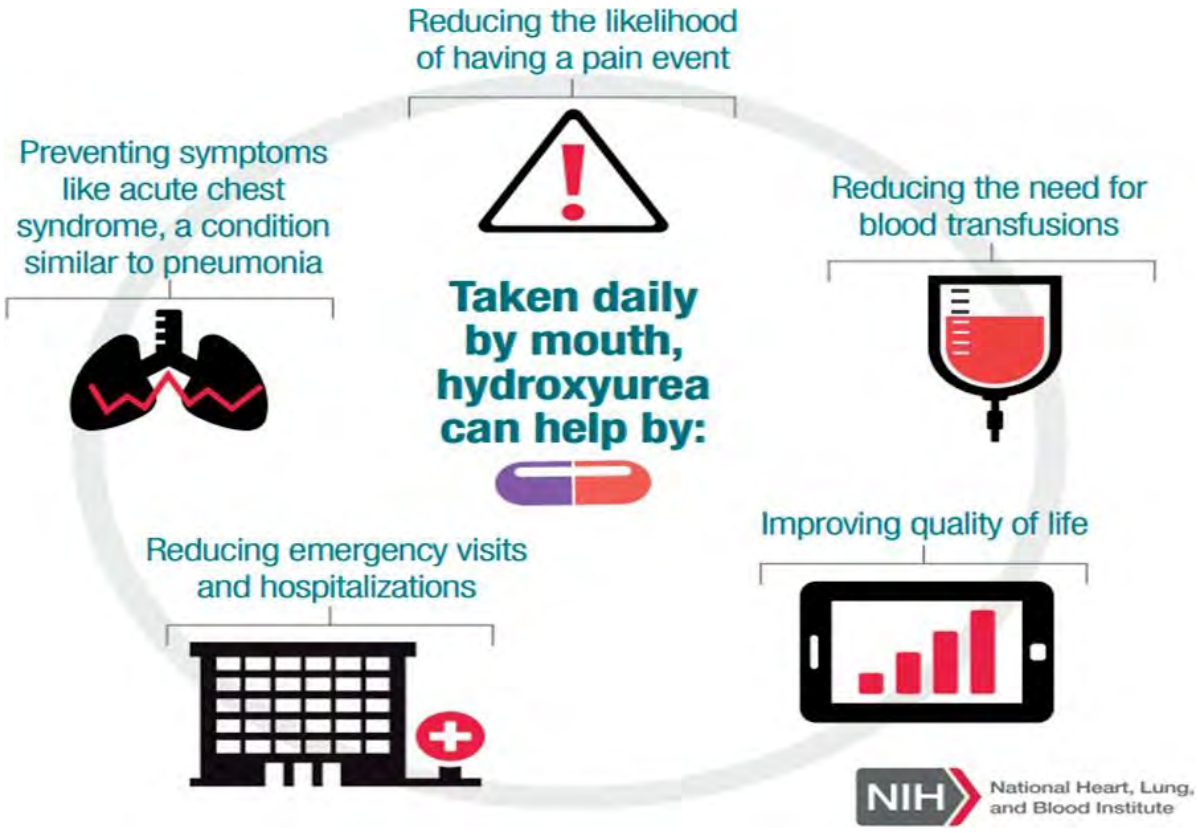
How does Hydroxyurea work?

Hydroxyurea helps red blood cells stay round and flexible, so they are able to travel more freely in the blood stream. The drug works by **increasing the amount of fetal hemoglobin (Hgb F)** in red blood cells. Hgb F is produced naturally in fetuses but turns off after birth. Hydroxyurea turns Hgb F back on. Hgb F prevents Hgb S from sticking together and sickling or red blood cells, thus is protective against symptoms caused by Hgb S. The higher the Hgb F, the better is the sickle cell patient. The higher percentage of Hgb F, the lower the percentage of Hgb S (sickle hemoglobin). People with sickle cell disease who haves higher level of Hgb F usually have fewer complications.

Hydroxyurea is a highly effective medicine for sickle cell disease. It is a safe medicine to take as prescribed by medical specialists treating sickle cell disease. When the dose is too low, it does not increase Hgb F enough to protect against sickle cell complications. When the dose is too high, the medicine can decrease the production of blood cells. For this reason, blood cell counts are checked regularly to optimize the dose of Hydroxyurea. Dose adjustments are expected as children grow.

Hydroxyurea in sickle cell disease

In 1995, a large collaborative study involving 21 different sites in the US and Canada treated sickle cell patients with either placebos or Hydroxyurea. The effects of Hydroxyurea were so beneficial that the study was terminated early, and all patients were given Hydroxyurea. In 2010, two large studies showed that **adult sickle cell patients who took Hydroxyurea for many years lived longer** than patients who did not take Hydroxyurea. In 2011, the Baby HUG study demonstrated that **babies with sickle cell disease who were treated with Hydroxyurea had fewer complications than babies who were given placebo and that Hydroxyurea was safe to use in babies.**
The NIH (National Institute of Health) guideline is to start Hydroxyurea in infancy.

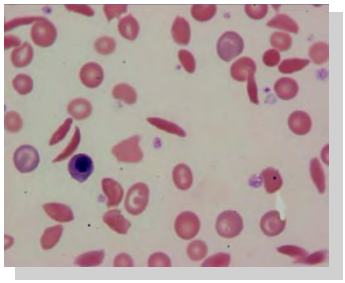


It is important to remember, Hydroxyurea is not a cure. If you/your child stops taking it, the red blood cells **will begin sickling again. Even if you/your child feels great or has never had issues related to sickle cell disease, remember, it is because the Hydroxyurea is working! Do not stop giving your child Hydroxyurea! **

Improvement with Hydroxyurea over time if starting after infancy

No Hydroxyurea

- Sickle cells
- Small RBCs
- Anemia–not many red cells
- Low Hgb F

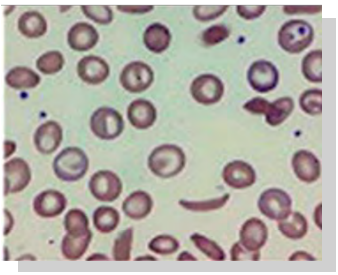


Symptoms:

- Fatigue
- Pain
- Acute chest

On Hydroxyurea 2 Months

- Larger RBCs
- Hgb F increasing

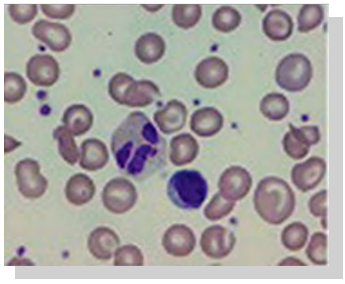


Patient has:

- More energy
- Less pain
- Less frequent acute chest

On Hydroxyurea 5 Months

- More red cells
- No sickle cells
- High Hgb F 20-30%

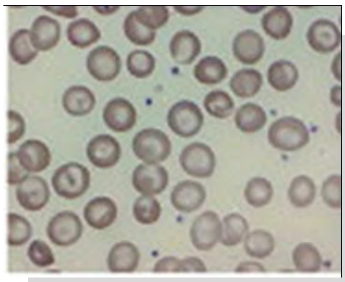


Patient has:

- Increased energy
- No pain
- No acute chest
- Feels good like everybody else

On Hydroxyurea 2 Years

- Normal appearing blood
- Large RBC size
- High Hgb F ~30% or higher



Patient has:

- Good energy
- Pain-free
- No acute chest
- Can't tell he/she has sickle cell disease

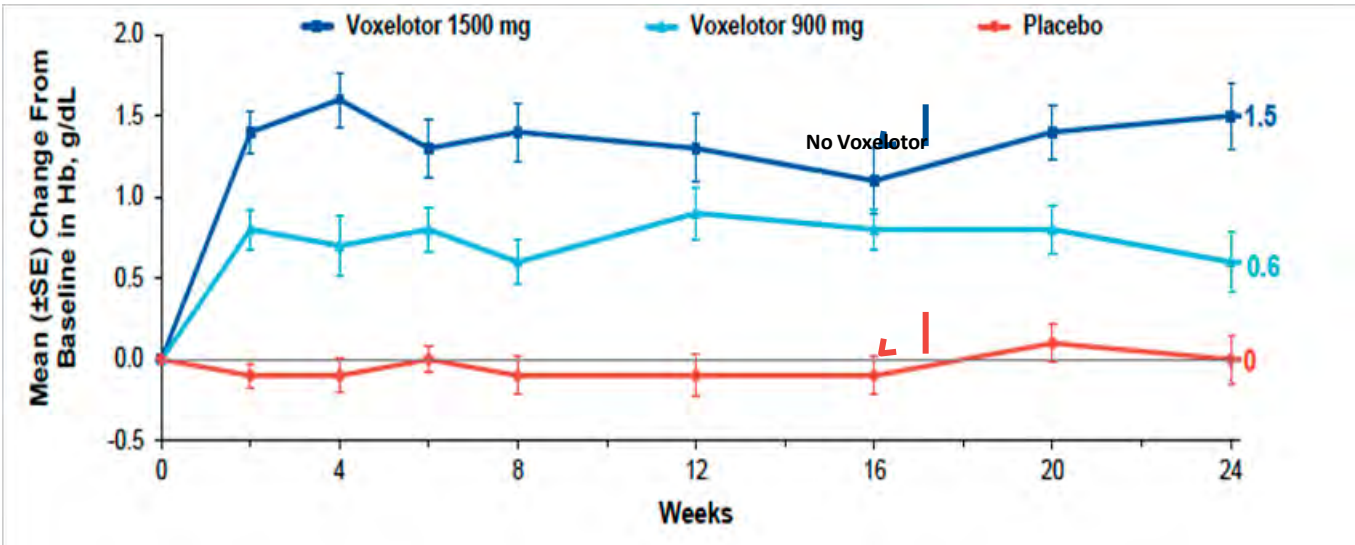
Babies' red blood cells start out beautiful like this, and we want to keep them this way by starting Hydroxyurea before sickling begins.

Voxelotor (Oxbryta™)

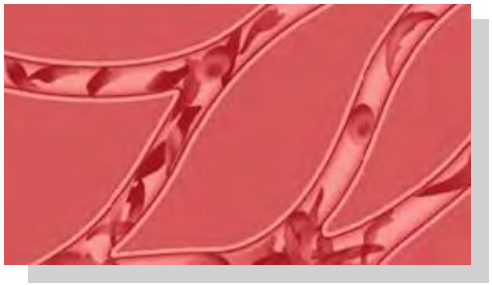
Voxelotor, also known as Oxbryta, is an oral medication approved by the FDA in November 2019 for patients with sickle cell disease who are 4 years and older. This medicine raises hemoglobin in patients with sickle cell disease. It also decreases the breakdown of red blood cells. However, unlike Hydroxyurea, Voxelotor does not prevent pain or acute chest syndrome or other sickle cell complications, and it may decrease oxygen delivery to tissues even if hemoglobin number is higher. Voxelotor can be taken together with hydroxyurea in some patients to further improve anemia. Some patients do not benefit from Voxelotor.

The graph below shows that hemoglobin is higher in patients who took voxelotor (blue lines) compared to those who did not take voxelotor (red line).

Higher Hgb on Voxelotor



Blood transfusions



Blood transfusions are used to treat some sickle cell complications, including difficulty breathing, aplastic crisis, acute chest, splenic sequestration, and strokes. This procedure involves transferring of red blood cells from one person to another. Transfusing normal red cells to a sickle cell patient may help deliver oxygen to the body and unblock vessels.

A sickle cell patient often needs transfusions due to:

Stroke: When a stroke occurs, the brain suffers damage when blood circulation is blocked to a portion of the brain. Transfusions are given to prevent further strokes and brain damage.

Acute chest syndrome: As anemia worsens, breathing is hard and often the oxygen level in the body is low. At these times, a blood transfusion may be needed to help oxygen delivery.

Aplastic Crisis: Since sickle cells break down so quickly and constant replenishment of new red blood cells is need to keep hemoglobin up, when sickle cell patients shut down production of new red blood cells, their red blood cell count and hemoglobn can quickly decrease to dangerously low levels. A blood transfusion of normal red blood cells will help keep hemoglobin level up until they recover.

Abnormal Transcranial Doppler Ultrasound (TCD): This test records sound waves that indicate blood flow. A very fast blood flow indicates an increased stroke risk. Hydroxyurea is very effective in decreasing TCD velocity, but if abnormalities persist, blood transfusions may be needed to decrease the risk of strokes, especially when abnormalities are seen on brain MRI/MRA.

Benefits of transfusion:

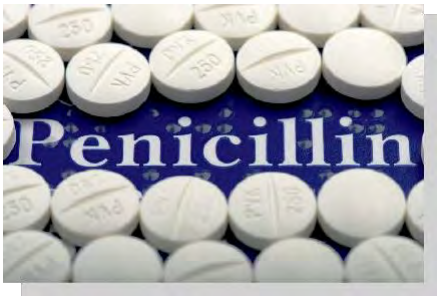
- Allows normal hemoglobin to deliver more oxygen in the body, prevents blockage of blood vessels.
- Suppresses the production of sickle cells since transfused blood cells live longer than sickle cells.

Complications of transfusion:

- Development of antibodies to transfused red blood cells, making matching blood for future transfusions difficult.
- Iron overload causing liver and other organ damage.

Infection prevention

Penicillin



Children with sickle cell disease are more susceptible to blood infections (septicemia) of certain types of bacteria due to the spleen not working properly. The spleen helps to fight against infection by filtering out bacteria and producing antibodies.
Damage to the spleen occurs early in life due to sickle cell disease. Scientific studies indicate that penicillin can prevent fatal cases of septicemia. When given two times each day, penicillin can kill bacteria before they grow in the blood and cause life-threatening septicemia.

Penicillin should be started once sickle cell anemia is diagnosed by newborn screen and stopped by age 5 after the child has been fully vaccinated.
Penicillin or its equivalents, such as Amoxicillin, is available in liquid, tablet, and chewable forms. The liquid penicillin must be stored in the refrigerator and be discarded every 14 days. Penicillin tablets should be stored in a cool, dry place and may be kept as long as 4 months.

Pneumococcal and Meningococcal Vaccines

Children with sickle cell disease are susceptible to infections, especially the types of bacteria called Pneumococcus and Meningococcus. Your child should be immunized against as many strains of the Pneumococcal bacteria as possible. The pneumococcal vaccine series administered by the primary pediatricians to all children is called Prevnar, and is given at 2, 4, and 6 months, as well as a booster. It is very important that your child receives the Prevnar series on time. The sickle cell clinic gives an additional pneumococcal vaccine, either Pneumovax 23 or Prevnar 20 to children with sickle cell disease after the primary Prevnar series. It is important that your child receives these immunizations.

There are 2 types of Meningococcal vaccines—one against serotypes A,C,W,Y and another against serotype B. Menactra or Menveo are for serotypes A,C,W,Y. which is given to all children but given to children with sickle cell disease at a younger age with regular boosters. Meningococcal B vaccine is given at age 10 as an initial 2 dose series then as single doses every 3 years.

Influenza and COVID Vaccines: When individuals with sickle cell disease get viral infections, such as the flu or COVID, they can become much sicker than other people. Therefore, it is recommended that all children receive the flu vaccine every year and the COVID vaccine/booster as directed by the CDC. All family members should also stay up to date with flu and COVID vaccines.

Hydration

With sickle cell disease, one of the best things you can do for yourself is drink water to stay well hydrated.

Carry a water bottle with you and keep drinking water throughout the day.

Use the chart to the right to guide you on the amount of water you should drink every day.

Dehydration (not having enough water in the body) can be caused by:

- Sweating in hot weather or during exercise
- Diarrhea or vomiting
- Frequent urination (sickle cell disease damages the kidneys, causing them to produce more urine than usual)
- Drinks with high caffeine (soda, energy drinks) and alcohol cause dehydration and are not a good idea.

Body Weight	Number of 8 oz. glasses of water needed
10 pounds	2-3
25 pounds	4-6
30 pounds	5-8
45 pounds	6-9
55 pounds	7-10
75 pounds	8-11
100 pounds	9-13
130 pounds	10-15
150 pounds	11-17
175 pounds	12-18

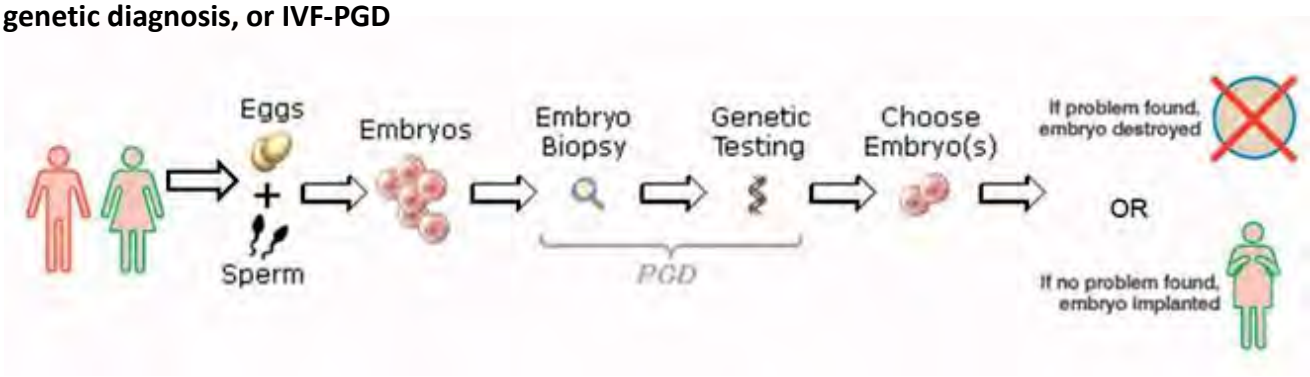
Tired of water? Add these to your hydration routine:

- Fruit
- Milk
- Juice
- Crystal Light drink mix
- Soup
- Gatorade
- Popsicles

Can sickle cell disease be prevented?

Sickle cell anemia is a genetic disease, meaning that it is inherited from parents who are carriers. People who are carriers, or who have the trait, and are planning to have children should consider genetic counseling. A hematologist or counselor can help you understand your chances of having a child with disease and explain the choices available to you. One way to ensure a baby does not have sickle cell disease if both parents are carriers is through in-vitro fertilization. Sperm and eggs are collected and fertilization occurs in the laboratory. Physicians can test an embryo to determine if the embryo has sickle cell disease or not. The embryos that do not have sickle cell disease are then implanted into mom's uterus to develop into a baby. This process is called :

in vitro fertilization/Preimplantation genetic diagnosis, or IVF-PGD



Can sickle cell disease be cured?

Bone Marrow Transplant

A bone marrow transplant can cure sickle cell anemia. It works best in young patients who have a full sibling donor. These are called **HLA-identical** or **match related donor (MRD)** transplants. Patients with severe forms of sickle cell disease, including Hgb SS or Hgb S/beta0 thalassemia, are eligible for transplant if a HLA-matched sibling is available. Transplants can also be done in some situations using donors other than a matched sibling. Transplants using half-matched donors, including parents or siblings with half matched HLA types, are called haplo-transplants.

These are the ideal criteria for bone marrow transplant:

- Bone marrow donor is a full sibling.
- Sibling is a full HLA match for bone marrow.
- Donor cannot have sickle cell disease but can have sickle cell trait.

Umbilical Cord Blood Banking

If you have a child with sickle cell disease and you are pregnant again with a full sibling of the child with sickle cell disease (same mother and same father), it is possible that the new sibling is a full HLA match with the child with sickle cell disease and can be a donor for curative transplant for the affected sibling in the future. The umbilical cord contains stem cells which can be used together with bone marrow cells to augment transplant in the future. Cord blood banking is a way of processing and storing newborn stem cells from umbilical cord, which is otherwise discarded. The stem cells are frozen and can be used in the future to help boost the number of stem cells in transplantation for full siblings with sickle cell disease. After the new baby is born, we will test the baby to see if the baby is HLA matched with the sibling with sickle cell disease. If so, cord blood is already stored. If not, the cord blood can be discarded. A cord blood bank called ViaCord has a program called Sibling Connection. The Sibling Connection program allows for free storage of cord blood for up to 5 years for a sibling of a child with a proven genetic disorder, such as sickle cell disease.

For more information, visit https://www.viacord.com/cord-banking/sibling-connection or call 1-866-668-4895.

THE SIBLING CONNECTION

ViaCord's Newborn Stem Cell Donor Program

Novel curative therapies

Gene Therapy

In December 2023, FDA approved gene therapy for sickle cell disease. Stem cells are collected from the bone marrow of the person with sickle cell disease. A gene for Hgb A is inserted into these stem cells, and the genetically modified stem cells are infused back into the patient to make red blood cells that contain some Hgb A, similar to sickle cell trait.

Gene Editing/CRISPR

In December 2023, FDA also approved gene editing for sickle cell disease. Stem cells are collected from the bone marrow of the person with sickle cell disease, and a technique called CRISPR is used to "turn on" the Hgb F gene to make red blood cells have more Hgb F without the need for Hydroxyurea. Ongoing studies are using gene editing to “correct” the Hgb S gene back into Hgb A gene.

Both gene therapy and gene editing/CRISPR are auto-transplantation processes and require clearing the body of the original bone marrow before infusion of the newly modified stem cells, a step that has associated toxicities. These genetic therapies are options for patients without donors for conventional bone marrow transplant.

Clinical Trials

Clinical trials are always taking place. To find out more about which trials are going on to find new treatments for sickle cell, visit www.clinicaltrials.gov

A child cured of sickle cell after a BMT with his sibling as a donor.
Photo credit: http://curesicklenow.org/more-stars/http://curesicklenow.org/more-stars/

Monitoring lab results

- **How is sickle cell disease blood work different?**
- **What lab work is monitored on Hydroxyurea treatment?**
- **What do lab results mean?**

Blood counts in sickle cell anemia

(Sickle cell "anemia" includes Hgb SS, S beta 0 thalassemia, Hgb SC^{Harlem}, SO^{Arab}, etc., which are genetically severe forms of sickle cell disease)

Labs at clinic visits:

CBC (complete blood count), **retic** count (new RBCs), **Hemoglobin F %**

Component	No SCA	SCA before HU	SCA after HU	Look for these changes in blood work
WBC 4.29 - 11.20 x10 3/uL	8.73	14.43 (H)	6.14	White blood cell count high in SCA due to inflammation, lower inflammation, and lower WBC with HU
Hemoglobin 10.6 - 13.3 g/dL	12.6	7.7 (L)	10.3	Hemoglobin low in SCA from sickle cells breaking down. HU protects red blood cells and increases hemoglobin
Hematocrit 32.3 - 39.7 %	35.8	21.6 (L)	27.8 (L)	
Platelet Count 203 - 368 x10 3/uL	269	523	156	Platelet count high in SCA due to inflammation. HU lowers inflammation and platelet count
RBC 3.93 - 5.00 x10 6/uL	4.25	2.49 (L)	2.62 (L)	
MCV 75.2 - 86.9 fL	84.2	86.7	106.1 (H)	HU increase red blood cell size, making them plump and juicy, less likely to sickle
MCH 24.8 - 29.4 pg	29.2	27.2	39.3 (H)	
MCHC 32.0 - 34.8 g/dL	33.9	33.0	37.1 (H)	
RDW 12 - 14 %	12	23 (H)	16 (H)	
MPV 8.9 - 12.5 fL	8.8	9.1	10.3	
Neutrophils None %	48.2	61.2	22.4	
Nucleated RBC 0.0 - 0.0 /100 WBC	0.0	4.9 (H)	0.5 (H)	
Neutrophils Absolute Count (ANC) 1.64 - 7.71 x10 3/uL	4.21	8.81 (H)	1.38	ANC high in SCA due to inflammation, comes back down with HU
Nucleated RBC Absolute 0.03 - 0.11 x10 3/uL	0.0	0.71 (H)	0.03	
Reticulocyte Count Automated 1.0 - 1.9 %	1.2	24.0 (H)	5.7 (H)	High % of new red cells in SCA, working hard to replace lost sickle cells; HU prevents sickling, less RBC loss, retic lower
Hemoglobin F (fetal hemoglobin) 0.0 - 2.0 %	2.0	5.4	32.1	HU raises fetal hemoglobin to protect red blood cells from sickling. Needs to be 20-30% to be effective. 30% or higher is best.
Bilirubin Total 0.2 - 1.4 mg/dL	0.4	5.5 (H)	1.4	Broken sickle cells release bilirubin, causing jaundice; HU prevents red blood cell destruction, decreasing bilirubin, no more jaundice

*In milder forms of sickle cell disease, including Hgb SC, Hgb S beta+ thalassemia, or Hgb SE, Hgb is usually ~ 10 – 11, retic usually 3-5%. MCV is lower than normal, usually between 60-70. Bilirubin varies, from normal to slightly high.

Interpreting lab results

Complete blood counts (CBC) are routinely performed during your visit. Labs that are evaluated are Hemoglobin (Hgb), reticulocyte count (retic), Mean Corpuscular Volume (MCV), Absolute Neutrophil Count (ANC), platelet count (plt), fetal hemoglobin (Hgb F), and bilirubin (bili).

Hemoglobin (Hgb) is the molecule inside red blood cells that carries oxygen throughout the body. When sickle red blood cells break down, hemoglobin is lost and oxygen carrying capacity is low. The hemoglobin level is tracked at each clinic visit to make sure that your hemoglobin is stable and in safe range.

The medication Hydroxyurea decreases sickling, which helps to raise the hemoglobin and the oxygen carrying capacity in the body.

Reticulocytes (retic) are young red blood cells that enter the bloodstream from the bone marrow. If you have sickle cell disease, you may have a higher reticulocyte count because your body is working hard to make more red blood cells to replace lost cells. People with sickle cell anemia have high reticulocyte counts. As the Hgb rises with treatment (such as Hydroxyurea), retic count becomes lower as fewer cells are lost and the bone marrow no longer needs to work so hard to make up for low Hgb. If the reticulocyte count drops too much, that means the body might not be making enough red blood cells to maintain a decent hemoglobin level. We like to keep retic count ~

Mean Corpuscular Volume (MCV) is the average volume of red cells. Children with Hgb SS usually have normal values, while children with Hgb S beta thalassemia or Hgb SC have lower MCV, because their red blood cells are smaller. Some children who might be iron deficient could have a lower MCV. When we place children on the medication Hydroxyurea, the red blood cells larger (plump and juicy) and the MCV becomes higher, which makes the red blood cell less likely to sickle. We look for high MCV in patients taking Hydroxyurea well.

Absolute Neutrophil Count (ANC) is the number of neutrophils in the blood. A neutrophil is a type of white blood cell that helps fight bacterial and fungal infection. In untreated sickle cell disease, white blood cell count and neutrophil counts are very high, which reflect inflammation that makes vaso-occlusion worse. Hydroxyurea decreases ANC, which is good, but we do not want to drop the ANC too low, so this value is routinely

Platelets (plt) are little cells in the blood that help us clot. A certain number of platelets is necessary to ensure against spontaneous bleeding. Treatment with Hydroxyurea lowers the platelet count, which is helpful, because platelets also contribute to vaso-occlusion. We check the platelet count to make sure it does not drop too low. We aim to maintain plt > 80 (80,000).

Fetal hemoglobin (Hgb F) is the hemoglobin in the fetus and helps carry oxygen from the mother's bloodstream to tissues in the fetus. After the baby is born, the amount of fetal hemoglobin starts to drop as the baby starts to make the adult form of hemoglobin (Hgb A or Hgb S). Hgb F interferes with sickling (or Hgb S polymerization), so high Hgb F is protective against sickle cell symptoms. Hydroxyurea works by increasing Hgb F. We dose Hydroxyurea to achieve high Hgb F. Hgb F 30% or higher is best.

Bilirubin (bili) is formed as an intermediate step when the body is getting rid of the hemoglobin that spilled out from sickle red cell breakdown. Bilirubin is yellow and causes jaundice. Children with untreated sickle cell disease have high bilirubin and often have jaundiced (yellow) eyes, because they have a lot of sickle cells that are constantly breaking down. If sickle cell anemia is treated, such as with Hydroxyurea, red blood cells do not sickle and break as much, the hemoglobin stays inside the red blood cells, bilirubin is not produced and the eyes stay clear without jaundice.

Health maintenance schedule

For sickle cell health, it is important to come to sickle cell clinic regularly. There are monitoring and preventive measures that guard against expected and unexpected complications. Even if your child is not displaying sickle cell complications that you can see, it is still important to maintain sickle cell clinic visits to make sure all immunizations and screening tests are up to date, and to keep up with sickle cell education and new information.

Clinic visits for patients on Hydroxyurea therapy should be seen no longer than 2 - 3 months apart. Patients with mild forms of sickle cell disease should maintain at least every 6 month checkups and more frequently as necessary.

Patients who come to clinic regularly do better with their sickle cell disease.

Pediatric Sickle Cell Clinic Health Maintenance Schedule

Age	Health Maintenance Schedule
0-1 years	• Every 2-3 month clinic visits. • Physical examination (especially spleen). • Labs: Complete Blood Counts (CBC), Reticulocyte Count (Retic), Hemoglobin electrophoresis at 6 months, annual CMP, LDH. • Start Hydroxyurea between 6-9 months of age, or when Hgb F falls below 30%. May begin earlier if anemia is severe.
0-5 years	• Penicillin or Amoxicillin prophylaxis—twice a day.
Age 1 and up	• Every 2 months clinic visits for genetically severe forms of sickle cell disease and for all patients on hydroxyurea. Monthly visits when initiating or adjusting hydroxyurea. • Physical examination. • Labs: CBc, retic, Hgb F every visit for patients on Hydroxyurea; annual CMP (complete metabolic panel), annual urinalysis starting age 5 • Ongoing Hydroxyurea monitoring every 2 months. • Menveo vaccine at 9 months and 12 months.
Age 2 and up	• Pneumovax 23 vaccine at age 2 and then 5 years later. • Menactra vaccine, then every 5 years. • For patients with mild disease (SC, Sbeta+) not on Hydroxyurea, visits may be spaced out to every 6 months after age 2.
Age 2-16	• Annual Transcranial Doppler Ultrasound (TCD) for patients with Hgb SS, Sbeta0, or other severe forms of sickle cell disease to screen for stroke risk. Brain MRI/MRA if abnormal.
Age 10	• Begin annual eye exams by ophthalmologist to screen for sickle cell retinopathy. • Begin annual urine microalbumin screening. • Meningococcal B vaccine: initial 2 dose series, then every 3 years.
Age 15	• Begin transition education.
Age > 18	• Transition to adult program as appropriate but may continue to be followed in our clinic through college.

Sickle Cell Organizations and Resources

Pediatric Specialists of Virginia Sickle Cell Program
Inova Schar Campus
8081 Innovation Park Dr. Suite 765, Fairfax, VA 22031
571-472-1717
https://psvcare.org/northern-virginia-comprehensive-pediatric-sickle-cell-program

Virginia Department of Health/VDH
P.O. Box 2448
Richmond, Virginia 23218-2448
109 Governor Street
Richmond, Virginia 23219
https://www.vdh.virginia.gov/sickle-cell-programs/

Center for Disease Control and Prevention
1600 Clifton Road
Atlanta, GA 30329-4027
800.CDC.INFO (800.232.4636) / TTY: 888.232.634
https://www.cdc.gov/sickle-cell/index.html

National Institutes of Health
9000 Rockville Pike
Bethesda, MD 20892
301.496.4000 / TTY: 301.402.9612
NIHinfo@od.nih.gov
https://www.nhlbi.nih.gov/health/sickle-cell-disease

Sickle Cell Information Center
Emory Center for Digital Scholarship
201 Dowman Drive
Atlanta, Georgia 30322 USA
404-727-7857
http://www.scinfo.org

Faces of Our Children
1920 L Street, NW, Suite 301,
Washington, D.C. 20032
Phone: 1-866-FACES11
http://www.facesofourchildren.org

Sickle Cell Disease Association of America
231 East Baltimore Street Suite 800
Baltimore, MD 21202
410.528.1555
scdaa@sicklecelldisease.org
http://www.sicklecelldisease.org

Sickle Cell Association of Virginia
Principal contact: Minni Powell
14506 Hockliffe Loop
Midlothian, VA 23112
minni.powell@gmail.com
804-743-0807
https://www.vpap.org/lobbying/client/370502-sickle-cell-association-of-virginia/

The Heart of Gold Sickle Cell Foundation of Virginia, Inc.
P.O. Box 23681 Alexandria, VA 22304
Phone: 703-370-3234
https://heartogold.org/

Sickle Cell Association of the National Capital Area, Inc / SCANCA.Inc
P.O. Box 41479
Washington, DC 20018-08792
(202) 271-5733
email@scancainc.org
http://scancainc.org/

Sickle Cell Transplant Advocacy & Research Alliance/STAR
PO Box 96
Great Barrington, MA 01230
https://curesicklenow.org/