

SICKLE CELL DISEASE:

A FAMILY GUIDE

(Fourth Edition)



Division of Family Services
Newborn Screening and Genetic Services

Services are funded in part by:
Special Child Health and Early Intervention Services



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Acknowledgments

We would like to extend special thanks to the following persons and groups:

- Children with sickle cell disease and their families for their constructive comments
- The Sickle Cell Advisory Committee of New Jersey

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This fourth edition, third printing was published through funding provided by:

New Jersey Department of Health and Senior Services
Division of Family Health Services
Newborn Screening and Genetic Services
Special Child Health and Early Intervention Services
P.O. Box 364
Trenton, New Jersey 08625-0364

Dedication

The members of the Sickle Cell Advisory Committee of New Jersey wish to dedicate this manual to the:

- approximately 80-90 infants born each year in New Jersey with sickle cell disease or other hemoglobinopathies;
- parents and family members who provide these children with the care, love and encouragement for an active and healthy childhood;
- dedicated staff of the statewide network of treatment centers which specialize in treating children with sickle cell disease;
- hospital emergency room staff who provide prompt emergency treatment to these children;
- pediatricians and family practice physicians who provide routine health care to these children and their families;
- local community organizations that advocate for individuals with sickle cell disease; and
- the educators and school personnel who care for the children during their school days.

How to Use This Guide

This guidebook is an introduction for families living with sickle cell disease. It provides an overview of sickle cell disease and what to expect. Please make marks in this book, fold the pages, and highlight sections. Share this guidebook with other people who will be a part of your child's life. But most importantly, speak with your sickle cell health care team about any questions or concerns that you have while reading this guidebook. Remember that this is just a guidebook and that each person's journey with sickle cell disease is different.

Introduction

Sickle cell disease is an inherited disorder of the red blood cells. It is especially common among African Americans. It can also affect those of Mediterranean, Caribbean, Latin American, Asian and Middle Eastern descent. This map gives a broad view of areas of the world where sickle cell disease is historically most common. As you can imagine in our global society, people with sickle cell disease live in all parts of the world today.



Children with sickle cell disease feel and look fine most of the time. But, they can become sick very quickly and with little warning. For this reason, the State Department of Health and Senior Services now screens all newborns in New Jersey for sickle cell disease. Identifying infants with sickle cell disease in the first weeks of life can help connect families to specialized medical teams.

To make specialty care available to more children, the Department of Health and Senior Services has established sickle cell treatment centers throughout New Jersey. Each center has a team of specialists who work with the child's regular doctor. Together, they help the family manage the child's health care needs.

Not every child with sickle cell disease develops every complication in this book. This information can seem overwhelming, and even frightening. That is not the purpose of this book. The purpose is to provide you with the information to recognize a health emergency right away. **The sooner you notice a problem, the sooner your child can get the treatment he/she needs.**

EMERGENCY TELEPHONE NUMBERS

Call 911 for Emergencies!

Dr. _____ (_____) _____

Regular Doctor/Pediatrician

Hematologist (Sickle Cell Specialist):

Dr. _____

From 9 a.m. to 5 p.m. (_____) _____

Evenings, Holidays, and Weekends (_____) _____

Names of Staff members at the Sickle Cell Treatment Center

Local Hospital

Address

Directions

Other Numbers: (_____) _____

(_____) _____

Warning Signs

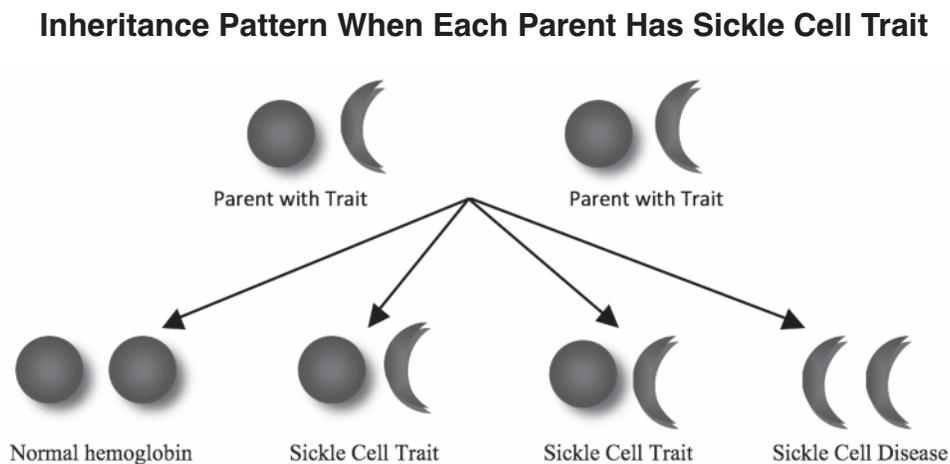
This table lists warning signs of sickle cell disease emergencies. This table is placed here in the front of the book so you can find it immediately. Each sign and emergency will be explained later on in this book. Find the symptoms in the table and call 911 or your sickle cell health care team as instructed:

SYMPTOM	Dial 911	Immediately Call Sickle Cell Team	Call Sickle Cell Team within 24 hours
FEVER	Difficult to wake up	<ul style="list-style-type: none"> • Temperature 101°F or above • Shaking chills (even without fever) 	
SWOLLEN SPLEEN	Difficult to wake up	<ul style="list-style-type: none"> • Enlargement (swelling) of the spleen and/or belly • Stomach pain • Refusing to eat • Paleness of lips, gums, nails • Sudden weakness 	
PAIN	Difficulty breathing or severe headache	<ul style="list-style-type: none"> • New or uncontrolled pain • Chest pain &/or cough • Fever • Nausea or vomiting • Unable to drink plenty of water • In boys, painful hardening of penis 	<ul style="list-style-type: none"> • Pain that is relieved by medications but lasts more than 2 -3 days • Before pain medications prescribed for your child run out
HEADACHE OR WEAKNESS	<ul style="list-style-type: none"> • Difficult to wake up • VERY severe head pain • Seizure (extreme shaking) • Fainting 	<ul style="list-style-type: none"> • Paleness of lips, gums, nails • Shortness of breath, racing heart beats • Sudden weakness, difficulty speaking or change in personality or behavior • Change in vision • Falling down and/or clumsiness • Weakness on one side or limb • Headache that does not get better or returns 	
VERY TIRED	<ul style="list-style-type: none"> • Extremely tired, difficult to wake up 	<ul style="list-style-type: none"> • Dizziness • Too tired to get out of bed or walk to the bathroom 	<ul style="list-style-type: none"> • Fatigue is getting worse • Staying in bed all day
TROUBLE BREATHING	Unable to catch breath	<ul style="list-style-type: none"> • Chest pain • Cough with fever and/or chest pain • Fast, noisy or difficulty breathing • Working hard to take deep breaths 	

Facts About Sickle Cell Disease

Sickle cell disease is an inherited condition. Just like the genes for eye color that are passed on from parents to babies, the gene for sickle cell disease is passed on by parents to babies. Both the mother and father must carry at least one affected gene. With each pregnancy, there is a one in four, or 25%, chance that the baby will inherit sickle cell disease. Sickle cell disease occurs when both parents pass along the affected gene. If one parent passes along an affected gene and the other passes along a normal gene, the baby will be a sickle cell carrier. This is also called, sickle cell trait. People living with trait do not have sickle cell disease and they do not experience the complications of sickle cell disease.

In the picture below, you will see how sickle cell disease is inherited when each parent carries the sickle cell trait.



When each parent has sickle cell trait, there is a one in four chance (25% chance) with each pregnancy that the baby could be born with sickle cell disease.

There are different forms of sickle cell disease. These depend on the types of genes that mothers and fathers carry. The types are described here:

- Hemoglobin SS (HbSS) is the most common form. In this case the mother and the father both carry the gene for sickle cell (S) and passed it along to their baby. HbSS is often a more severe form of sickle cell disease.
- Hemoglobin SC (HbSC) is also common in the United States. In this case, one parent, the mother or father, carry the trait for sickle cell (S). The other parent carried the trait for hemoglobin C disease (C). HbSC is often a moderately severe form of sickle cell disease.
- Hemoglobin S-beta thalassemia (HbS-thalassemia) occurs when one parent carries the trait for sickle cell (S) and the other carries the trait for a condition called beta thalassemia. HbS-thalassemia may be very mild or very severe depending on the beta thalassemia gene that is inherited. Your hematologist will use a blood test to determine if your child has HbS-thalassemia +, a milder form, or HbS-thalassemia-0, a more severe form.

Testing Other Family Members

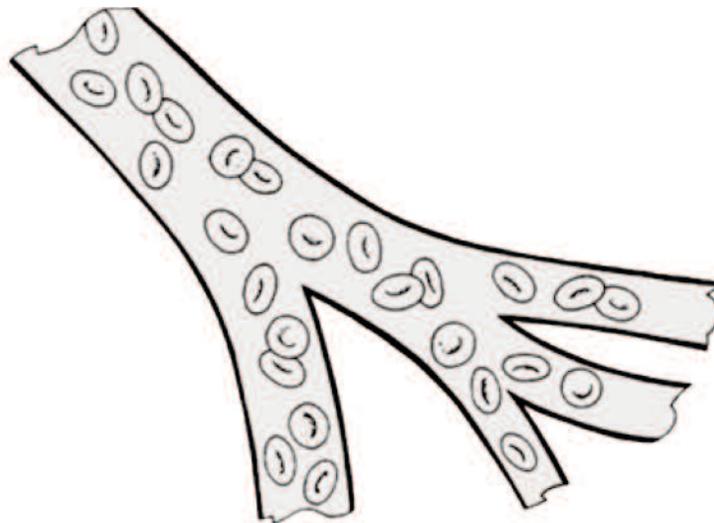
The trait for sickle cell disease runs in families. Sickle cell trait is not a disease. People who carry sickle cell trait will not develop sickle cell disease later in life. This means that your relatives may be carrying the trait and not be aware of it. This information will be very important to a person when he or she is planning to have children. In the case where both partners have sickle cell trait there is a 25 percent chance their child could be born with sickle cell disease.

Genetic Counseling

Prenatal testing is available. The testing is quite accurate and can tell whether the baby will be born with sickle cell trait, sickle cell disease, or neither. If you are pregnant or considering becoming pregnant, and would like additional information, you can meet with a specialist in genetics. Genetics is the science of how diseases and conditions are passed through families.

What Does Sickle Cell Disease Do To Blood?

Sickle cell disease affects red blood cells. Red blood cells contain a protein called hemoglobin. Hemoglobin has the job of carrying oxygen we inhale to all the parts of the body. Normal hemoglobin is called hemoglobin A, ("A" is for adult). Red blood cells containing hemoglobin A are round, soft, and flexible. These red blood cells can squeeze through small blood vessels allowing blood to flow easily.

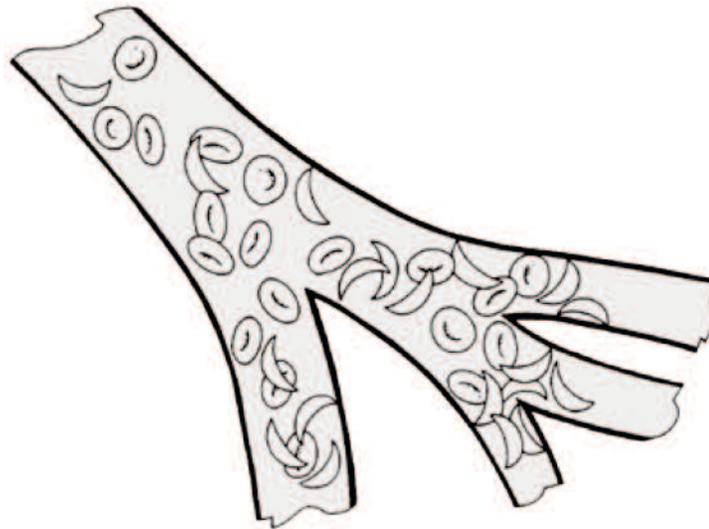


Blood Vessels with round red blood cells containing normal hemoglobin, this is also called Hemoglobin A.

The red blood cells of sickle cell contain an abnormal type of hemoglobin called sickle hemoglobin, or hemoglobin S. Sickle hemoglobin is not good at carrying oxygen. Under certain stressful conditions such as illness, fever, and dehydration, sickle hemoglobin changes its shape into a half moon or "sickle" shape. The red blood cells then become stiff and break apart easily.

These fragile and abnormally shaped cells are not able to flow easily through blood vessels. In fact, these sickle shaped red blood cells get caught on each other and the walls of the blood vessels causing a jam. The body cannot get the oxygen it needs from the red blood cells. This lack of oxygen can cause complications, depending on the part of the body affected. For example, this can cause the severe pain that is common in sickle cell disease.

Red blood cells containing sickle hemoglobin only live 10-20 days. Compare that with normal red blood cells which can live as long as 120 days. This shortened life span of the red blood cell results in an amount of hemoglobin that is lower than normal. Anemia is a term that is used to describe lower than normal hemoglobin levels. People living with sickle cell disease often have life-long anemia. They are often described as “pale”. They tire more easily than others.



Blood Vessels with mostly sickled cells containing hemoglobin S

Complications of Sickle Cell Disease

Blood flows to and from almost every body part. This means that sickle shaped red blood cells can cause a problem in the way oxygen is being delivered to all organs and tissues in the body. Sickle shaped red blood cells can also affect the blood vessels carrying them. This lack of oxygen and damage to blood vessels can result in complications in many different body parts. Complications can be acute or chronic.

Acute complications happen suddenly and require immediate medical attention. Some acute complications can be avoided but most of the time they are unexpected and come without warning. These types of complications may occur at any age. But some are more common in different age groups.

Chronic complications develop slowly over time. The damage of chronic complications may result in an organ or organ system no longer working correctly. Organ systems that can be affected by chronic complications include the kidneys, lungs, heart, eyes and joints. Luckily, many chronic complications can be prevented. Others may be reversed or slowed down if caught early.

Recognizing and treating acute complications quickly is important to treating sickle cell disease. Treatment is also aimed at avoiding chronic complications. Treatment will be discussed in more details later in this guidebook.

Acute Complications of Sickle Cell Disease

The following is a list of acute, or sudden, complications that require immediate medical attention:

Fever and Infection

Infection is an acute complication of sickle cell disease. Unlike other children, those living with sickle cell disease are unable to fight infections well. This is especially true if it is a bacterial infection. Bacteria can multiply in the blood and quickly overwhelm the body's ability to fight back. This condition, known as sepsis, is life-threatening. Infections may also take place in the lungs (pneumonia) or bones (osteomyelitis).

The child with a serious infection may not seem very sick at first. It is important to learn the early warning signs of an infection and know when to take your child right away to the doctor or hospital. A group of medications called antibiotics fight bacterial infections. **It is extremely important that there are no delays in your child receiving antibiotics when there is a fever.**

- **Signs and Symptoms**

- Fever is the most common symptom of infection.

- **Special Care for Fever**

- Do not give any medicine such as ibuprofen (Motrin®) or acetaminophen (Tylenol®) before checking your child's temperature. These medications will lower the temperature and may hide or "mask" an infection.
- Use a thermometer to check for fever. This is more reliable than touching your child to feel for fever.
- Do not wait for a fever to go down. Call your health care team right away.

- **Preventing Infections**

- *Penicillin*

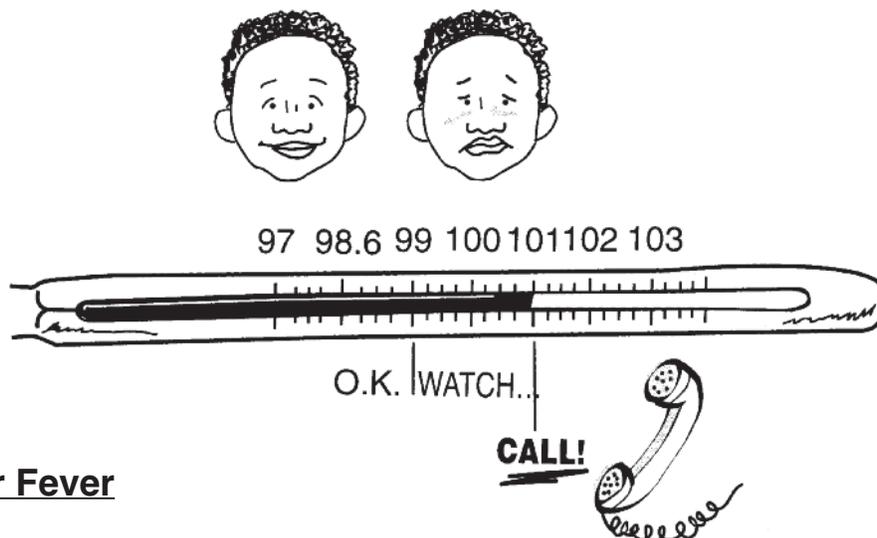
Penicillin is an antibiotic. Antibiotics help prevent dangerous infections. Your child's doctor will prescribe penicillin shortly after he/she is born with sickle cell disease. Penicillin will most likely be stopped by your doctor at a later age. In the meantime, your child will take penicillin twice a day, every day, to prevent infections. Some bacteria can cause infections even when penicillin is taken correctly. For this reason, **please remember that fever is always an emergency.**

- *Pneumococcal polysaccharide vaccine*

The Pneumococcal vaccine (Pneumovax®) is an immunization that provides added protection from a type of bacteria. This is first given at age 2 years and then there will be at least one booster given at a later age.

- *Other Immunizations*

Children with sickle cell disease should receive all immunizations recommended in childhood. This should include a yearly flu shot as well.



Checking for Fever

Pain Crisis

Pain crisis is also called vaso-occlusive crisis. It is the most common acute complication of sickle cell disease and the number one reason people are hospitalized. Pain is the result of the sickle shaped red blood cells blocking the flow of blood to a body part.

While not always avoidable, some precautions can be taken to avoid a pain crisis. These include drinking plenty of water, wearing clothes appropriate for the weather, and taking time to rest.

Pain can often be treated at home with medications prescribed by your health care team. But at times, the pain is just too severe and hospitalization is necessary. At the hospital intravenous (IV) fluid and pain medicine will be given directly into a vein intravenously. Your health care team will give your child strong medications for pain. This is necessary to make your child comfortable. Your doctors, nurses, and pharmacists are very well trained and experienced in using these strong pain medications safely.

Aplastic Crisis

Normal red blood cells live about four months before they break down. But this is not the case with sickle red blood cells that live less than one month. Even though the body tries, it cannot make new red blood cells fast enough to replace the old ones. This causes the child's hemoglobin to be low. Low hemoglobin is called anemia. Children with sickle cell disease usually have anemia. In most cases, the body adjusts to the anemia.

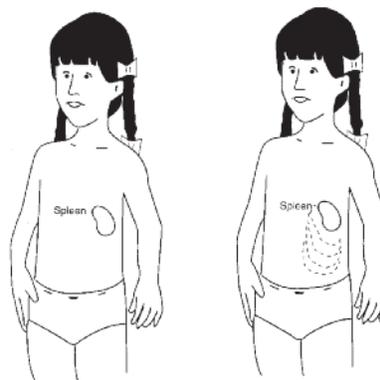
There are some common childhood viruses that may cause the body to temporarily stop making red blood cells. This shut off of a supply of new red blood cells, combined with the short life span of the red blood cells of sickle cell disease, results in a severe anemia. This is called aplastic crisis.

The virus most likely to cause aplastic crisis, is parvovirus B19. The common name for this viral infection is "Fifth disease". Fifth disease may, or may not, cause a mild fever, a fine red rash on the arms and a rash on the cheeks that looks like the child has been slapped. This rash, known as the "slapped cheek rash" is not always noticeable.

Aplastic crisis will cause a child to feel extremely tired. He or she may become cranky and irritable. The lips and nail beds will be pale. Hemoglobin can drop to a dangerously low level. Symptoms of aplastic crisis require emergency medical attention.

Splenic Sequestration Crisis

The spleen is an organ on the left side of the belly under the rib cage. It helps the body fight infection. In children with sickle cell disease, the spleen may get plugged with sickle cells. This can cause the spleen to rapidly fill with blood and swell. This complication is called splenic sequestration. A child with splenic sequestration may complain of belly pain. He or she may be very tired, irritable, and the lips and nail beds will be pale. Your health care team will teach you how to feel the spleen. It is important to practice this so that you will be calm and comfortable should a sequestration happen. Splenic sequestration is extremely dangerous and life threatening. There can be no delay in seeking emergency care.



Normal spleen and swelling spleen

Acute Chest Syndrome

Acute chest syndrome occurs when red blood cells become sickle shaped and clog the small blood vessels of the lungs. Acute chest syndrome can very quickly become a life-threatening complication. It often happens with pneumonia or pain crisis. The signs of acute chest syndrome include: chest pain, fever, increased effort needed to breathe, and cough. An x-ray of the chest will show changes in the lungs. If your child is experiencing any combination of these symptoms, you must seek medical care immediately. A person who has had acute chest syndrome is more likely to have this again. Always let your caregiver, especially in emergency rooms, know if there is a history of acute chest syndrome.

Stroke

Stroke is the most long-lasting acute complication of sickle cell disease. A stroke happens when sickle shaped red blood cells block the blood flowing in the brain. This blockage leaves a part of the brain without vital oxygen. Stroke is most common in those with HbSS and HbS-thalassemia-0.

Signs of stroke include a severe headache, changes in vision, weakness (especially on one side of the body), trouble speaking, vision changes, clumsiness, confusion and seizures. If your child experiences any of these signs or is just not acting normal you must seek emergency medical attention right away. The effects of stroke are often life-long. Rapid medical treatment can limit the effects of the stroke.

Priapism

Males, especially teenagers and young adults, can have a long-lasting, unwanted and painful erection (hardening) of the penis. This acute complication is called priapism. This problem happens when sickle shaped cells block the circulation of blood in the penis.

If priapism lasts more than two hours or there is an inability to urinate, notify the doctor. Boys may find it embarrassing, so they may not tell you. Talk to them and tell them this problem might happen. Tell them to let you know right away if it does. Erections that are not painful and long lasting are normal.

If a male has had priapism, they are likely to have this complication again. It may be recommended that a urologist, a doctor who specializes in this area, be seen to develop a plan for preventing priapism from returning.

Chronic Complications of Sickle Cell Disease

The following is a list of chronic, or slowly developing, complications of sickle cell disease:

Eye Problems

The retina is where the eye takes what we see and changes it into messages read by the brain. There are very small and fragile blood vessels in the retina that can be permanently damaged over time by sickle shaped blood cells. Yearly eye exams can find these changes before vision is affected. Any loss or change in vision should be reported to the health care team immediately.

People with sickle cell disease often have a yellow color to the white part of the eye (sclera). The red blood cells contain a yellow substance called bilirubin, which gets released as the blood cells breakdown. This yellow tinge, also called icterus or jaundice, does not cause harm to the eyes.

Aseptic Necrosis

When sickle shaped cells block the blood vessels of the joints (where bones meet) over and over again, the joint can become damaged. Unlike a pain crisis, the pain and stiffness of aseptic necrosis (also called avascular necrosis or AVN) does not go away and tends to be almost daily. This problem happens most commonly in the hip joint but may affect the knee and shoulder joints as well. Your health care provider should be told about frequent pain and stiffness in the joints.

Skin Problems

Some people develop open skin wounds called ulcers. This happens especially around the ankles. If there is an open wound that is not healing normally, notify your health care provider. Although it is rare, poorly healing ulcers usually affect teenagers and young adults with sickle cell disease.

Gallstones

The gall bladder is a small sac near the liver. Its job is to store bile, a liquid needed by the stomach to digest food. Sickle shaped red blood cells break down easily releasing a substance called bilirubin. Bilirubin is needed to make bile. The increased bilirubin results in an increase in bile. When this bile sits in the gall bladder it can become thickened and eventually harden into stones. These stones cause pain. This pain is usually in the right side of the belly. Gallstones may also cause stomach bloating and nausea. People can live without a gallbladder. Surgery to remove this is often needed to relieve the symptoms and avoid serious complications.

Kidney Problems

One role of the kidneys is to maintain the balance of fluid in the body by making urine. Normally, the kidneys will help remove extra fluid in the body by concentrating it into urine. The sickle shaped red cells can damage the ability of the kidneys to perform this job. As a result, the kidneys will produce more urine that is less concentrated. This extra urine can cause a child to experience bedwetting. Bedwetting can cause a great deal of stress in children. They may avoid staying overnight with family, sleepovers and sleep away camp. The disruption to sleep is upsetting to the whole family.

It is important to speak openly with your child about the medical cause of bedwetting. Do not use words that shame or embarrass the child. Bedwetting affects children without sickle cell disease so pediatricians can be very helpful with this. The same strategies used in children without sickle cell disease who wet the bed may be useful. This includes limiting fluids in the evening. Alarms are made that wake the child as soon as moisture is felt. By using these alarms regularly, a child begins to make the connection between feeling the moisture and being awoken. Bedwetting alarms can be very successful.

The kidneys also filter toxins and waste products from the blood. Sickle cell disease can damage this filtering system eventually leading to kidney failure. Your health care team will routinely screen the urine and blood for early signs of changes to the filtering system. This early detection can prevent long-term serious kidney damage.

Treatment of Sickle Cell Disease

Bone Marrow Transplant

Bone marrow transplant (or stem cell transplant) is currently the only cure for sickle cell disease. All of the body's blood cells are made within the bone marrow. The goal of bone marrow transplant is to replace the bone marrow that makes red blood cells affected by sickle cell disease with that of a healthy donor. The donor's bone marrow will then make healthy red blood cells and sickle cell disease is cured.

To prepare for this procedure, the person with sickle cell disease receives chemotherapy, and sometimes radiation, in high enough doses to wipe out the bone marrow. Then an infusion of bone marrow cells from a tissue-matched donor is delivered. The new bone marrow takes over producing the donor's healthy blood cells and sickle cells are no longer made. This procedure can potentially have very serious side effects. Because of this risk, bone marrow transplant is currently being used in the most severe cases of sickle cell disease.

Not just anyone can be a bone marrow donor. Everyone is born with a unique set of tissue markers. Think of these markers, called HLA, as a genetic fingerprint. Half of the HLA markers are inherited from the father and half from the mother. In each family, full siblings have a 25% chance, or one in four, of inheriting the same HLA fingerprint as each other.

There is also a possibility that someone outside of the family may potentially be an HLA matched-donor. There are large databases known as banks that keep HLA profiles on potential volunteer donors. If it is determined that a person would benefit from bone marrow transplant, there is a search for potential donors in these banks. People with similar racial and ethnic backgrounds tend to share HLA markers. Although progress is being made, the number of potential volunteer donors does not adequately reflect racial diversity. This can make finding a donor outside of the family very difficult.

Hydroxyurea

Hydroxyurea is a medication used to treat sickle cell disease. It is not a cure for sickle cell disease. Hydroxyurea is a type of chemotherapy medicine that has also been used to treat some cancers. Hydroxyurea works by increasing the amount of fetal hemoglobin in the blood. Fetal hemoglobin is the type of hemoglobin inside the red cells of babies before they are born. Red blood cells containing fetal hemoglobin do not become sickle shaped. Even in small amounts, fetal hemoglobin can protect the body from the more severe complications of sickle cell disease.

Large research studies have proven that hydroxyurea is a safe and valuable medication. Research has proven that adults and children who take hydroxyurea have decreased hospital stays and complications, including pain crisis and acute chest syndrome. Based on these studies, it is recommended that all children with severe forms of sickle cell disease (HbSS and HbS-thalassemia-0) be considered for hydroxyurea treatment starting at age 9 months.

There are side effects associated to hydroxyurea. There may be some mild stomach upset or darkening of skin and nails. The side effect that is most serious is hydroxyurea's potential to lower other blood cells needed by the body. These side effects must be closely watched for through regular checkups and blood tests. So far studies have not shown that hydroxyurea has long-term side effects. Hydroxyurea should not be taken by someone who is pregnant, or planning to become pregnant.

As with all medications that are chemotherapy, caution should be used when handling hydroxyurea. Gloves should be worn when touching the medicine. Diapers should be thrown right away, preferably outside. Anyone who may be pregnant should not be preparing the medication. Capsules should not be opened or crushed. A liquid form is available for children who cannot swallow the capsules whole yet.

Blood Transfusions

Transfusions of red blood cells from healthy donors can be life saving for people having severe sickle cell complications. Transfusions are sometimes used on a regular basis to avoid severe complications in people with a strong history of these. Your sickle cell doctor will not give a transfusion unless it is absolutely necessary.

Many families are very anxious about blood transfusions. These concerns are real and should be discussed with your health care team. If you have religious beliefs or concerns about receiving blood transfusions, please discuss them with your child's doctor or nurse. It is important that these concerns and beliefs be discussed openly before there is a medical emergency that may require a blood transfusion.

In the past, serious infections, especially Human Immunodeficiency Virus (the cause of AIDs) and hepatitis (a liver infection), were spread by blood transfusions. Because of this donors and the blood they donate are carefully screened. Please discuss any concerns you have about the safety of donated blood with your health care team.

Laboratory and Screening Tests

Laboratory tests and screening tests will tell the health care team a great deal of information. Some tests can help diagnose acute complications as they are taking place so immediate interventions can be done to begin the recovery process. Other tests can show if chronic complications are starting to develop. This allows the health care team to step in and, hopefully, stop further damage to the body.

Laboratory tests

Blood tests involve the insertion of a needle into a vein, usually on the arm. A vacuum tube is attached to the end of the needle that pulls a small amount of blood into the tube. This tube is then sent to the laboratory where tests are performed. The purpose of blood tests is determine what is normal and detect when abnormal things are happening in the body. Urine tests are also sent to the laboratory and can give important information as well.

The most common laboratory tests are discussed here. There will be times that other tests may be performed. Do not ever hesitate to find out what tests are being done and why.

- ***CBC (Complete Blood Count)***

The CBC measures the type and quality of blood cells in the body. The three types of blood cells are:

- WBC (white blood cells) help the body to fight infection. When WBCs are elevated there is a concern that there may be an infection.
- Hemoglobin is inside the red blood cells and it carries oxygen to the body tissues. Because hemoglobin is abnormal in sickle cell disease, this value will be lower than normally expected. It is important to know what your child's baseline hemoglobin level is so that changes in this can be detected quickly.
- Platelets clot blood and makes bleeding stop. Platelets may be low in number if they are trapped during splenic sequestration.

- ***Reticulocyte Count (Retic)***

Reticulocytes are young red blood cells. Because red cells affected by sickle cell disease have a short lifespan the body is required to make more young red cells. Because of this, it is common for the reticulocyte count to be higher than normal.

- *Type and Crossmatch*

This test is done before blood transfusions to make sure that the blood being given matches your child's blood type. It is done before every transfusion is given.

- *Hemoglobin Electrophoresis*

The hemoglobin electrophoresis measures the types of hemoglobin the body is making.

People who are unaffected by sickle cell disease only make normal hemoglobin known as hemoglobin A.

- Sickle Cell hemoglobin is known as Hemoglobin S. If a person has sickle cell, HbSS, the body will only make hemoglobin S.
- If a person has sickle cell type HbSC, the body will make hemoglobin S and hemoglobin C.
- If a person has sickle cell type HbS-thalassemia-0, the body will make mostly hemoglobin S, and some hemoglobin F (fetal hemoglobin).
- If a person has sickle cell type HbS-thalassemia+, the body will make mostly hemoglobin S, some hemoglobin F and a little bit of normal hemoglobin (A).

- *Blood Chemistries*

This test can tell a lot about the health and function of the kidneys and liver. It also gives information about nutritional status.

- *Urine Tests*

Urine samples are collected periodically. These samples are then tested to determine the health of the kidneys. Urine tests can also let health care providers know how well hydrated your child is or if he or she has an infection in the kidneys and bladder.

- *Blood Culture*

If your child has a fever, a blood culture will be drawn. This blood sample is sent to the lab where it is observed for up to five days. This is the amount of time needed for bacteria, if it is present in the body, to grow in the lab setting. This test is also used to guide the choice of antibiotics (infection fighting medicines) needed.

Screening Tests

Depending on the type of sickle cell disease your child has or the symptoms he or she is experiencing, other tests may be done. Here is a list of the most common tests.

- *Ultrasound*

An ultrasound is a test that uses sound waves to create a picture of what is happening in the body. Ultrasounds are painless. A clear jelly is put on the skin. A wand, known as a transducer, is rubbed over the jelly sending out harmless sound waves. The way these waves behave when they hit the body part below is made into a black and white computer image. Ultrasounds are often done of the kidneys to make sure they are healthy. The abdominal (belly) ultrasound is used to measure the size of the spleen or to look for stones in the gall bladder.

- *Transcranial Doppler*

Also known as the TCD, the transcranial doppler is a special ultrasound used to determine if there is an increased risk of having a stroke caused by sickle cell disease. During the TCD, the

transducer is moved over the neck and head to create a picture of the flow of blood to and from the brain. All children with sickle cell disease, types HbSS and HbS-thalassemia-0 should have a yearly TCD between the ages of 2 and 16 years. The TCD is a very important test.

- *Echocardiogram*

An echocardiogram or “echo” is an ultrasound of the heart. The transducer is moved on the chest to show the heart structure and how blood is moving through the heart. Depending on your child’s type of sickle cell disease, this test will be done periodically to monitor for changes in the heart.

- *X-Rays*

X-rays are the simplest and quickest way to find out what is going on in the body. They are most often done when someone is experiencing symptoms such as pain or cough. X-rays can detect injuries or breaks in bones. Often children with sickle cell disease and fever will have a chest x-ray to find out if pneumonia is causing the fever.

- *MRI and CAT Scans*

These two tests take a bit more time than x-rays but each gives a better picture of the inside of the body, especially the soft organs of the body. Each uses a machine to take many pictures from different angles. A computer pieces these pictures together to give a detailed image. CT, often called “CAT”, scan, uses x-rays and the MRI uses magnetic and radio waves. MRI of the joints is often done to see if there is damage from sickle cell disease. An MRI or CT scan of the brain may be done if there is a worry about a stroke.

- *Pulmonary Function Tests*

Pulmonary function tests, or PFTs, are done to measure the health of the lungs. The lungs have the job of bringing oxygen into the body and transferring it to the blood. Red blood cells will become sickle-shaped if the oxygen in the body is decreased. For this reason, PFTs will be done beginning in school aged children and then periodically to detect changes in the lungs. PFTs are also helpful in diagnosing asthma, often when there have been no symptoms of this in the past. During this test, a person is asked to breathe in and out deeply and measurements are taken.

- *Sleep Study*

A sleep study takes place in the hospital setting. The purpose is to measure the quality of sleep. A sleep study can detect if there is a drop in the level of oxygen being inhaled during sleep. Apnea, a brief period without taking breaths during sleep, can also be detected during a sleep study. Just as is the case with PFTs, this test is important in detecting drops in oxygen in the body that can lead to red cells becoming sickle shaped.

- *Neuropsychological Testing*

This is a series of tests done by a trained psychologist to determine how someone thinks and learns information. These tests help to identify a child’s strengths and weaknesses. This information is then shared with the school where it can be used to maximize the child’s educational experience. This testing is often performed after a parent or educator has expressed concerns with school performance. It can be helpful if neuropsychological testing is done before problems arise at key times in a child’s education. Examples of these key times are the start of kindergarten, middle school and high school.

Staying Well

Routine Medical Care

As it has been mentioned, sickle cell disease is a health condition that has acute or sudden, complications and chronic, or lifelong, complications. But, for the most part, people with sickle cell disease feel well the other times. As with any other health condition such as high blood pressure, diabetes, or asthma, there are steps one must take to stay well.

- Taking all medications prescribed, according to the instructions given by your doctor or nurse practitioner is very important. This is especially true with penicillin.
- Be prepared by keeping important health records close at hand. When seeing a health care provider or visiting the emergency room, this information will be very valuable in providing an accurate picture of your child's health.

These records should include:

- Names and contact information for all health care providers
 - A copy of your child's insurance card
 - List of allergies,
 - Type of sickle cell disease
 - List of sickle cell disease complications experienced
 - List of other health problems
 - All medications that have been prescribed (bring labeled bottles to doctors' appointments and the emergency room)
 - Most recent lab results (you can get this from your health care team)
- Provide materials such as this book to others who will spend time with your child. This may include babysitters, grandparents, schoolteachers, etc.
 - Take your child to your regular doctor (pediatrician or family doctor) for childhood shots and regular check-ups.
 - Keep appointments with the sickle cell health care team.
 - Make sure to see the dentist twice a year.
 - Update your pharmacist to any new allergies or side effects of medications.
 - Let every doctor, nurse or dentist who sees your child know that sickle cell disease has been diagnosed.
 - Make your health care providers aware of any dietary supplements, complementary medicines, and herbal remedies. Sometimes, these types of treatments can interfere with prescribed medications. It is the goal of your health care team to work with you to include complementary treatments into your child's plan but this must be done safely.
 - Practice safety rules of childhood. This includes using car seats and seat belts, bicycle helmets, keeping medications out of reach, etc.

Hospital Stays

Unfortunately, living with sickle cell disease means that some time will be spent in the hospital. It is important to consider these things to make the transition to the hospital and back home go as smoothly as possible.

- Hospital stays can be frightening for a child. As much as possible, make arrangements for you, or others with whom your child is comfortable, to be at the bedside. Understandably, this is not always possible. When leaving, make it very clear that you will return, perhaps by leaving something personal with your child.
- Child Life Specialists are trained to work with hospitalized children through play. Be sure to introduce your child to the Child Life Specialist.
- Be honest with your child about what will take place in the hospital. Use clear and age appropriate words to maintain a trustful relationship.
- Remind hospital staff members that painful procedures take place in a room specially designated for this purpose, often called the “treatment room”. This will help your child always feel safe in his or her hospital bed.
- You, as the parent or caregiver, need to establish relationships with hospital staff members. Quickly seek reassurance if you have concerns or questions. This can be done by directly speaking with the staff, your child’s sickle cell team, a social worker or counselor, or the manager of the hospital unit. Your child will trust those taking care of him or her, only if you trust them as well.

Anesthesia and Surgery

Let your sickle cell team know before your child receives anesthesia or sedation for any type of surgery or medical procedure. It is important for your child's sickle cell specialist to know if anesthesia is planned even for dental work. Special precautions are required for a child with sickle cell disease before he/she receives any type of anesthesia. This must be discussed with your doctor before any procedure is done.

Growth and Development

Children with sickle cell anemia may be shorter than their friends of the same age. They also may have a delay to the start of puberty (sexual development). Children’s final height is determined by the height of their parents. Most children with sickle cell disease will reach this expected height but this might be at a later age when compared to other children. This may be a source of concern or embarrassment for some children. Maintain the conversation on the topic of growth with your Sickle Cell health care team.

A peer support group of other children with sickle cell disease may help your child deal with feelings and questions. Camps for children with sickle cell disease offer a great opportunity to be around others in a fun environment. Ask your child's health care team about these types of activities.

Nutrition and Diet

Your child should:

- Eat a well-balanced diet appropriate for the child's age group. There is no special diet for a child with sickle cell disease
- Drink plenty of fluids to avoid dehydration
- Water is always best. Sugary drinks such as sports drinks, soda and juice can decrease a child’s appetite
- Drink extra fluids during increased physical activity, when temperatures are warm, and during fevers

- Avoid caffeinated drinks like regular coffee, iced tea and cola as they can lead to dehydration
- During an acute complication, it is more important to drink fluids than it is to eat
- Speak to your doctor or nurse before adding vitamins or supplements to the treatment plan

Physical Activity

- Children should take part in any physical activities that interests them. Let your child set his/her own pace and rest when the body tells him/her to rest. Children learn at an early age to read the signals their bodies send them. It is important to trust your child so he/she learns to manage his/her health independently.
- Light aerobic exercise like swimming, walking, and biking are great for keeping fit and healthy. The body loses more fluid during exercise so it is important to drink more fluids.
- Swimming in very cold water may trigger a painful crisis in some children. Before the child goes swimming, make sure the water is not too cold. If the water feels cold to the touch, it is probably too cold for swimming. Children should dry off with a towel as soon as they come out of the water.
- Any adult overseeing your child during physical activity should be made aware of the fact that your child has sickle cell disease.

Weather

- Extremes of heat and cold temperatures should be avoided. Extreme temperatures may trigger a crisis.
- Too many clothes in hot weather should be avoided. Warm clothes are needed in cold weather but the child should not be overdressed. Dressing in layers allows your child to take off items as the day warms up or he or she goes inside.

Travel

- Pack a thermometer, up-to-date medical information, insurance card, and medications into your carry-on bags. This way you will have them should your luggage become lost. Medications should be in the original labeled bottles to avoid confusion.
- People living with sickle cell disease can have trouble during flights at high altitudes in a non-pressurized airplane. Since commercial airlines are pressurized, flights on these planes are not a problem.
- It is a good idea to discuss travel plans with the Sickle Cell Team. They can often suggest a hospital or doctor who may be helpful if your child has a problem far from home.

Discipline

It is very important to remember that a child is first and foremost, a child. Consistent discipline is needed and should never be compromised because of sickle cell disease. Children require limitations and consistency to develop a secure view of life. As parents and guardians, you must determine what your parenting style will be and discipline should be considered in this decision.

Pregnancy

Females with sickle cell disease can become pregnant. But, there are risks to both the mother and developing baby that will require very close follow up with an obstetrician trained in high risk pregnancy. Contraceptives (birth control) should be used to avoid unplanned pregnancy by all sexually active teenage girls and young women.

Sickle Cell Disease and School

Your child can grow and become almost anything he or she wants to be in life. There are people with sickle cell disease who have become doctors, lawyers, teachers, nurses, etc.

Your child should be urged to fully participate in school. It is important for you and school staff to understand, however, that your child may miss some school because of sickle cell disease. There are laws that exist to make sure your child receives a fair and equal education, despite living with sickle cell disease. You, your health care team and the school will work together to make sure your child succeeds throughout his or her educational career.

It is important to talk to your child's teachers and school nurse about sickle cell related problems your child may have at school. The nurses, social workers and counselors at your sickle cell treatment center are available to help the teachers, nurse, principal, and your child's classmates understand sickle cell disease.

The School Should Know:

- How to reach you or another adult if your child becomes ill at school
- The telephone number of the Sickle Cell Treatment Center and pediatrician
- Your child will need to drink water throughout the day to stay well
- Your child will need to take frequent restroom breaks
- Your child may tire more easily in physical education and recess
- Your child should not be exposed to hot or cold temperatures without proper clothing when walking to and from school, waiting for a school bus, or during gym class and fire drills
- Your child should be sent to the school nurse if he/she does not feel well

You Should Know That It Is Important to:

- Call the school when your child is going to be absent
- Arrange for homework to be sent home when your child must miss school
- Urge your child to keep up with schoolwork that is missed during absences
- Talk to the guidance counselor or child study team leader about tutoring and other services that may be available to your child
- Talk to the school nurse about developing an individualized health plan (IHP) for your child

Information on the Internet

There is an enormous amount of very helpful information available on the internet. It is important to look to sites that are accurate and up-to-date. Information from government agencies such as the National Institutes of Health or the Center for Disease Control, and state health agencies can be considered reliable. Hospitals often have information on their websites that is dependable as well. Avoid sites where products described as treatments or cures are mentioned, as they may not be trustworthy. When in doubt, email or print the information to share with your health care team.

Medical Words

Acute Chest Syndrome - a pattern of chest or back pain, cough and difficulty breathing that may signal the start of a sickle cell crisis in the chest; a pneumonia

Anemia - hemoglobin that is lower than normal values

Aplastic Crisis - bone marrow temporarily stops making red blood cells

Aseptic Necrosis - gradual breakdown or destruction of bones usually involving the hips, knees or elbow joints (Also known as Avascular Necrosis or AVN)

Bilirubin - yellow substance that is the result of the breakdown of red blood cells

Bone Marrow - the spongy red center of bones where blood cells are made

Carriers - persons who carry a gene for a disease but do not have the disease

Chemotherapy - medicines that are used to fight cancer

Chronic Transfusions - a blood transfusion that is given about once each month to hopefully prevent further complications from sickle cell disease

Crisis - an emergency related to sickle cell disease; often used to describe an episode of sickle cell pain (vaso-occlusive crisis)

Dehydration - loss of body fluids

Diarrhea - frequent and watery bowel movement

Genes - message carriers that are passed on to children through the mother's egg and the father's sperm. Genes carry the messages for things like eye color, hair color, blood type, and the kind of hemoglobin a person will have

Genetic Centers - Medical facilities where Genetic specialists can arrange for genetic tests and can answer questions on how genetic conditions may be passed down in families

Geneticist - a doctor who specializes in hereditary conditions

Hematologist - doctor who specializes in blood disorders

Hematuria - blood in the urine

Hemoglobin - part of blood within the red blood cells that carries and delivers oxygen to all parts of the body

Hemoglobin SC Disease - a type of sickle cell disease in which one parent passed the gene for sickle cell disease and the other parent passed the gene for hemoglobin C disease

Hemoglobinopathy - a disease or condition that causes the body to make abnormal hemoglobin

Inherited - passed on from the father and/or mother to their child

Jaundice - yellow pigment (color) to eyes and skin

Meningitis - brain/central nervous system infection

Osteomyelitis - bone infection

Platelets - part of the blood that helps make blood clot

Pneumonia - lung infection

Priapism - prolonged, unwanted painful erections

Sepsis - serious blood infection

Sickle BetaThalassemia - a type of sickle cell disease in which one parent passed the gene for sickle cell disease and the other parent passed the gene for beta thalassemia

Sickle Shaped Cells - red blood cells that look sickle-shaped (“half moon”, “frowns”) under a microscope

Hemoglobin SS Disease (Sickle Cell Anemia) – type of sickle cell disease which results from both parents passing along the gene for sickle cell disease

Sickle Cell Trait - a carrier condition in which a normal hemoglobin gene is inherited from one parent and a sickle hemoglobin gene from the other

Spleen - an organ on the left side of the belly that filters and removes bacteria and old blood cells from the blood

Splenic Sequestration - spleen swells with trapped blood

Stroke - blood becomes trapped in the vessels of the brain causing permanent brain damage from the lack of oxygen

Beta Thalassemia - an inherited blood disorder that results in the production of abnormal hemoglobin

Urination - passing water or peeing

Vessels - the tubes that blood flows through such as veins, arteries, and capillaries

