

Sickle Cell Disease



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SICKLE CELL DISEASE

A HANDBOOK FOR FAMILIES

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■ WHAT IS SICKLE CELL DISEASE?

Sickle cell disease is an inherited blood disorder that affects the ability of red blood cells to carry oxygen throughout the body. Normally, red blood cells are round and flexible. Sickle cell disease causes red blood cells to change into a curved, or sickle, shape. These sickle-shaped red blood cells stick together, blocking blood flow to the hands, feet, joints, bones, and major organs. When blood flow to these areas decreases, the tissues do not get enough oxygen, causing pain and other problems. To understand how sickle cell disease affects the body, it helps to understand a little more about blood.

■ WHAT IS BLOOD?

Blood is a mixture of cells, proteins, and a watery substance called plasma. The cells in blood are made in our bone marrow, the sponge-like center of the bones in our skull, spine, pelvis, and ribs. Like water being pumped through a hose, our blood is pumped by our heart through small tubes called blood vessels. There are two types of blood vessels: arteries and veins. The arteries carry blood away from the heart and bring oxygen to the body's tissues. The veins then return the oxygen-poor blood from the tissues back to the heart.



Blood has many different functions. It carries nutrients and gases throughout the body to our tissues and organs. Blood carries waste products away from the tissues and organs as well. Blood also helps fight infection and heal wounds.

Blood has four major components:

- Plasma—a yellowish liquid consisting mostly of water. Plasma acts as a river, helping the white blood cells, red blood cells, and platelets flow freely through the blood vessels. It also carries nutrients, hormones, proteins, and waste products around the body.
- White blood cells—the fighter cells, also known as leukocytes. White blood cells, which are part of the immune system, protect the body from infection and disease.
- Platelets—the component that helps stop bleeding. Platelets form a scab after skin or tissue has been injured.
- Red blood cells—making up the majority of the cells in our blood, these small cells, also known as erythrocytes, are slightly flattened, so they look like a doughnut with the hole filled in. The function of red blood cells is to carry oxygen, carbon dioxide, and nutrients throughout the body. Red blood cells contain a protein called hemoglobin, which carries oxygen throughout the body. The red blood cells pick up oxygen in the very small vessels of the lungs and carry it to the body's tissues and organs. The cells of the tissues use the oxygen and create carbon dioxide, a waste product. After red blood cells drop off oxygen at the tissues, they pick up carbon dioxide and carry it back to the lungs to be exhaled from the body.

■ HOW DOES SICKLE CELL DISEASE AFFECT THE BLOOD?

Red blood cells normally contain hemoglobin A, or adult hemoglobin. In sickle cell disease, the child inherits an abnormal gene for the production of hemoglobin, which is called a gene mutation. This gene mutation causes the body to make hemoglobin S, or sickle hemoglobin, instead of hemoglobin A. Sickle hemoglobin is not efficient at carrying oxygen. Under certain stressful conditions, such as fever and dehydration, the red blood cells change into a rigid sickle shape, like a crescent moon. When this happens, the sickle-shaped cells get caught on each other and on the blood vessel walls, instead of flowing easily through the vessels like soft, round, normal blood cells. When these sickle-shaped cells become caught on each other, they block the flow of blood through that vessel and prevent the tissues from getting fresh, oxygenated blood. This is what causes pain, the most well-known symptom of sickle cell disease. However, this lack of oxygen is also responsible for many other problems, which will be addressed later.

Red blood cells that contain hemoglobin A live for almost 120 days. However, because they can sickle and become rigid, cells containing hemoglobin S live only 10–20 days before breaking apart. This breaking apart of red blood cells is called hemolysis, and the result is a low hemoglobin level in people living with sickle cell disease. Anemia is the medical term for a low hemoglobin level.

■ WHO GETS SICKLE CELL DISEASE?

Sickle cell disease is a genetic disorder, meaning it can be inherited from a parent who carries the gene mutation for sickle cell disease. Many people associate sickle cell disease with Africans. Although it is extremely common in Africa, sickle cell disease actually began in four areas of the world with a high incidence of malaria: Africa, India, the Mediterranean, and the Middle East. It is believed that the sickle cell mutation developed as a way to protect people living in these areas from malaria. Throughout the years, intermarriage between ethnicities and population migration to other countries has made sickle cell a global disease. Today, there are people with sickle cell disease living in almost every country.



■ HOW IS SICKLE CELL DISEASE INHERITED?

Sickle cell disease is inherited in the same way that many of our genetic traits, such as eye color, are inherited from our parents. Sickle cell disease is a recessive condition that occurs when a child inherits the sickle cell gene from both parents, one from the mother and one from the father. If a child only inherits one sickle cell gene from either parent, then he or she will have sickle cell trait and be referred to as a “carrier.” People who live with sickle cell trait do not experience the symptoms or problems that those who live with sickle cell disease experience.

■ WHAT ARE THE TYPES OF SICKLE CELL DISEASE?

Although there are many types of sickle cell disease, four types are most common: sickle cell anemia (HbSS), sickle hemoglobin C disease (HbSC), sickle beta-plus thalassemia (HbS- β^+ thal), and sickle beta-zero thalassemia (HbS- β^0 thal).

Sickle Cell Anemia, or Hemoglobin SS (HbSS)

HbSS is the most common and severe form of sickle cell disease. In this form, each parent has passed on a mutated gene. As a result, the body produces abnormal hemoglobin.

Sickle Hemoglobin C (HbSC)

Another gene mutation that can cause the body to make abnormal hemoglobin is hemoglobin C. If one parent passes along a mutated hemoglobin S gene and the other passes along a mutated hemoglobin C gene, the child is diagnosed with hemoglobin SC, or HbSC. Patients with HbSC tend to experience milder symptoms than those diagnosed with HbSS. However, some patients may experience the same severe symptoms as those with HbSS.



Sickle Beta Thalassemia

Beta (β) thalassemia is another inherited disorder that affects the amount and quality of hemoglobin made by the body. The combination of the beta thalassemia gene with the sickle cell gene leads to a diagnosis of sickle beta thalassemia, which is a form of sickle cell disease. Sickle beta-zero thalassemia (HbS- β^0 thal) means there is no normal hemoglobin being made by the body. It is usually a serious form of sickle cell disease. Sickle beta-plus thalassemia (HbS- β^+ thal) means there is a decreased amount of normal hemoglobin being made and is usually a milder form of sickle cell disease.

■ WHAT ARE THE COMPLICATIONS OF SICKLE CELL DISEASE?

Because sickle cell disease affects blood, and blood travels to every part of the body, problems can occur almost anywhere in the body. The problems of sickle cell disease are a result of the damage caused by the sickle-shaped red blood cells and the chronic anemia caused by hemolysis. These sickle-cell problems can be acute, which means they have a rapid onset and are immediately obvious, or chronic, which means they occur over time and are subtler.

Acute Complications

INFECTION AND FEVER

People with sickle cell disease are at an increased risk for life-threatening infections. The spleen is an organ that serves as a filter to help remove bacteria from the bloodstream and also helps produce some white blood cells. Sickle-shaped red blood cells can damage the spleen early in life. The damaged spleen is no longer able to filter bacteria out of the bloodstream. As a result, bacteria can travel through the

bloodstream and, in just a few hours, cause an overwhelming infection. These infections can cause shock and even death. Consequently, every time a person with sickle cell disease has a fever, it must be considered an emergency—a healthcare provider must be contacted and medical attention must be sought immediately. Your child will need laboratory work, including a blood culture and, possibly, a chest X ray. Antibiotics will be given either intravenously or by injection. Your child may be hospitalized, but in most cases he or she will receive antibiotics in the clinic or emergency department.

It is impossible to know just by looking at your child whether his or her fever is caused by bacterial sepsis or by another, less serious reason. However, bacteria can quickly and severely affect the body, which is why it must be assumed the fever is caused by bacteria, and there should be no delay in seeking treatment.

SPLENIC SEQUESTRATION

Oxygen levels in the spleen are low and the blood moves through it slowly. Because of this, red blood cells tend to sickle in the spleen. In some cases, this can result in a blockage of blood flow out of the spleen and trap red blood cells and platelets. Normally, the spleen is tucked under the left side of the rib cage. When blood becomes trapped, the spleen swells and there is a rapid fall in hemoglobin and platelets as the amount of blood circulating in the rest of the body decreases. This condition is known as splenic sequestration and is a medical emergency. Sequestration is most common in children with HbSS and HbS- β^0 thal. It can occur, however, in the less severe diseases of HbSC and HbS- β^+ thal, but usually at a later age. Your hematology team will show you how to recognize what feels normal and what feels swollen for a spleen. Often, splenic sequestration will happen repeatedly. It may be necessary for your child's spleen to be removed surgically in an operation called a splenectomy. If this is the case, your hematology team will discuss all of the risks and benefits of having a splenectomy.

PAIN CRISIS (VASO-OCCLUSIVE CRISIS)

Under certain conditions—such as exposure to extreme temperatures, fever, dehydration, infection, or stress—red blood cells can become sickle-shaped. This may cause a blockage of blood vessels, preventing oxygenated blood from getting to the body's tissues. This lack of oxygen can be very painful. Damage to the blood vessels and swelling in the area of the blockage can increase pain. Pain crisis, also called vaso-occlusive crisis, is the number one reason people living with sickle cell disease



seek medical attention, and it is the most common reason they are admitted to hospitals. Dactylitis, or “hand and foot syndrome”, is often the first type of pain babies and toddlers with sickle cell disease experience. Dactylitis is swelling and extreme tenderness in the hands, feet, or both. Older children and adolescents will mainly experience pain in their long bones (arm and legs), chest, and back.

Your child's healthcare provider will prescribe pain medications and give instructions on their administration. Sometimes home treatment is not enough to relieve the pain. In these cases, admission to a hospital may be necessary.

ACUTE CHEST SYNDROME

Acute chest syndrome is a very serious complication of sickle cell disease and is the second most common reason people are hospitalized. Acute chest syndrome is the result of sickle-shaped red blood cells blocking the tiny vessels of the lungs. It is characterized by chest pain, difficulty breathing, fever, and changes to the lungs as seen on an X-ray. Acute chest syndrome may be caused by pneumonia. A pain crisis may lead to acute chest syndrome, because pain makes it difficult to expand the lungs with deep breaths. Pain medications can make a child drowsy, which also interferes with taking deep breaths. For this reason, you and your child will be taught deep-breathing exercises, called incentive spirometry, to be used during a pain crisis to help prevent acute chest syndrome.

Often, individuals experiencing acute chest syndrome will need a red blood cell transfusion. Given early, a transfusion may prevent transfer to an intensive care unit or the need to use a ventilator.

APLASTIC CRISIS

Parvovirus B19 infection, known as fifth disease, is a very common childhood infection that causes mild fever, cold symptoms, and a rash on the arms. The cheeks also may be bright red. Parvovirus B19 infection can shut down the production of red blood cells in the bone marrow for as long as 10 days. For most children, this does not cause any serious problems. However, the red blood cells of children with sickle cell disease live only a short time, so parvovirus B19 infection can result in severe anemia or a drop in hemoglobin. Because the bone marrow has shut off red blood cell production, there is a very low or absent reticulocyte count (reticulocytes are young red blood cells). If your child develops signs of worsening anemia, such as fatigue, severe headache, or extremely pale lips and nail beds, your hematologist will suspect an aplastic crisis.



STROKE

Over time, sickle cell disease may damage the blood vessels of the brain, making them narrower. Sickle-shaped red blood cells can block these damaged vessels and prevent oxygen from getting to the brain tissue, causing a stroke which can result in permanent brain damage. A child who is suffering a stroke may have any of these symptoms: severe headache, weakness on one side, inability to speak or understand language, seizures, drooping facial features, or trouble swallowing. A child may have a transient ischemic attack (TIA), during which these symptoms resolve in fewer than 24 hours. If you suspect your child is having a stroke or TIA, seek immediate medical attention. The sooner medical treatment begins, the less damage the stroke can cause.

Approximately 10% of children with HbSS and HbS- β^0 thal are at risk of suffering a serious neurological event. The average age for experiencing such an event is 8 years old. A child who has suffered one stroke or TIA has an extremely high risk of having another, possibly more serious, one. For this reason, regular red blood cell transfusions are started. The goal of regular red blood cell transfusions is to replace the child's blood, which can sickle and cause further blockages, with the normal red blood cells of a donor. Transcranial Doppler (TCD) imaging is now used routinely as a screening tool to determine if a child is at risk for stroke. TCDs detect abnormal flow through vessels of the brain that have suffered damage from sickle-shaped red blood cells. An abnormal TCD is a strong predictor of stroke. Children found to have this should be offered regular transfusions. Annual screening with TCD should begin at 2 years of age for children with HbSS and HbS- β^0 thal.

PRIAPISM

Priapism is a complication experienced by some males with sickle cell disease. Sickle-shaped red blood cells can block the blood from flowing out of the penis. The result is a long-lasting and unwanted erection of the penis that is often extremely painful. Priapism is a medical emergency, especially if it lasts more than 2 hours. It is important that caregivers and health professionals instruct boys to let someone know when they are experiencing priapism. If left untreated, priapism may lead to an inability to have a normal erection in the future. Priapism treatments vary, but always include intravenous fluids and pain medication. Sometimes blood transfusions and medications that open up blood vessels are needed. If these measures do not work, a surgeon trained in this field, known as a urologist, will use a surgical procedure to release the trapped blood. It is common for priapism to happen again. Preventive measures, such as taking hydroxyurea and medications that dilate, or open, blood vessels, may need to be used on a daily basis.

Chronic Complications

EYES

Bilirubin is a yellow substance inside red blood cells. Because sickle-shaped red blood cells are fragile, they can break open easily and release bilirubin into the circulation, causing the yellow tint to the eyes that is often seen in people with sickle cell disease.

The retina is the thin lining on the inside of the eye that receives images the eye sees and changes them into messages the brain can read. The blood vessels of the retina are extremely tiny. Over time, the small blood vessels of the retina can become blocked by sickle-shaped red blood cells. To help the blood flow, the body creates new blood vessels to carry blood to the retina. However, these new vessels are weak and tend to break open, causing bleeding and damage to the retina, which is called sickle cell retinopathy. Sickle cell retinopathy has no symptoms in its early stages, which is when it is most easily treated by laser surgery. For this reason, children should have yearly eye exams, beginning in kindergarten. Sickle cell retinopathy is most common in people with HbSC disease and in older adolescents and adults.



HEART AND LUNGS

The heart is a muscle. Its job is to pump blood to the lungs, which is where the blood picks up oxygen and carries it to all the tissues in the body. Because people with sickle cell disease have lower hemoglobin levels and less oxygenated blood, their hearts must work harder. As a result, it is very common for them to have cardiomegaly, or an enlarged heart. Individuals with sickle cell disease are often told that their hearts have a murmur, or unusual sound. These murmurs are most often the result of chronic anemia.

Nearly one-third of adults with sickle cell disease will develop a condition known as pulmonary hypertension. Pulmonary hypertension is high blood pressure in the artery that supplies the lungs with blood from the heart. Pressure builds up as this artery becomes damaged by sickle-shaped red blood cells and becomes narrower. This increased pressure makes the heart's job much more difficult. The right side of the heart, which is the side that pumps blood into the lungs, can become enlarged and weak. The symptoms can be shortness of breath and dizziness. These symptoms often are not felt until pulmonary hypertension has progressed. Fortunately, pulmonary hypertension can be detected early by a simple test called an echocardiogram.

Conditions that affect the lungs and the amount of oxygen that comes into the body, such as asthma and sleep apnea, must be controlled in a person living with sickle cell disease to prevent sickling and, thus, long-term problems. If your child is diagnosed with one of these conditions, it is very important that he or she is under the care of the appropriate specialist and all of the treatment advice is followed.

KIDNEYS

The main job of the kidneys is to filter waste out of the blood. More than 1 million tiny filters inside the kidneys, called nephrons, remove the waste. The collected waste combines with water, which is also filtered by the kidneys, to make urine. The urine then leaves the kidneys and travels down the ureters into the bladder. When the bladder is full enough, the pressure causes a person to feel like he or she has to go to the bathroom. Urine then leaves the body through the urethra.

Sickle-shaped red blood cells can block the tiny vessels of the kidneys and damage the nephrons, causing the urine to be less concentrated than it should be. This results in an increased amount of urine and makes it very easy to become dehydrated. It is also the reason many children with sickle cell disease experience bed-wetting, known as nocturnal enuresis. These children can be treated the same as children who wet the bed and do not have sickle cell disease. Treatment for bed-wetting includes limiting fluids in the evening, using bed-wetting alarms, and, in some cases, taking medications.

If the nephrons continue to be damaged by sickle cell disease, it is possible for the kidneys to lose their ability to filter the blood. In the worst-case situations, kidney failure may occur.

Early detection of kidney damage only requires a simple urine test. The test looks for protein the filtering system was supposed to catch but instead escaped into the urine. This test will also look for blood in the urine, which is called hematuria. The first urine sample of the morning gives the best indication of the kidneys' health. A nephrologist, a kidney specialist, may be consulted if there is blood or protein in the urine.

GALLBLADDER

The gallbladder is an organ in the digestive system that stores bile, which is needed to help digest fats. Bile is made from bilirubin, a substance contained in red blood cells. When red blood cells break open, or hemolyze, the bilirubin is released into the bloodstream. Because people with sickle cell disease have a high rate of hemolysis, they have an excess of bile that can cause gallstones to form in the gallbladder. Gallstones may cause sharp, sudden pains in the upper-right abdomen, which may worsen after eating fatty or spicy foods. These stones are diagnosed by an abdominal ultrasound. Sometimes the gallbladder can become inflamed, resulting in fever, pain, or both. When this happens, the gallbladder may need to be surgically removed.

JOINTS

The blood supply to the body's joints is limited. This is especially the case in "ball-and-socket" joints like those found in the shoulders and hips. Avascular necrosis is a problem that occurs in these joints after the blood flow is repeatedly blocked by sickle-shaped red blood cells. The femoral head, located at the top of the thigh bone, or femur, is at greatest risk for this problem. Avascular necrosis feels like arthritis pain, with stiffness and achiness, especially in the morning or after sitting for a while. If a child is still growing, physical therapy can be very helpful in healing the joint. In people who have finished growing, physical therapy can help relieve the pain, and surgery may possibly be postponed. In some cases, a joint-replacement surgery is needed.

LEARNING ISSUES

It is possible for children living with sickle cell disease to have some brain damage that goes undetected. This damage can be narrowing of the blood vessels that supply the brain with blood or even silent strokes. Silent strokes are strokes that cause no physical symptoms but can be seen on an MRI scan or CT scan of the brain. Also, like children who live with other chronic illnesses, children with sickle cell disease may miss many days of school and have days when they are not at their best. This can lead to learning challenges. It is recommended that children with sickle cell disease periodically undergo neuropsychological testing, a type of evaluation that determines a child's strengths and weaknesses when it comes to learning.

GROWTH AND DEVELOPMENT

Chronic anemia (low hemoglobin) and high-calorie needs may cause children with sickle cell disease to experience delayed growth. They may also have a later onset of puberty. It is important to keep in mind, however, that your child will experience the changes of puberty, although it may be at an older age than children who do not have sickle cell disease. Your sickle cell healthcare team will weigh and measure your child regularly. If your child does not have any signs of puberty or begins to fall behind the growth curve, they may refer your child to an endocrinologist. An endocrinologist is a growth and hormones specialist.



SKIN

Leg ulcers are poorly healing skin sores typically found on the lower leg. They can occur in 10%–15% of older teen and adult patients, usually with HbSS or HbS-β⁰ thal. These ulcers are the result of poor blood flow and usually develop from an injury to the skin. Leg ulcers are very difficult to treat, but many methods used to treat ulcers associated with other health conditions, such as diabetes, are often used to treat ulcers associated with sickle cell disease. In many cases, regular transfusions of red blood cells are given to raise the hemoglobin, dilute the sickle cells, and, hopefully, promote healing. It is important to make your healthcare providers aware of any poorly healing wounds, especially on the legs.

PREGNANCY

A woman with sickle cell disease may get pregnant. However, it is common for the disease and its complications to become more severe during pregnancy, because it is a very stressful time for a woman's body. There are risks to the developing baby as well. Pregnancy in a woman living with sickle cell disease is considered high-risk and must be closely monitored by both a high-risk pregnancy specialist and a hematologist.

IRON OVERLOAD

Iron is needed by the body to make hemoglobin. Hemolysis results in the release of iron into the body so that it may be used to make new red blood cells. Because of the increased rate of hemolysis in individuals living with sickle cell disease, there is the potential for iron to build up in the body. Blood transfusions also can contribute additional iron to the body. Your child's hematology team will monitor the results of a laboratory test called ferritin to determine if iron levels are becoming too high. The body has no way of getting rid of iron naturally. If the ferritin level is very high, medications called iron chelators may be used to remove excess iron to prevent organ damage.

■ WHEN WILL MY CHILD NEED TO BE IN THE HOSPITAL?

From time to time, children diagnosed with sickle cell disease will need to spend time in the hospital. The most common reasons for hospitalization are fever and pain that cannot be relieved by medications at home. Other reasons include aplastic crisis, splenic sequestration, and respiratory symptoms.

Some children with sickle cell disease may need surgery. Because of chronic anemia and the potential for sickling of the red blood cells, these children are at greater risk for complications from surgery and anesthesia. These complications include infection, acute chest syndrome, and stroke. Before surgery can take place, children often will need to receive fluid by vein, and possibly a blood transfusion. Following surgery, they will need to be observed closely for complications.

■ WHAT TESTS AND PROCEDURES WILL MY CHILD NEED?

Blood Tests

Often, it will be necessary for your child to have blood tests to monitor his or her sickle cell disease, look for complications, and check responses to treatment. The following are some common blood tests:

- Hemoglobin electrophoresis—diagnoses sickle cell disease. This test measures the different types of hemoglobin to determine if your child has sickle cell disease, and if so, which type.
- Complete blood count (CBC)—determines the number of white blood cells (infection-fighting cells) and platelets (clot-forming cells) and the level of hemoglobin (degree of anemia) in your child's body. A reticulocyte count, which measures the rate at which the body is producing new red blood cells, is often done with a CBC.
- Chemistry panel—checks that your child's body has the correct amount of important substances such as electrolytes, protein, and sugar. It also checks the liver and kidney function, as well as nutritional status.
- Blood cultures—observed in the lab for bacteria growth. These cultures will be drawn each time your child has a fever.
- Blood type and antibody screen—helps identify the correct donor blood for your child, in case he or she needs a blood transfusion.



Urine Tests

At times, your child will be asked to give a urine sample. This sample provides information about the health of the kidneys and whether there might be an infection in the urinary tract.

Radiology Tests

Radiology tests are used to detect changes that may indicate a future problem, diagnose a complication, or monitor response to a treatment plan. The following are examples of radiology tests:

- X-rays—the most commonly used radiology test. X-rays take pictures of the inside of the body to help diagnose problems. A chest X-ray is often done to check for pneumonia and acute chest syndrome in children with sickle cell disease.
- Computed tomography (CT) scans—used less frequently but may be helpful at times. A CT scan takes multiple cross-sectional pictures that are then put together by a computer to give a better idea of what soft tissues, bones, and organs look like. The results are sometimes more helpful than an X-ray's results.
- Magnetic resonance imaging (MRI)—uses a magnetic field and radio waves to produce images similar to those of a CT scan. MRI is used when a CT scan or X-ray is unable to give enough detailed information.
- Ultrasound—a noninvasive test that uses sound waves to produce images of the inside of your child's body. An abdominal ultrasound may be performed to determine the presence of gallstones.
- Transcranial Doppler (TCD)—an ultrasound technique that looks at the blood vessels of the brain. TCD is a very powerful screening tool used to determine the risk of stroke in a child with sickle cell disease.

Other Tests

- Echocardiography (ECHO)—similar to an ultrasound, ECHO is a noninvasive way to look at the structure of the heart as well as how the blood flows through it. It is helpful in the early detection of pulmonary hypertension.
- Lung function tests (also called pulmonary function tests, or PFTs)—show how well your child's lungs function.
- Polysomnography (also called a sleep study)—diagnoses sleep disorders. This test can also determine if your child is having trouble getting enough oxygen while asleep.
- Neuropsychological testing—measures your child's strengths and weaknesses in areas such as learning abilities, language, memory, and attention span.

■ HOW IS SICKLE CELL DISEASE TREATED?

Penicillin

A major research study conducted in the 1980s found that penicillin taken twice a day until the age of 5 cuts the risk of death from infection by 84% in infants and toddlers. As a result of this study, many states began screening newborns for sickle cell disease so penicillin could be started right away. These newborn-screening programs have drastically improved the survival rate in early childhood. Penicillin taken twice a day, caregiver education, and immunization with the pneumococcal vaccine polyvalent (Pneumovax 23) have made a huge difference in the health of babies born with sickle cell disease.

Folic Acid

Folic acid is a vitamin needed to make hemoglobin. Because children with sickle cell disease need to make more hemoglobin, they need more folic acid. Often, the hematologist will prescribe a folic acid pill to take once a day.

Pain Medication

Pain crisis is the number one reason children living with sickle cell disease come to the hospital. However, some pain can be managed at home with medications prescribed by your child's hematology team.

There are two main types of pain medications prescribed. The first is nonsteroidal anti-inflammatory drugs (NSAIDs), such as ibuprofen (Motrin[®], Advil[®]), ketorolac (Toradol[®]), and naproxen sodium (Aleve[®], naprosyn). These medications are helpful in reducing some of the swelling associated with pain crisis, as well as helping to relieve the pain. The second group of medications is narcotic pain relievers, such as acetaminophen (Tylenol[®]) with codeine, morphine, and oxycodone (Percocet[®], OxyContin[®]).

Powerful medications such as codeine, morphine, and oxycodone are often necessary in treating pain crisis. Although these medications have the potential to become addictive, they are safe when used as prescribed.

In the hospital, a patient-controlled analgesic (PCA) pump is often used to deliver a continuous, small amount of pain medicine (usually morphine). A PCA pump allows the child to deliver his or her own "as-needed" dose. The nurse programs and changes the settings of the PCA pump based on your child's pain level and use of the pump.

Pain medications may have side effects. The most common are itching, upset stomach, constipation, and sleepiness. If your child is experiencing any of these side effects, speak with your hematology team, and they will help your child with these side effects or even prescribe a different medication.



Transfusion

Occasionally, a transfusion of red blood cells may be necessary to treat certain situations and complications of sickle cell disease. This transfusion will be given into a vein over the course of 2–4 hours. Transfusions may be planned in advance, as is the case before some surgeries; however, a transfusion may also be needed in an emergency situation. Individuals who have had strokes or an abnormal transcranial Doppler (TCD) will likely require chronic transfusions. These are usually given every 2–4 weeks. If you have any questions or reservations about transfusions, talk to your healthcare team before an emergency happens.

Hydroxyurea

Before birth, red blood cells contain fetal hemoglobin (hemoglobin F). If a child is born with sickle cell disease, this hemoglobin F is replaced by hemoglobin S in the first few months of life. Some people inherit the ability to keep making a small amount of hemoglobin F. As they get older, these people tend to have fewer complications of sickle cell disease. A cancer medication, hydroxyurea, was found to stimulate the production of hemoglobin F. Hydroxyurea is a medication that has been found to turn on the body's production of hemoglobin F. When taken correctly and monitored carefully, hydroxyurea does not have serious side effects. Hydroxyurea has been shown in research studies to decrease the number of pain episodes and acute chest syndrome as well as the need for transfusions and hospitalizations.

Bone Marrow Transplant

Bone marrow transplant is the only cure for sickle cell disease. Because bone marrow transplant has the potential for very serious side effects, it is most often used in those living with severe sickle cell disease. Before bone marrow transplant, the child will be given chemotherapy and radiation to destroy his or her own bone marrow (where blood cells are made). He or she then receives an infusion of the donor's bone marrow or stem cells. In the weeks to months following the transplant, the donor cells take over and produce blood cells that are unaffected by sickle cell disease.

Bone marrow transplant does have risks, including serious infection, bleeding, and organ damage from the chemotherapy and radiation. Graft-versus-host disease, in which the donor's new marrow begins to attack the patient's body cells, may also develop. In some cases, these complications can be fatal. Finally, the bone marrow may not "take" or replace the patient's marrow, which is a condition called graft failure.

Currently, the standard of care is to use only HLA-matched sibling donors. However, researchers are exploring the use of nonrelated bone marrow and cord blood donors. They are also working to minimize the side effects of transplant.

■ WHAT RESEARCH IS TAKING PLACE?

Fortunately, there is a great deal of research being done in the area of sickle cell disease. Studies are taking place to improve and limit the side effects of hydroxyurea and bone marrow transplant. The following are some additional research interests.

Endothelial Dysfunction and Nitric Oxide

Research is showing that sickle cell disease is not simply a disease of red blood cells, but also a disease of the blood vessels and their lining, or endothelium. When the sickled red blood cells come into contact with the endothelium, they cause inflammation. If the endothelium becomes inflamed, the space in which the blood has to travel becomes narrower and makes a sickle-cell blockage more likely.

Nitric oxide is an important substance in the body. It helps keep blood vessels open, allowing blood to flow freely. Nitric oxide also decreases certain proteins that cause red blood cells to stick to the endothelium. Scientists have found that people with sickle cell disease have less nitric oxide than those without the condition.

Researchers are looking at ways to decrease the inflammation of the endothelium and to increase the levels of nitric oxide in people with sickle cell disease.

Gene Therapy

In 1997, researchers figured out how to insert the gene for sickle cell disease into mice. This was the first time a human disease had been inserted into a mouse. Then, in 2001, scientists had another breakthrough and were able to correct the sickle cell gene in the affected mice. Today, research continues to move toward the next extremely difficult step, which is to take that corrected gene and insert it into the DNA of a human with sickle cell disease.

Genetic Factors

Although every person with sickle cell disease inherits the same gene mutation, they do not all have the same symptoms and complications. People from the same part of the world and same racial background seem to have similar health problems, which is probably because of other genetic traits that impact sickle cell disease. As scientists learn more about the genetic traits associated with different complications, they will know more about predicting how an individual's sickle cell disease might "behave." In the future, treatment may be customized based on the genes a person has inherited along with sickle cell disease. Treatments such as bone marrow transplant and hydroxyurea therapy may then be initiated before complications, and not because of them.

■ HOW CAN I HELP MY CHILD STAY WELL?

Newborn screening has resulted in children being referred shortly after birth to centers where care is delivered by a team approach. Experienced hematologists, nurse practitioners (or physician assistants), nurses, nutritionists, social workers, and others address all aspects of care. Access is provided to the screening tests and other specialists who will be involved in your child's care. These centers are most often located within the hospital your child will be admitted to ensure seamless care.

It is important for your child to see his or her hematology team on a regular basis, even if he or she is doing well. The team will provide the education you need to care for your child in the best way. Also, the ongoing screening and testing done at these well visits is needed for the prevention and early detection of long-term complications.

Your child will continue to be monitored by his or her primary care doctor for all routine care that is unrelated to sickle cell disease. This includes immunizations, checkups, and treatment for minor illnesses or injuries. It is very important to keep up-to-date with immunizations because these will help your child avoid serious infections. In addition to the immunizations all children receive, your child should also receive the pneumococcal vaccine polyvalent (Pneumovax 23) at ages 2 and 5. The meningococcal vaccine will also be given. This will provide extra protection against the bacteria that threaten children with sickle cell disease who do not have well-functioning spleens. Your child also should have an annual flu shot.

Avoiding situations that can lead to a sickle cell crisis is very important. This means making sure your child rests as needed, stays well hydrated, and dresses properly for the weather. Teach your child good hand-washing and hygiene techniques to avoid the spread of infection.



■ IS MY CHILD'S DIET IMPORTANT?

Your child should eat a well-balanced diet that has the number of healthy calories advised for his or her age. He or she should also take a daily folic acid supplement, if prescribed. During each visit to your hematology team, your child will be weighed and measured. Your child will need the support of a nutritionist if he or she is not growing or gaining weight. It is important to maintain a healthy weight because obesity can lead to stress on the joints and heart, along with conditions such as hypertension and diabetes.

Drinking fluids, especially water, is very important for your child because dehydration can lead to sickling of red blood cells. This is especially true during stressful times, such as pain crisis, illness, or being in hot weather. Caffeinated and sugary beverages, however, should be avoided.

■ ARE MY FEELINGS NORMAL?

Feelings of guilt, fear, worry, and helplessness, among others, are common when you have a child living with sickle cell disease. Even though sickle cell disease is a serious condition, you should know there are many steps you can take to help improve your child's quality of life.

It helps to develop strong relationships with healthcare providers who can answer questions and provide treatment for your child. Your healthcare team may be able to introduce you to other parents and children affected by sickle cell disease, so you can share information, offer support, and give practical advice to one another.

Sometimes parents are anxious about what might happen to their child because they do not have sufficient or accurate information about sickle cell disease. Many parents find it helpful to learn as much as they can about sickle cell disease. Learning how to help prevent pain and to palpate the spleen are essential for keeping your child healthy. You should take your child for regularly scheduled checkups with a hematologist. You can work with your child's hematologist and sickle cell disease team to learn more about what you can do to help your child stay healthy. This way, you will also be able to help your child receive care quickly if problems develop.

Having a child with a chronic illness can trigger many emotions. Because sickle cell disease is inherited, some parents feel guilty about having "given" the disease to their child. Some parents find support in their church or other faith community, as well as their child's school counselor, social worker, and nurse. You might want to attend support groups with other parents of children with sickle cell disease. Don't be shy about asking questions or seeking help for your child.

You are an important role model for your child. When your child sees how you get information and guidance for making decisions about keeping him or her healthy, your child learns good self-care habits. Seeing you take good care of your own health and enjoy your life helps to motivate your child to follow in your footsteps.

The unknown can be frightening to children. Be honest with your child when he or she asks questions about sickle cell disease, its treatment, and its complications. Educate your child about sickle cell disease in age-appropriate words. This will help him or her feel empowered and in control. Keeping your child's developmental level in mind, allow him or her to assume responsibility for some aspects of care. This will help prepare your child for the transition of having to care for himself or herself as adulthood approaches.

■ CAN MY CHILD ATTEND SCHOOL?

School is an important part of every child's life. In addition to learning about reading, writing, and math in school, children also learn how to get along with others. Living life in ways that are similar to their siblings and peers also gives children an important feeling of being included in the "regular" world and helps them feel safe. Children who feel included and safe grow up feeling comfortable asking for help and wanting to contribute to their community.

You can help your child participate in activities that build self-esteem and self-reliance. Your child's hematologist may recommend limiting some kinds of activities; however, you can explore alternatives that will give your child opportunities to develop skills and talents outside of the classroom.

At the start of the school year, consider meeting with your child's teacher, principal, and school nurse. Beginning with preschool and kindergarten, you can help educate school personnel about sickle cell disease by giving them educational materials provided by your sickle cell disease team. Sometimes nurses and social workers from your team can call or visit your child's school and talk to your child's teachers about sickle cell disease. Ask them to share the information with others in the school. They can learn about the disease and provide support to your child during the school day.

Although children with sickle cell disease may miss some school because of doctors' visits and times when they do not feel well, it is beneficial for them to attend school as much as possible. After an illness or painful episode, it is best to send your child back to school as soon as possible. In general, children are better off when they attend school regularly.

If your child is not doing well in school, talk with his or her teachers. Your child may need to be tested for learning difficulties. Children with learning difficulties need individualized educational plans to help them succeed in school.

You can talk to members of your child's healthcare team about information they might want to share with the school to make sure your child's educational and health needs are addressed. For instance, children with sickle cell disease should be allowed to have water at their desk and to go to the bathroom more frequently than their classmates. They should also be encouraged to participate in all class activities unless advised otherwise by their sickle cell team. Each child is unique, and there may be other ways an educational plan can be designed to help your child. Your child may need tutoring to help catch up after missing school because of illness. Ask the school and your healthcare team if there are tutoring programs at school or in the hospital. Aim for your child to be healthy, happy, and successful, and work with your child's healthcare team to provide the kinds of support your child deserves.

ONLINE RESOURCES

Information for School Personnel from the State of New Jersey Department of Health and Senior Services
www.state.nj.us/health/fhs/sicklecell/index.shtml

KidsHealth for Parents—Sickle Cell Disease
http://kidshealth.org/parent/medical/heart/sickle_cell_anemia.html

National Heart, Lung, and Blood Institute; National Institutes of Health—Sickle Cell Disease
www.nhlbi.nih.gov/health/dci/Diseases/Sca/SCA_WhatIs.html

Sickle Cell Disease Association of America, Inc.
www.sicklecelldisease.org

The Sickle Cell Information Center at Grady Health System, Atlanta, GA
www.scinfo.org

Sickle Cell Slime-o-Rama Game
<http://sicklecell.starlight.org/>

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NOTES



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