

SICKLE CELL DISEASE AND OTHER HEMOGLOBINOPATHIES (HE-ma-GLO-been-NOP-a-thees)

What is it?

Hemoglobinopathies are inherited disorders involving the abnormal structure of the hemoglobin molecule, red blood cells. The hemoglobin molecules in each red blood cell carry oxygen from the lungs to the body organs and tissues. In sickle cell disease, the hemoglobin is defective. After the hemoglobin molecules give up their oxygen, some of them may stick together in long, rigid rods. These rods cause the red blood cells to become hard and to assume a sickle (banana) shape. Normal red blood cells are very flexible and move easily through small blood vessels. The sickle-shaped cells are unable to squeeze through narrow blood vessels and cause blockages that deprive the body's cells and tissues of blood and oxygen. This produces periodic episodes of pain and ultimately can damage tissues and vital organs and lead to other serious medical problems. There are hundreds of different hemoglobin types and as a result many different forms of hemoglobinopathies exist. Sickle cell anemia (Hb SS) is the most common type of sickle cell disease. There are other hemoglobin types such as hemoglobin C or hemoglobin E, which in combination with the gene for sickle hemoglobin can result in different forms of sickle cell disease (Hb SC, Hb SE, Hb S/beta thalassemia).

How do you get it?

Sickle cell disease and other sickling hemoglobin disorders are inherited in an autosomal recessive pattern. As an autosomal recessive disorder, the parents of a child with one of these conditions are unaffected, healthy carriers of the condition and have one normal hemoglobin gene and one abnormal hemoglobin gene. With each pregnancy, carrier parents have a 25 percent chance of having a child with two copies of the abnormal gene, resulting in a hemoglobinopathy. Carrier parents have a 50 percent chance of having a child who is an unaffected carrier and a 25 percent chance of having an unaffected, non-carrier child. These risks hold true for each pregnancy.

How common is it?

Sickle cell disease affects more than 50,000 Americans, primarily those of African heritage, but also those whose ancestors come from Spanish-speaking regions (South American, Cuba, Central America); Saudi Arabia;

India; and Mediterranean countries, such as Turkey, Greece, and Italy. Sickle cell disease is one of the most prevalent genetic diseases in the United States. In Missouri the disease occurs in approximately 1 in every 400 African American births.

How is it treated?

If your child needs additional testing or diagnostic evaluation, it is important that you follow through with the pediatrician's and/or specialist's recommendations for additional testing and referrals.

Any sign of illness in an infant with sickling disease is a potential medical emergency. Acute and chronic tissue injury can occur when sickled cells cause vascular occlusion. Sickling diseases can cause severe pain anywhere in the body, but most often in the hands, arms, chest, legs and feet. Complications may include, but are not limited to, the following:

- Sepsis – The first sign of infection may be an elevated fever above 101 degrees Fahrenheit. These children require immediate medical attention. They are very susceptible to pneumococcal infections.
- Acute chest syndrome – A serious condition caused by infection and/or trapped sickled red blood cells in the lungs. Symptoms may include dyspnea, coughing and chest pain.
- Hand-and-foot syndrome – When the small blood vessels in the hands or feet are blocked, pain and swelling can result along with fever. They may be the first symptom of sickle cell anemia in infants.
- Stroke – Cerebral vascular occlusion due to sickled cells can affect even very young children. Any loss of consciousness or weakness in an extremity should be evaluated promptly.
- Aplastic Crisis – The bone marrow stops producing red blood cells, resulting in severe anemia. The child may appear tired and less active than usual.
- Painful episodes – The pain of sickling disorders is acute and prescription medications for pain relief may be required even for very young children.
- Splenic sequestration - Early signs include pallor, enlarged spleen and pain in the abdomen due to accumulation of sickled cells in the spleen. This complication can result in circulatory collapse or shock, with sudden death, if not recognized and treated immediately.

The National Institutes of Health clinical guidelines for management of sickle cell disease state, "Penicillin prophylaxis should begin by 2 months of age for

infants with suspected sickle cell anemia, whether or not the definitive diagnosis has been established." Antibiotic therapy should continue until at least 5 years of age. An alternative antibiotic is available for children who are allergic to penicillin therapy. Prescription pain medication also may be indicated during sickling crises. Health care monitoring and maintenance with appropriate immunizations is imperative to the health of the baby, and pneumococcal conjugate vaccine immunizations also are recommended, beginning at 2 months of age.

Regular visits to the pediatric hematologist, strict compliance in medication administration and blood tests as ordered by the hematologist are crucial to the health and future well being of the baby.

Parents of infants with sickle cell disease should be instructed in all aspects of routine childcare. They should be able to accurately check the infant's temperature. They must be able to recognize early symptoms of complications, including the warning signs of inactivity, fever, pallor and respiratory distress. Parents should be taught to palpate the infant's spleen and to recognize splenic enlargement. Parents must understand the importance of prompt assessment of the infant by a pediatric hematologist when fever, pallor, unexplained irritability, diarrhea, vomiting or other signs of illness are present. Fever above 101 degrees Fahrenheit requires immediate medical evaluation.

Genetic counseling services are recommended for individuals with hemoglobinopathies and for those who carry the abnormal traits, particularly concerning future pregnancies. These individuals may have questions about the disorders that are best answered by hematology specialists and genetic counselors.

Where can I get services?

Cardinal Glennon Memorial Hospital for Children
St. Louis, MO
314-577-5639

Children's Mercy Hospital
Kansas City, MO
816-234-3265

St. Louis Children's Hospital
St. Louis, MO
314-454-6018

University Hospital and Clinics
Columbia, MO
573-882-3961

What does DHSS offer?

SickleCell/Publications/Facts

Special Health Care Needs

AdultGenetics/SickleCell/Publications/Facts

Related Links



Medline Plus (National Library of Medicine and the National Institute of Health) www.medlineplus.gov

National Institutes of Health www.nih.gov

National Newborn Screening and Genetics Resource Center
<http://genes-r-us.uthscsa.edu>

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

The Sickle Cell Information Center www.scinfo.org

Save Babies Through Screening Foundation
4 Manor View Circle Malvern, PA 19355-1622 Toll Free Phone: 1-888-454-3383
Fax: (610) 993-0545 Email: email@savebabies.org
Website: <http://www.savebabies.org/diseasedescriptions.php/>

American College of Medical Genetics

Newborn Screening ACT Sheets and Confirmatory Algorithms
<http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm>

Cardinal Glennon Children's Hospital

St. Louis, Missouri 314-577-5639
Website: <http://pediatrics.slu.edu/index.phtml?page=geneticsdiv>

Children's Hospital at University Hospital and Clinics

Columbia, Missouri 573-882-6991
Website: <http://www.genetics.missouri.edu/>

Children's Mercy Hospital

Kansas City, Missouri

816-234-3290

Website: <http://www.childrens-mercy.org/content/view.aspx?id=155>

St. Louis Children's Hospital

St. Louis, Missouri

314-454-6093

Website: <http://www.peds.wustl.edu/genetics/>