STATE OF TENNESSEE NEWBORN SCREENING PROGRAM

SICKLE CELL AWARENESS MONTH

Special Edition Sickle Cell Awareness Month
September 2011

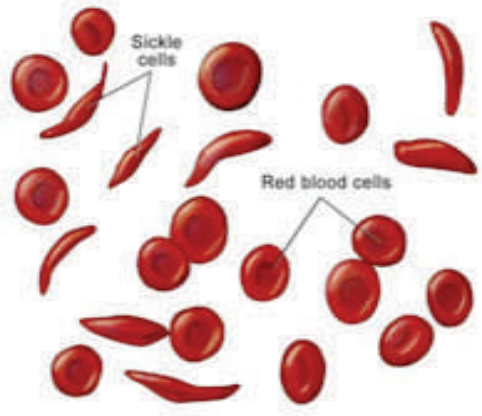
Sickle Cell Disease

Hemoglobinopathies are genetic conditions that affect the kind and amount of hemoglobin a person has in their red blood cells. Hemoglobin is the part of the red blood cell that carries oxygen throughout the body and gives the red blood cell its color and shape. Hemoglobiniopathies imply structural abnormalities in the globin proteins themselves. Thalassemias, in contrast, usually result in underproduction of normal globin proteins. Hemoglobinopathies can be found in all ethnic groups but are more common in African Americans.

The most common hemoglobinopathy is sickle cell. Sickle cell anemia is an inherited form of anemia – a condition in which there aren't enough healthy red blood cells to carry adequate oxygen throughout your body.

Normally, your red blood cells are flexible and round, moving easily through your blood vessels. In sickle cell anemia, the red blood cells become rigid and sticky and are shaped like sickles or crescent moons. These irregularly shaped cells can get stuck in small blood vessels, which can slow or block blood flow and oxygen to parts of the body.

There's no cure for most people with sickle cell anemia. Individuals affected with sickle cell disease have lifelong hemolytic anemias with acute and chronic tissue damage secondary to the blockage of blood flow. These hard, pointed cells go through small blood vessels and clog and break apart. This causes pain, delayed growth, a low blood count (anemia) and other serious problems. Additional clinical manifestations include episodic vaso-occlusive crises, functional asplenia, sepsis, infections, splenic sequestration, and



bone marrow aplasia. Sickle cell can be lethal, especially in early infancy or childhood (10% mortality) because of overwhelming sepsis or splenic sequestration. However, treatments can relieve pain and help prevent further problems associated with sickle cell anemia.

When sickle cell disease is suspected on newborn screening, an extremely high priority is placed on contacting the family so their child can have confirmatory testing.

If the second blood test confirms the diagnosis of sickle cell disease, an educational program is initiated. This begins with the patient being seen and evaluated by a sickle cell nurse and pediatric hematologist. Detailed discussions with family center on the inheritance and clinical significance of sickle cell disease and emphasize the need for two daily doses of liquid penicillin to prevent pneumococcal infection. Penicillin should be given on a regular basis at least until 5 years of age. Discussion also includes other significant complications of sickle cell disease that can occur early in

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Special Points of Interest

- Sickle Cell– the disease, the signs and symptoms, and the complications to watch for.
- Hematology Centers for your area!
- Spotlights on key personnel in Centers.
- Promotional Events for Sickle Cell.

State of Tennessee
Department of Health
Newborn Screening
630 Hart Lane

Phone: 615-262-6304
Fax: 615-262-6458

Signs and Symptoms to Watch For

Signs and symptoms of sickle cell anemia usually show up after an infant is 4 months old and may include:

Anemia. Sickle cells are fragile. They break apart easily and die, leaving you chronically short on red blood cells. Red blood cells usually live for about 120 days before they die and need to be replaced. However, sickle cells die after only 10-20 days. The result is a chronic shortage of red blood cells, known as anemia. Without enough red blood cells in circulation, your body can't get the oxygen it needs to feel energized. That's why anemia causes fatigue.

Episodes of pain. Periodic episodes of pain, called crises, are a major symptom of sickle cell anemia. Pain develops when sickle-shaped red blood cells block blood flow through tiny blood vessels to your chest, abdomen and joints. Pain can also occur in your bones. The pain may vary in intensity and can last for a few hours to a few weeks. Some people experience only a few episodes of pain. Others experience a dozen or more crises a year. If a crisis is severe enough, you may need to be hospitalized.

Hand-foot syndrome. Swollen hands and feet may be the first signs of sickle cell anemia in babies. The swelling is caused by sickle-shaped red blood cells blocking blood flow out of their hands and feet.

Frequent infections. Sickle cells can damage your spleen, an organ that fights infection. This may make you more vulnerable to infections. Doctors commonly give infants and children with sickle cell anemia antibiotics to prevent potentially life-threatening infections, such as pneumonia.

Delayed growth. Red blood cells provide your body with oxygen and nutrients you need for growth. A shortage of healthy red blood cells can slow growth in infants and children and delay puberty in teenagers.

Vision problems. Some people with sickle cell anemia experience vision problems. Tiny blood vessels that supply your eyes may become plugged with sickle cells. This can damage the retina – the portion of the eye that processes visual images.

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Complications of Sickle Cell Disease

Sickle cell anemia can lead to a host of complications, including:

Stroke. A stroke can occur if sickle cells block blood flow to an area of your brain. Signs of a stroke include seizures, weakness or numbness of your arms and legs, sudden speech difficulties, and loss of consciousness. If your baby or child has any of these signs and symptoms, seek medical treatment immediately. A stroke can be fatal.

Acute Chest Syndrome. This life-threatening complication of sickle cell anemia causes chest pain, fever and difficulty breathing. Acute chest syndrome can be caused by a lung infection or by sickle cells blocking blood vessels in your lungs. It may require emergency medical treatment with antibiotics and other treatments.

Pulmonary hypertension. People with sickle cell anemia can also develop high blood pressure in their lungs (pulmonary hypertension). Shortness of breath and difficulty breathing are common symptoms of this condition, which can be fatal.

Organ damage. Sickle cells can block blood flow through blood vessels, immediately depriving an organ of blood and oxygen. In sickle cell anemia, blood is also chronically low on oxygen. Chronic deprivation of oxygen-rich blood can damage nerves and organs in your body, including your kidneys, liver and spleen. Organ damage can be fatal.

Blindness. Tiny blood vessels that supply your eyes can get blocked by sickle cells. Over time, this can damage the retina – the portion of the eye that processes visual images – and lead to blindness.

Skin ulcers. Sickle cell anemia can cause open sores, called ulcers, on your legs.

Gallstones. The breakdown of red blood cells produces a substance called bilirubin. A high level of bilirubin in your body

For any of these symptoms please seek immediate medical attention.

St Jude Children's Research Hospital

The identification of Sickle Cell Disease (SCD) or sickle cell trait (SCT) in newborns is done at birth through the NBS Program at the state public health screening laboratory. Follow-up of SCD and SCT involves confirmatory diagnosis, genetic counseling, education and clinical management. St Jude is one of our 4 designated sickle cell centers in the state of Tennessee that works with the NBS Program to help accomplish these goals of follow up. St Jude has two dedicated Health Educators to help accomplish the follow up needed. Christy Matthews and Gloria Brunson are responsible for providing genetic education to parents of newborns with sickle cell disease and other hematological disorders. They also provide genetic education for teens at risk for having a child with sickle cell trait/disease. They are also joined by Yvonne Carroll who is the Director of

St Jude Children's Research Hospital
www.stjude.org
Clinical Programs/Treatment Programs

Patient Services of the Department of Hematology.

Christy has been an employee for 6 years with St Jude. She has a Masters in Education. Christy has also worked for the last 8 months as the Newborn Case Manager to initiate care for **all** newborns with sickle cell in West Tennessee. She will soon be processing and initiating care for newborns that have other Hemoglobin disease (i.e., C Disease, E Disease, etc.) as they have hired a new Newborn Nurse Case Manager, Kristen "Shay" Crook. Kristen will be taking over the responsibility of processing and initiating care for the newborns with sickle cell disease.

Christy is the author of two abstracts:

1) Families of Infants with Sickle Cell Disease Can Retain Genetic Counseling Information. 2007
2) Measuring Long-Term Retention of Genetic Education Provided To Teenagers With Sickle Cell Disease. 2009

Thanks to Christy, Gloria, and Yvonne for all your hard work...we love working with you... and welcome to Shay! We look forward to working with you.

UT Medical Center of Knoxville Hematology Department

The University of Tennessee is another Hematology Center that assists the NBS Program in locating and confirming diagnoses. Our key contact there is the East Tennessee Newborn Screening Coordinator, **Diana "Dee" White**.

As the newborn screening coordinator she provides education, counseling and social work services to families of infants diagnosed with sickle cell disease as well as provides outreach education and free hemoglobinopathy testing to at risk populations.

Dee is joined in her efforts by:

Dr. Karla J Matteson, Dr. Ilse J. Anderson, Dr. Carmen B. Lozzio, Darla J Smith, Amanda Noyes, and Abbey Putnam, all of whom work very hard to insure that appropriate rescreening, diagnostic testing, and follow-up is provided as needed.

http://gsm.utmck.edu/genetics/utgeneticscenter.cfm

Sickle Cell Disease (continued)

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life, including pneumonia, anemia, enlargement of the spleen, swelling of the hands and feet, infection of the bone, and pain crises affecting various parts of the body. Children with sickle cell disease are seen in Hematology Clinic every 3 months for the first 2 years of life and every 6-12 months afterwards.

Immunization against pneumococcus, *H. influenza*, and hepatitis infection should be given. Parent and patient education should include information regarding rapid access to appropriate medical care for aggressive pain management, prompt recognition and early treatment of infections, management of dehydration and acidosis, blood transfusion therapy for selected problems (e.g., anemia and cerebrovascular accidents), and judicious use of oxygen reduce morbidity and mortality.

For more information go to: http://health.state.tn.us/nbs/information.htm

Meharry Sickle Cell

The Meharry Sickle Cell Center provides comprehensive services including clinical care, education, genetic counseling, laboratory services, and research. In this capacity, blood samples from all children born in Tennessee suspected to have a hemoglobinopathy and from all adults seeking evaluation, undergo state of the art testing to confirm or establish a hemoglobinopathy. It is the MSCC responsibility to notify promptly the health care agencies and parents of the patients with positive results so that proper medical care and management can be instituted. Additionally, the MSCC provides follow-up services to newborns with sickle cell anemia to ascertain that all children are undergoing appropriate preventive care such as receiving prophylactic antibiotics within three months of birth. The MSCC staff is composed of the following:

The **Center's Director**, Steven N. Wolff, M.D. provides comprehensive medical and diagnostic care for adults with sickle cell disease residing in the 40 County Middle Tennessee regions.

The **Center's Associate Director and Laboratory Director**, Pilar Aguinaga, MT (ASCP), M.Sc., PhD. CLDir (NCA) heads the State of Tennessee Newborn Screening Hemoglobinopathy Confirmatory Laboratory at the MSCC.

Other Clinical Laboratory scientists in the Confirmatory Laboratory are Robert Mushi, MT (ASCP), PhD (Laboratory Supervisor), and Celestine Mgbemere, MT (ASCP), MD. They perform an array of sophisticated laboratory methods for the accurate and timely diagnosis of hemoglobin disorders. Annually, over 6,000 blood samples for suspected hemoglobin disorders undergo laboratory evaluation.

The **Genetic Counselor**, Mrs. Tené Hamilton Franklin, M.Sc. provides genetic counseling and education to the families of children who have sickle cell disease. She also leads a Sickle Cell Support Group for patients and their families, and works closely with the community through various outreach activities.

The **Program Coordinator**, Ms. Lanique Woodson, MSP administers the Center's budget and monitors the daily MSCC business.

The **Administrative Assistant**, Ms. Lisa Walker bridges the MSCC with our clients, faculty and staff at Meharry. She sends electronic patient results to the Health Care Agencies and providers who originated the blood sample, through our secure network. She also tends to the daily activities of the MSCC.

Ronlanda Foley BS, LPN
Meharry Sickle Cell Center

The **Follow-Up Nurse/Educator**, Mrs. Ronlanda Foley BS, LPN does the newborn follow-up of babies diagnosed with sickle cell disease or other severe Hemoglobinopathies. She also provides hemoglobinopathy education to adults and the families of the affected newborns. The nurse usually informs the parties involved that the infant needs retesting at the pediatrician office, local health department or, free testing at the Meharry Sickle Cell Center; and the specimen needs to be sent to the Meharry Sickle Cell Center for result confirmation.

For infants with suspected disease the nurse follows up for repeat specimen collection as soon as the demographic information is sent. The nurse is responsible for contacting the pediatrician and parents to adhere to proper repeat confirmation testing. Once results are confirmed, the nurse notifies (by faxing) the state, pediatrician, and Vanderbilt Pediatric Hematology Clinic for a Sickle Cell appointment. When the nurse receives a response from Vanderbilt, the nurse will notify the pediatrician of the appointment date, and make a suggestion of starting penicillin by two months of age. A typical appointment date is usually a couple of weeks after the confirmation test. Once all medical professionals involved in the treatment of the infant and in correspondence with care of the infant have completed their part: the nurse then turns the information over to the Genetic Counselor for counseling and support.

Mrs Foley's duties also include free trait testing to individuals in the Middle Tennessee area. Testing usually occurs during outreach events (health fairs, back to school day….), in the Center's lab by appointment, or a home visit for an individual who is not able to attend a health fair or come into the Sickle Cell Center. In addition, the nurse is also responsible for ordering, maintaining and sending testing supplies to agencies in the state of Tennessee; sending weekly reports of all confirmed infants to the Sickle Cell treatment Centers in the state of Tennessee. She is also responsible for assisting the Adult Hematologist in Clinic once a week, attending a monthly support group, assisting in the planning and organizing of community events, and educating agencies and health care professionals on proper specimen collection and

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Meharry Sickle Cell (continued)

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basic hemoglobinopathy education. She reports confirmed results (via telephone or fax) to agencies, develops health information related to Sickle Cell disease and trait, and is actively involved with health organizations on health disparities among minorities.

Mrs. Foley has also been involved in an assortment of outreach activities while at Meharry. Meharry has continued their partnership with the Martha O'Brien's Center's "Tied Together Program". The Tied Together Program concentrates its efforts on families in the Cayce Homes; which offers a 10-week session on parenting. MSCC also participates in various community support programs, namely Glencliff and Overton High Schools health fairs, lectures, and screenings in the community. We had a successful state audit in which we were recognized by the State of Tennessee for Outstanding Service for 21 years. Our 6th Annual Walk-A Round Campus, an event, focused on raising awareness, promoting health and fitness, and acknowledging individuals in the community dealing with Sickle Cell or a family member caring for someone with Sickle Cell Disease.

http://www.mmc.edu/research/centers/sicklecell/index.html

T. C. THOMPSON CHILDREN'S HOSPITAL HEMATOLOGY

Dr. Jennifer Keates-Baleeiro is an Assistant Professor of Pediatrics at the Center for Childhood Cancer and Blood Disorders at T.C. Thompson Children's Hospital at Erlanger, Chattanooga, TN. She currently leads the Comprehensive Sickle Cell Clinics for babies to young adults, who were identified through the State of Tennessee Department of Health Newborn Screening Program. Children ages 2 months-5 years are seen quarterly; school aged children are seen biannually and teenagers are seen yearly. Appropriate penicillin prophylaxis, hydroxyurea supplementation and screening with TCD (trancranial Doppler), ECHO, eye exam, audiology and pulmonary testing are conducted. Chronically transfused patients who have experienced stroke are also managed through the clinic.

Educational outreach to promote health and well-being in sickle cell patients is encouraged in comprehensive clinics and through minority health fairs. School visits are offered by childlife specialist Ashley Williams, nurse educator, Deborah Gourley and social worker, Sugar Wheeler. In an effort to increase awareness regarding bone marrow transplantation for a cure for sickle cell disease, a joint effort with Blood Assurance has resulted in >150 minority registrants for the National Bone Marrow registry through a church based initiative program. Stomp out Sickle Cell continues annually to also encourage blood donation for our chronically transfused patients.

Current challenges include developing a transition care clinic for the adolescent and young adult sickle cell population at Erlanger. In addition to the sickle cell population, Dr. Keates-Baleeiro also sees a robust pediatric oncology patient population.

(L to R) Deborah Gourley, RN, Jennifer Keates-Baleerio, MD, Sugar Wheeler, LCSW, Avery Mixon, MD, Sonya Martin, CMS, Ashley Williams, Childlife

Dr. Keates-Baleeiro received her Bachelor of Science from Tufts University and her Masters in Psychology from New York University before receiving her medical degree from MCP-Hahnemann University. She completed her residency at Children's Hospital of Michigan. Dr. Keates pursued fellowship at the University of Michigan Medical Center and Vanderbilt University. Her first academic appointment was as Instructor in Pediatric Hematology-Oncology at Vanderbilt University Medical Center, before her current appointment at University of TN, Chattanooga division. Dr. Keates-Baleeiro is Board Certified in Pediatrics and Pediatric Hematology-Oncology.

Dr. Keates-Baleeiro lives in Chattanooga with her husband, Dr. Carlos E. Baleeiro and two children, Sophia and Carlos Paxson. A black Labrador completes the family.

A new addition to the academic practice includes Dr. Benjamin Avery Mixon, who is Board certified in Pediatrics and Board Eligible in Pediatric Hematology-Oncology. He sees all hemoglobinopathies identified by the Newborn Screening Program in addition to pediatric oncology patients. Dr. Mixon received both his Bachelor of Science and Medical Degree from the University of Mississippi. He completed his residency at the University of South Florida. Dr. Mixon completed his fellowship at Vanderbilt University. His first academic appointment is at the University of Tennessee, Chattanooga division.

Dr. Mixon lives in Chattanooga with his wife, Jennifer and their three children, Avery Jr., Parker, and Maryellen.

http://www.erlanger.org/body.cfm?id=73

PROMOTIONAL EVENTS FOR SICKLE CELL AWARENESS MONTH

September 3-4, 2011
39th Annual Walker Evans Memorial Sickle Cell Golf Tournament
Cottonwood Golf Course, Harrah's Casino Resort
~~Tunica, MS~~
http://www.sicklecellgolf.com/index.htm

September 27—October 1, 2011
39th Annual Sickle Cell Disease Association of America Conference
Memphis, TN
www.sicklecelldisease.org

September 15 & 16
MOVIE NIGHT!
Darkhorse Theater
Sponsored by Meharry Sickle Cell Center 615-327-6849
Nashville, TN

September 30, 2011
Dinner & Divas….A Sickle Cell Benefit
Cook Convention Center, Memphis, TN

OCTOBER 1, 2011
ZOO DAY
NASHVILLE ZOO
Sponsored by Meharry Sickle Cell Center 615-327-6849
10AM-2PM
Nashville, TN

September 17, 2011
5K Run-Walk-Fun
Memphis, TN
www.sicklecellwalk.org

We're on the Web
health.state.tn.us/NBS/index.htm