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From the Director's Desk

Clinton H. Joiner, MD, PhD

Everyone has two genes that make hemoglobin, the red protein in the blood that carries oxygen. Most people have two genes that both make the usual hemoglobin A (Hb A), one from each parent. Persons who have one gene for Hb A and one for sickle hemoglobin (Hb S) have sickle cell trait, and are said to be a "carrier" of the sickle gene. They are sometimes described as having Hb AS because they make both Hb A and Hb S, in roughly equal amounts. If both parents have trait, there is a 25% chance they may pass both S genes to a new baby, who would have sickle cell disease (Hb SS). They could also have a child who had only Hb A (25% chance) or have trait (Hb AS, 50% chance).

About one in every 12 African Americans has sickle cell trait, which is about 8% or 800 people for every 10,000. Taking all Americans, it's about 5 people per 10,000. For every one person in the US with sickle cell disease there are about 50 people with sickle cell trait. Although sickle cell trait is more common in people of African descent, it is also found among Caucasians with ancestors from the eastern Mediterranean, the Middle East and India. About 4% of the people in the US with sickle cell trait are white.

Sickle cell trait generally does not cause major health problems. Blood counts are normal; there is no anemia. Overall lifespan is normal. People with sickle cell trait have the same risk as everybody else to get common ailments such as high blood pressure, diabetes, cancer, heart disease, arthritis, etc. However, there are a few health conditions that affect sickle cell carriers because of their trait status. People with trait normally don't have pain episodes, although they can have sickling, pain and spleen complications at very high altitudes; this has occurred when flying at high altitudes in un-pressurized airplanes. (Normal air travel is perfectly safe.) Some people with trait have hematuria – blood in the urine – which can sometimes (though rarely) cause serious problems. Most people with trait have a mild limitation of their ability to concentrate urine under conditions of severe water deprivation. This rarely causes a problem in everyday life, but might make one more likely to get dehydrated under very hot conditions if water was not available. Another potential (but rare) problem is that eye injuries may need to be treated specially in people with sickle cell trait.

Recently there has been a lot of publicity about sudden death in athletes with sickle trait during conditioning training. The NCAA has recommended that all athletes be screened and those with trait have a special, lower intensity training regimen. Personally, I disagree with this recommendation for several reasons. First, this is a very rare occurrence, and it is not clear how much this risk is increased in athletes with sickle cell trait. Second, the vast majority athletes with sickle trait participate in sports safely and without any impairment in performance. Third, I believe that trainers and coaches should modify training regimens to reduce the risk for all athletes, because only a fraction of those who have died have had sickle trait. Finally, I worry that reduced intensity conditioning for athletes with trait may cause them to be viewed as less fit for play and thereby lessen their chances of success in competitive athletics.

Parents of children with sickle cell trait should NOT discourage them from participating in athletics. Parents of ALL children should be aware of the exercise regimens that their children participate in, to make sure that adequate water and rest periods are provided and that coaches do not push young athletes beyond their physical limits.

Becoming An Agent of Change for Sickle Cell Disease SCDAA Convention 2009

By: Nicole W. Barnes

Mother of a 20-month old child with sickle beta plus thalassemia



What a phenomenal experience I had in Orlando, Florida at the 37th Annual Sickle Cell Disease Association of America (SCDAA) convention "Building Bridges for Change." Over 300 patients, families, advocates and healthcare professionals from Utah to Toronto, Canada came to the convention, held September 30 - October 3, 2010.

The entire staff and volunteers treated us with such hospitality. Each day was unique with presenters covering topics from clinical updates and practices, innovative programs and services to public health issues and advocacy. Whether we were having our morning breakfast of fruit & bagels or participating in a Hawaiian luau extravaganza, or being entertained by Ruben Studdard, who performed at the Dorothy Boswell Gala, each activity allowed me to participate, learn and grow in knowledge of sickle cell disease.

Parenting a child is always a challenging responsibility and adding a chronic illness into the family dynamics magnifies and intensifies this challenge.

This convention allowed me to see that everyday we must work to connect patients, families, doctors, volunteers and researchers to resources and programs that will meet the needs and challenges of people of every racial and ethnic background so that sickle cell patients can live longer, healthier lives. If parents empower each other, we will be able to provide hope and inspiration to those living with and affected by sickle cell disease. Our children need lots of hugs and kisses. Your child, and my child are truly special and brave to fight this disease.

I met college professors, local Cincinnatians, and the mother of Dale Lloyd II, who was a 19 year - old freshman athlete at Rice University that passed away unexpectedly due to sickle cell trait in 2006. Many more people have sickle cell trait than sickle cell disease but many people with sickle cell trait are not aware they have it. Individuals with sickle cell trait are generally asymptotic and have no abnormal physical findings. As researchers continue to foster new treatments and hopefully a cure, patients and their families should be counseled about their risk of having a child with sickle cell disease based on their partner's genotype. Many new advances in therapy have been utilized to treat sickle cell disease such as hydroxyurea, penicillin prophylaxis, stroke prevention with transcranial doppler ultrasound, and bone marrow transplants with increasing success rates.

I was taken aback to discover how little funding is allocated to programs like sickle cell disease. The federal budget expires this year and the The Sickle Cell Anemia Prevention and Treatment Act is an earmark, which Democratic Congressman Danny Davis (Chicago-IL) is reintroducing this year. As parents, we have to be proactive in order for transformation to take place. For more information on this issue, you can log onto www.opensecrets.org and make your voice count to bring about change to the sickle cell community. As one speaker said about national funding, "We have to stop allowing soup to run free in the streets, if only I had a spoon." We must have the right tools in place. What will your spoon be? A massive educational outreach? Requesting legislative or congressional appropriation for programs that benefit your community?

Next year will mark the 100th year celebration since the discovery and publication of "peculiar elongated and sickle cell shaped red blood corpuscles" in a case of severe anemia, by Dr. James Herrick. Having a voice in community education and awareness will advance care, fund raising efforts and research for sickle cell disease. Knowledge and treatment of sickle cell disease can mean a brighter future for our children.

Again, I thank the U.S Department of Health and Human Services, HRSA Maternal and Child Health Bureau and Cincinnati Comprehensive Sickle Cell Center for the opportunity to know more and become more. In order for things to change, I must change. Be an agent of change.

8th Annual Research Day Features Pain Management and Youth Empowerment Activities

By: Lori Crosby, PsyD, Janelle Hines, MA & Monica Mitchell, PhD

The Eight Annual Sickle Cell Disease Research and Education Day was held on August 29, 2009 and focused on pain management and youth empowerment. Over 450 people attended the event held in the Sabin Auditorium. As always, families were treated to a buffet luncheon, invited to participate in educational and fun games and received school supplies. Families and youth learned about pain management techniques including yoga, massage, distraction activities, and relaxation. This year, Dr. Punam Malik provided an overview of the state of gene therapy research in pediatric sickle cell disease and families were given information about how to reduce risks related to the H1N1 virus (or the swine flu). Throughout the event, families and children were able to participate in open research studies. Every eligible family chose to participate in at least one study this year (100% participation). Parents and teens completed a survey to evaluate the day's events and participants indicated that they would like to continue to learn about additional research studies at future Research and Education Days. According to the survey, 100% of participants reported that they would recommend the event to others and plan to attend next year.





This year's Teen Symposium featured a special event, a Sickle Cell Disease Teen Art Show. During last year's Teen Symposium, adolescents worked with a graphic designer to develop educational brochures and posters. This year six of the posters were featured during the Art Show. Teens were asked to identify their favorite poster and to rate the content and format of all of the posters. This information will be used to develop a series of educational materials that youth can hand out to family members, friends, teachers, and community members. Community support for the event was even greater than in previous years. Several local companies and businesses donated volunteers, supplies and gifts to the event. The event ended with a musical performance by Lyric 513, an all female teen group. Next year's event is planned for August 2010. Save the Date Cards will be sent out in June.

If you would like additional information about open research studies in Psychology, please contact Venita Robinson at (513) 803-2040. For information about open studies in the Sickle Cell Center, please contact Tammy Nordheim at (513) 636-7374.

PhotoVoice Project and Photo Exhibit Gives Voice to Youth with Sickle Cell Diseases

Jessica Valenzuela, PhD, Lori Crosby, PsyD & Monica Mitchell, PhD



Fifteen youth ages 8 to 17 participated in a PhotoVoice Program. On December 6th, they featured their photographic works and shared their experiences with family members and the audience in a *Photo Gallery and Exhibit*. The PhotoVoice program involved

having youth take pictures based on themes they developed during a group meeting. They would then take pictures all week and share their photographs at the next group session. Some of the themes were "Living My Life," "Things I Like and Don't Like," and "People, Places and Things in My Life." During the final week, youth chose their favorite pictures and quotes and made a display board to showcase their work. They also made audio recordings and a PowerPoint presentation that was shown during the Photo Galley and Exhibit. The event kicked off with a reception during which time exhibit attendees talked with participants about their display boards, received autographs, and viewed pictures that participants took during the 5 week program. Youth gave presentations about their PhotoVoice experiences and participated in a question and answer panel. Over 100 people attended the event and information from participants, parents and attendees suggests that they found it valuable. Participants reported that PhotoVoice helped them learn about sickle cell and many reported that it was an empowering experience.

It's Not Too Late for School Intervention

David Kathman

Can you believe that the school year is already almost half over? It is not too late to ensure your child has a successful school year, and the School Intervention Program (SIP) is here to work with parents and schools to help make that happen. There are several things to remember when working with the SIP program:

Keep SIP involved with your child throughout the school year.
SIP cannot effectively work with your child's school if you contact us for help at the end of the school year.

 If your child currently has an IEP or 504 plan, ask the school to add the "sickle cell disease plan of care" to their plan.

This helps to provide the classroom and medical accommodations necessary for your child to stay healthy at school.

Communicate with your child's school about illnesses.

It is important for every child to attend school every day, but for a child sickle cell disease, that is not always possible. If your child is sick and cannot attend school, it is important for the parent to call and inform the school about why your child is not at school that day. Not only will this help to minimize truancy issues, but it also keeps communication open with school staff about your child's illness.

SIP is here for your support to help your child succeed at school. If you have any questions, please feel free to contact SIP at 513-636-6741.



Winter Tips

It's important to remember that that chilly winter weather can cause a sickle cell disease pain crisis. Cold weather can cause blood vessels to shrink, which can cause to more blockages and pain

The Comprehensive Sickle Cell Center Hospital suggests several winter tips to help prevent a pain crisis:

- Always make sure your child dresses appropriately for the weather - a warm coat, hat, gloves are a must; earmuffs and scarves when needed. Dressing in warm layers of clothing is important as well. Children with sickle cell disease are sensitive to extreme temperature changes, and going from a warm house into cold air outside could possibly cause a pain crisis
- Remove children's wet clothing immediately when they come in from outdoors and make sure they change quickly into warm, dry clothes.
- Have a "recess" plan in advance with your child's teacher to make sure that there is a plan for staying indoors for recess during cold weather.
- Keep children hydrated with plenty of fluids.

It's Not Too Late for Flu Shots

Seasonal flu shots and H1N1 shots are still available in the clinic. Doctors are recommending that all patients with sickle cell disease be vaccinated because they are at higher risk of complications from either flu.

For more information, please contact your child's nurse coordinator:

Ages Newborn - 6 years old: Lisa Ovesen - 513-803-0005

Ages 7 years old - 14 years old: Pat Boyd - 513-636-1749

Ages 14 years old - 21 years old: Tracy Mahaney - 513-803-0066



Check Out The New Cincinnati Comprehensive Sickle Cell Center Website:

http://www.cincinnatichildrens.org/svc/alpha/c/ cancer-blood/blood-disease/sickle-cell/default.htm



Gene Therapy Raises Hope for Sickle Cell Anemia Cure

Peggy O'Farrell, Cincinnati Enquirer

Kameron Kinebrew, 12, spent a month hooked to a morphine pump to control the sickle-cell anemia pain, something he compared to "a couple of knives stabbing you in your back, over and over."

His twin brother, Kaleb, rattles off pain management techniques without thinking: slow breathing, plenty of fluids, distraction. "We know what to do," he said. "It's normal for us."

People with sickle-cell disease face a lifetime of pain and fatigue. They also risk stroke, blindness, organ failure and bone loss. The only effective treatments are bone marrow transplants and chemotherapy, and they don't work for everyone with the disease. But researchers at Cincinnati Children's Hospital Medical Center offer hope for a cure for the tens of thousands of Americans afflicted by the life-shortening blood disease.

After a decade of research, scientists led by Punam Malik have developed a cutting-edge gene therapy that - in lab animals and human tissue samples - made the body stop producing the malformed red blood cells that characterize sickle-cell disease. They want to start testing it next year if federal regulators sign off. "If it works, it's a cure," said Malik, a hematology-oncology specialist who heads up all of the gene and molecular therapy research at Cincinnati Children's.

Sickle cell affects 600 to 800 people in Greater Cincinnati and Northern Kentucky, and some 70,000 to 100,000 people in the United States. One in 500 African-Americans has the disease. Worldwide, the disease affects millions.

In sickle cell, a genetic defect makes red blood cells carry an abnormal type of hemoglobin, called hemoglobin S, that changes their shape, making it harder for the blood cells to pass through blood vessels. The cells can't efficiently carry oxygen to tissue, and the tissues become damaged and eventually die. Over several years, people with sickle cell suffer organ damage and organ failure. One study found that organ failure, especially kidney failure, killed about 20 percent of adult sickle-cell patients. The damage starts early. Kameron Kinebrew's doctors have already identified spots of dead bone tissue in his legs because of sickle cell.

The therapy developed at Cincinnati Children's would implant a gene that counteracts hemoglobin S, allowing the body to make the correct type of hemoglobin to form normal, doughnut-shaped red blood cells.

In the proposed clinical trial, researchers will collect bone marrow stem cells from the bone marrow of patient volunteers. In the lab, they'll implant engineered viruses containing a new gene in those stem cells, which make red blood cells. Then, they'll return the stem cells containing the engineered viruses to the patient volunteers' bodies. Each patient volunteer will get his own stem cells back.

The viruses used in the experimental therapy have been altered so that they can't cause disease. Giving patients back their own stem cells means patients and doctors won't have to worry about organ rejection, Malik said.

Researchers hope the implanted gene would allow sickle cell patients to make hemoglobin F, which lets red blood cells form normally. The engineered virus would continue to reproduce in volunteers' bone marrow and let volunteers permanently make normal red blood cells if the therapy works.

Federal health regulators are reviewing the therapy, which took a decade of groundwork to develop. If the feds give the go-ahead, Malik and her team could begin the first phase of human trials in adults with sickle cell next year, possibly in the spring or summer. Malik expects to recruit 10 people for the first phase.

Testing the new therapy will be a slow process that could take another five years or longer. Regulators and researchers will monitor volunteers who undergo the therapy to make sure it's safe and to see if it works.

The experimental therapy might not work in people, Malik warned. Some therapies don't translate from mice to humans. Federal regulators could pull the plug at any time if there are signs the therapy could be dangerous. In a French gene therapy trial aimed at treating a rare immune disorder in children in 2003, some patients who received the therapy developed leukemia.

Malik and her colleagues, though, remain cautiously optimistic. "If it works, the impact would be significant," said Clinton Joiner, director of the Comprehensive Sickle Cell Center and the Hematology Program at Cincinnati Children's. "This is a bad, bad disease to have."

About 500 people die of sickle cell and its complications each year in the U.S., Joiner said, and about half of sickle-cell patients die by age 45. Sickle-cell patients don't have many treatment options now, he said.

Bone marrow transplants work well, but only about 10 percent of people with sickle cell get them because it's so hard to find a donor match if a sibling can't donate. There's also a time limit for bone marrow transplants, Joiner said. By the time sickle cell patients are 16 or 17, the disease has caused enough damage that the risks from the transplant are too high to make it worthwhile, and the transplant itself can cause problems even in younger patients.

Some sickle-cell patients benefit from hydroxyurea, a drug that lets the body make hemoglobin F and reduces the number of sickle cells, but doesn't remove them completely. Hydroxyurea doesn't work for everyone with sickle cell, though, and it's a form of chemotherapy, so it has its own side effects, including immune suppression.

The disease affects the entire family. The Kinebrews of Colerain Township never know when they're going to have to take Kaleb or Kameron to the emergency room for a pain crisis. Both boys have suffered acute chest syndrome, a condition similar to pneumonia that occurs when lung tissue isn't getting enough oxygen. It's extremely painful and dangerous as lung function is lost. Both have undergone blood transfusions, one temporary remedy for the disease.

"A lot of times, people look at the boys, and you can't see sickle cell. They live with it every day, and they don't look sick," said Kevin Kinebrew, the twins' father. "The reality is depending on the day, you never know if there's going to be a pain crisis or an emergency or a call from school that they're having problems."

Kinebrew and his wife, Robyn, both have the sickle cell gene, although Robyn was originally told she didn't have it. When the boys were diagnosed at birth with sickle cell, doctors re-tested Robyn and found out she carried the trait.

The boys participate in ongoing research at Cincinnati Children's, including a study looking at what factors contribute to acute chest syndrome, she said. "It's important to learn more, to find something that might help them and other families," she said.

Additional Facts

Sickle-cell anemia is a hereditary disease affecting red blood cells, which carry oxygen to the body's tissues. Both parents have to carry the genetic trait for the disease for a child to develop it.

There is no cure, and few treatments. Average life expectancy for Americans with the disease is in the mid-40s, though sickle cell patients can live into their 60s.

Normal red blood cells are doughnut-shaped and can easily slip through arteries, veins and tiny capillaries to carry oxygen to tissue throughout the body. In sickle cell, the cells are shaped like crescent moons or sickles, making it harder for them to get through the blood vessels.

Sometimes, the misshapen cells get stuck or cause other damage that narrows the blood vessels. If the blood vessels are in the brain, stroke can result. If the blood vessels are in the eyes, blindness may result.

The cells become misshapen because the body makes the wrong kind of hemoglobin, the chemical that makes blood red.

Babies can be diagnosed with sickle cell at birth, but symptoms don't start until they're about 4 to 6 months old.

That's when the body stops making fetal hemoglobin, or hemoglobin F. At that point, a baby without sickle cell disease starts making hemoglobin A, which lets their bodies make normal red blood cells.

Babies with sickle cell make hemoglobin S, which deforms the red blood cells. The cells become fibrous and form sickles, especially when they're low in oxygen.

People with sickle cell suffer chronic pain, including severe episodes that may require narcotics. When blood flow to the organs is disrupted, permanent damage can result, including organ failure. Infections are common, as is damage to bones and joints. Sickle cell is the leading cause of stroke in children.

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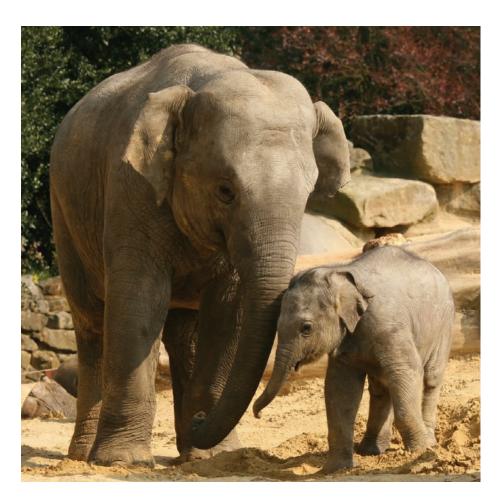
Researcher's Remarks

Punam Malik, MD

This Cincinnati Enquirer article has generated excitement and interest in our gene therapy research throughout the country. The article was syndicated in newspapers across the United States and has led to numerous phone calls to the Cincinnati Comprehensive Sickle Cell Center from interested patients with sickle cell disease from all over the country.

As you read this article, there are several things to keep in mind about this study as it begins to move forward. Studies to prove the safety and success of using gene therapy in sickle cell disease will begin late next year. The study will first be piloted in adults (ages 18-30 years old).

Also, to prevent the spread of flu, especially to our vulnerable sickle cell patients, focus groups to discuss gene therapy are on hold until late spring 2010. However, if you are interested in being considered as a focus group participant, please contact 513-636-6779 or sicklecell@cchmc.org for more information.



Save the Date

Zoo Day 2010 will be held on Saturday, April 24, 2010 at the Cincinnati Zoo and Botanical Gardens. This event is especially for families with children ages newborn – 6 years old with sickle cell disease.

Watch your mailboxes for more information!

Mark Your Calendar!

Sickle Cell Parent Support Group Canceled

Canceled until further notice due to visitor restrictions at the hospital. Contact: Lisa Leace at (513) 636-1747 or Cheryl Blair at (513) 636-8315

February 16-19, 2010

The Fourth Annual Sickle Cell Disease Research and Educational Symposium and Annual National Sickle Cell Disease Scientific Meeting will be held in Ft. Lauderdale, Florida. For more information, please email symposium@floidasicklecell.org or visit www.floridasickle.org.

April 24, 2010

Save the Date for Zoo Day Watch for more information, or contact Lisa Shook at 513-636-7541.

May 6-7, 2010

Sickle Cell Education Symposium and Hemoglobinopathy Counselor Training Course for Healthcare Providers. Nursing and social work CEU's will be available.

Contact Lisa Shook at 513-636-7541 or lisa.shook@cchmc.org for more information.

This newsletter is made possible by a grant from the Ohio Department of Health. Center Talk is published twice yearly by the Cincinnati Comprehensive Sickle Cell Center at Cincinnati Children's Hospital Medical Center. It informs consumers of our education/counseling, community outreach and treatment services, individuals and families affected by a hemoglobinopathy, and health professionals that care for persons affected by sickle cell disease and other hemoglobinopathies. Our goal is to provide a forum to communicate news and other items of interest in regards to sickle cell disease and other hemoglobinopathies. Feedback, ideas, information and news can be directed to; Lisa Shook, 513-636-7541, lisa.shook@cchmc.org