New Hope on the Horizon

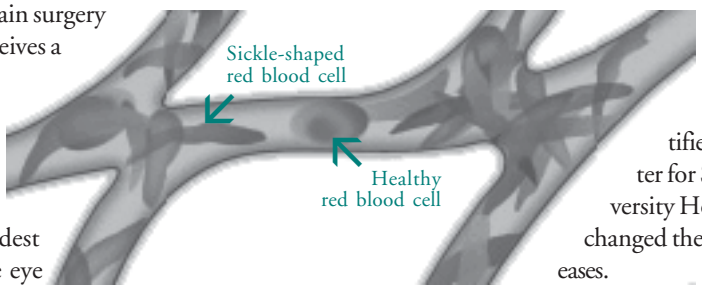
Screening for Sickle Cell Disease

By Kauthar B. Umar, M.A.

Closing the Gap, Maternal Health • January/February 2004

When Andrew Chase suffered a stroke four years ago, his family was devastated. It wasn't the severity of the stroke or the grueling rehabilitation that left them stunned, it was his age. Andrew was just three years old. But for this particular toddler, such complications were not completely unexpected.

Like more than 72,000 Americans of predominately African and Mediterranean decent, Andrew was born with sickle cell disease (SCD), a type of genetic blood disorder. The March of Dimes reports that 10 percent of children living with SCD suffer a stroke and can experience damage to blood vessels in the brain. By the time Andrew was five years old, he underwent bilateral brain surgery and today, at seven years old, he receives a blood transfusion every month.



"When you have a child with sickle cell and you see him in pain, it's the absolute worst feeling in the world," said Andrew's mother, Syreeta Chase. "The hardest thing is looking your child in the eye and knowing that you can't do anything."

But now, thanks to federal dollars and advancing medical technology, there's new hope for families affected by SCD.

The Sickle Cell Disease Association of America reports that 1,000 babies are born with SCD each year, making it one of the most prevalent genetic disorders in the United States. Largely found in the minority community, sickle cell disease occurs in about one in every 600 African American births and one in every 1,000-1,400 Hispanic American births, according to the National Institutes of Health (NIH).

Three common types of sickle cell disease in the United States are hemoglobin SS, better known as sickle cell anemia, hemoglobin SC disease and hemoglobin sickle beta-thalassemia.

The disease is caused by a mutation in one of the two genes that determines the structure of hemoglobin, a critical molecule found in red blood cells that transports oxygen from the lungs to other parts of the body. In patients with this disease, abnormal hemoglobin molecules stick to one another and form long, rod-like structures.

Under a microscope, these abnormal cells may look like the C-shaped farm tool called a sickle. Sickle cells tend to become trapped and destroyed in small blood vessels, the spleen, liver and other organs. The result is a shortage of red blood cells, or anemia.

While Andrew suffers from the most severe complications from SCD, the effects of the disease vary greatly from one person to the next. Sufferers can develop chronic severe anemia, infections and periodic episodes of pain.

To improve services for infants with sickle cell and their families, the Health Resources and Services Administration (HRSA) awarded $3.6 million in October 2003 to projects that link comprehensive sickle cell treatment centers, state newborn screening programs and health care professionals to broad community-based organizations. The 17 two-year grants are funded under HRSA's SCD and Newborn Screening Program through its Maternal and Child Health Bureau. For families affected by the disease, new preventive opportunities, advanced treatment and better outreach can save lives.

Chase admits that access to outreach and education would have helped a great deal—approximately 2 million Americans carry the sickle cell trait and 1 in 12 African Americans have the trait.

Like many parents, she was uneducated about and ill-prepared for the reality of SCD. She was unaware that Andrew's father was a carrier of sickle cell disease and it wasn't until her pregnancy with Andrew that she discovered that she also had the sickle cell trait (see next page).

Today, Barbara Harrison, a certified genetics counselor from the Center for Sickle Cell Disease at Howard University Hospital, says that genetic testing has changed the face of SCD and other genetic diseases.

Couples who are planning to have a baby can get carrier testing at medical centers and sickle cell treatment facilities throughout the country. A blood test called the hemoglobin electrophoresis, performed on both partners, is the most complete test for sickle cell trait and related conditions.

"If they find out that both have the trait, then they need to know their options, and there are many," said Harrison. "The first thing the couple should do is to appreciate the 25 percent chance that the offspring will not have SCD, instead of focusing on the 75 percent chance that they could. It's quite possible for a couple to have kids that do not have the disease."

With the proper genetic testing, a couple with the trait can also undergo specific procedures to ensure that their baby will be free from SCD. In vitro fertilization and pre-implantation genetic diagnosis involve harvesting several eggs and sperm. After allowing the cells to grow for a number of days, genetic testing can be completed on the group of cells. Those cells that do not carry the disease or the trait are isolated and then implanted back into the mother. The process is expensive though, and doesn't work all the time, Harrison warned.

Once the baby is conceived, there are two main options for prenatal diagnosis, according to Harrison. Chorionic villus sampling, CVS, which has a one percent chance of miscarriage, is performed between the 10th and 12th weeks of pregnancy. A sample of the placenta is removed to allow doctors to test DNA and determine if the fetus has SCD. Amniocentesis, which involves sampling the amniotic fluid, is done after 15 weeks and has a 0.5 percent risk of miscarriage.

"One sure thing is that you don't have to unknowingly have a child with SCD in this day and age," said Harrison.

Over the years, as this disease has become a legitimate

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national health concern, postnatal testing has become routine. Today, nearly every state in the U.S., as well as the District of Columbia, Puerto Rico and the Virgin Islands, performs hemoglobin screenings on all newborns, regardless of race, to test for SCD. Children are usually diagnosed within their first few months, allowing for proper treatment to begin.

NIH recommends that patients with sickle cell disease seek consistent treatment in order to manage the symptoms and limit the frequency of medical crises. Supplementation with folic acid, an essential element in producing cells, is recommended because of the rapid red blood cell turnover. Such treatment, in combination with newborn screening programs, parent education and penicillin prophylaxis to prevent infection, has aided in a drastic reduction in mortality rates in recent years.

Currently, researchers are studying a number of new drug treatments to possibly reduce the severity and frequency of complications of the disease. These include treatment with an antifungal drug called clotrimazole, which may help prevent red blood cells from sickling. Hydroxyurea—approved in 1998 by the Food and Drug Administration for use in treating patients over the age of 18 with SCD—has improved the survival rate of some of the most severe patients, according to NIH. About 100 children with sickle cell disease have been cured through a bone marrow transplant, using donated bone marrow from an immunologically matched sibling. However, this approach carries a high risk—about 10 percent of children have died as a result of the transplant.

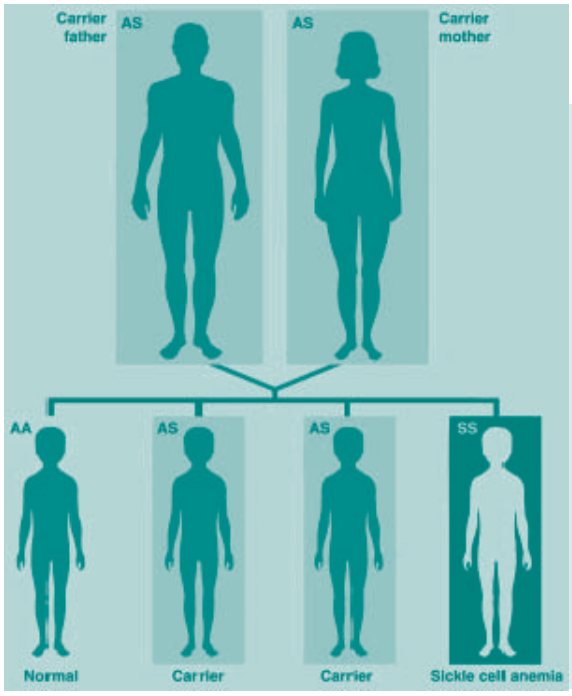
For all of the advanced research and new treatments, though, Andrew's mother still thinks there is work to be done to educate the public about the disease.

"I think about Andrew and I want people to know about this disease," she said. "I want them to be sensitive. I want them to be knowledgeable."

For more information, contact the Sickle Cell Disease Association of America at 800-421-8453 or go to http://www.sicklecelldisease.org ❖

For more information on the Sickle Cell Disease and Newborn Screening Program at HRSA, go to http://mchb.hrsa.gov/grants/default.htm#sickle❖

For more information, contact the National Heart, Lung, and Blood Institute at 301-592-8573 or go to http://www.nhlbi.nih.gov/health/public/blood/index.htm❖



Signs and Symptoms

The signs and symptoms of sickle cell anemia are different in each person. Some people have mild symptoms. Others have very severe symptoms and are often hospitalized for treatment. The most common symptoms or signs include, but are not limited to:

- Anemia
- Pain when sickle-shaped red blood cells block the flow of blood to an organ
- Fatigue (feeling very tired)
- Paleness
- Yellowing of the skin and eyes (jaundice)
- Shortness of breath
- Swelling in hands and feet
- Eye problems
- Infections
- Pneumonia and influenza
- Hepatitis
- Delayed growth and puberty
- Sores (ulcers) on the legs.
- Stroke
- Gallstones

Source: National Heart, Lung, and Blood Institute

Trait vs. Disease

When two people with sickle cell trait have a baby, there is a:

❖ One in four chance (25 percent) the baby will inherit two sickle cell genes and have the disease.

❖ One in four chance (25 percent) the baby will inherit two normal genes and not have the disease or trait.

❖ Two in four chance (50 percent) the baby will inherit one normal gene and one sickle cell gene. The baby will not have the disease, but will have sickle cell trait like the parents.

For more information, visit the National Institutes of Health Diseases and Conditions Index at http://dci.nhlbi.nih.gov/Diseases/Sca/SCA_Causes.html ❖

Information and charts/National Heart, Lung, and Blood Institute