What is Sickle Cell Disease?

Sickle cell disease, also known as sickle cell anemia, is inherited. People who have the disease inherit two copies of the sickle cell gene—one from each parent. The gene codes for the production of an abnormal hemoglobin, a protein that carries oxygen in the blood. The gene is passed from parent to child through the germline, the sex cells that give rise to sperm or egg cells.

Normal red blood cells are flexible and disk-like, allowing them to easily pass through blood vessels. Diseased red blood cells are misshapen and rigid, taking on a characteristic “C” shape that is the hallmark of sickle cell disease. Sickled cells tend to be sticky and form little clumps that can block blood vessels, especially blood vessels in the brain, causing severe pain and organ damage. People who have sickle cell disease can experience episodes of extreme pain called “sickle cell crises.” Some people have very severe symptoms while others have mild symptoms. People with sickle cell trait have one normal and one abnormal (sickle) copy of the sickle cell gene, so they do not have sickle cell disease; however, they have a greater risk of developing sickle cell anemia if they inherit the abnormal gene from a family member.

Bone marrow transplants provide a cure for some people with sickle cell disease. However, people who have sickle cell trait have a greater risk of developing sickle cell anemia if they inherit the abnormal gene from a family member.

The National Heart, Lung, and Blood Institute

The National Heart, Lung, and Blood Institute (NHLBI), which began as the National Heart Institute in 1936, has supported sickle cell research since 1940. The NHLBI was founded as the National Heart Institute. Since 1972, when the National Sickle Cell Disease Program began, the NHLBI has spent more than $1 billion on sickle cell research.

The NHLBI has playd a crucial role in not only funding basic research but also developing and implementing large clinical trials, and conducting workshops and consensus meetings to guide the research agenda. Research on sickle cell disease and other diseases that affect hemoglobin has played a central role in the advancement of genetics and molecular biology and has sparked innovations in other areas of medicine.

Future

The past 100 years of sickle cell research have resulted in landmark discoveries that ushered in the era of molecular genetics. The NHLBI continues to look ahead to find new and better treatments. Its translational research portfolio of basic, clinical, and translational research addresses the genetic factors affecting disease manifestation, regulation of hemoglobin synthesis, development of drugs to increase fetal hemoglobin production, and the development of animal models for preclinical studies. The institute supports research on translation of blood-forming stem cells, gene therapy, a better understanding of new treatments for pain, optimal uses of blood transfusions, and management of iron overload related to blood transfusions.

The Institute is also leading an effort to develop evidence-based clinical practice guidelines for the care of people who have sickle cell disease, which are expected to be released in 2011. The NHLBI is committed to working with other agencies within the Department of Health and Human Services to disseminate the clinical guidelines with an emphasis on care by primary care practitioners. To ensure that the new guidelines reach their intended audience, the NHLBI will launch a public awareness and education campaign to focus nationwide attention on sickle cell disease as a serious public health issue.

The NHLBI recognizes that actively engaging patients, families, practitioners, and communities is essential to improving the lives of persons affected by sickle cell disease, and will continue to work with them, community-based groups and scientific organizations to do so.

Sickle Cell Disease and Clinical Trials

The NHLBI sponsors a number of important clinical trials designed to advance the search for better treatments of sickle cell disease. These studies would not be possible without the participation of volunteers who help researchers determine which treatments work. For information on current clinical trials, please visit http://www.sicklecellclinicaltrials.gov.


The NHLBI’s Health Information Center

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Discrimination Prohibited: Under provisions of applicable public laws enacted by Congress since 1964, no person in the United States shall, on the grounds of race, color, sex, national origin, handicap, age, or any other basis prohibited by law, be excluded from participation in, be denied the benefits of, or be subjected to discrimination under any program or activity, on the basis of race, color, or any other condition. In addition, Executive Order 12254 prohibits discrimination on the basis of sex, race, color, national origin, handicap, age, and handicap. Executive Order 12254 states that if federally funded contractors may not discriminate against any employee or applicant for employment because of race, color, religion, sex, national origin, age, or handicap. Therefore, the Heart, Lung, and Blood Institute must be operated in compliance with these laws and Executive Orders.

A Century of Progress: Milestones in Sickle Cell Disease Care

Introduction

In 1910, Chicago physician James B. Herrick published a description of oddly shaped blood cells taken from dental student Walter Clement Noel, providing the first clue to what is now known as sickle cell disease. The disease is named for the sickle-shaped red blood cells that carry oxygen throughout the body. People who have sickle cell disease can experience a variety of symptoms, from mild to very severe, and the severity of symptoms can vary from person to person. People who have sickle cell disease can also experience life-threatening complications, including stroke, heart attack, and pneumonia.

One hundred years later we know that the sickle-shaped cells are a defect in hemoglobin, the protein in red blood cells that carries oxygen throughout the body. A gene alteration in the hemoglobin molecule causes the body to produce misshapen red blood cells, many of which take on the characteristic “C” shape that is the hallmark of sickle cell disease.

Sickle cell disease occurs in approximately one out of every 600 African Americans. All states screen newborns for sickle cell disease. Sickle cell disease affects an estimated 70,000 to 100,000 people, the majority of whom are African Americans. In the United States, sickle cell disease affects an estimated 100,000 to 150,000 people, the majority of whom are African Americans.

In the United States, sickle cell disease is one of the most common genetic disorders, affecting about 1 in 363 African American newborns. Sickle cell disease is the most common genetic disorder among African Americans. In addition, about 2 million people in the United States have sickle cell trait. People who have sickle cell trait do not have the disease, but they carry one of the genes that cause it. Similar to people who have sickle cell disease, people with sickle cell trait can pass the gene to their children.

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Researchers suggest exchange of blood vessels.

In 1949-50, the University of Chicago physician James B. Herrick described sickle cell disease for the first time. Herrick performed a description of sickled Chicago physician James B. Herrick (1879-1950). Herrick performed a description of sickled blood samples from a 20-year-old woman. Herrick was the first to publish a description of sickled blood samples from a 20-year-old woman. Herrick was the first to publish a description of sickled blood samples.

The shape of the reds was very irregular, but what especially caught the eye of Dr. Linus Pauling and others was the presence of sickled blood cells. The level of fetal hemoglobin in their red blood cells was found to be 1.5 times normal.

In 1961, Dr. Roland Scott established the sickle cell branch at the National Heart, Lung, and Blood Institute (NHLBI). Scott and his colleagues began funding studies on sickle cell disease and other conditions. In 1963, the three-dimensional structure of the hemoglobin protein was deciphered by F. H. C. Crick and M. H. F. Perutz. Perutz received the Nobel Prize for this work in 1967.

In 1968, the National Heart Institute established First round of grants includes $8,640 to the University of Chicago to develop a diagnostic tool developed to identify the nature of sickle cell disease, raising research funding to $10,000 per grant. In 1969, Dr. Marclan A. Walker, a 21-year-old West Virginia college student, describes having lived with sickle cell disease in the public eye.

“Sickle Cell Research for Treatment and Cure,” NHLBI sponsored study shows once again the challenges of sickle cell disease. The NHLBI convenes workshop of NIH Consensus Development Panel Report. Dr. Otis Brawley, an oncologist, brings together researchers, health professionals, and the public to discuss key public outreach issues. Dr. Brawley shares the compelling benefits of hydroxyurea and suggests as possible determinant of disease. Newborn screening for sickle cell disease encouraged in-state, the District of Columbia, Puerto Rico, and the U.S. Virgin Islands.


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