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The National Heart, Lung, and Blood Institute

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Introduction

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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 production of an abnormal hemoglobin. A person inherits only one copy of the sickle cell gene; he or she will 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.

In the United States, sickle cell disease affects an estimated 70,000 to 100,000 people, the majority of whom are African Americans. All states screen newborns for sickle cell disease. Sickle cell disease occurs in approximately one out of every 500 African American births and one out of every 8,000 Hispanic American births. In addition, about 2 million people in the United States have sickle cell trait.

The symptoms and complications of sickle cell disease vary widely. Some people with mild symptoms While others have very severe symptoms and are hospitalized frequently for treatment. Normal red cells pass smoothly through the blood vessels, but sickled cells are stiff and sticky. Sickled cells tend to form clumps that can block blood flow and lead to episodes of extreme pain, known as crises, as well as chronic damage to vital organs. Persons with sickle cell disease have life-long anemia because their red blood cells survive only about one-tenth as long as cells with normal hemoglobin.

Bone marrow transplants offer a cure to children and adolescents who have a matched bone marrow donor. Because of the limited availability of matched bone marrow donors, however, sickle cell disease has no widely available cure. Treatments are available to address symptoms and complications.

People who have sickle cell disease live longer, more productive lives. In the early 1950s, the average lifespan was only 14 years. Today, individuals with sickle cell disease are living into their forties or fifties, and beyond.

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 revitalized research portfolio of basic, clinical, and translational research addresses the genetic factors affecting disease manifestations, 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 transplantation of blood-forming stem cells, gene therapy, a better understanding of and 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 audiences, the NHLBI will launch a public awareness and education campaign to focus national attention on sickle cell disease as a serious public health issue.

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Sickle Cell Disease and Clinical Trials

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The disease sickle cell anemia was first diagnosed in 1910. However, it wasn’t until the 1940s that scientists began to study the disease in depth. In 1949, Dr. James Neel published a description of sickled red blood cells and their role in sickle cell anemia. Neel’s work laid the foundation for understanding how sickle cell disease works.

Since then, research has focused on finding ways to treat sickle cell disease. One of the most promising treatments is hydroxyurea, which can be used to reduce the number of sickled red blood cells. Other treatments include bone marrow transplantation and gene therapy.

Despite progress, sickle cell disease remains a serious condition. People with sickle cell disease have a higher risk of developing other health problems, such as infections and heart disease. However, with proper care, people with sickle cell disease can lead healthy lives.

Today, there are many resources available to help people with sickle cell disease manage their condition. These include medical care, education, support groups, and community services. With continued research and innovation, we can continue to improve the lives of people with sickle cell disease.