What is Sickle Cell Disease?

Sickle cell disease is a genetic disorder that affects the production and function of hemoglobin, the protein in red blood cells that carry oxygen. People with sickle cell disease inherit a gene mutation that causes their red blood cells to become misshapen and rigid, resembling a sickle or crescent moon shape. This misshapen shape makes the cells less efficient at carrying oxygen and more prone to forming clumps, which can block blood flow and cause pain and organ damage.

The symptoms and complications of sickle cell disease vary widely. Some people with mild symptoms only have occasional pain episodes, while others experience chronic pain, organ damage, and a shortened lifespan. Treatment options range from pain management and blood transfusions to bone marrow transplantation and gene therapy.

What Causes Sickle Cell Disease?

Sickle cell disease is caused by a genetic mutation in the HBB gene, which is located on chromosome 11. The mutation affects the production of a protein called hemoglobin, which helps red blood cells carry oxygen. In sickle cell disease, the hemoglobin is abnormal and causes the red blood cells to become misshapen and rigid, leading to pain and other complications.

Epidemiology of Sickle Cell Disease

Sickle cell disease is more common in people of African ancestry, but it can occur in people of any race. An estimated 1 in 363 African Americans carry the sickle cell trait, and about 1 in 12 African Americans has sickle cell disease. In other populations, sickle cell disease is less common, but it still affects people of Hispanic American birth.

How Is Sickle Cell Disease Diagnosed?

Sickle cell disease is diagnosed based on a combination of symptoms, physical examination, and laboratory tests. A complete blood count (CBC), which measures the number and types of cells in the blood, can reveal anemia and other signs of sickle cell disease. A blood smear, which visually examines the red blood cells, can also be used to diagnose sickle cell trait or disease.

How Is Sickle Cell Disease Treated?

There is no cure for sickle cell disease, but there are treatments and interventions that can help manage symptoms and prevent complications. Pain management is a key aspect of care, and patients may receive medications, oxygen therapy, and blood transfusions as needed. Other treatments include hydroxyurea, a medication that reduces blood cell production and may help prevent pain crises, and bone marrow transplantation, which can be used for people with severe disease.

Future

The NHLBI continues to support research that is essential for the prevention and treatment of sickle cell disease. We have supported the development of new drugs, therapies, and interventions that can help manage symptoms and prevent complications. We have also supported research on the genetics of sickle cell disease, which is essential for understanding the disease and developing new treatments.

The NHLBI is committed to advancing research and care for people with sickle cell disease. We will continue to support research that is essential for the prevention and treatment of this disease, and we will work with other federal agencies and organizations to coordinate the use of available resources to the greatest advantage of the people we serve.
The dual role of the red blood cell
is...to carry oxygen to the body, and to
return carbon dioxide to the lungs.

In the late 19th century, Dr. James
Herrick first described the sickle
shape of red blood cells from
sickle cell disease patients. He
called these cells “sickled cells”
because they appeared as
irregular, but what especially
struck Dr. Herrick was the
brittle nature of the sickled cells.

Sickle cell disease is a genetic
condition characterized by
abnormal red blood cells that
become rigid and sticky, blocking
blood flow and causing acute
medical conditions such as
pain crises and organ failure.

In 1910, Dr. James Herrick
published a paper describing
the sickle shape of red blood cells
from patients with sickle cell disease.

In 1911, Dr. Walter Clement
Noel, a dental student from
Memphis, published a paper
determining sickle cell trait and
disease. Remarked upon paper.

In 1914, the term “sickle cell
disease” was coined based on a
paper.

From Grenada, the real diagnosis is
sickle cell disease. Receiving gene from one
parent only produces sickle cell trait.

1916 In 1916, a study showed the
carriage of sickled cells in babies from newborns
to age 70. First study to demonstrate that
hydroxyurea reduces number of sickled cells.

1922 In 1922, the first sickle cell
transfusion performed. Transfusion was
done to a child with sickle cell disease.

1924 In 1924, a study showed that
hydroxyurea reduces number of sickled cells.

1927 In 1927, a study demonstrated the
effectiveness of hydroxyurea in reducing
the number of sickled cells in mice.

1932 In 1932, a study showed that
hydroxyurea reduces number of sickled cells.

1936 In 1936, a study showed the
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1939 In 1939, a study showed that
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2017 In 2017, a study showed that
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2020 In 2020, a study showed that
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2023 In 2023, a study showed that
hydroxyurea reduces number of sickled cells.

Fame and distinction await the
National Medical Association
– Dr. Robert B. Scott, Journal of the American
Medical Association

In 1972, Dr. Robert B. Scott,
Journal of the American
Medical Association, declared
sickle cell disease to be a
community health problem.

In 1994, the National Association
for Sickle Cell Disease, Inc. was
founded by K. John Marcellus
Walker, a 21-year-old West
African American male, to
advocate for sickle cell disease
awareness and treatment.

The National Association for Sickle
Cell Disease, Inc. recommends
the adoption of Sickle Cell Newborn
Screening and the establishment
of comprehensive sickle cell centers
and programs in each state.

The Newborn Screening Saves Lives Act of 2007 establishes
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receive screening for sickle cell disease.

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