



Sickle Cell Anemia: Genetic Code, Symptoms, and Advances in Diagnosis

Aram Jamin*

Department of Molecular Genetics, University of Florida, Florida, United States of America

DESCRIPTION

Sickle Cell Anemia (SCA) is a hereditary blood disorder characterized by abnormal hemoglobin, the protein responsible for carrying oxygen in red blood cells. This condition is prevalent in certain populations, particularly those with ancestry from sub-Saharan Africa, the Middle East, India, and regions with high malaria prevalence. Sickle cell anemia is a complex and challenging disease that affects millions of people worldwide. This article delves into the genetic basis, symptoms, diagnosis, and treatment of sickle cell anemia, bright on the impact it has on individuals and the ongoing efforts to manage and eventually cure the disease. The core problem of sickle cell anemia lies in the genetic code. Hemoglobin, the oxygen-carrying protein in red blood cells, is composed of four subunits—two alpha globin and two beta globin. In individuals with sickle cell anemia, a single mutation in the beta globin gene leads to the production of abnormal hemoglobin, known as hemoglobin. This mutated hemoglobin causes red blood cells to take on a distinctive sickle shape when oxygen levels are low, leading to various complications. The beta globin gene mutation responsible for sickle cell anemia is inherited in an autosomal recessive manner. This means that an individual must inherit a copy of the mutated gene from both parents to develop the disease. If an individual inherits one normal gene and one mutated gene, they are carriers of the sickle cell trait but do not manifest symptoms of the disease. However, carriers can pass the mutated gene to their offspring.

Symptoms and complications

Sickle cell anemia manifests in a variety of symptoms and complications, ranging from mild to severe. The severity of symptoms can vary among individuals and may be influenced by factors such as the presence of other genetic variants, environmental factors, and the individual's overall health.

Chronic anemia: One of the primary symptoms of sickle cell anemia is chronic anemia, which results from a reduced number of healthy red blood cells. Anemia can lead to fatigue, weakness, and shortness of breath.

Pain crises: Sickle cell anemia is characterized by recurrent pain crises, also known as vaso-occlusive crises. These episodes occur when sickle-shaped red blood cells block blood vessels, leading to pain, tissue damage, and potential organ dysfunction.

Organ damage: Over time, repeated blockages of blood vessels can result in organ damage. Organs commonly affected include the spleen, kidneys, liver, lungs, and brain.

Infections: Individuals with sickle cell anemia are more susceptible to infections, particularly those caused by bacteria such as *Streptococcus pneumoniae*. This increased susceptibility is due to the compromised function of the spleen, an organ vital for fighting infections.

Stroke: The abnormal shape of sickle cells increases the risk of stroke, especially in children. Blockages in blood vessels can lead to reduced blood flow to the brain, causing long-term neurological damage.

Diagnosis

Hemoglobin electrophoresis: This laboratory test separates and identifies different types of hemoglobin based on their electrical charge. Individuals with sickle cell anemia will show an abnormal hemoglobin pattern.

Genetic testing: Genetic testing is used to confirm the presence of the beta globin gene mutation responsible for sickle cell anemia. This can be particularly valuable for identifying carriers and providing genetic counseling for at-risk couples.

Complete Blood Count (CBC): A CBC can reveal anemia by assessing the number of red blood cells, hemoglobin levels, and other related parameters.

Peripheral blood smear: Microscopic examination of a blood smear can reveal the characteristic sickle-shaped red blood cells.

Newborn screening programs have been implemented in many countries to identify infants with sickle cell anemia early in life. Early diagnosis allows for timely intervention and management to improve outcomes for affected individuals.

Correspondence to: Aram Jamin, Department of Molecular Genetics, University of Florida, Florida, United States of America, E-mail: jamin.aram@gmail.com

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Treatment and management

While there is currently no cure for sickle cell anemia, various treatment strategies aim to alleviate symptoms, prevent complications, and improve the quality of life for individuals with the disease. Pain crises are a symbol of sickle cell anemia, and managing pain is a central aspect of treatment. This may involve the use of pain medications, hydration, and other supportive measures. Hydroxyurea is a medication that can stimulate the production of fetal hemoglobin, which inhibits the formation of sickle cells. This drug has been shown to reduce the frequency of pain heads and other complications. In cases of severe anemia or to prevent complications such as stroke, blood transfusions may be recommended. Regular transfusions can help maintain a higher percentage of normal red blood cells. For select individuals, especially children, bone marrow transplantation may deal a potential cure. However, this procedure is complex and carries risks, making it suitable for a limited number of cases. Managing the overall health of

individuals with sickle cell anemia is vital. This includes vaccinations to prevent infections, adequate hydration, and regular monitoring of organ function.

Sickle cell anemia remains a significant global health challenge, particularly in regions with a high prevalence of the disease. Advances in our understanding of the genetic and molecular basis of the condition have led to improved diagnostic methods and treatment options. While current therapies focus on managing symptoms and preventing complications, ongoing research for more targeted and curative interventions. Addressing the multifaceted aspects of sickle cell anemia requires a collaborative effort involving healthcare professionals, researchers, policymakers, and the affected communities. Through increased awareness, support, and scientific advancements, the hope is that the burden of sickle cell anemia can be alleviated, ultimately leading to better outcomes and an improved quality of life for those living with this challenging condition.