**Sickle-Cell Anemia**

**Name**

**Institutional Affiliation**

**Date**

SICKLE-CELL ANEMIA

**Introduction**

Sickle-cell anemia is a genetic disorder of red blood cells. It occurs when there are insufficient red blood cells to carry blood to all parts of the body. The red blood cells in this condition contort into crescent or sickle shape. The rigid cells get stuck in small blood vessels; thus, they slow or block blood flow to all parts of the body. In other words, the disorder results from mutated hemoglobin. The condition leads to early cell death, causing a shortage of healthy red blood cells; this becomes a challenge in the supply of blood to all parts of the body—the inability to supply sufficient oxygen results in pain episodes (Williams & Thein 2018).

The sickle-celled red blood cells clog blood vessels; thus, preventing tissues from getting enough blood; this results in episodes of pain. When these cells move across the spleen, they get destroyed, and the body cannot replace the lost blood cells early enough; this leads to insufficient red blood cells in the body.

**History and causes of sickle cell anemia**

Sickle cell anemia was first discovered in 1910. Herrick identified the disease in a dental student who presented pulmonary symptoms; thus, he termed it sickle cell. He did this after seeing the peculiar appearance of the patient's red blood cells. Edward irons examined the blood of the patient in Chicago Presbyterian Hospital. Sickle cell anemia is common in people of African, but it present in Portuguese, Spanish, Turks, Sardinians, Greek, and Mainland Italians. In 1949, Linus Pauling and colleagues demonstrated that sickle cell anemia occurs due to red blood cells' abnormality. It was during this time that a genetic disease was linked to a mutation of a protein.

**Causes of sickle-cell anemia**

The disorder results from the mutation HBB gene. These genes have blueprints for cells to make hemoglobin protein. Hemoglobin is made up of iron-containing heme that gives blood its color—the same iron helps hemoglobin to release oxygen. HBB gene in sickle-cell changes one of the amino acids in the hemoglobin; in this case, the hemoglobin protein stick together and form stick fibers. These fibers affect the shape of the red blood cells, making them develop the peculiar sickle or crescent shape. People with sickle cell anemia have hemoglobin S molecule (HbS).

Additionally, children are at risk of the disorder if all parents have the sickle-cell anemia trait. When both parents carry the gene for sickle-cell anemia, the child inherits a copy of the condition from each parent; thus, the child develops the condition. Notably, if only one parent has sickle-cell trait, all of their children will have sickle-cell trait, but none of these children will have sickle-cell disease.

**Diagnosis of sickle-cell anemia**

Blood test-Doctors use a blood test to detect a defective form of hemoglobin that identifies the presence of sickle-cell anemia. Genetic screening is done for new bone babies. Hemoglobin electrophoresis is a blood test used to diagnose newborn babies to determine if the child has a sickle-cell trait or a sickle-cell carrier. Doctors examine transcranial Doppler ultrasound screening to assess the risk of stroke and start treatment immediately.

Doctors should examine the four types of sickle-cell anemia; they include sickle-cell anemia SS, Sickle-hemoglobin-C disease, Sickle-Beta Plus, and Sickle- Beta Zero Thalassemia. The HbSS occurs when a person inherits two sickle-cell traits from the parents. It is the most severe type of condition. HbSC occurs when a person inherits the sickle-cell gene from one parent resulting in abnormal hemoglobin; this type is less severe than HbSS. HbS Beta Thalassemia is inherited from one parent and a gene for Beta Thalassemia from another parent. Lastly, the sickle-cell trait occurs when a person has only one faulty gene.

**Biological characteristics and symptoms of sickle-cells**

The cells of sickle-cell anemia break down prematurely. The cells are sickle-shaped; thus, it leads to shortness of breath. The condition also leads to fatigue, delayed growth, and development in children. The rapid breakdown of red blood cells can cause yellow skin and eyes. Painful episodes can occur when the sickle cell blood cells stiffen and become inflexible. The deprivation of sufficient oxygen to body organs can lead to organ damage. Sickle cell anemia could increase blood pressure to the blood vessels that supply oxygen to the heart leading to heart failure.

Additionally, the condition is characterized by hypertension.

The symptoms of sickle-cell anemia include swelling of hands and feet, frequent infection, delayed growth, and vision problem. Other symptoms of the condition include joint pain and chronic neuropathic pain. The pain induces when sickle-shaped blood cells prevent the flow of blood into small vessels should be treated as a medical emergency.

The early signs of sickle-cell anemia include jaundice, icterus, fatigue, and dactylitis. Sickle-cell anemia patients do not cause learning difficulties, but pain episodes can cause the inability to concentrate in class. When a stroke occurs, this can lead to loss of consciousness, speech problem, and seizures.

**Prognosis of sickle-cell anemia**

Those diagnosed with sickle-cell anemia have a short life expectancy. Some of those infected with this condition remain without the symptoms for one year. Some people do not survive beyond infancy or early childhood. There are new treatments that have been invented to help improve life expectancy and life quality. Lesha Thomas found stem cell transplant as the cure for sickle-cell anemia.

Additionally, an allogeneic bone marrow transplant can cure the diseases. Preventing pain episodes can help improve sickle-cell anemia. Treatment of the disease includes blood transfusion and bone marrow transplant. Those with sickle-cell anemia should drink plenty of water, dress in warm clothes when it is cold, and avoid swimming in cold water. Other ways of preventing infection include regular hand washing, following food safety guidelines, and receiving flu and pneumococcal disease vaccination.

The treatment usually involves preventing pain crises, preventing complications, and relieving symptoms of the disease. The treatment involves using drugs like L-glutamine oral powder, Crizanlizumab, voxelotor (can help with anemia), and pain-relieving medication (Quinn, 2018). The pain-relieving medications include ibuprofen and the use of opioids if pain persists. A patient with a stroke should undergo a blood transfusion.

Parents with sickle-cell anemia trait should undergo genetic testing and in vitro fertilization to prevent the passing of the gene to children. Preimplantation genetic testing helps to test the embryo and prevents the disorder. Sickle-cell anemia worsens over time; thus, treatments are available to reduce complications and increase the lifespan of patients.

References

Quinn, C. T. (2018). l-Glutamine for sickle cell anemia: more questions than answers. *blood*, *132*(7), 689-693.

Williams, T. N., & Thein, S. L. (2018). Sickle cell anemia and its phenotypes. *Annual review of genomics and human genetics*, *19*, 113-147.