SICKLE CELL DISEASE

Research Paper, Part 1

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Hemoglobin (Hemoglobin A), a crucial cellular component of red blood cells in the human body, aids in transmitting oxygen from the blood to the many regions of the body where it is required. When the shape of the red blood cell alters from round and smooth to stiff, sickle-shaped, and sticky, the faulty red blood cell cannot provide an adequate amount of oxygen to vital organs and tissues. This inherited blood disorder called sickle cell disease manifests itself in sickle-shaped red blood cells due to abnormal hemoglobin, or hemoglobin S. Because of its sensitivity to low oxygen levels, the faulty Hemoglobin S causes red blood cells to alter their shape.

Sickle cell disease is a homozygous autosomal recessive condition. This indicates that each parent must pass the child on one copy of the defective hemoglobin (Hemoglobin S). The parents are typically heterozygous, which implies that they carry the defective gene along with the normal hemoglobin gene (Hemoglobin A) but do not have the condition. This is known as the sickle cell trait. Since a person with sickle cell trait has enough normal hemoglobin to prevent the sickling of red blood cells, signs and symptoms do not usually manifest.

In contrast to other nations with high mortality rates, such as France and the UK, where 50–90% of children with sickle cell disease die within the first five years of life, over 94% of those born in the US live into adulthood. Since sickle cell disease is genetic, an individual is born with the condition. Sickle cell disease screening is done as standard care in the US, while the countries with higher mortality rates due to low resources, newborn screening is not routine care.

According to the CDC, one in twelve African Americans living in the United States has sickle cell traits, frequently passed on to their offspring. Sickle cell disease affects about 100,000

Americans (CDC, 2022). The genetic disorder often impacts people with ancestors from the Middle East, Asia, the Caribbean, and the Eastern Mediterranean.

Normal red blood cells are round, smooth, and concave because they lack a nucleus, which enables them to move quickly through the body's vessels. Hemoglobin, a protein that aids in maintaining the structure of red blood cells, helps it carry oxygen. When exposed to a hypoxic environment, red blood cells with faulty Hemoglobin S and other abnormal Beta alleles undergo polymerization and develop rigidity. Hemolysis is a risk associated with the rigid RBC, and the increased density impacts the blood flow. Due to enhanced hemolysis, red blood cells carrying hemoglobin S may only live 10 to 20 days as opposed to the usual RBC, which can last up to 120 days. The most prevalent and severe form of sickle cell disease is sickle cell anemia, caused by the short-lived RBC and the bone marrow's failure to make enough RBCs.

Sickle cell disease leads to many complications, known as sickle cell crisis. Depending on the induvial, several forms of sickle cell crises, including vaso-occlusive, hyper hemolytic, aplastic, and splenic sequestration. A vaso-occlusive problem happens when the sickled red blood cells restrict blood flow, depriving the tissues of oxygen. This may result in a fever, pain, edema, and kidney and stroke problems. A hyper hemolytic crisis occurs when an individual's hemoglobin level drops abruptly, which can cause organ failure, jaundice, gallstones, anemia, etc. Red cell production temporarily stops during an aplastic crisis, which causes anemia. A splenic sequestration crisis occurs when the spleen becomes clogged with sickled red blood cells, causing the organ to enlarge and cease functioning. This situation causes the risk of infection and potential spleen removal. Other complications, seen more in common with adults, include stroke, cognitive dysfunction, leg ulcers, chronic pain, retinopathy, gallstones, pulmonary hypertension, chronic kidney disease, etc.

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