Sickle Cell Disease: What You Need to Know

By Ada Lynda Ijeozor

Introduction

Sickle cell anaemia is a disease passed down through families. It is an inherited condition of the blood and it is considered the most common genetic disease. It is a group of disorders that affects haemoglobin. Haemoglobin is the protein, which carries oxygen to the tissues.

Sickle cell disease is most common with Africans and African-Americans. It is also found in other ethnic and racial groups, including South and Central America, the Caribbean, Mediterranean countries (such as Turkey, Greece, and Italy), India, and Saudi Arabia. It is estimated that around one baby in every 1,900 born in the UK and one out of every 835 African American babies have sickle cell disease. Although current data on Sickle cell disease in Africa is inadequate, it is estimated that 3 out of every 100 babies in African have sickle cell disease and this is associated with the very high rate of childhood mortality in Africa.

Sickle cell disease causes the haemoglobin protein to be altered making the surface of the protein sticky and causing the different sickle cell haemoglobin molecules to stick together, forming long fibres or rods. These fibres distort the shape of the red blood cells into sickles and cause these cells to be less flexible or stiffer in traveling through the red blood vessels, preventing free travel of the red blood cells and thereby depriving the organs and tissues in the body of oxygen. When the organs and tissues are deprived of oxygen, it can often result to periodic pain, organ damage and stroke.

Red blood cells (RBC's) that have undergone the sickling process rupture more quickly than regular RBC's as the sickling process makes them more fragile and less malleable and this leads to anaemia. The damaged sickle red blood cells also clump together and stick to the walls of blood vessels, blocking blood flow.

Causes

Sickle cell disease is not contagious, but is acquired at birth. It occurs when a child inherits two sickle haemoglobin genes, one from each parent. The disease is caused by a genetic abnormality in the gene for haemoglobin, which results in the production of sickle haemoglobin. When oxygen is released from sickle haemoglobin, it sticks together and forms long fibres or rods, which damage and change the shape of the red blood cell.

Symptoms

The signs of sickle cell disease usually begin a few months after birth. Although in some cases, the signs may not appear until childhood. The early signs of sickle cell include;

- Sleeping longer or more often

- Tiredness, difficulty breathing
- Pain or swelling in the hands or feet
- Cold hands or feet and pale skin
- Yellowing of the eyes and skin (jaundice)
- Rapid heart rate
- Delayed growth and puberty
- Painful joints caused by arthritis
- Heart or liver failure due to too much iron (from blood transfusions)
- Lung infection (pneumonia)
- Urinary tract infection
- Anaemia
- Bone infection (osteomyelitis).

Almost all people with sickle cell anaemia have painful episodes called crises. These can last from hours to days.

Key facts

- Sickle cell disease is an inherited blood disorder and is passed down through families.

- Sickle Cell Anaemia (SCA) is the most common of the sickle cell diseases and is defined by the presence of the abnormal haemoglobin HbS. This is caused by having two copies of the beta-globin gene containing the genetic alteration for HbS. The other forms of sickle cell disease result from inheriting a genetic alteration for HbS in one gene and another abnormal beta-globin chain variant in the second beta-globin gene.

- Sickle Cell Anaemia is characterised by episodes of pain, chronic haemolytic anaemia and severe infections, usually beginning in early childhood.

- Sickle cell disease is common in people of African, Mediterranean, Middle Eastern, and Indian ancestry, and in people from the Caribbean and parts of Central and South America.

- The highest incidence sickle cell disease is in West Africa, where 1 in 4 of the population are carriers.

- Sickle red blood cells live only 10-20 days instead of 120 days for normal red blood cells.

- Today, the life expectancy for patients who have Hb-SS has increased to a median of 45 years and for HbSC patients to 65 years, a significant improvement over the median survival to age 14.3 years for patients having Hb-SS 3 decades ago.

- The improvement in life expectancy is related, in part, to early identification and prevention of death from pneumococcal sepsis in young children and to improved education regarding SCD complications and early intervention.

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