Perspective
Volume 12:S2, 2022

Journal of Blood & Lymph

ISSN: 2165-7831 Open Access

Sickle Cell Disease-An Overview

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About the Study

Sickle cell disease, which is an autosomal recessive disease, is the most common genetic blood ailment in the United States, with African Americans being the most affected. One in every 12 African Americans and one in every 100 Hispanic Americans has the sickle cell trait, which means they are sickle cell disease carriers.

A mutation in the hemoglobin-gene on chromosome causes sickle cell disease. Normal haemoglobin (haemoglobin-A) red blood cells are smooth and spherical, and they glide through blood arteries. Abnormal Haemoglobin Molecules (Hbs) cling together and form long, rod-like formations in persons with sickle cell disease. These formations cause red blood cells to stiffen and take on a sickle shape, causing blockages and causing damage to important organs and tissue. Sickle cells are rapidly destroyed in the bodies of sickle cell patients, resulting in anaemia. Sickle cell anaemia is the name given to the disease because of this anaemia.

When compared to normal erythrocytes, sickle cells adhere better to the endothelium. Endothelial cell activation, as a result of inflammation-associated proinflammatory cytokines such tumour necrosis factor-, interleukin-1, and interferon-, increases this. Endothelial cells express leukocyte adhesion molecules, and procoagulant factors such as Von Willebrand factor and thrombospondin are released.

Sickle cells also obstruct blood flow through vessels, causing lung tissue damage and symptoms such as acute chest syndrome, pain episodes, stroke, and priapism (painful, prolonged erection). The spleen, kidneys, and liver are also affected. Patients, particularly young children, are readily overwhelmed by bacterial infections due to spleen loss.

A simple blood test can be used to diagnose sickle cell disease. Sickle solubility testing, hemoglobin electrophoresis, High-Performance Liquid Chromatography (HPLC), and Isoelectric Focusing (IEF) are currently the most prevalent screening approaches, each with its own set of benefits and drawbacks. The sickle solubility test is a low-cost assay that detects turbidity or crystal formation following lysis of Hbs-containing erythrocytes by relying on the relative insolubility of Hbs in the presence of a reducing agent, such as sodium dithionite. The solubility test cannot distinguish between persons with sickle cell disease and sickle cell trait because, it simply identifies the presence or absence of sickle haemoglobin, and it can be mistakenly negative, necessitating confirmatory tests.

Hemoglobin electrophoresis, HPLC, and IEF are employed as either primary or confirmatory testing for sickle cell trait. These approaches can distinguish sickle cell trait from sickle cell disease syndromes by providing hemoglobin discrimination and relative quantification. Hemoglobin electrophoresis, for example, is a low-cost and widely used technique that uses gel electrophoresis principles to separate haemoglobin molecules by size and charge. Because comigration of certain rare haemoglobin variations with Hbs can make conventional electrophoresis difficult to interpret, alternative gels such as citrate agar or cellulose acetate, as well as IEF techniques, are frequently used for further haemoglobin differentiation.

In the scientific setting, molecular hemoglobinopathies procedures are frequently employed to detect sickle cell trait carriers, especially in investigations where haemoglobin electrophoresis samples are not collected. Exome sequencing, direct genotyping for the SNP that encodes the sickle mutation and even GWAS have all been made possible thanks to rapid improvements in NGS.

How to cite this article: Farooqi, Awj. "Sickle Cell Disease-An Overview ." *J Blood Lymph* 12 (2022) : 010.

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Received: 03-Jan-2022, Manuscript No. JBL-22- 50891; **Editor assigned:** 06-Jan-2022, Pre QC No. JBL-22- 50891 (PQ); **Reviewed:** 17-Jan-2022, QC No JBL-22-50891; **Revised:** 20-Jan-2022, Manuscript No. JBL-22- 50891 (R); **Published:** 27- Jan-2022, DOI: 10.37421/ 2165-7831.2022.12. 010