

Sickle Cell Disease

All cells of the human body require a regular supply of oxygen to carry out their basic functions. Red blood cells (RBCs) are instrumental in carrying oxygen to the different parts of the body. This action is mediated by the help of a protein in the RBCs, called hemoglobin, which binds oxygen and transports it throughout the body. Normally, RBCs are smooth, doughnut-like cells that are extremely flexible and are able to easily pass through the narrowest blood vessels.

Sickle cell disease results when the molecules of the structurally abnormal hemoglobin, Hemoglobin S (HbS), aggregate and lead to inefficient and sickle shaped RBCs. While normal RBCs have a life span of 120-days, these sickled RBCs die within 20 days. Since the bone marrow cannot replenish the dying cells quickly enough, the RBC count is drastically lowered; a condition known as anemia. Furthermore, sickled RBCs are very rigid and tend to get stuck in the narrow capillaries, causing blockage of blood flow to organs. This may result in ischemia (reduced blood flow), pain, and/or organ damage (especially in the spleen).

Globally, sickle cell disease affects millions of people worldwide. It is particularly common among people from Africa, Mediterranean countries (such as Greece, Turkey, and Italy), the Arabian Peninsula, India, and Hispanic regions (South America, Central America, and parts of the Caribbean). This geographical distribution of the disease is explained by the high prevalence of malaria in these regions. The malarial parasite completes part of its life cycle inside the human RBC and is not able to survive inside a sickle RBC, even if the individual is only carrying the gene for sickle cell disease without actually been clinically affected. Therefore, such individuals are immune from malaria, enabling them more fit to survive than normal individuals.

Risk Factors

Sickle cell disease is caused by a mutation in the beta globin gene, and is an autosomal recessively transmitted disorder, which means that a person needs to inherit the defective gene from both parents to develop the disease. An individual with only one copy of the defective gene (heterozygous carrier) is clinically asymptomatic and is said to possess a sickle cell trait.

Additionally, in individuals with the genetic defect, the painful sickle cell crisis can be precipitated by certain medical and environmental factors including infection, bleeding, exposure to cold weather, leg ulcers, and blockage of the blood vessels.

Diagnosis and Management

Newborns can and should be checked for sickle cell disease or sickle cell trait using any of the available diagnostic options. This is especially important if the parents happen to be carriers or affected. Prenatal genetic testing is commonly practiced in many laboratories around the world.

The only permanent form of treatment or cure in sickle cell disease is a bone marrow transplant. However, this requires a suitable, immunologically matched donor. Additionally, the condition is managed through alleviation of symptoms and avoiding complications. It is important to prevent infections like pneumonia, which is the leading cause of mortality in children with the disease. Antibiotics like penicillin are, therefore, a must for affected children. Painful vaso-occlusive crises, where small blood vessels are blocked with sickle cells, are countered by pain management methods and require prompt medical attention.

Sickle Cell Disease in Arab Populations

Sickle cell disease is highly prevalent in the Arab states. Some of the world's highest frequencies of the disease are seen in Saudi Arabia (5.2%), Oman (3.8%), UAE (1.9%). Lower values are observed in Yemen (0.95%), Bahrain (0.7%), and other Arab countries. Studies in population genetics revealed that the extent of clinical severity in sickle cell disease is related to the genetic background (haplotype) of the affected individual. In the Arabian Peninsula, sickle cell disease occurs in association with two backgrounds: the African haplotype, a severe type, mostly occurring in patients residing in the western regions of the Arabian Peninsula, and the Arabian/Asian haplotype, a moderate type, mostly occurring in patients residing in the eastern regions of the Arabian Peninsula.

The high prevalence of the disease has prompted extensive studies on it, and almost all Arab countries have undertaken vast amounts of work on the clinical features, genetics, and management of sickle cell disease. Several Arab states have taken extensive measures to reduce the rates of sickle cell disease. Screening programs are one of the most effective of such strategies. Especially in the countries of the Gulf Cooperation Council (GCC), screening of students, infants, and mandatory screening of couples before their marriage have been very effective. In Bahrain, for instance, these programs have led to a significant drop in the rates of sickle cell disease from 2.1% in year 1985 to less than 1% in year 2002. With regard to treatment, countries like Sudan and Saudi Arabia have established guidelines on the standard case management of sickle cell disease. In the United Arab Emirates, sickle cell disease is the second most prevalent hemoglobin disorder. Its preponderance has been delineated through population and premarital screening; the latter has been made mandatory effective 2006. The co-inheritance of the sickle gene with beta-thalassemia has led to a patient population with markedly varying phenotypes.

Tips to Prevent Sickle Cell Crises

A vaso-occlusive crisis in patients with sickle cell disease is a condition where blood flow to organs is severely restricted, causing damage to the organ. A little bit of special care can go a long way in making better the life of patients with sickle cell disease. The following are some of the things to remember when caring for patients with sickle disease, all aimed at reducing the incidence of painful crisis.

- Follow a healthy lifestyle pattern. Avoid smoking, second-hand smoking, alcohol consumption, and emotional stress. Eat a balanced healthy diet, and do moderate low intensity exercise regularly. Doctors may suggest taking folic acid supplements to help make new red blood cells.
- Try to avoid situations where you may get injured.
- Increase your liquid intake. Take at least 3-4 liters of liquids daily. Have fluids on hand at home and away. Avoid too much exposure to the sun.
- Avoiding infections is important. Get yourself vaccinated against viral diseases, such as influenza, pneumonia, meningitis, and hepatitis B. Generally, infants with sickle cell disease are often placed on daily penicillin therapy until at least aged 5 years.
- Consult with your physician regularly and go for regular checkups every 3-6 months. Regular eye exams are also recommended.
- Avoid extreme heat or cold. Wear warm clothes outside in cold weather and inside air-conditioned spaces during hot weather. Do not swim in cold water.
- Avoid high altitudes and non-pressurized airplane flights, where oxygen pressure might be low.
- Avoid demanding physical activities, especially if the spleen is enlarged.
- If pregnant, take care that you get good and early prenatal care.
- Consider having the child with the disease wear a Medic Alert bracelet.