# 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 different parts of the body. This action is mediated by the help of a protein in the RBCs, called hemoglobin, which binds to oxygen and transports it throughout the body. Normally, RBCs are smooth, doughnut-shaped cells that are extremely flexible and able to easily pass through the narrowest blood vessels.

Sickle cell disease occurs when molecules of 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, the sickled RBCs die within 20 days. Since the bone marrow cannot replenish the dying cells as quickly, 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 the organs. This may result in ischemia (reduced blood flow), pain, and/or organ damage (especially in the spleen).

Sickle cell disease affects millions of people worldwide. It is particularly common in Africa, the Mediterranean countries (such as Greece, Turkey, and Italy), the Arabian Peninsula, India, and the 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. The malarial parasite completes part of its life cycle inside the human RBC and is unable to survive inside a sickle RBC; this is true even when the individual has only one defective gene for the sickle cell disease (carrier) without been clinically affected. Therefore, such individuals are immune to malaria, making 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 inherited 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 have a sickle cell trait.

Additionally, in individuals with this genetic defect, a painful sickle cell crisis can be triggered 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 be tested for sickle cell disease or sickle cell trait using any of the available diagnostic options. This is especially important if the parents are affected or happen to be carriers. Prenatal genetic testing is commonly practiced in several laboratories around the world.

The only permanent form of treatment or cure for sickle cell disease is a bone marrow transplant. However, this requires a suitable immunologically-matched donor. The condition can be managed by alleviating symptoms and avoiding complications. Folic acid supplements may be prescribed to enhance the production of new red blood cells in patients. It is important to prevent infections like pneumonia, which is the leading cause of mortality in children with

this disease. Antibiotics like penicillin are, therefore, a must for affected children. Painful vaso-occlusive crises, where small blood vessels are blocked with sickle cells, require prompt medical attention and are countered by pain management methods.

## SICKLE CELL DISEASE IN THE ARAB WORLD

Sickle cell disease is highly prevalent in the Arab states. Some of the world's highest frequencies of the disease are seen in Oman (3.8%), Saudi Arabia (1%-2%), and UAE (1.9%). Lower values are observed in Yemen (0.95%), Bahrain (0.4%), 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 individuals. 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 widespread measures to reduce the rates of sickle cell disease. Screening programs are one of the most effective of such strategies. A few of the Gulf Cooperation Council (GCC) countries have initiated screening of students, infants, as well as mandatory screening of couples before marriage, which has 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 0.6% in the year 2009. Saudi Arabia has 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 high prevalence has been lowered through population and premarital screening; the latter has been made mandatory effective in 2006. The co-inheritance of the sickle gene with beta-thalassemia has led to a patient population with markedly varying phenotypes.