HEMOPHILIA

Hemophilia is the earliest known hereditary bleeding disorder. When injured, affected individuals tend to have a slow clotting (coagulation) process because of the lack of necessary blood factors (proteins). Consequently, those patients have prolonged bleeding after injury or surgery.

Hemophilia is clinically classified into two types: Hemophilia A (Classic Hemophilia) associated with the deficiency of coagulation factor VIII, and Hemophilia B (also known as Christmas Disease) in which coagulation factor IX is deficient. Hemophilia A and B have very similar signs and symptoms even though they are caused by mutations in different genes. It is estimated that 1 in 5,000 to 10,000 males are born with hemophilia A worldwide accounting for 85% of global hemophilia cases. The remaining cases (15%) exhibit type B hemophilia at the rate of 1 in 30,000 males worldwide.

The amount of the clotting factor in the blood identifies the severity of both types of hemophilia. Patients with less than 1% level of the normal clotting factor have severe hemophilia. Unfortunately, most hemophiliac patients (70%) have this severe form with serious complication of spontaneous bleeding into joints and muscles. In contrast, moderate hemophilia with rare bleeding events will result when the corresponding clotting factor is about 5% of the normal level.

RISK FACTORS

Mutations in the genes encoding for blood clotting factors VIII and IX result in Hemophilia A and B, respectively. Both genes are located on the X chromosome which presents as two copies in females (XX) but one copy only in males (XY). Therefore, if a female has a mutation in one copy, she usually does not develop the disease, but becomes a silent carrier of the trait since the other copy of the gene compensates for the defective one. Having two defective copies of a gene is the only cause of hemophilia in females, though it occurs rarely. On the other hand, males are more affected than females because they have only one copy of the X chromosome. As male patients do not pass on their X chromosome to their sons, the sons do not inherit the disease, but all the daughters of male patients would be carriers.

DIAGNOSIS AND MANAGEMENT

Patients with severe hemophilia are usually diagnosed after few months of birth due to serious bleeding problems. On the contrary, patients with the milder form of the disease may be diagnosed only when they accidentally have a severe injury.

Tests are widely available for hemophilia. Since both Hemophilia types (A and B) cause the same complications but are treated differently, distinguishing the type of hemophilia via special blood test is extremely important in deciding a proper treatment regimen. This is because Hemophilia A patients are treated with a supply of factor VIII, whereas type B hemophiliacs necessitate factor IX compensation.

Previously, treatment method for hemophilia involved pooling of plasma products from a very large number of donors, thus carrying the potential risk

of transmitting infectious diseases, such as hepatitis and AIDS, through blood transfusion. Factors deficient in Hemophilia A and B are now being synthesized using advanced biotechnology which has resulted in removing these risks. Difficulties sometimes arise in some patients when the transfused clotting factor is recognized as a foreign substance and produce an immunological reaction. Furthermore, managing irreversible joint damage and life-threatening hemorrhage may present a serious challenge for both healthcare providers and the patients.

Daily exercises are recommended for affected individuals to strengthen their muscles and joints, particularly the elbows, knees, ankles, to prevent bleeding problems. Many recommended exercises include standard sports warm-up and training exercises such as stretching of the calves, ankle circles, elbow flexions, and quadriceps sets.

HEMOPHILIA IN THE ARAB WORLD

Although hemophilia is not as prevalent as other genetic blood disorders (G6PD deficiency, thalassemia, sickle cell disease) in Arab countries, it is the most prevalent form of inherited bleeding syndromes (IBS) in the region. Hemophilia A and B account for 30% and 28% respectively of all rare bleeding disorders worldwide. In agreement with world data, Hemophilia A accounts for the majority of Arab patients with rare bleeding disorders (Saudi Arabia 73.3%, Egypt 70.6%, Jordan 17.1%), whereas Hemophilia B occurs less frequently (Saudi Arabia 26.7%, Egypt 13.9%, Jordan 4.2%). Although Hemophilia A cases have been reported in other Arab countries such as Algeria, Bahrain, Kuwait and Iraq, regional prevalence data is unavailable.