

THROMBOPHILIA

Blood contains cellular components such as red blood cells (RBCs), white blood cells (WBCs), platelets (thrombocytes) and several proteins, suspended in a yellowish liquid called plasma. Some of these proteins play an important role in maintaining the semi-fluid consistency (viscosity) of the blood that aids proper circulation, preventing blockages and stroke. These proteins along with platelets also support the clotting process (blood coagulation) that is required to control bleeding in case of injuries. This balanced clotting mechanism is vital to regulate normal blood function. Thrombophilia is a term covering a range of conditions that result in having 'sticky blood' due to increased blood clot formation (thrombosis) which can occur in different parts of the body. Based on where it develops, it can be classified as: cardiovascular thrombosis - affecting the heart and blood vessels around it; cerebrovascular thrombosis - clot formation in the brain; venous thrombosis - affecting a vein, and further classified into deep venous thrombosis (DVT, usually in the legs) and pulmonary embolism (obstruction of blood vessels in the lung).

RISK FACTORS

Thrombophilia can be either congenital (inborn) or acquired as a result of certain conditions.

Congenital thrombophilia is mainly caused by mutations (changes) in the genes responsible for the production of components involved in the clotting pathway. These mutations may cause alterations in the amount, structure, and/or function of the clotting factors leading to excessive thrombosis. Inheritance of congenital thrombophilia is considered complex

and follows different patterns, involving two or more genes. A positive family history is an important indication for an individual to be at high risk, increasing the chances of developing the condition two to twenty times compared to the general population.

Thrombotic events are found to affect individuals of all ages, but the chance of developing thrombophilia increases with age. Obesity is also a known risk factor. Other possible risk factors include surgery, trauma, use of oral contraceptives, hormone replacement therapy, smoking and prolonged immobilization. Certain autoimmune diseases, kidney disorders, cancers, etc. are also considered to increase the risk of developing thrombosis.

DIAGNOSIS AND MANAGEMENT

Diagnosis of inherited thrombophilia can be established by blood tests that measure the amount and function of the clotting factors. Inherited thrombophilia is usually suspected when at least two episodes of thrombosis occur separately or if a positive family history exists. Some of the important genetic tests for thrombophilia may include analyzing genes that are responsible for the proteins Factor V Leiden, Prothrombin, Protein C or Protein S.

Treating thrombophilia is based on using blood-thinning drugs (anticoagulants), such as heparin or warfarin. The duration of treatment is based on the nature of the thrombotic event. However, side effects of these drugs can include excessive bleeding and/or other complications, so treatment should be carefully discussed with a physician. In addition, regular

exercise and weight management may reduce the risk of developing thrombosis.

THROMBOPHILIA IN THE ARAB WORLD

Thrombophilia cases in the region are mostly 'acquired'. A multi-centre study conducted with patients from Bahrain, Kuwait, Oman and the UAE shows that 85% of thrombosis cases developed post-surgery.

Although most cases of thrombophilia are acquired, genetic studies have been widely undertaken in

several Arab countries to identify mutations that cause the congenital form. For instance, in Jordan, the prevalence rate of congenital thrombophilia accounts to 1: 25,000. This study also identified several different types of genetic defects associated with hereditary thrombophilia.

Protein C deficiency has been found to be one of the common defects, as reported in Jordan, Saudi Arabia, Kuwait, Oman and Palestine. Another contributing genetic factor in the region is Factor V Leiden, with reported cases in Bahrain, Iraq, Lebanon, Tunisia and Saudi Arabia.

