Thrombophilia

Blood contains many proteins regulating its substance. Some help keep it fluid to avoid blockage or strokes, while others help it to clot or thicken to stop bleeding in cases of wounds and cuts. Platelets are cellular fragments that play an important role in clot formation. The balance between clot formation and clot breakdown in blood vessels is normally very efficient in preventing the development of a blood clot (thrombus) in the circulation. If this balance is impaired, as in the case of excess clotting (coagulation), the signs and symptoms of thrombophilia will result.

Thrombophilia is a term covering a range of conditions that result in having 'sticky blood' due to increased thrombus formation or thrombosis. Thrombosis can occur in different places and is classified accordingly:

- Cardiovascular thrombosis affects the heart and the blood vessels around it and includes angina, myocardial infarction, and peripheral vascular disease.
- Cerebrovascular thrombosis is a clot formation in the brain and includes the transient ischemic attack and stroke.
- Venous thrombosis occurs in a vein and include deep venous thrombosis (DVT) and pulmonary embolism (obstruction of blood vessels in the lung).

Cardiovascular and cerebrovascular thromboses are highly dependent on platelet collection, while venous thromboembolic thrombosis is related to abnormalities in proteins of the clotting system.

Risk Factors

Thrombophilia can be either inherited or acquired during life. The risk factors of developing thrombophilia include:

Genetic: Congenital thrombophilia is mainly caused by mutations in the genes responsible for the production of clotting factors. These mutations may cause alterations in the amount, structure, and/or function of the clotting factors leading to excessive thrombosis.

Age: The chance of developing thrombophilia increases with age.

Weight: Obesity increases the risk to have the disease.

Other factors: Recovering from a major surgery, having a period of immobilization (like; pregnancy), using oral contraceptives, and smoking are also possible risk factors.

Obviously, these risk factors are cumulative. For instance, an elderly individual who smokes and has bone fracture that requires immobilization would have a very high risk of developing a dangerous clot.

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, and/or MTHFR.

Treating thrombophilia is based on using blood-thinning drugs, such as aspirin, heparin or warfarin. However, side effects of these drugs can include blood loss and/or potentially dangerous blood changes, so treatment should be carefully discussed with a physi-

cian. In addition, regular exercise and avoiding obesity may reduce the risk to develop thrombosis.

Thrombophilia in Arab Populations

Although most thrombophilia cases are acquired, genetic studies have been widely undertaken in Egypt, Jordan, Kuwait, Lebanon, Morocco, Oman, Saudi Arabia, and Tunisia in order to identify the causative mutations of the congenital form of the disease. The prevalence rate of genetic thrombophilia in Jordan, for example, has been calculated at 1:25,000. In Oman, patients with inherited thrombophilia due to Protein C, a coagulation inhibitor, deficiency have their first thrombotic event at an average of 38 years (34 years in females and 43 years in males).