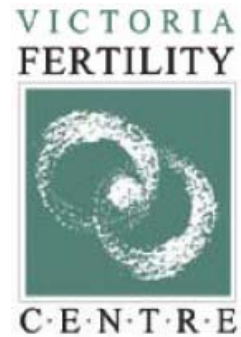


BLOOD CLOTTING ABNORMALITIES



Blood clotting factors are biochemical substances in our bloodstream that help the blood to clot when bleeding occurs. Blood clotting, known as coagulation, is a very complicated process, requiring a balance between factors that promote clot formation and factors that either prevent clot formation or dissolve it when it occurs. These are referred to as factors which are procoagulants and anticoagulants. When an imbalance between the two mechanisms occurs, there will be either an increased risk of clotting inside the blood vessels (thrombosis) or alternatively, excessive bleeding.

WHAT CONDITIONS ARE ASSOCIATED WITH AN INCREASED RISK OF THROMBOSIS?

There are many different causes for thrombosis – which may be both genetic and non-genetic. Examples of non-genetic causes which increase the tendency to coagulation include decreased physical activity (such as long distance air travel), major surgery, oral contraceptive use, hormone replacement therapy, pregnancy and obesity.

WHAT ARE THE GENETIC CAUSES FOR THROMBOSIS?

The most common genetic cause for an increased risk of thrombosis is an abnormality in the gene for one of the blood clotting factors known as Factor V. This abnormality is found in 3-5% of the general population, and in 20-50% of people who have had either a personal or family history of thrombosis. When an individual has one copy (heterozygous) of the mutated Factor V gene, he/she will have a five to tenfold increased risk of having at least one episode of thrombosis in his/her lifetime. If an individual has two copies of the mutated gene (homozygous), this risk is increased to eighty to one hundredfold.

The second most common genetic cause is a mutation in the blood clotting Factor II – also known as prothrombin. When an individual has one copy of the mutated Factor II gene (heterozygous), he/she will have a three-fivefold increased risk of having at least one episode of thrombosis. It is rare to find individuals with two copies of the mutated gene – but it is presumed that they will have a significantly increased risk of thrombosis.

There are also other genetic causes for thrombosis – specifically, for deficiencies in the naturally occurring antithrombotic factor such as Protein S, Protein C and Antithrombin III. Elevated levels of homocysteine may be also associated with some increased risk of thrombosis.

Abnormalities in these coagulation factors may be associated with the following clinical conditions:

1. Pregnancy complications such as preeclampsia, stillbirth, poor fetal growth (fetal growth restriction) and recurrent spontaneous abortions.
2. There is also evidence that these clotting factor abnormalities may be associated with recurrent in vitro fertilization failures. In other words, it is presumed that they may be associated with a lower incidence of embryo implantation.

HOW CAN THESE ABNORMALITIES BE DETECTED?

Laboratory tests are available to test for these abnormalities.

WHO SHOULD CONSIDER TESTING?

1. Individuals with a past history of thrombosis.
2. Individuals with a family history of thrombotic abnormalities.
3. Women with adverse pregnancy outcomes.
4. Possibly also women who have had repeated IVF failures.

HOW IS THE CONDITION MANAGED?

If an abnormality indicates that you have an increased tendency for thrombosis, your physician may consider prophylactic treatment. This treatment may include the use of a blood thinner such as heparin. In some cases, a low dose combination of low dose aspirin and heparin will be used – however, the treatment will be individualized and explained to you by your physician.