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Factor V Leiden

Factor V Leiden thrombophilia is an inherited disorder of blood clotting. Factor V Leiden is the name of a specific mutation (genetic alteration) that results in an increased tendency to form abnormal blood clots in blood vessels. People who have the factor V Leiden mutation are at somewhat higher than average risk for a type of clot that forms in large veins in the legs (deep venous thrombosis, or DVT) or a clot that travels through the bloodstream and lodges in the lungs (pulmonary embolism, or PE). Factor V Leiden is the most common inherited form of clotting (thrombophilia). Between 3 and 8 percent of the Caucasian (white) U.S. and European populations carry one copy of the factor V Leiden mutation, and about 1 in 5,000 people have two copies of the mutation. A mutation in the factor V gene increases the risk of developing blood clots. The Factor V protein is involved in a series of chemical reactions that hold blood clots together. A molecule called activated protein C (APC) prevents blood clots from growing too large by inactivating factor V. In people with the factor V Leiden mutation, APC is unable to inactivate factor V normally. As a result, the clotting process continues longer than usual, increasing the chance of developing abnormal blood clots.

The use of hormones, such as oral contraceptive pills (OCPs) and hormone replacement therapy (HRT), including estrogen and estrogen-like drugs) taken after menopause, increases the risk of developing DVT and PE. Healthy women taking OCPs have a three- to four-fold increased risk of developing a DVT or PE compared with women who do not take OCP. **Women with factor V Leiden who take OCPs have about a 35-fold increased risk of developing a DVT or PE** compared with women without factor V Leiden and those who do not take OCPs. Likewise, postmenopausal women taking HRT have a two- to three-fold higher risk of developing a DVT or PE than women who do not take HRT, and **women with factor V Leiden who take HRT have a 15-fold higher risk**. Women with heterozygous factor V Leiden who are making decisions about OCP or HRT use should take these statistics into consideration when weighing the risks and benefits of treatment.

The factor V Leiden mutation is associated with a slightly increased risk of pregnancy loss (miscarriage). Women with this mutation are two to three times more likely to have multiple (recurrent) miscarriages or a pregnancy loss. Some research suggests that the factor V Leiden mutation may also increase the risk of other complications during pregnancy, including pregnancy-induced high blood pressure (preeclampsia), slow fetal growth, and early separation of the placenta from the uterine wall (placental abruption). However, the association between the factor V Leiden mutation and these complications has not been confirmed. Most women with factor V Leiden thrombophilia have normal pregnancies.

How is factor V Leiden thrombophilia treated?

The management of individuals with factor V Leiden depends on the clinical circumstances. People with factor V Leiden who have had a DVT or PE are usually treated with blood thinners, or anticoagulants. Anticoagulants such as heparin are given for varying amounts of time depending on the person's situation. It is not usually recommended that people with factor V Leiden be treated lifelong with anticoagulants if they have had only one DVT or PE, unless there are additional risk factors present. Having had a DVT or PE in the past increases a person’s risk for developing another one in the future, but having factor V Leiden does not seem to add to the risk of having a second clot. In general, individuals who have factor V Leiden but have never had a blood clot are not routinely treated with an anticoagulant. Rather, these individuals are counseled about reducing or eliminating other factors that may add to one’s risk of developing a clot in the future. In addition, these individuals may require temporary treatment with an anticoagulant during periods of particularly high risk, such as major surgery.

Factor V Leiden increases the risk of developing a DVT during pregnancy by about seven-fold. Women with factor V Leiden who are planning pregnancy should discuss this with their obstetrician and/or hematologist. Most women with factor V Leiden have normal pregnancies and only require close follow-up during pregnancy. For those with a history of DVT or PE, treatment with an anticoagulant during a subsequent pregnancy can prevent recurrent problems.Factor V Leiden is the most common inherited form of thrombophilia. The risk of developing a clot in a blood vessel depends on whether a person inherits one or two copies of the factor V Leiden mutation. Inheriting one copy of the mutation from a parent increases by fourfold to eightfold the chance of developing a clot. People who inherit two copies of the mutation, one from each parent, may have up to 80 times the usual risk of developing this type of blood clot. Considering that the risk of developing an abnormal blood clot averages about 1 in 1,000 per year in the general population, the presence of one copy of the factor V Leiden mutation increases that risk to 1 in 125 to 1 in 250. Having two copies of the mutation may raise the risk as high as 1 in 12.