



Genetics Uncoded:



Facts about

Factor V Leiden Thrombophilia



What Your Test Results Mean

Factor V Leiden thrombophilia is an adult-onset condition that can modestly increase risk for blood clots. These results are most relevant to pregnancy management.

● Factor V Leiden Thrombophilia Explained

Factor V Leiden thrombophilia is the most common inherited defect of coagulation, present in about 5% of Caucasian Americans and 40% of pregnant women presenting with venous thrombosis. The severity of thrombophilia is dependent on the number of copies of the variant as well as other factors. Individuals with one copy of R506Q and no personal history have less than a 1% chance for venous thromboembolism during pregnancy; with a personal history, risk increases to 10%. Individuals with two copies of G1691A and no personal or family history have a 1-2% risk for venous thromboembolism during pregnancy. In the presence of family or personal history, risk approaches 17%.

Coexisting thrombophilias and other risk factors including travel, use of oral contraceptives, use of hormone replacement therapy, use of selective estrogen receptor modulators, pregnancy status, age, and surgery also play a role in risk of venous thrombosis. Alternative methods to oral contraceptives should be considered in women with factor V Leiden thrombophilia due to the increased risk of thromboembolism.

● How the Genetics Work

The clinical features of Factor V Leiden can be explained by a variant, c.1691 G>A (R506Q), in the *F5* gene. All individuals have two copies of the *F5* gene. Individuals with factor V Leiden have either one or two copies of *F5* variant. The variant causes resistance to activated protein C (APC), a protein that normally decreases the rate of thrombin generation in plasma. APC resistance causes the increased risk for thrombosis or blood clots.

Questions?

Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.