



INTERPRETATION GUIDELINES

Factor V Leiden and Factor II Prothrombin by Real-Time PCR

Methodology: This Factor V Leiden and Factor II Prothrombin Test screens for both G1691A and G20210A mutations in factor V and factor II genes correspondingly. This assay is performed in a Real-Time PCR format utilizing several PCR reactions.

Positive Result Interpretation Report

Result	Comment	Interpretation
Positive	G1691A heterozygote	One copy of the G1691A mutation was identified in the Factor V Leiden gene indicating that this individual is heterozygous on G1691A mutation. Individuals heterozygous for the G1691A mutation have increased risk of venous thrombosis. Appropriate genetic counseling is recommended.
Positive	G1691A homozygote	Two copies of the G1691A mutation were identified in the Factor V Leiden gene indicating that this individual is homozygous on G1691A mutation. Individuals homozygous for the G1691A mutation have highest risk of venous thrombosis. Appropriate genetic counseling is recommended.
Positive	G20210A heterozygote	One copy of the G20210A mutation was identified in the Factor II Prothrombin gene indicating that this individual is heterozygous on G20210A mutation. Individuals heterozygous for the G20210A mutation have increased risk of venous thrombosis. Appropriate genetic counseling is recommended.
Positive	G20210A homozygote	Two copies of the G20210A mutation were identified in the Factor II Prothrombin gene indicating that this individual is homozygous on G20210A mutation. Individuals homozygous for the G20210A mutation have highest risk of venous thrombosis. Appropriate genetic counseling is recommended.
Positive	G1691A and G20210A compound heterozygote	One copy of the G1691A mutation was identified in the Factor V Leiden gene and one copy of the G20210A mutation was identified in the Factor II Prothrombin gene indicating that this individual is compound heterozygous on G1691A mutation. Individuals heterozygous for the G1691A mutation have highest risk of venous thrombosis. Appropriate genetic counseling is recommended.

Clinical Significance: Factor V Leiden is a missense G1691A mutation in the clotting factor V gene. This results in a protein product that is resistant to cleavage by activated protein C (APC), a key step in the anticoagulation system. The resulting phenotype of APC resistance is characterized primarily by increased risk of venous thrombosis, but has also been associated with recurrent pregnancy loss and complications, as well as myocardial infarction in young adult female smokers. It is the most common inherited thrombophilic condition, found in approximately 5% of Caucasians. The prothrombin G20210A mutation in the factor II gene was described in patients with a family history of thrombosis. The prothrombin G20210A mutation is present in approximately 0.7% to 4.0% of the Caucasian population and in approximately 6% of patients with a first venous thromboembolism. Both the factor V Leiden G1691A mutation and the prothrombin G20210A mutation exhibit semidominant expression in that both heterozygotes and homozygotes are at increased risk of occurrence/recurrence of venous thrombosis.

This test was developed and its performance characteristics determined by this laboratory. It has not been cleared or approved by the Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. It should not be regarded as investigational or for research. The laboratory is regulated by the Clinical Laboratory Improvement Act of 1988.