**Factor V Leiden**

The diagnosis of the etiologic causes of thrombophilia can be challenging. Thrombotic disease is typically characterized by the presence of any number of acquired and genetic risk factors. The Factor V Leiden allele represents a majority of identifiable genetic causes of thrombotic disease and should be considered in the diagnosis and thrombotic risk assessment in patients with a personal or familial history of recurrent thrombosis. The Factor V Leiden is typically present in 5-10% of the population. In one study of local interest, 7.9% of the South Central Pennsylvania population was identified as having the Factor V Leiden allele.

The Factor V Leiden assay allows the detection and genotyping of a single point mutation (G to A at position 1691) of the human Factor V gene (Factor V Leiden mutation) from DNA isolated from human whole peripheral blood. The test is performed on the LightCycler instrument utilizing real-time polymerase chain reaction (PCR) for the amplification of Factor V DNA recovered from clinical specimens and fluorogenic target-specific hybridization for the detection and genotyping of the amplified Factor V DNA. The 1691 point mutation of the Factor V gene causes an arginine to glutamine substitution at position 506 in the Factor V protein and renders it partially resistant to inactivation by Activated protein C (APC). APC resistance is regarded as the most prevalent coagulation abnormality associated with venous thrombosis.

**How are the results reported?**

*The results for Factor V Leiden testing are reported as one of three possible results:*  

**NORMAL:** A normal result means that the Factor V Leiden allele (or mutation) was not detected on either version/copy of the Factor V gene.  

**HETEROZY:** This result (Heterozygous) indicates that one copy of the Factor V allele has been identified with the Leiden mutation.  

**HOMOZYGO:** This result (Homozygous) indicates that both copies of the Factor V allele have been identified with the Leiden mutation.

**Important Additional Notes:**

- This assay is a germline genetic assay and should only be ordered once on any particular patient. The Factor V Leiden genotype is constitutional and does not change.  
- The Factor V Leiden assay is typically ordered in conjunction with the Prothrombin 20210A mutational assay and other markers of hypercoagulability, in patients with thrombosis.
References:


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