CPAL
Central **P**ennsylvania **A**lliance **L**aboratory

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Factor V Leiden (Revised)

Starting Date:
Currently Available

Mnemonic: FACT V MUT

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Reference Value: NORMAL

Summary:
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.

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For questions about this and other information, call Central Pennsylvania Alliance Laboratory at 1-888-480-1422.

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.

How often is the Factor V Leiden assay performed?
The Factor V Leiden assay is typically performed and reported at CPAL on Tuesdays and Thursdays.

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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4. Press, RD, et al. (2003) Clinical Utility of Factor V Leiden (R506Q) Testing for the Diagnosis and Management of Thromboembolic Disorders. CAP Consensus Conference XXXVI: *Diagnostic Issues in Thrombophilia*.
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6. Wisotzkey, J.D., Bell, T. and Monk, J.S. *Simultaneous Polymerase Chain Reaction Restriction Fragment Length Polymorphism (PCR-RFLP) Identification of the Factor V Leiden Allele and the Prothrombin 20210A Mutation.* **Diagnostic Molecular Pathology**, 7:180-183, 1998.
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