



**CPAL**

Central Pennsylvania Alliance Laboratory

# Technical Bulletin

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## Factor V Leiden

**Starting Date:**

Currently Available

**Mnemonic: FACT V MUT**

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**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 APC. Activated protein C (APC) resistance is regarded as the most prevalent coagulation abnormality associated with venous thrombosis.

**Reference Ranges:**

The “normal” result for this assay is reported as WILDTYPE. This indicates that the mutation, better termed “polymorphism”, is not present on either of the two copies of the factor V gene (allele).

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

### **Important Additional Notes:**

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- 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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