



CPAL

Central Pennsylvania Alliance Laboratory

Technical Bulletin

No. 57A

July 5, 2006

Factor V Leiden (Revised)

Starting Date:

Currently Available

Mnemonic: FACT V MUT

Contact:

Dr. Jeffrey Wisotzkey, 717-851-1422
Technical Director, CPAL

Reference Value: WILDTYPE

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.

Originally Issued on: May 16, 2006

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:

WILDTYPE: This is a genetic term that means the most common or typical genotype. CPAL uses this term for the reporting of “normal” Factor V Leiden results since this is the standard practice in the field. A **WILDTYPE** 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 has 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:

1. Burick, A., Wisotzkey, J.D., Najarian, M.P., Monk, J.S., Rhoads, J.E., *The Role of Preoperative Factor V Leiden Screening in Different Geographic Populations. The American Surgeon.* 63:547-550, 1997.
2. Ridker, PM, et al. (1997) Ethnic Distribution of Factor V Leiden in 4047 Men and Women: Implications for Venous Thromboembolism screening. *JAMA*, 277, 1305-1307.
3. Grody, WW, et al. (2001) American College of Medical Genetics Consensus Statement of Factor V Leiden Mutation Testing. *Genetics in Medicine*, 3, Vol.2, 139-148.
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.*
5. Wisotzkey, J.D., P. Bayliss, E. Rutherford and T. Bell. *Placental Genotyping of the Factor V Leiden, Prothrombin 20210A and the Methylene tetrahydrofolate Reductase (MTHFR) C677T Alleles in IUGR Pregnancies (Letter). Thrombosis and Haemostasis*, 81:844-845, 1999
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.

Originally Issued on: May 16, 2006

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