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Factor V Leiden Mutation Analysis

Factor V Leiden (*F5*) point mutation G1691A (Formerly FVL)

Indications for Molecular Testing

- Family history of venous thrombosis
- Unprovoked thrombotic event at <45 years of age
- Women with multiple stillbirths or spontaneous abortions
- Confirmation of diagnosis of *F5* by non-molecular means

Testing Methodology

The assay utilizes the Invader Plus® chemistry manufactured by Hologic for the detection of the FVL(F5) c.1601G>A, R506Q mutation in genomic DNA (NM_000130.4 transcript isoform). Invader and allele-specific probes match the mutant and the wild-type alleles and have overlapping 5'-ends that are cleaved upon perfect hybridization to the amplified DNA. The cleaved 5'-end of the primary probes transiently hybridize with a corresponding fluorescence resonance energy transfer (FRET) cassettes triggering the cleavage of the fluorophore from the cassette by the cleavase enzyme and allowing signal release and detection.

Interpretation of DNA analysis

Factor V Leiden, a point mutation (G1691A) in the gene coding for coagulation Factor V, has been associated with an increased risk of venous thrombosis due to increased resistance to degradation of factor V by activated Protein C. For individuals presenting with venous thrombosis, FVL occurs in 11 – 20% of those in all age groups and 50% of individuals under 50 years of age. Heterozygosity for this mutation produces a 7 – fold increase relative risk of venous thrombosis. Approximately 5% of Caucasians are heterozygous for this mutation. The homozygous occurrence of this mutation has been associated with an 80 – fold increased risk for venous thrombosis.

Specimen Requirements

Peripheral blood--1 lavender-top (EDTA) tube. Invert several times to mix blood.
Do not freeze. Forward at ambient temperature to:

Molecular Diagnostic Laboratory
Barnes-Jewish Hospital, Institute of Health
Mail Stop 90-28-344
425 South Euclid Avenue, Room 5970
St. Louis, MO 63110

Clinical information must be provided with specimen referral in order to correctly interpret test results.

Current Pricing

Contact Lab Customer Service for current pricing 314 362-1470.
CPT code: 81241

Kujovich, JL. Gene Reviews: Factor V Leiden Thrombophilia. Pagon RA, Adam MP, Ardinger HH, et al., editors. Seattle (WA): University of Washington, Seattle; 1993-2015. ISSN: 2372-0697
Grody WW, Griffin JH, Taylor AK, Korf BR, Heit JA; ACMG Factor V. Lei. den Working Group. American College of Medical Genetics consensus statement on factor V Leiden mutation testing. Genet Med. 2001 Mar-Apr;3(2):139-48.
Rosendaal FR, Reitsma PH. Genetics of venous thrombosis. J Thromb Haemost. 2009;7 Suppl 1:301–4.
Ridker PM, Hennekens CH, Lindpaintner K, et al. Mutation in the gene coding for coagulation factor V and the risk of myocardial infarction, stroke, and venous thrombosis in apparently healthy men. N Engl J Med 1995; 332: 912-17.