

Factor V Leiden, Molecular Methods

Test Code: FVLM (individual order) or HCOG (Hyper-coagulation Panel)

Use: Factor V Leiden test is a diagnostic test for the detection and genotyping of the Factor V Leiden mutation as an aid to diagnosis in the evaluation of patients with suspected thrombophilia.

Clinical Significance: Genetic alterations predisposing to thrombosis are common in the general population. Activated protein C (APC) resistance is an autosomal dominant condition that is responsible for approximately 50% of hereditary thrombophilia. Factor V Leiden mutation is one of the key genetic risk factors for thrombosis with the R506G mutation, resulting in a substitution of glutamine for arginine at position 506, accounting for 85-95% of cases. Heterozygous carriers of this mutation have a three-fold to eight-fold increased risk of venous thrombosis. Individuals homozygous for the mutation carry an 80 – to over 100 – fold increase of thrombosis.

Methodology: Extraction of genomic DNA from whole blood is followed by amplification using polymerase chain reaction (PCR) and detection using a melting curve analysis. During the melting curve analysis, fluorescence is continuously monitored while the temperature is increased. The increasing temperature will result in a dissociation of the amplified product at a specific temperature (melting temperature, T_M). At the T_M , the fluorescence properties decrease rapidly. In the presence of a mutation, the resulting T_M will be lower than the wild type.

Normal Range: No Mutation Detected

Reportable Range: Results for the Factor V Leiden test are reported as homozygous mutation (two copies for the mutation), heterozygous mutation (one copy of the mutant gene and one copy of the normal (wild type) gene), or no mutation detected (two copies of the normal (wild type) gene).

Assay Availability: Batched once weekly on Tuesday

Results Reported: 7 days

Specimen: Whole Blood: 1 lavender top (EDTA) tube
Specimens should be labeled with patient name, medical record number, date and time of collection, and then sent at room temperature to the laboratory. Specimens used for testing in other departments are unacceptable due to possible contamination.

Volume: 4 mL (minimum volume: 1 mL)

Storage: If not tested immediately, store whole blood refrigerated.

Causes for Rejection: Heparinized specimens; Quantity not sufficient for analysis; specimen grossly contaminated; frozen whole blood specimens; specimen leaky or tube broken.

Laboratory Contact: For further information, please call the Molecular Diagnostics Laboratory at (501) 526-6439.