Factor V (Leiden), Mutation Analysis

CPT Code: 81241
Order Code: 1089
ABN Requirement: No
Specimen: Whole Blood
Volume: 5.0 mL
Minimum Volume: 3.0 mL
Container: EDTA (lavender-top) (preferred) EDTA (royal blue-top) Sodium heparin (green-top) ACD solution A (yellow-top) ACD solution B (yellow-top) Lithium heparin (green-top)

Collection:

1. Collect and label sample according to standard protocols.
2. Gently invert tube 8-10 times immediately after draw. DO NOT SHAKE.
3. Do not centrifuge.

Please Note: This germline genetic test requires physician attestation that patient consent has been received if ordering medical facility is located in AK, DE, FL, GA, IA, MA, MN, NV, NJ, NY, OR, SD, or VT or test is performed in MA.

Special Instructions: Extracted DNA: Please call 1-(866)-GENE-INFO or 1-866-436-3463 for additional information. Frozen shipping is acceptable.

Transport: Store EDTA whole blood at 2°C to 8°C after collection and ship the same day per packaging instructions included with the provided shipping box.

Stability:

Ambient (15-25°C): 8 days
Refrigerated (2-8°C): 8 days
Frozen (-20°C): 30 days

Methodology: Polymerase Chain Reaction & Detection

Turn Around Time: 4 to 7 days

Reference Range: See Laboratory Report

Clinical Significance: This test detects the factor V Leiden variant, the most common cause of inherited thrombophilia; it may be used to evaluate individuals with a strong personal or family history of venous thromboembolism (VTE) and inform treatment or preventive decisions [1].

Factor V Leiden refers to the c.1691G>A variant in the FV gene, which encodes coagulation factor V. This variant results in resistance to factor V protein degradation by activated protein C and increases the risk of VTE 6 to 8 fold in heterozygous carriers and 80 fold in homozygous carriers [1]. The mean age of symptom onset is 31 to 44 years, but some heterozygous carriers can be asymptomatic [2]. In the United States, Factor V Leiden is most prevalent in White individuals, with an estimated frequency of 5% [1].

Factor V Leiden testing may be indicated in clinical scenarios where results can help guide clinical decisions for the patient and family members. These clinical scenarios include first VTE developed under 50 years of age, VTE at an unusual site (eg, cerebral veins), recurrent VTE, a strong family history of VTE, and low activated protein C resistance activity [1].

Routine testing for factor V Leiden is not recommended for prenatal carrier screening, newborn screening, or individuals taking an oral contraceptive [2]. A negative result of this test does not rule out inherited thrombophilia. Other than factor V Leiden, variants in the genes that encode coagulation factor II, protein C, protein S, and antithrombin can also cause inherited thrombophilia.

The results of this test should be interpreted in the context of pertinent clinical and family history and physical examination findings.

References

1. Zhang S, et al. Genet Med. 2018;20(12):1489-1498.
2. Vnencak-Jones CL, et al. Genetics. In: Rifai R, et al, eds. Tietz Textbook of Laboratory Medicine. 7th ed. Elsevier Inc; 2022.

The CPT codes provided are based on AMA guidelines and are for informational purposes only. CPT coding is the sole responsibility of the billing party. Please direct any questions regarding coding to the payer being billed.