Factor V Leiden

Description:

Factor V Leiden (FVL) is the most common hereditary blood coagulation disorder in the United States. The Factor V Leiden R506Q variant (c.1691G>A) is associated with resistance to activated protein C (APC) which predisposes individuals to venous thrombosis. Individuals who are heterozygous for FVL have a 2–8 fold risk of developing venous thrombosis. Individuals who are homozygous for FVL have a 50 fold risk of developing venous thrombosis. The frequency of FVL in individuals of Caucasian descent is about 5%, and is about 1% in individuals of African-American descent.

Multiple professional societies recommend testing for Factor V Leiden only when results would affect clinical management.

Indications:

When results would affect clinical management, consider testing with the following indications:

- Unprovoked first or recurrent venous thromboembolism, especially when manifests as deep vein thrombosis (DVT) or pulmonary embolism
- Family members of those known to have Factor V Leiden or recurrent thrombosis
- Females with venous thromboembolic event during pregnancy
- Females with venous thromboembolism in association with use of oral contraceptives or hormone replacement therapy

Specimen:

At least 2mLs of whole blood in lavender top (EDTA) tube. Label tube with patient's name, birth date, and date of collection. Phlebotomist must initial tube to verify patient's identity.

We are unable to accept blood samples collected within two (2) weeks of a transfusion.

Testing Methodology:

Allelic Discrimination TaqMan Assay (Applied Biosystems) is used to determine the genotype at the Factor V Leiden locus. End-products are analyzed using the ABI 7500 Real-Time PCR System for genotype detection.

*Analysis of Factor V Leiden variant is also offered as part of our Thrombophilia Panel which includes the Prothrombin 20210G>A variant.

Sensitivity:

This test methodology detects >99% of instances of the variant. Approximately 90–95% of APC-resistance is due to Factor V Leiden mutation.

Turn-Around Time:

7 days

CPT Codes:

- 81241

Please call 1-866-450-4198 for current pricing, insurance preauthorization or with any billing questions.

Results:

Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for the clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.

Cincinnati Children's®
changing the outcome together

Genetics and Genomics Diagnostic Laboratory
CLIA#: 36D0656333
Phone: (513) 636-4474
Fax: (513) 636-4373
Email: LabGeneticCounselors@cchmc.org
www.cincinnatichildrens.org/genetics

Shipping Instructions:

Please enclose **test requisition** with sample.
All information must be completed before sample can be processed.

Place samples in styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Saturday.

Ship to:

Genetics and Genomics Diagnostic Laboratory
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229
513-636-4474

References:

ACOG. ACOG Practice Bulletin No. 138: Inherited thrombophilias in pregnancy. Obstet Gynecol. 2013b; 122:706-17.

Dentali F, Ageno W, Bozzato S, Malato A, Gianni M, Squizzato A, Prisco D. Role of factor V Leiden or G20210A prothrombin mutation in patients with symptomatic pulmonary embolism and deep vein thrombosis: a meta-analysis of the literature. J Thromb Haemost. 2012;10:732–7.

EGAPP Working Group. Recommendations from the EGAPP Working Group: routine testing for Factor V Leiden (R506Q) and prothrombin (20210G>A) mutations in adults with a history of idiopathic venous thromboembolism and their adult family members. Genet Med. 2011;13:67–76.

Stevens SM, Woller SC, Bauer KA, Kasthuri R, Cushman M, Streiff M, Lim W, Douketis JD. Guidance for the evaluation and treatment of hereditary and acquired thrombophilia. J Thromb Thrombolysis. 2016;41:154–64.

Rosendaal FR, Reitsma PH. Genetics of venous thrombosis. J Thromb Haemost. 2009;7 Suppl 1:301–4

Chong LY, Fenu E, Stansby G, Hodgkinson S. Management of venous thromboembolic diseases and the role of thrombophilia testing: summary of NICE guidance. BMJ 2012;344:e3979.