

# Thrombophilia Panel (Factor V Leiden and Prothrombin G20210A)

## Description:

Inherited Thrombophilia: The Factor V Leiden (FVL) R506Q variant (c.1691G>A) and the Factor II (Prothrombin) G20210A variant are the two most common inherited disorders of blood clotting that predispose individuals to venous thrombosis.

The Factor V Leiden R506Q variant (c.1691G>A) is associated with resistance to activated protein C. Individuals who are heterozygous for FVL have a 2–8 fold risk of developing venous thrombosis. When coupled with oral contraceptive use or estrogen therapy, heterozygotes have an estimated 7-25-fold risk of venous thrombosis. Individuals who are homozygous for FVL have a 50-fold risk of developing venous thrombosis. Up to 20% of individuals with venous thrombosis have FVL. The frequency of heterozygous FVL in individuals of Caucasian descent is about 5%, and the frequency is around 1% in individuals of African-American descent.

The Prothrombin G20210A variant is the second-most common cause of inherited thrombosis and results in elevated levels of prothrombin, which mildly increases the risk of venous thrombosis. An increased risk of venous thrombosis at certain unusual sites such as the cerebral, mesenteric, portal, or hepatic veins may also be associated with this variant. The G20210A variant results in approximately 30% higher levels of prothrombin, which can cause a mild hypercoagulable condition associated with deep vein thrombosis. Heterozygosity for the G20210A mutation results in a 2 to 5-fold higher risk of thrombosis. Combined heterozygosity for Factor V Leiden and Prothrombin G20210A results in up to a 20-fold increased risk of thrombosis. The population frequency of Prothrombin G20210A variant is between 1% and 4% in Caucasians and about 0.2% in African-Americans.

Multiple professional societies recommend testing for Factor V Leiden and Prothrombin 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
- Family members of those known to have Factor V Leiden or Prothrombin G20210A
- Females with venous thromboembolic event during pregnancy or 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.

## Testing Methodology:

Allelic Discrimination TaqMan Assay (Applied Biosystems) is used to determine the genotype at each of the above loci. End-products are analyzed using the ABI 7500 Real-Time PCR System for genotype detection.

\*Analysis of the Factor V Leiden variant and the Prothrombin G20210A variant are also offered as single tests.

## Sensitivity:

This test methodology detects >99% of instances of these variants.

## Turn-Around Time:

7 days

## CPT Codes:

- 81240, 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.

## 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 Friday.

## Ship to:

Cytogenetics and Molecular Genetics Laboratories  
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.

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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.