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 thrombo-embolism
- 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 2 mLs whole blood in a lavender top (EDTA) tube, 6 cytobrushes or saliva in an Oragene saliva kit. Please call 513-636-4474 for a free saliva collection kit.

Note: Label tube with patient's name, birth date, and date of collection.

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

Testing Methodology:

Genomic DNA is isolated, quantified and amplified by polymerase chain reaction using specific oligonucleotide primers in a multiplex assay. Factor V and Factor II genotypes are determined using iPLEX® chemistry combined with the MassARRAY® System from Agena Bioscience to detect the presence of the Factor V Leiden 1691G/A (rs6025) and Prothrombin 20210G/A (rs1799963) polymorphisms.

*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.

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

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 Saturday.

Ship to:

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

References:

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