Thrombotic Risk Profile (FVL+PT, DNA)

The American College of Medical Genetics (ACMG) recommended that Factor V Leiden and Factor II DNA testing should be performed in patients with any type of venous thrombosis (hepatic, mesenteric, cerebral and recurrent), with a strong family history of thrombotic disease. Also indicated for pregnant women with venous thrombosis, a history of pregnancy difficulties such as miscarriages, placental abruption, intrauterine fetal growth retardation or stillbirth, or women taking oral contraceptives and female smokers (under age 50) with myocardial infarction.

Testing Method and Background

This test utilizes the Xpert® Factor II & Factor V Assay (Cepheid) an in vitro diagnostic for the genotyping of Factor II (Prothrombin) G20210A and Factor V (Factor V Leiden) G1691A using genomic DNA. The Xpert® technology uses a real-time Polymerase Chain Reaction (PCR) method for determining the genotyping status.

Factor V c.1601G>A (p.Arg534Gln) is commonly referred to as Factor V Leiden (FVL). FVL (G1691A) refers to the G to A transition at nucleotide position 1691 of the Factor V gene, resulting in the substitution of arginine by glutamine in the Factor V protein, causing resistance to cleavage by Activated Protein C (APC). The factor II (G20210A) mutation refers to the G to A transition at nucleotide 20210 in the 3' untranslated region of the gene and is associated with increased plasma levels of prothrombin. Carriers of the 20210A allele have higher prothrombin levels than persons with the wild-type allele (20210G) and have an increased risk of venous thrombosis.

Limitations of this test include rare Factor V and Factor II mutations or SNPs in probe-binding region that may interfere with target detection/yield. The performance of this test has not been evaluated with samples from pediatric patients.

Inherited Thrombosis is associated with congenital predisposing risk factors such as Factor II (Prothrombin, FII) and Factor V (Leiden, FV) proteins involved in the blood coagulation enzyme activity cascade. The FII and FV mutations are present in ~2% and 5% of individuals with N. European ancestry respectively, but at much lower levels in other populations.

Highlights of Factor V and Factor II Analysis

FV: Genotyping of the 1691G>A mutation (Normal, Heterozygous, Homozygous)
FII: Genotyping of the 20210 G -->A mutation (Normal, Heterozygous, Homozygous)

Ordering Information

Get started (non-HFHS): Print a Genetic Hereditary Disorder requisition form online at www.HenryFord.com/HFCPD

Get started (HFHS): Order through Epic using test "Thrombotic Risk Profile (FVL+PT, DNA)" (DNA2100)

Specimen requirements:

- Peripheral Blood - 1-3ml in lavender top tube (EDTA) **Specimen stability: Ambient - 72 hours; Refrigerated - 2 weeks**

Cause for Rejection: Clotted, hemolyzed, or frozen specimens, improper anticoagulant, tubes not labeled with dual patient identification, non-dedicated tubes.

TAT: 5-7 business days (after Prior Authorization obtained)

Mail test material to:
Henry Ford Center for Precision Diagnostics
Pathology and Laboratory Medicine
Clinic Building, K6, Core Lab, E-655
2799 W. Grand Blvd., Detroit, MI 48202

CPT Codes: 81240, 81241, G0452

Contact us: Client Services, Account and Billing Set-up, and connect with a Molecular Pathologist at (313) 916-4DNA (4362)

For more information on Comprehensive Molecular Services, visit our website
www.HenryFord.com/HFCP

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