

ANNOUNCEMENT

Date: May 18, 2012

Dear Valued Clients:

Foundation Laboratory is pleased to announce that effective May 17, 2012 Factor V Leiden R506Q Mutation assay will be performed in-house. This FDA approved assay is performed using multiplex PCR with solid phase electrochemical methodology on GenMark Dx eSensor platform.

Factor V Leiden variant is one of the elements responsible for hypercoagulability disorder. In normal individuals, factor V functions as a cofactor to allow factor Xa to activate thrombin. Thrombin in turn cleaves fibrinogen to form fibrin, which polymerizes to form dense meshwork that makes up the majority of clot. Activated protein C (APC) is a natural anticoagulant that acts to limit the extent of clotting by cleaving and degrading factor V. Factor V Leiden is an autosomal dominant condition that exhibits incomplete dominance and results in a factor V variant that cannot be easily degraded by activated protein C (APC). Factor V Leiden results from a G>A substitution at nucleotide position 1,691 (R506Q) in the *F5* gene.

Factor V Leiden is the most common inherited cause of Thrombophilia. The Leiden allele of the factor V gene contains a G>A substitution.

Heterozygotes have a slightly increased risk for venous thrombosis (3-7 times) but Homozygote's have a much greater thrombotic risk (80 times). Most prevalent in the U.S. and European Caucasian populations are reported heterozygote's of approximately 5-7% and homozygosity of 0.02% in one study. Found in 5.27% of Caucasian Americans and progressively less common on Hispanic-Americans (2.21% heterozygotes), African-Americans (1.23% heterozygotes), and Asian-Americans (0.45% heterozygotes).

Factor V Leiden testing is indicated for the following patients:

Age <50, any type of venous thrombosis, venous thrombosis in unusual sites (hepatic, mesenteric, cerebral veins), recurrent venous thrombosis, venous thrombosis and strong family history of thrombotic disease, venous thrombosis in pregnant women taking oral contraceptives, relatives of individuals with venous thrombosis under age 50, and myocardial infarction in female smokers under age 50. Also, Factor V testing may be considered in the following patients:

Venous thrombosis, age >50, relatives of individuals known to have factor V Leiden, women with recurrent pregnancy loss of unexplained severe preeclampsia, placental abruption, intrauterine fetal growth retardation, or stillbirth.

Additional recommendations also include molecular genetic testing for the other most common risk factors such as Factor II and MTHFR for patient testing positive for Factor V.



Specimen Requirements:

- 5 ml whole blood collected in EDTA (lavender top) tube, refrigerate.

Turn Around Time:

- 10 days

For supplies and other needs please contact your Foundation Laboratory representative.

Sincerely,

Reza M. Massoumi, Ph.D.
Director of Technical Operations