THROMBOPHILIA (ASSOCIATED WITH FACTOR V DEFICIENCY)

London Health Sciences Centre | St. Joseph's Health Care London

Pathology and Laboratory Medicine

Orderable - FVL

Turnaround Time: 4 weeks

Laboratory:
Molecular Diagnostics Lab

Requisition:
MOLECULAR DIAGNOSTICS REQUISITION

Method of Analysis:
Analysis of FV Leiden mutation at the 1601G locus utilizes an Agena iPLEX custom panel followed by SpectroCHIP® Array detection using the MassARRAY System (Agena)

Test Schedule:
As required,
Monday to Friday 0800-1600 hours

Alternate Name(s):

Factor V
Familial Thrombophilia
Prothrombin/Factor V Prothrombin

Specimen:

1 x 4 mL K_2 or K_3 EDTA Lavender top Vacutainer tube-may be a frozen aliquot

Collection Information:

Blood samples may be maintained at room temperature or a frozen aliquot is also acceptable.

Reference Ranges:

See report

Interpretive Comments:

Resistance to activated protein C (APC) is a major cause of familial thrombophilia, and can be corrected by an anticoagulant activity expressed by purified factor V. It has been suggested that a point mutation in the gene coding for factor V is responsible for APC resistance (PMID:7909098, 8208267). This point mutation, (F5:c.1601G>A), occurring towards the 3' end of exon 10 of the factor V gene, is predicted to cause a missense mutation in the APC cleavage site, (F5:p.Arg534Gln) and confers an increase in the relative risk of a first episode of venous thrombosis of x7 for heterozygote carriers, x35 for heterozygote carriers taking oral contraceptive medication, and x80 for individuals homozygous for the Factor V Leiden mutation(PMID:11529700).

Storage and Shipment:

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Must be received in testing laboratory within 5 days of collection, shipped at room temperature by courier/overnight delivery.