THROMBOPHILIA (PROTHROMBIN)

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

Pathology and Laboratory Medicine

Orderable - PV

Turnaround Time: 4 weeks

Laboratory:
Molecular Diagnostics Lab

Requisition:
MOLECULAR DIAGNOSTICS REQUISITION

Method of Analysis:
A simple method of analysis of mutations at the "G20210" locus (numbered F2:c.*97G>A RefSeq NM_000506.3) in the prothrombin gene, utilizes 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):

Prothrombin Gene

Specimen:

Whole blood-1 x 4 mL Lavender EDTA top Vacutainer tube

Collection Information:

Blood samples must be maintained at room temperature **or** if prior arrangements have been made with the lab, other tissue samples may be accepted for DNA extraction (i.e. CVS, amniotic fluid, skin etc.)

Reference Ranges:

See report

Interpretive Comments:

At least five genetic defects, (PMID:9299960) accounting for approximately 15% of families with inherited thrombophilia, have been established as risk factors for venous thrombosis. There are protein C, protein S, and antithrombin deficiencies, and represent defects in the anticoagulant pathways of blood coagulation. Two other genetic risk factors, resistance to activated protein C associated with the factor V Leiden mutation and increased prothrombin associated with the prothrombin "20210" A allele, are much more prevalent and together can be found in 63% of the thrombophilia families. Heterozygous carrier status of the prothrombin "20210" A allele confers an 2.8-fold increase in the relative risk of a first episode of venous thrombosis (PMID:11529700).

Storage and Shipment:

THROMBOPHILIA (PROTHROMBIN)

London Health Sciences Centre
St. JOSEPH'S HEALTH CARE LONDON
Pathology and Laboratory Medicine

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