University of Nebraska Medical Center

Lab Alert

REGIONAL PATHOLOGY SERVICES

DEPARTMENT OF PATHOLOGY AND MICROBIOLOGY

THROMBOPHILA MUTATION DETECTION (FACTOR II & V AND MTHFR)

Test Overview: The Molecular Diagnostics Laboratory will change the method of mutation testing for Factor II- prothrombin (G20210A) and Factor V- Leiden (G1691A). The new method will employ multiplex PCR, allele specific primer extension (ASPE) and bead hybridization.

The laboratory will begin testing for methylenetetrahydrofolate reductase (MTHFR) mutations C677T and A1298C. The method will employ multiplex PCR, allele specific primer extension (ASPE) and bead hybridization.
MTHFR mutation analysis can be ordered separately or as part of the Thrombophilia mutation panel (Factor II, Factor V and MTHFR).

Also part of a Hypercoaguable Panel.

Clinical Significance: These tests can be used to help assess the risk of thrombotic disease.

Method: Multiplex PCR, allele specific primer extension (ASPE) and bead hybridization.

Availability: Once a week; Results in one week.

Specimen: Blood

Collect: One 5.0 mL EDTA (Lavender) tube.

Volume: Minimum 2.0 mL whole blood

Transport: Refrigerated

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Reference Range: With report

CPT Code: MTHFR: 83890;83900;83914;83912;83896x4
Thrombophilia Panel: 83890;83900;83901;83914;88384;83912x2

Test performed by The Nebraska Medical Center Laboratory.

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