

Inherited Thrombophilia Assessment: Combined Factor V Leiden (FVL), Prothrombin Polymorphism, and MTHFR Thermolabile Variant

DESCRIPTION:

- Factor V Leiden Arg506Gln: causes resistance to cleavage by Activated Protein C (APC), which allows Factor V to continue to function in the thrombotic cascade. This mutation is present (heterozygous) in 20-40% of patients with thrombosis (frequency 2 to 7% in the Caucasian population).
- Prothrombin (Factor II) G20210A: an alteration in the 3' untranslated region of the gene is associated with increased prothrombin levels predisposing to thrombosis. G20210A (heterozygous) is present in 6% of patients with thrombosis (frequency 1 to 2% in the Caucasian population).
- Methylene tetrahydrofolate reductase (MTHFR) C677T: A thermolabile variant in the gene is associated with increased plasma homocysteine levels. The thrombophilic risk conferred by homozygosity for C677T alone is controversial. However, it appears to confer increased risk in combination with other genetic mutations (homozygote frequency 5 to 15% in the Caucasian population).
- A complete work-up may include functional studies not available at Shodair: Antithrombin III, Proteins C and S, anticardiolipin, and lupus anticoagulant.

REASONS FOR REFERRAL:

- Patients with a history of or current thrombosis (DVT), pulmonary embolism, or thrombotic stroke. Patients with thrombosis associated with pregnancy, oral contraceptive usage, or surgery.
- Patients with a positive family history of hypercoagulability, especially if mutations have been identified in the family.
- Patients with pregnancy complications including fetal loss (especially if second / third trimester, recurrent, or if loss associated with placental thrombosis), pre-eclampsia (especially if severe, including HELLP), and abruption.

METHOD OF ANALYSIS:

- DNA amplification by PCR and specific primers, specific endonuclease cleavage, and high-resolution gel electrophoresis (see individual test descriptions).
- Results are reported within 2 weeks of receipt of sample, expedited upon request.

REFERENCE RANGES:

- Heterozygous or homozygous for gene change, or homozygous normal (see individual test descriptions). Presence of more than one mutation may multiply risk.

SAMPLE REQUIREMENTS:

- For DNA testing, 5 to 10 milliliters of blood (minimum 1 ml) in EDTA (purple top) tubes should be sent by overnight carrier at room temperature.

TEST CPT CODES:

CPT 83890 DNA extraction
CPT 83892 DNA enzymatic digestion
CPT 83894 DNA separation
CPT 83898 DNA amplification X 2
CPT 83912 DNA interpretation and report

Discounts from list price are available for institutional billing under contractual arrangement with the laboratory. Contact Ellen Livers at 800-447-6614 ext 7523.