

Prothrombin (Factor II) Polymorphism Testing

DESCRIPTION:

- An alteration in the 3' untranslated region of the prothrombin gene (G20210A) is associated with increased prothrombin levels and increased function of the thrombotic cascade.
- G20210A is present in 6% of patients with thrombosis (heterozygote frequency 1-2% in the Caucasian population).
- Heterozygotes are at a three-fold increased risk of serious thrombosis during their lifetime. Homozygotes are at higher risk.
- Patients heterozygous for both Factor V and Prothrombin alterations are at significantly increased risk of thrombosis.

REASONS FOR REFERRAL:

- Patients with hypercoagulability, evidenced by thromboses, should be evaluated for a panel of inherited thrombophilic factors, of which Factor V Leiden is the most common, with Factor II polymorphism the second most common. This includes patients with thrombosis associated with pregnancy, birth control pill usage, surgery, and those with leg thromboses, pulmonary embolism, or thrombotic stroke.
- Patients with a positive family history of hypercoagulability, Factor V Leiden, and patients with pregnancy complications (recurrent loss, pre-eclampsia).
- Because many patients with thrombosis have multiple risk factors, combined genetic testing for Factor V Leiden and MTHFR variant is available (see "Factor V / Prothrombin multiplex" and "Inherited Thrombophilia Assessment in Obstetrics").

METHOD OF ANALYSIS:

- DNA from the patient is amplified by PCR using primers flanking the DNA coding for the G to A polymorphism at nucleotide 20210 of the prothrombin gene 3' untranslated region. The mutation is detected by high resolution gel electrophoresis after digestion of the PCR product with HindIII.
- Results are reported within two weeks or less of receipt of sample and can be expedited upon request.

REFERENCE RANGES:

- Homozygous for G20210A (greatest risk for thrombosis).
- Heterozygous for G20210A (3-fold increased lifetime risk of thrombosis).
- Homozygous for G20210 (normal, not affected).

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.
- Prenatal testing: Contact the laboratory.

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