

Factor V Leiden (FVL) Mutation Testing

DESCRIPTION:

- The Leiden mutation in Factor V (FVL) causes resistance to cleavage by Activated Protein C (APC), an anti-thrombolytic factor. The resistance of Factor V to inactivation allows it to continue to function in the thrombotic cascade.
- FVL is present in 20-40% of patients with thrombosis (heterozygote frequency 2-7% in the Caucasian population).
- 10% of heterozygotes and almost 100% of homozygotes experience serious thrombosis during their lifetime.

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. 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 Prothrombin polymorphism and MTHFR variant is available (see "Inherited Thrombophilia Assessment in Obstetrics".)

METHOD OF ANALYSIS:

- DNA from the patient is amplified by PCR using primers flanking the DNA coding for Arg506, which is mutated to Gln (Factor V Leiden). The mutation is detected by high resolution gel electrophoresis after digestion of the PCR product with MnlI.
- Results are reported within 2 weeks or less of receipt of sample and can be expedited upon request.

REFERENCE RANGES:

- Homozygous for Gln506 (up to 80- fold increased risk for thrombosis).
- Heterozygous for Gln506 (up to 7-fold increased risk of thrombosis).
- Homozygous for Arg506 (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.

TEST CPT CODES:

CPT 83890 DNA extraction
CPT 83894 DNA separation
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.