Factor V Leiden Mutation Test

Summary and Explanation of the Test:

The University of Toledo Medical Center molecular diagnostics laboratory offers a test for Factor V Leiden utilizing polymerase chain reaction. Factor V Leiden is a single point mutation in the factor V gene (c.1691G>A; p.R506Q) on chromosome 1q23 which synthesizes a factor V molecule that is not properly inactivated by Activated Protein C (APC). Because of this dysregulation, the procoagulant affects of factor V are enhanced in the coagulation cascade. The Factor V Leiden mutation is the most common hereditary disorder contributing to the risk of thrombophilia. The resulting APC resistance is associated with a 5-10X and 50-100X increased risk of deep venous thrombosis (DVT) in the heterozygous and homozygous forms respectively. This test is particularly useful in assessing patients with DVT or at risk for DVT.

Genotype of the mutation is determined by a liquid bead-based assay on the Luminex 100/200 flow cytometer. After genomic DNA extraction from whole blood, the target is amplified by PCR and the product is hybridized to two different polystyrene beads (mutant and wild type) bearing complimentary oligonucleotide sequences. After adding fluorescent reporter streptavidin-phycoerythrin (SAPE), beads are washed and read on the Luminex 100/200 instrument. Genotyping is determined by analysis of signal generated from the wild type and mutant beads.

Turn-Around-Time: 7-10 days

Sample Requirements:

Whole blood collected in EDTA (purple top) or ACD (yellow top) vacutainer tubes is the specimen of choice. *Samples collected in a green top tube (heparin anticoagulated) are not acceptable.

Results Reporting:

A report is issued containing the results of the test (normal, heterozygous, or homozygous) and interpretation with reference to the associated risk.

References:

1) Ridker PM, et al. N Engl J Med. 1995 Apr 6;332(14):912-7. 2) Simioni P, et al. Semin Thromb Hemost. 2006 Oct;32(7):700-8 3) Muriel G, et al. Blood. 1998 Nov 1;92(9):3478-9. 4) Bosler D, et al. J Mol Diagn. 2006 Sep;8(4):420-5. 5) Frosst P, et al. Nat Genet. 1995 May;10(1):111-3.

For any questions regarding coagulation factor testing, please contact the Molecular Diagnostics laboratory at 419-383-5636 or the director at 419-383-6444. Further information can also be found on the Molecular Diagnostics web site at:
http://www.utoledo.edu/med/depts/path/moldx/index.html