Factor II (prothrombin) 20210G>A Mutation Test

Summary and Explanation of the Test:

The University of Toledo Medical Center molecular diagnostics laboratory offers a test for the Factor II (prothrombin) 20210G>A mutation utilizing polymerase chain reaction. Prothrombin is a precursor of the enzyme serine protease thrombin which has procoagulant, anticoagulant and antifibrinolytic activities. The prothrombin gene is located on chromosome 11 (11p11-q12) and has 14 exons. A single point mutation has been identified in exon 14 at position 20210G>A which results in increased transcription of the gene and elevated protein levels. This allele has a frequency of 1.2% and is associated with a 2.8 fold increased risk of venous thrombosis in individuals of Northern European ancestry. The homozygous state is rare; it is expected that the risk of thrombosis would be significantly greater than 3 fold, but no data exist to define the actual risk.

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