Thrombophilia Risk Genotyping Assay: Test Ordering Information

Test Information (v1_020112):
Thrombophilia Risk Genotyping Assay (CMGDL test code 4002)
CPT Codes 83891x1, 83892x2, 83900x1, 83901x5, 88384x1, 83912-Report & Interpretation
For additional information please refer to the CMGDL website www.medgen.med.miami.edu.

Indications for Testing
This test may be indicated or recommended if
-A patient has a personal or family history of venous thrombosis and/or related risk factors;
-An asymptomatic individual has a family history of venous thrombosis and/or related risk factors.

For a detailed list of test indications please refer to the individual Test Ordering Information documentation for the Factor V Leiden, Factor II, and MTHFR Genotyping Assays (visit www.medgen.med.miami.edu). See also References.

This test is not indicated or recommended as:
- General population screening or routine test;
- Prenatal or newborn testing;
- Prenatal testing is not performed by the CMGDL.

Specimen Requirements
2.0 ml of whole blood (minimum)
Collection Tube: Lavender Top Tube /w EDTA
Handling: Room Temp- specimen processed within 72 hours

Turn-Around Times
3 to 5 business days.

Methodology and Assay Characteristics
This test is performed using the eSensor® Technology, a solid-phase electrochemical method for determining the genotyping status of a defined panel of mutations.
This test is performed by the CMGDL according to the eSensor Thrombophilia Risk product insert. The eSensor Trombophilia Risk Test is an in vitro diagnostic for the detection and genotyping of Factor II (Prothrombin, 20210 G>A), Factor V (Leiden, 1691 G>A) and MTHFR (5, 10 methylenetetrahydrofolate reductase, 677C>T and 1298 A>C) gene mutations.

Panel of Alleles Detected
This test is validated to detect the following mutations:
-the F5 gene 1691G>A R506Q mutation, also described as Factor V Leiden mutation (standard nomenclature of the mutation: c.1601G>A, p.Arg534Gln; NM_000130.4);
-the F2 gene 20210G>A mutation (also designated as G20210A, colloquial nomenclature). This mutation should be described as g.21538G>A (AF478696.1), c.*9G>A being the mutation located 97 nucleotides downstream of the stop codon. The designation of 20210 is based on a historical reference sequence;
-the MTHFR 677C>T (Ala222Val, dbSNP rs1801133) and 1298A>C (Glu429Ala, dbSNP rs1801131) mutations.

Analytical Sensitivity and Specificity
99 percent of analytical sensitivity and specificity.

Test Limitations
- This test is validated to detect the F5 gene 1691G>A R506Q mutation, the F2 gene 20210G>A mutation, and the MTHFR 677C>T (Ala222Val, dbSNP rs1801133) and 1298A>C (Glu429Ala, dbSNP rs1801131) mutations. Additional allelic variants in the F5 gene or other genes will not be detected;
- We cannot exclude the possibility of rare mutations located in close proximity to the mutation tested that could interfere with the genotyping results and be the cause of an incorrect result;
- Results of this test should be interpreted in the context of clinical presentation and in consultation with a licensed physician and/or geneticist;
- Genetic testing using the methods applied by the CMGDL is expected to be highly accurate. However, the chance of a false positive or a false negative result, due to laboratory errors incurred during any phase of the testing, cannot be completely excluded.

Special Considerations
- Molecular diagnosis of Thrombophilia requires analysis of non-hematopoietic tissue in a patient after hematopoietic stem cell transplantation;
- Molecular diagnosis of Thrombophilia in liver transplant recipients requires molecular genetic testing of donor tissue;
- Molecular genetic tests are reliable in individuals on warfarin or heparin anticoagulation, and independent of thrombotic episodes.

Related Tests (visit our website at www.medgen.med.miami.edu)
Factor II G20210A and Factor V Leiden Genotyping Assay (CMGDL test code 4102)
Factor V Leiden Genotyping Assay (CMGDL test code 4202)
Factor II G20210A Genotyping Assay (CMGDL test code 4302)
MTHFR Genotyping Assay (CMGDL test code 4402)

References