Factor II G20210A Genotyping Assay: Test Ordering Information

**Test Information (v1_020112):**
Factor II G20210A Genotyping Assay (CMGDL test code 4302)
CPT Codes 83891x1, 83892x2, 83900x1, 83901x5, 88384x1, 83912-Report & Interpretation
For additional information please refer to the CMGDL website [www.medgen.med.miami.edu](http://www.medgen.med.miami.edu).

**Indications for Testing**

**Introductory Considerations**
The diagnosis of prothrombin-related Thrombophilia requires 20210G>A mutation analysis. This condition is suspected in individuals with a history of VTE that manifested as deep vein thrombosis or pulmonary embolism, especially with a history of VTE during pregnancy or associated with use of oral contraceptives. It is also suspected in individuals with a personal or family history of recurrent thrombosis at young age.

**Appropriate 20210G>A testing circumstances**
- A first unprovoked VTE before age 50 years;
- A history of recurrent VTE;
- Venous thrombosis at unusual sites such as the cerebral, mesenteric, portal, or hepatic veins;
- VTE during pregnancy or the puerperium;
- VTE associated with the use of estrogen-containing oral contraceptives or hormone replacement therapy;
- A first VTE at any age in an individual with a first-degree family member with a VTE before age 50 years;

**Testing may also be considered in case of**
- Asymptomatic adult family members of probands with heterozygous or homozgyous 20210G>A mutation, especially those with a strong family history of VTE at a young age;
- Asymptomatic female family members of probands with known prothrombin-related thromophilia who are pregnant or considering estrogen contraception or pregnancy;

(For a comprehensive list of circumstances, see [www.genetests.org](http://www.genetests.org).

**Factor II testing is not 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 Thrombophilia 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.

**Alleles Detected**
This test is validated to detect the F2 gene 20210G>A mutation (also designated as G20210A, colloquial nomenclature). This mutation should be described as g.21538G>A (AF478696.1), c.*97G>A being the mutation located 97 nucleotides downstream of the stop codon. The designation of 20210 is based on a historical reference sequence.

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

**Test Limitations**
- This test is validated to detect the F2 gene 20210G>A mutation (also designated as G20210A, colloquial nomenclature). Additional allelic variants in the F2 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 prothrombin-related Thrombophilia requires analysis of non-hematopoietic tissue in a patient after hematopoietic stem cell transplantation;
- Molecular diagnosis of prothrombin-related Thrombophilia in liver transplant recipients requires molecular genetic testing of donor tissue, the site of prothrombin synthesis;
- 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/)
Thrombophilia Risk Genotyping Assay (CMGDL test code 4002)
Factor II G20210A and Factor V Leiden Genotyping Assay (CMGDL test code 4102)
Factor V Leiden Genotyping Assay (CMGDL test code 4202)
MTHFR Genotyping Assay (CMGDL test code 4402)

References