**MHTFR Genotyping Assay: Test Ordering Information**

**Test Information (v1_020112):**
*MHTFR* Genotyping Assay (CMGD test code 4402)

**CPT Codes**
83891x1, 83892x2, 83900x1, 83901x5, 88384x1, 83912-Report & Interpretation

For additional information please refer to the CMGD website [www.medgen.med.miami.edu](http://www.medgen.med.miami.edu).

**Indications for Testing**
- Evaluation of individuals with a family history positive for venous thrombosis and/or Factor V Leiden, prothrombin G20210A, *MTHFR* 677C>T and 1298A>C mutations;
- Patients with venous thrombosis, coronary artery disease or stroke of unknown etiology;

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

**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 CMGD 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 methylene tetrahydrofolate reductase, 677C>T and 1298 A>C) gene mutations.

**Panel of Alleles Detected**
This test is validated to detect 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 *MTHFR* 677C>T (Ala222Val, dbSNP rs1801133) and 1298A>C (Glu429Ala, dbSNP rs1801131) mutations. Additional allelic variants in the *MTHFR* 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 CMGD 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.

**Related Tests** (visit our website at [www.medgen.med.miami.edu](http://www.medgen.med.miami.edu/))
Thrombophilia Risk Genotyping Assay (CMGD test code 4002)
Factor II G20210A and Factor V Leiden Genotyping Assay (CMGD test code 4102)
Factor V Leiden Genotyping Assay (CMGD test code 4202)
Factor II G20210A Genotyping Assay (CMGD test code 4302)

**References**
- van der Put, N. M., et al. (1998). A second common mutation in the methyltetrahydrofolate reductase gene: An additional risk factor for...