**CYP2C19 Genotyping Assay: Test Ordering Information**

**Test Information:**
*CYP2C19* Genotyping Assay (CMGDL test code 4001)

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

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

**Indications for Testing**
- Patients candidate for or undergoing drug therapy with drugs metabolized by the CYP2C19 isoenzyme;
- Patients with personal or family history positive for clinical inefficacy or adverse effects to drugs metabolized by the CYP2C19 isoenzyme.

In particular, the test may be indicated if the following drugs are considered:

- **Clopidogrel (Plavix®)** (see “CMGDL Clopidogrel Sensitivity Genotyping assay”, test code 4101)
- **Antidepressants such as Amitriptyline and escitalopram**
- **Tamoxifen**

*Note:* the clinical impact of the CYP2C19 genotype is influenced by whether a drug is activated (e.g. clopidogrel, tamoxifen) or inactivated (e.g. amitriptyline, escitalopram) by the CYP2C19 enzyme.

Involvement of other metabolic pathways, and other non-genetic factors such as concurrent intake of other medications may also influence the clinical impact of the CYP2C19 genotype (see the “Genetics and Clinical Overview” at [www.medgen.med.miami.edu](http://www.medgen.med.miami.edu) for additional information).

**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**
- 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 (read more at [www.medgen.med.miami.edu](http://www.medgen.med.miami.edu)).

**Panel of Alleles Detected**

This test is validated to detect the most common *CYP2C19* alleles, *2, *3, and *17. Additional alleles such the *4, *5, *6, *7, *8, *9, *10, *13 alleles are reported for informational purposes only (see Test Limitations).

**Clinical Sensitivity**

Estimated at 99 and 87 percent in Asians and Caucasians respectively, based on the prevalence of pathogenic allelic variants detected; sensitivity is unknown in other ethnicities.

**Analytical Sensitivity and Specificity**

99 percent of analytical sensitivity and specificity.

**Test Limitations**

- Although 11 alleles are targeted by this assay, due to the scarcity of DNA samples for rare (minor) *CYP2C19* alleles such the *4, *5, *6, *7, *8, *9, *10, *13 alleles, the CMGDL has clinically validated and determined accuracy and precision only for the most common alleles, *CYP2C19*2, *3, and *17.
- Additional allelic variants in *CYP2C19* (other than the above stated 11 alleles) or other genes will not be detected.
- For all the alleles we cannot exclude the possibility of rare mutations located in close proximity to the mutations tested that could interfere with the genotyping results and be the cause of a diagnostic error such as a false positive or incorrect result or false negative result (eg reporting a *1/*1 genotype in presence of one or two mutated alleles).

- Mutation detection is not a substitute for therapeutic drug monitoring.
- Non-genetic factors may also affect drug metabolism. Results of this test should be interpreted in the context of clinical presentation and in consultation with a licensed geneticist and/or pharmacist.

- This test was developed and its performance determined by the CMGDL. It has not been cleared or approved by the U.S. Food and Drug Administration. This test is used for clinical purposes. Pursuant to the requirements of CLIA ‘88, this laboratory has verified the test accuracy and precision. 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.

**Related Tests** (visit our website at [www.medgen.med.miami.edu](http://www.medgen.med.miami.edu))

Clopidogrel Sensitivity *CYP2C19* Genotyping assay (CMGDL test code 4101)