Pharmacogenetics: Expanded Panel

Test Code: MPG01
Turnaround time: 10 days
CPT Codes: 81225 x1, 81226 x1, 81227 x1, 81230 x1, 81231 x1, 81328 x1, 81355 x1, 81240 x1, 81241 x1

Condition Description

Pharmacogenomics (or pharmacogenetics), abbreviated PGx, is the study of how genes affect a person’s response to certain drugs. PGx testing uses genetic information to predict how individual patients will metabolize some prescription medications. This prediction allows an effective dose to be tailored to a person’s genetic makeup, helping to prevent adverse drug reactions. Pharmacogenetics Extended Test targets variants to genotype for various haplotypes among the following 14 genes.

Genes and Alleles Tested:
- **ANKK1/DRD2 DRD2:Taq1A**
- **COMT NM_000754.3:c.472G>A(p.V158M); rs4680**
- **C2** NM_000506.3:c.*97G>A; rs1799963
- **F5 NM_000130.4:c.1601G>A(p.R534Q); rs6025**
- **OPRM1 NM_000914.3:c.118A>G(p.N40D); rs1799971**
- **SLCO1B1** *5
- **VKORC1 NM_002046.4:c.-1639G>A; rs9923231**

Note that “*” (read as “star”) denotes the common name of a haplotype (or combination of alleles within a gene on the same chromosome).

Indications

Individuals who meet the defined clinical criteria for being prescribed specific drugs for cardiovascular conditions, cancer treatments, psychotropic medications, and pain management.

Methodology

The test uses custom manufactured TaqMan® drug metabolism and SNP genotyping assays in 384-well format that are run on the QuantStudio™ 6 Flex platform. The complete test consists of two components: SNP genotyping and copy number quantification. The SNP genotyping component consists of 64 TaqMan® SNP Assays for 63 loci, which include single base and short insertion/deletion polymorphisms. The copy number component consists of one TaqMan® Copy Number assay for the CYP2D6 gene.

Specimen Requirements

**Type: Liquid Buccal Swab**

Specimen Requirements:
- ORAcollect-DX (OCX-100)|Assisted saliva collection kit
- DNA Genotek ORAcollect-DX kit used according to manufacturer instructions.
- Please contact EGL for a saliva collection kit for patients that cannot provide a blood sample.

Specimen Collection and Shipping:
- Please do not refrigerate or freeze saliva sample. Please store and ship at room temperature.

**Type: Whole Blood (EDTA)**

Specimen Requirements:
- EDTA (Purple Top)
- Infants and Young Children (2 years of age to 10 years old): 3-5 ml
- Older Children & Adults: 5-10 ml
- Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:
- Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

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### Related Tests

- Pharmacogenetics Cardiovascular Panel (MPG02)
- Pharmacogenetics Pain Management and Psychotropic Panel (MPG03)