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**
- **COMT** NM_000754.3:c.472G>A(p.V158M); rs4680
- **F2** 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_020414.3:c.118A>G(p.N40D); rs1799971

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 (OCD-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.
Related Tests

Pharmacogenetics Cardiovascular Panel (MPG02)

Pharmacogenetics Pain Management and Psychotropic Panel (MPG03)