Pharmacogenomics Test List

Test Name	Test No.
Cytochrome P450 Testing	
Cytochrome P450 2D6/2C19 Genotyping	**512255**
Cytochrome P450 2D6 Genotyping	**512150**
Cytochrome P450 2C19 Genotyping	**512212**
Cytochrome P450 3A4/3A5 Genotyping	**512260**
Cytochrome P450 2C9 Genotyping	**512143**
HLA Testing	
HLA B*58:01, Allopurinol Hypersensitivity	**167351**
HLA B*57:01, Abacavir Hypersensitivity HLA Association Test	**006926**
Carbamazepine sensitivity HLA Associations (HLA B*15:02, HLA A*31:01)	**167443**
Other	
TPMT and *NUDT15* Genotyping	**512300**
UGT1A1 Irinotecan Toxicity	**511200**
DPYD Genotyping	**512275**
GeneSeq® Neuro: Malignant Hyperthermia Susceptibility Panel	**630700**

Gene variants tested

- ***CYP2D6*:** *2, *3, *4, *5 (deletion), *6, *7, *8, *9, *10, *11, *12, *13 (hybrid) *14, *15, *17, *29, *31, *35, *36 (hybrid), *36 + *10 (hybrid), *40, *41, *42, *49, *53, *59, *68 (hybrid), copy number determination
- ***CYP2C19*:** *2, *3, *4, *5, *6, *7, *8, *9, *10, *17, *35
- ***CYP2C9*:** *2, *3, *5, *6, *8, *11, *13
- ***CYP3A4*:** *22
- ***CYP3A5*:** *3, *6, *7
- ***TPMT*:** *2, *3A, *3B, *3C
- ***NUDT15*:** *2 or *3, *4
- ***UGT1A1*:** *28 , *36, *37
- ***DPYD*:** c.299_302del (*7, rs72549309), c.557A>G (rs115232898), c.703C>T (*8, rs1801266), c.868A>G (rs146356975), c.1129-5923C>G (HapB3, rs75017182), c.1236G>A (HapB3, rs56038477), c.1314T>G (rs186169810), c.1475C>T (rs72549304), c.1679T>G (*13, rs55886062), c.1774C>T (rs59086055), c.1905+1G>A (*2A, rs3918290), c.2279C>T (rs112766203), c.2639G>T (rs55674432), c.2846A>T (rs67376798).

  Note: These are all the Association for Molecular Pathology (AMP) Tier 1 and Tier 2 recommended *DPYD* variants. c.1236G>A is a normal function variant linked to the decreased function variant c.1129-5923C>G in HapB3. If both these variants are detected, only the c.1129-5923C>G is reported.
- **GeneSeq Neuro: Malignant Hyperthermia Susceptibility Panel:** *CACNA1S, RYR1* and *STAC3* genetic variants

Note: *1 in genotype results denotes detection of the normal (reference) sequence at all the variant sites assessed.

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Visit **clinpgx.org/** and **pharmdog.clinpgx.org/** to access ClinPGx curated pharmacogenomic guideline recommendations.

Pharmacogenomics Test List

Result Interpretation

Pharmacogenomic result interpretations vary depending on the test/genes involved.

Cytochrome P450 Enzymes

Genetic variation in cytochrome P450 (CYP450) genes can affect metabolic activity. CYP450 drug metabolizing enzyme activity can range from the total absence of metabolism to ultrarapid metabolism of certain drugs. Results include genotype and predicted metabolic activity.

Metabolic Activity

Depending on the CYP450 gene, metabolic activity categories include all or several of the following:

- **"Ultrarapid":** Ultrarapid metabolizer (UM) – Increased activity
- **"Rapid":** Rapid metabolizer (RM) – Slightly increased activity
- **"Normal":** Normal metabolizer (NM) – Normal activity
- **"Intermediate":** Intermediate metabolizer (IM) – Reduced activity
- **"Poor":** Poor metabolizer (PM) – Significantly reduced or absent activity

Note: "Rapid" is a metabolic activity category for CYP2C19 only. For CYP2C19 there are also "Likely Intermediate" (LIM) and "Likely Poor" (LPM) categories. For CYP2D6, predicated metabolic activities may also be reported as a range or may be categorized as indeterminate.

HLA

Positive or negative for allele(s) associated with adverse events from specific drug therapies

TPMT and *NUDT15*

TPMT: Genotype (including *2, *3A, *3B and *3C) and predicted metabolic activity: Normal, Intermediate, Poor.
NUDT15: Genotype (including *2 or *3, and *4) and predicted metabolic activity: Normal, Intermediate, Possible Intermediate, Poor, Indeterminate.

For both *TPMT* and *NUDT15,* decreased metabolic activity is associated with increased risk of adverse events (myelosuppression) from thiopurine drugs.

UGT1A1

One copy (heterozygous), two copies (homozygous), or negative for the *28 allele associated with reduced *UGT1A1* enzyme activity and increased risk for irinotecan toxicity. *36 and *37 variant alleles are also detected.

DPYD

Genotype (including variants noted above) and predicted metabolic activity (Normal, Intermediate and Poor). Intermediate metabolizers have partial DPD enzyme deficiency and Poor metabolizers have complete DPD deficiency. Both have increased risk for severe or even fatal drug toxicity when treated with fluoropyrimidine drugs.

GeneSeq® Neuro: Malignant Hyperthermia Susceptibility Panel

Next generation sequencing to identify genetic variants, including single nucleotide variants (SNVs), insertions, deletions and copy number variants (CNVs) in the following genes: *CACNA1S, RYR1* and *STAC3*.

For more information, please contact us at **800-777-0177** or **Monogram@Labcorp.com**. Visit **Labcorp.com** for full test information, including CPT codes and specimen requirements.

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