

Specimen Collected: 12-Sep-23 13:29

| CYP2D6 | Received: 12-Sep-23 13:30 | Report/Verified: 12-Sep-23 15:40 | |
|------------------------|---------------------------|----------------------------------|--------------------|
| Procedure | Result | Units | Reference Interval |
| 2D6GENO Specimen | Whole Blood | | |
| CYP2D6 Genotype | *1/*1 | | |
| CYP2D6 Phenotype | Normal | | |
| 2D6GENO Interpretation | See Note ^{f1} i1 | | |
| EER CYP2D6 | See Note ^{f2} | | |

Result Footnote

f1: 2D6GENO Interpretation
The following CYP2D6 allele(s) were detected: *1/*1. This result predicts the normal metabolizer phenotype with an activity score estimated at 2 of 2.

Recommendation: Guidelines for genotype-based dosing are published by the Clinical Pharmacogenetics Implementation Consortium (CPIC) and can be found at: <https://cpicpgx.org/> and <https://www.pharmgkb.org/>.

This result has been reviewed and approved by [REDACTED]

f2: EER CYP2D6
Authorized individuals can access the ARUP Enhanced Report using the following link:

Test Information

i1: 2D6GENO Interpretation
BACKGROUND INFORMATION: CYP2D6

Characteristics: The cytochrome P450 (CYP) isozyme 2D6 is involved in the metabolism of many drugs. Variants in the gene that code for CYP2D6 may influence pharmacokinetics of CYP2D6 substrates, and may predict or explain non-standard dose requirement, therapeutic failure or adverse reactions.

Inheritance: Autosomal codominant.

Cause: CYP2D6 gene variants and copy number affect enzyme expression or activity function.

Variants Tested: (Variants are numbered according to M33388 sequence.)

*1: Indicative of no detected targeted variants and an assumption of functional allele.

CYP2D6*2: rs16947, g.2850C>T; rs1135840, g.4180G>C

CYP2D6*2A: rs1080985, g.-1584C>G; rs16947, g.2850C>T; rs1135840, g.4180G>C

CYP2D6*3: rs35743686, g.2549del

CYP2D6*4: rs1065852, g.100C>T; rs3892097, g.1846G>A; rs1135840, g.4180G>C

CYP2D6*5: gene deletion

CYP2D6*6: rs5030655, g.1707del; rs1135840, g.4180G>C

CYP2D6*7: rs5030867, g.2935A>C

CYP2D6*8: rs5030865, g.1758G>T; rs16947, g.2850C>T; rs1135840, g.4180G>C

CYP2D6*9: rs5030656, g.2615_2617del

CYP2D6*10: rs1065852, g.100C>T; rs1135840, g.4180G>C

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

Unless otherwise indicated, testing performed at:

ARUP Laboratories

500 Chipeta Way, Salt Lake City, UT 84108

Laboratory Director: Jonathan R. Genzen, MD, PhD

ARUP Accession: 23-255-900129

Report Request ID: 18466496

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Test Information

i1: 2D6GENO Interpretation
 CYP2D6*11: rs1080985, g.-1584C>G; rs201377835, g.883G>C; rs16947, g.2850C>T; rs1135840, g.4180G>C
 CYP2D6*13: a CYP2D7-derived exon 1 conversion
 CYP2D6*14: rs5030865, g.1758G>A; rs16947, g.2850C>T; rs1135840, g.4180G>C
 CYP2D6*15: rs774671100, g.137_138insT
 CYP2D6*17: rs28371706, g.1023C>T; rs16947, g.2850C>T; rs1135840, g.4180G>C
 CYP2D6*29: rs16947, g.2850C>T; rs59421388, g.3183G>A; rs1135840, g.4180G>C
 CYP2D6*35: rs769258, g.31G>A; rs16947, g.2850C>T; rs1135840, g.4180G>C; rs1080985, g.-1584C>G
 CYP2D6*36: a CYP2D6*10 carrying a CYP2D7-derived exon 9 conversion
 CYP2D6*36-*10: a CYP2D6*36 and a CYP2D6*10 in tandem
 CYP2D6*40: rs28371706, g.1023C>T, rs16947, g.2850C>T; rs1135840, g.4180G>C; rs72549356, c.1863_1864ins TTTCGCCCTTTCGCCCC
 CYP2D6*41: rs16947, g.2850C>T; rs28371725, g.2988G>A; rs1135840, g.4180G>C
 CYP2D6*42: rs16947, g.2850C>T; rs1135840, g.4180G>C; rs72549346, g.3260_3261insGT
 CYP2D6*49: rs1065852, g.100C>T; rs1135822, g.1611T>A; rs1135840, g.4180G>C
 CYP2D6*69: rs1065852, g.100C>T; rs16947, g.2850C>T; rs28371725, g.2988G>A; rs1135840, g.4180G>C
 CYP2D6*114: rs1065852, g.100C>T; rs5030865, g.1758G>A; rs16947, g.2850C>T; rs1135840, g.4180G>C
 DUP: complete gene duplications

Clinical Sensitivity: Drug-dependent.

Methodology: Polymerase chain reaction (PCR) and fluorescence monitoring. Sequencing is only performed if needed to characterize a duplicated CYP2D6 gene.

Analytical Sensitivity and Specificity: Greater than 99 percent.

Limitations: Only the targeted CYP2D6 variants will be detected by this panel, and assumptions about phase and content are made to assign alleles. Publicly available sources such as the www.pharmvar.org or www.pharmgkb.org provide guidance on phenotype predictions and allele frequencies. A combination of the *5 (gene deletion) and a gene duplication cannot be specifically identified. This combination is not expected to adversely affect the phenotype prediction. Diagnostic errors can occur due to rare sequence variations. Risk of therapeutic failure or adverse reactions with CYP2D6 substrates may be affected by genetic and non-genetic factors that are not detected by this test. This result does not replace the need for therapeutic drug or clinical monitoring.

Please note the information contained in this report does not contain medication recommendations, and should not be interpreted as recommending any specific medications. Any dosage adjustments or other changes to medications should be evaluated in consultation with a medical provider.

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Patient Age/Sex:

Female

Test Information

i1: 2D6GENO Interpretation

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

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