

Client: Example Client ABC123 123 Test Drive Salt Lake City, UT 84108 UNITED STATES

Physician: Doctor, Example

**Patient: Patient, Example** 

**DOB** Unknown Gender: Female

**Patient Identifiers:** 01234567890ABCD, 012345

**Visit Number (FIN):** 01234567890ABCD **Collection Date:** 00/00/0000 00:00

## CYP<sub>2</sub>D<sub>6</sub>

ARUP test code 3001513

2D6GENO Specimen

Whole Blood

CYP2D6 Genotype

## Heterozygous

## 2D6GENO Interpretation

## See Note

The following CYP2D6 allele(s) were detected: \*35/Negative. 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://www.pharmgkb.org/.

This result has been reviewed and approved by Ph.D.

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.
VARIANTS TESTED: (Variants are numbered according to M32388)

VARIANTS TESTED: (Variants are numbered according to M33388

sequence.) Negative: No variants detected is predictive of the \*1 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 CYP2D6\*11: rs1080985, g.-1584C>G; rs201377835, g.883G>C;

H=High, L=Low, \*=Abnormal, C=Critical

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rs16947, g.2850C>T; rs1135840, g.4180G>C
CYP2D6*12: rs5030862, g.124G>A; 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 TTTCGCCCCTTTCGCCCC CYP2D6*41: rs16947, g.2850C>T; rs28371725, g.2988G>A; rs1135840,
g.4180G>C
CYP2D6*45: rs28371710, g.1716G>A; rs16947, g.2850C>T; rs1135840,
g.4180G>C
CYP2D6*46: rs28371696, g.77G>A; rs28371710, g.1716G>A; rs16947, g.2850C>T; rs1135840, g.4180G>C
CYP2D6*49: rs1065852, g.100C>T; rs1135822, g.1611T>A; rs1135840,
g.4180G>C
g.4180G>C
CYP2D6*53: rs1135822, g.1611T>A
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.
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METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitorina.

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. Publically available sources such as the www.pharmvar.org or www.pharmgkb.org provide guidance on phenotype predictions and allele frequencies. 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.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US 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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VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
2D6GENO Specimen	21-271-107604	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
CYP2D6 Genotype	21-271-107604	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
2D6GENO Interpretation	21-271-107604	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, \*=Abnormal, C=Critical

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