

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 12/31/1752
Gender: Unknown
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 01/01/2017 12:34

CYP2D6

ARUP test code 3001513

2D6GENO Specimen whole Blood

CYP2D6 Genotype *41/*41

2D6GENO Interpretation

See Note

Interpretation: Two decreased function CYP2D6 alleles were detected. This result predicts the intermediate to normal metabolizer phenotype with an activity score of 1. Impaired metabolic phenotypes may confer sensitivity to drug-drug interactions with CYP2D6 substrates. Depending on the metabolic pathway for the drug(s) of interest, the impact on dosing may depend on phenotype predictions for other genes.

This result has been reviewed and approved by [REDACTED]

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 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

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

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Tracy I. George, MD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 19-324-400745
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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CYP2D6*11: rs1080985, g.-1584C>G; rs201377835, g.883G>C;
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*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
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.

METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.

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. 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.

See Compliance Statement C: www.aruplab.com/CS

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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
2D6GENO Specimen	19-324-400745	8/20/2019 12:00 00 AM	11/20/2019 2:22:26 PM	11/26/2019 9:16:00 AM
CYP2D6 Genotype	19-324-400745	8/20/2019 12:00 00 AM	11/20/2019 2:22:26 PM	11/26/2019 9:16:00 AM
2D6GENO Interpretation	19-324-400745	8/20/2019 12:00 00 AM	11/20/2019 2:22:26 PM	11/26/2019 9:16:00 AM

END OF CHART

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