

500 Chipeta Way, Salt Lake City, Utah 84108-1221

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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

**Client:** ARUP Example Report Only

500 Chipeta Way

Salt Lake City, UT 84108-

USA

**Provider:** .108 -TEST,**Patient:** 2D6 GENO, POSITIVE**DOB:****Sex:** Male**Patient Identifiers:** 40662**Visit Number (FIN):** 40987**Client Supplied ID:****Specimen Collected:** 19-Sep-22 16:31

CYP2D6	Received: 19-Sep-22 16:38	Report/Verified: 20-Sep-22 12:40	
Procedure	Result	Units	Reference Interval
2D6GENO Specimen	Whole Blood		
CYP2D6 Genotype	*13/*41		
CYP2D6 Phenotype	<b>Intermediate *</b>		
2D6GENO Interpretation	See Note <sup>f1 i1</sup>		

**Result Footnote**

f1: 2D6GENO Interpretation  
The following CYP2D6 allele(s) were detected: \*13/\*41. This result predicts the intermediate metabolizer phenotype with an activity score estimated at 0.5 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 Yuan Ji, Ph.D.

**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.)

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

\*=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:** 22-262-900235**Report Request ID:** 16423084**Printed:** 20-Sep-22 16:59

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**Patient:** 2D6 GENO, POSITIVE**DOB:****Patient Identifiers:** 40662**Test Information**

i1: 2D6GENO Interpretation  
 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; 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](http://www.pharmvar.org) or [www.pharmgkb.org](http://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

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**Patient:** 2D6 GENO, POSITIVE

**DOB:**

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**Test Information**

i1: 2D6GENO Interpretation

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 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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