Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108
UNITED STATES

Physician: arup, arup

**Patient: Test, 2D6GENO Neg**

**DOB**
**Sex:** Male
**Patient Identifiers:** 46479
**Visit Number (FIN):** 46808
**Collection Date:** 3/3/2023 11:05

## CYP2D6

ARUP test code 3001513

| | |
|---|---|
| 2D6GENO Specimen | Whole Blood |
| CYP2D6 Genotype | Neg/Neg |
| CYP2D6 Phenotype | Normal |
| 2D6GENO Interpretation | See Note |

The following CYP2D6 allele(s) were detected: Neg/Neg. 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 other organizations. See: https://www.pharmgkb.org/

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 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
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 TTTCGCCCCTTTCGCCCC
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.

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.

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

VERIFIED/REPORTED DATES

| Procedure | Accession | Collected | Received | Verified/Reported |
|---|---|---|---|---|
| 2D6GENO Specimen | 23-062-105125 | 3/3/2023 11 05:00 AM | 3/3/2023 11:05:40 AM | 4/6/2023 2:33:00 PM |
| CYP2D6 Genotype | 23-062-105125 | 3/3/2023 11 05:00 AM | 3/3/2023 11:05:40 AM | 4/6/2023 2:33:00 PM |
| CYP2D6 Phenotype | 23-062-105125 | 3/3/2023 11 05:00 AM | 3/3/2023 11:05:40 AM | 4/6/2023 2:33:00 PM |
| 2D6GENO Interpretation | 23-062-105125 | 3/3/2023 11 05:00 AM | 3/3/2023 11:05:40 AM | 4/6/2023 2:33:00 PM |

END OF CHART

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