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

PATIENT REPORT

Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108-
USA

Provider: .108 -TEST,

Patient: **CYP GD, POSITIVE**
DOB:
Sex: Female
Patient Identifiers: 40673
Visit Number (FIN): 40998
Client Supplied ID:

Specimen Collected: 19-Sep-22 16:37

CYP450 Genotyping Panel, with GeneDose | **Received: 19-Sep-22 16:38** **Report/Verified: 20-Sep-22 14:55**

Procedure	Result	Units	Reference Interval
CYP PANEL Specimen	Whole Blood		
CYP2C19 Genotype	*2/Neg		
CYP2C19 Phenotype	**Intermediate** *		
CYP2C8 Genotype	Neg/Neg		
CYP2C8 Phenotype	Normal		
CYP2C9 Genotype	Neg/Neg		
CYP2C9 Phenotype	Normal		
CYP2C Cluster Geno	**Heterozygous** *		
CYP2C Cluster Pheno	**See Note** *		
CYP2D6 Genotype	*2A/*4		
CYP2D6 Phenotype	**Intermediate** *		
CYP3A4 Genotype	Neg/Neg		
CYP3A4 Phenotype	Normal		
CYP3A5 Genotype	*3/*3		
CYP3A5 Phenotype	**Poor** *		
CYP2B6 Genotype	*6/Neg		
CYP2B6 Phenotype	**Intermediate** *		
CYP PANEL Interpretation	See Note f1 i1		
CYP PANEL,GeneDose Link	See Note i2		

Result Footnote

f1: CYP PANEL Interpretation
The following CYP2C19 allele(s) were detected: *2/Neg. This result predicts the intermediate metabolizer phenotype.

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

The following CYP2C8 allele(s) were detected: Neg/Neg. This result predicts the normal metabolizer phenotype.

The following CYP2C9 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/

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

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Result Footnote

f1: CYP PANEL Interpretation
One copy of the 2C cluster rs12777823 was detected. This variant is associated with reduced warfarin dose requirement in some individuals of African ancestry.

The following CYP2D6 allele(s) were detected: *2A/*4. This result predicts the intermediate metabolizer phenotype with an activity score estimated at 1 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/.

The following CYP3A4 allele(s) were detected: Neg/Neg. This result predicts the normal metabolizer phenotype.

The following CYP3A5 allele(s) were detected: *3/*3. This result predicts the poor metabolizer phenotype.

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

The following CYP2B6 alleles were detected: *6/Neg. This result predicts the intermediate metabolizer phenotype.

Recommendation: Guidelines for gene-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 Yuan Ji, Ph.D.

Test Information

i1: CYP PANEL Interpretation
BACKGROUND INFORMATION: Cytochrome P450 Genotyping Panel

Characteristics: The cytochrome P450 (CYP) isozymes 2B6, 2C19, 2C8, 2C9, 2D6 and the CYP3A subfamily are involved in the metabolism of many drugs. Variants in the genes that code for CYP2B6, CYP2C19, CYP2C8, CYP2C9, CYP2D6, CYP3A4, and CYP3A5; and CYP2C cluster (rs12777823) loci, will influence pharmacokinetics of respective substrates, and may predict or explain non-standard dose requirements, therapeutic failure, or adverse reactions.
Inheritance: Autosomal codominant.
Cause: Gene variants affect enzyme function.
Variants Tested:
(Variants are numbered according to the following transcripts:
CYP2C19 NM_000769, CYP2C8 NM_000770, CYP2C9 NM_000771, 2C cluster rs12777823, CYP2D6 M33388 sequence, CYP3A4 NM_017460 and CYP3A5 NM_000777, CYP2B6 NM_000767).
Negative: No variants detected is predictive of the *1 functional alleles.
CYP2C19*2: rs4244285, c.681G>A; rs12769205, c.332-23A>G

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i1: CYP PANEL Interpretation

CYP2C19*3: rs4986893, c.636G>A
CYP2C19*4A: rs28399504, c.1A>G
CYP2C19*4B: rs28399504, c.1A>G; rs12248560, c.-806C>T
CYP2C19*5: rs56337013, c.1297C>T
CYP2C19*6: rs72552267, c.395G>A
CYP2C19*7: rs72558186, c.819+2T>A
CYP2C19*8: rs41291556, c.358T>C
CYP2C19*9: rs17884712, c.431G>A
CYP2C19*17: rs12248560, c.-806C>T
CYP2C19*35: rs12769205, c.332-23A>G

CYP2C8*2: rs11572103, c.805A>T
CYP2C8*3: rs10509681, c.1196A>G
CYP2C8*4: rs1058930, c.792C>G

CYP2C rs12777823, g.96405502 G>A

CYP2C9*2: rs1799853, c.430C>T
CYP2C9*3: rs1057910, c.1075A>C
CYP2C9*4: rs56165452, c.1076T>C
CYP2C9*5: rs28371686, c.1080C>G
CYP2C9*6: rs9332131, c.818del
CYP2C9*8: rs7900194, c.449G>A
CYP2C9*11: rs28371685, c.1003C>T
CYP2C9*12: rs9332239, c.1465C>T

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

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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

CYP2B6*4: rs2279343, c.785A>G
CYP2B6*6: rs3745274, c.516G>T; rs2279343, c.785A>G
CYP2B6*7: rs3745274, c.516G>T; rs2279343, c.785A>G; rs3211371, c.1459C>T
CYP2B6*9: rs3745274, c.516G>T
CYP2B6*18: rs28399499, c.983T>C
CYP2B6*22: rs34223104, c.-82T>C
CYP2B6*36: rs34223104, c.-82T>C; rs3745274, c.516G>T; rs2279343, c.785A>G

CYP3A4*1A: rs2740574, c.-392G>A
CYP3A4*22: rs35599367, c.522-191C>T

CYP3A5*3: rs776746, c.219-237A>G
CYP3A5*6: rs10264272, c.624G>A
CYP3A5*7: rs41303343, c.1035dup

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.

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i1: CYP PANEL Interpretation

Limitations: Only the targeted 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 CYP2D6*5 (gene deletion) and a CYP2D6 gene duplication cannot be specifically identified; however, 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 gene 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.

i2: CYP PANEL, GeneDose Link

INTERPRETIVE INFORMATION: CYP PANEL, GeneDose Link

GeneDose LIVE content is provided by Coriell Life Sciences and not by ARUP Laboratories.

Any dosage adjustments or other changes to medications should be evaluated in consultation with a medical provider.