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PATIENT REPORT

Patient Age/Sex: Female

Specimen Collected: 12-Sep-23 13:26

Cytochrome P450 Genotyping Panel | **Received: 12-Sep-23 13:30** **Report/Verified: 12-Sep-23 15:21**

Procedure	Result	Units	Reference Interval
CYP PANEL Specimen	Whole Blood		
CYP2C19 Genotype	*1/*1		
CYP2C19 Phenotype	Normal		
CYP2C8 Genotype	*1/*1		
CYP2C8 Phenotype	Normal		
CYP2C9 Genotype	*1/*1		
CYP2C9 Phenotype	Normal		
CYP2C Cluster Geno	Negative		
CYP2C Cluster Pheno	Normal		
CYP2D6 Genotype	*1/*1		
CYP2D6 Phenotype	Normal		
CYP3A4 Genotype	*1/*1		
CYP3A4 Phenotype	Normal		
CYP3A5 Genotype	*1/*1		
CYP3A5 Phenotype	Normal		
CYP2B6 Genotype	*1/*1		
CYP2B6 Phenotype	Normal		
CYP PANEL Interpretation	See Note [f1 i1]		
EER CYPP450 Panel	See Note [f2]		

Result Footnote

f1: CYP PANEL Interpretation

The following CYP2C19 allele(s) were detected: *1/*1. This result predicts the normal metabolizer phenotype.

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

The following CYP2C8 alleles were detected: *1/*1
The metabolizer phenotype is drug-dependent.

The following CYP2C9 allele(s) were detected: *1/*1. This result predicts the normal metabolizer phenotype, with an activity score of 2 of 2.

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

The 2C cluster variant (rs12777823) was not detected. This result predicts a normal phenotype and is not expected to contribute to warfarin dosing estimates.

The following CYP2D6 allele(s) were detected: *1/*1. 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://cpicpgx.org/ and https://www.pharmgkb.org/.

Result Footnote

f1: CYP PANEL Interpretation
The following CYP3A4 allele(s) were detected: *1/*1. This result predicts the normal metabolizer phenotype.

The following CYP3A5 allele(s) were detected: *1/*1. This result predicts the normal metabolizer phenotype.

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

The following CYP2B6 alleles were detected: *1/*1. This result predicts the normal metabolizer phenotype.

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

This result has been reviewed and approved by [redacted]

f2: EER CYPP450 Panel
Authorized individuals can access the ARUP Enhanced Report using the following link:

[redacted]

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 nonstandard 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).

*1: Indicative of no detected targeted variants and an assumption of functional allele.

CYP2C19*2: rs4244285, c.681G>A; rs12769205, c.332-23A>G
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

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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
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

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i1: CYP PANEL Interpretation

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.
Analytic Sensitivity and Specificity: Greater than 99 percent.
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 nongenetic 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.

Test Information

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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.