

Jane Doe
Biological Sex: Female
Date of Birth: 11/13/2001
Sample ID: SM12805
Sample Type: BLOOD
Collection Date: 11/13/2024
Received Date: 11/14/2024

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Requisition ID:
RQ12345
Report Number:
RP1234
Report Date:
12/05/2024

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PHARMACOGENOMIC SUMMARY

To facilitate the use of pharmacogenomic guidance, we report known variants in genes associated with response to common medications. Our test examines 15 genes with alleles with known pharmacogenomic implications. For additional guidance on how the findings in this report might affect drug response please refer to the FDA. **You should always consult your healthcare provider prior to making any changes to your medication regimen.**

GENE	DIPLTYPE	GENE PHENOTYPE
CYP2B6	*6 / *22	Intermediate Metabolizer
CYP2C19	*1 / *1	Normal Metabolizer
CYP2C9	*1 / *1	Normal Metabolizer
CYP2D6	*1 / *4	Intermediate Metabolizer
CYP3A4	*1 / *36	Variant Present
CYP3A5	*1 / *3	Intermediate Metabolizer
CYP4F2	*1 / *1	Variant Absent
DPYD	*1 / *1	Normal Metabolizer
IFNL3	rs12979860 T/T	Variant Present
NUDT15	*1 / *1	Normal Metabolizer
SLCO1B1	*1 / *1	Normal Function
TPMT	*1 / *1	Normal Metabolizer
UGT1A1	*1 / *1	Normal Metabolizer
VKORC1	rs9923231 C/C	Variant Absent

GENES AND ALLELES ANALYZED

CYP2B6: *4; *6; *9; *18; *22 **CYP2C9:** *2; *3; *4; *5; *6; *8; *11; *12; *13; *15; *16; *26; *28; *29; *30; *31; *42; *55 **CYP2C19:** *2; *3; *4; *5; *6; *7; *8; *9; *10; *17; *35 **CYP2D6:** *2; *3; *4; *6; *7; *8; *9; *10; *11; *12; *14; *15; *17; *21; *31; *40; *41; *42; *49; *56; *59; *100; *114 **CYP3A4:** *22 **CYP3A5:** *3; *6 **CYP4F2:** *3 **DPYD:** rs115232898; rs3918290; rs55886062; rs75017182+rs56038477; rs146356975; rs67376798; rs59086055; rs183385770; rs112766203 **F5:** rs6025 **IFNL3:** rs12979860 **NUDT15:** *3; *4; *9 **SLC01B1:** *5; *9; *14; *20 **TPMT:** *2; *3A; *3B; *3C; *4; *11; *29 **UGT1A1:** *6; *27 **VKORC1:** rs9923231 See limitations in subsequent page(s).

TEST METHODS

- Specimen receipt, accessioning, data analysis, and interpretation is performed by MyOme Inc., 1455 Adams Drive, Suite 1150, Menlo Park, CA 94025, CLIA# 05D2203070. Blended Genome-Exome Sequencing, excluding data analysis and interpretation, is performed by Broad Clinical Labs LLC, 27 Blue Sky Dr, Burlington, MA 01803, CLIA#22D2055652.
- Genomic DNA obtained from the submitted sample was used to construct a PCR-free whole genome library and an exome library and sequenced using Illumina technology generating low-coverage whole-genome and higher coverage exome data. Reads were aligned to the NCBI GRCh38 reference assembly.
- Sequencing data is then analyzed through MyOme's small variant calling process and a modified version of Aldy¹ is used to infer PGx diplotypes. For variants that fall outside of exome target regions genotype likelihoods are estimated for bases covered by at least one sequencing read. Genotypes at additional sites are imputed based on a genotype reference panel.
- Star alleles are based on PharmVar star allele definitions when available. CYP2D6, CYP2C9, and DPYD gene phenotypes are assigned based on the diplotype level activity score, using published thresholds²⁻⁷.

TEST LIMITATIONS

- This test is designed to provide information only for the genes and alleles denoted in the Genes and Alleles Analyzed section of the report. A *1 allele indicates that no test alleles were found for one or both haplotypes. There may be other variants that impact your response to medications not examined here. This test does not detect CYP2D6 copy number changes or hybridizations.
- There may be variants in this test that cannot be detected with the current technology. Sensitivity to detect variants may be reduced in low complexity regions and segmental duplication regions.
- A history of stem cell or bone marrow transplantation, or recent blood transfusion may impact the accuracy of the results.
- Like most tests, this test carries a risk of false negative or false positive results, which may be caused by, without limitation, sample contamination from biological or non-biological sources, specimen marking issues, rare genetic variants interfering with analysis, and other technical issues and limitations.

DISCLAIMERS

This test was developed, and its performance characteristics were determined, by MyOme, Inc., a clinical laboratory certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) and College of American Pathologist (CAP) accredited to perform high complexity clinical laboratory testing. This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). Along with factors such as age and diet, genetic variants can impact your response to certain medications. Test results should always be interpreted by a clinician in the context of clinical and familial data with the availability of genetic counseling when appropriate. You should always consult your healthcare provider prior to making any changes to your medication regimen. Predicted phenotype and allele functionality implications may change depending upon the emergence of new research and/or guideline updates. Drug-drug, drug-gene, and drug-diet interactions, as well as other factors, that are not within the scope of this test may impact the pharmacogene phenotypes reported in this test. Like most tests, this test carries a risk of false negative or false positive results. Testing is unavailable for samples damaged by human error or lost/destroyed due to weather, transit issues or other problems beyond the control of MyOme. MyOme is not responsible for the content of third-party websites referenced in this report.

REFERENCES

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