

Pharmacogenomic Variant Screening

Background

Pharmacogenomic (PGx) testing is a precision medicine tool that empowers patients and doctors with information that can be used to help minimize adverse drug events. By identifying genomic variations that influence drug metabolism, drug efficacy, and drug toxicity, this test provides information that your healthcare team can use to optimize and personalize medication therapies. Your healthcare provider(s) can integrate your PGx test results with the multiple factors that influence each individual's response to medication.

The Alamy Health team updates the gene list based on current scientific knowledge. Drug-gene pairs are selected for analysis based upon strong and consistent scientific evidence linking them to defined drug responses and for which the benefits clearly outweigh the risks. Based on this analysis and available testing, the table below lists the genes and variants that are screened and has links to relevant resources.

Limitations

This analysis does not identify all genetic variants that influence enzyme activity and expression. Additionally, it does not account for genetic variations in other metabolic pathways that could affect drug metabolism, non-genetic factors, or possible interactions between medications.

Individuals undergoing PGx testing should be aware of the potential for rare diagnostic errors. These may arise from factors such as sample mix-ups or genetic testing inaccuracies due to limitations of the sequencing technology and/or interpretation process. This test does not determine the specific arrangement of gene copies (aka allele phasing). Rather, it predicts the most likely combination based on information gleaned from the general population via test databases. Typically, minor differences in allele combinations do not affect the overall result, but if a combination difference is detected that could change a drug response, this information and the potential impact will be included in the report.

This test may be less accurate for individuals with leukemia, recent blood transfusions, or organ or stem cell transplants. If you have experienced any of these, please be sure to let your healthcare provider know so that the test results can be interpreted appropriately for you.

The interpretation of genetic changes is based on current medical knowledge and may evolve as new information becomes available. We recommend that you review the results of your PGx genetic test along with your clinical information with your healthcare providers including your pharmacists to guide drug selection and drug dosing decisions.

PHARMACOGENOMIC VARIANTS SCREENED

Gene Symbol	Information Resources: CPIC	Information Resources: PharmGKB
ABCG2	ABCG2 CPIC guidelines	Gene-specific Information Tables for ABCG2
CACNA1S	CACNA1S CPIC guidelines	Gene-specific Information Tables for CACNA1S
CYP2B6	CYP2B6 CPIC guidelines	Biogeographical Groups for Population Classification
CYP2C19	CYP2C19 CPIC guidelines	Gene-specific Information Tables for CYP2C19
CYP2C9	CYP2C9 CPIC guidelines	Gene-specific Information Tables for CYP2C9
CYP2D6	CYP2D6 CPIC guidelines	Gene-specific Information Tables for CYP2D6
CYP3A5	CYP3A5 CPIC guidelines	Gene-specific Information Tables for CYP3A5
CYP4F2	CYP4F2 CPIC guidelines	Gene-specific Information Tables for CYP4F2
DPYD	DPYD CPIC guidelines	Gene-specific Information Tables for DPYD
G6PD	G6PD CPIC guidelines	Gene-specific Information Tables for G6PD
HLA-A *31:01	HLA-A and HLA-B CPIC guidelines	Gene-specific Information Tables for HLA-A and HLA-B
HLA-B *58:01	HLA-A and HLA-B CPIC guidelines	Gene-specific Information Tables for HLA-A and HLA-B
HLA-B *57:01	HLA-A and HLA-B CPIC guidelines	Gene-specific Information Tables for HLA-A and HLA-B
HLA-B *44	HLA-A and HLA-B CPIC guidelines	Gene-specific Information Tables for HLA-A and HLA-B
HLA-B *15:02	HLA-A and HLA-B CPIC guidelines	Gene-specific Information Tables for HLA-A and HLA-B
MT-RNR1	MT-RNR1 CPIC guidelines	Gene-specific Information Tables for MT-RNR1
NUDT15	NUDT15 CPIC guidelines	Gene-specific Information Tables for NUDT15
RYR1	RYR1 CPIC guidelines	Gene-specific Information Tables for RYR1
SLC01B1	SLC01B1 CPIC guidelines	Gene-specific Information Tables for SLC01B1
UGT1A1	UGT1A1 CPIC guidelines	Gene-specific Information Tables for UGT1A1
VKORC1	VKORC1 CPIC guidelines	Gene-specific Information Tables for VKORC1