alamya health

A Future Where Precision Medicine is the Standard

Pharmacogenomic Variant Screening

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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 Alamya 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	Variants/Alleles	PharmGKB Resource
ABCG2	rs2231142	pharmGKB
CACNA1S	rs1800559, rs772226819	pharmGKB
CYP2B6	*2, *4, *6, *7, *8, *9, *12, *13, *18, *19, *20, *22, *24, *26, *28, *34, *35, *36, *37, *38	pharmGKB
CYP2C9	*2, *3, *4, *5, *6, *8, *11, *12, *13, *14, *15, *16, *23, *24, *25, *26, *28, *29, *30, *31, *33, *35, *37, *38, *39, *42, *43, *44, *45, *46, *50, *52, *55, *61	pharmGKB
CYP2C19	*2, *3, *4, *5, *6, *7, *8, *9, *10, *16, *17, *19, *22, *24, *25, *26, *35, *38	pharmGKB
CYP2D6	*2, *3 ,*4, *6, *7, *8, *9, *10, *11, *12, *14, *15, *17, *18, *19, *20, *21, *29, *31, *35, *36, *38, *40, *41, *42, *44, *47, *51, *56, *60, *62, *81, *92, *96, *99, *100, *101, *120, *124, *129, *144, *161	pharmGKB
CYP4F2	*3	pharmGKB
CYP3A5	*1D, *3, *6, *7	pharmGKB
DPYD	rs183385770, rs143154602, rs75017182 and rs56038477 (HapB3), rs78060119 (*12), rs186169810, rs72549304, rs111858276, rs55886062 (*13), rs59086055, rs138616379, rs145773863, rs72549303 (*3), rs3918290 (*2A), rs137999090, rs112766203, rs55674432, rs67376798, rs141044036, rs72547601, rs1801268 (*10), rs72549309 (*7), rs115232898, rs72549308, rs72549310, rs72549307, rs1801266 (*8), rs146356975	pharmGKB
F5	rs6026	pharmGKB
IFNL3/IFNL4	rs12979860, rs8099917	pharmGKB pharmGKB
MT-RNR1	rs267606617	pharmGKB
NUDT15	*2, *3	pharmGKB
RYR1	rs118192178, rs121918592, rs121918593, rs121918594, rs121918595, rs121918596, rs1801086, rs193922747, rs193922748, rs193922753, rs193922762, rs193922764, rs193922768, rs193922770, rs193922772, rs193922802, rs193922803, rs193922807, rs193922809, rs193922816, rs193922818, rs193922832, rs193922843, rs193922876, rs193922878, rs28933396, rs28933397~~, rs63749869~~	pharmGKB
SLCO1B1	*5, *9, *14, *15, *20, *23, *31, *37, *46, *47	pharmGKB
TPMT	*2, *3A, *3B, *3C, *4	pharmGKB
UGT1A1	*6, *28, *36, *37	pharmGKB
VKORC1	rs9923231	pharmGKB