Reported Genotypes:

The PGx cardiology focus panel performs genotyping analysis on key genes and variant hot spots with defined evidence from pharmacogenomics-centric guidelines and databases such as Clinical Pharmacogenetics Implementation Consortium (CPIC), ClinPGx, FDA biomarker labeling recommendations, and the and Dutch Pharmacogenetics Working Group (DPWG). Specific loci documented as informing effectiveness of drug metabolism evaluated on the current PGx cardiology panel (v 1.0) are detailed below. The report is limited to the following SNP reference IDs (rsIDs), star-allele configurations, and variants (see summary of markers tested). There are no inherited diseases or conditions strongly linked to germline genetic variants in CYP2C19, ABCG2, and CYP2C9 independent of drug metabolism and response. Germline variants in SLCO1B1 can cause Rotor syndrome (OMIM:237450), where a porphyrin compound found in human urine is markedly increased. Of note, this PGx test is not designed to detect these germline variants for this syndrome.

Summary of Markers Tested

Gene	rsID	HGVS (RefSeq Select transcript)	Star allele	CPIC function
ABCG2	rs2231142	NM_004827.3:c.421C>A, (p.Gln141Lys)	Not assigned	Decreased function
CYP2C9	rs1799853	NM_000771.4:c.430C>T, (p.Arg144Cys)	*2	Decreased function
	rs1057910	NM_000771.4:c.1075A>C, (p.Ile359Leu)	*3	No function
	rs28371686	NM_000771.4: c.1080C>G, (p.Asp360Glu)	*5	Decreased function
	rs9332131	NM_000771.4:c.818del, (p.Lys273fs)	*6	No function
	rs7900194	NM_000771.4: c.449G>A, (p.Arg150His)	*8	Decreased function
	rs28371685	NM_000771.4:c.1003C>T, (p.Arg335Trp)	*11	Decreased function
	rs9332239	NM_000771.4: c.1465C>G, (p.Pro489Ser)	*12	Decreased function
CYP2C19	rs4244285; rs12769205	NM_000769.4:c.681G>A, p.Pro227; NM_000769.4:c.332-23A>G	*2	No function
	rs4986893	NM_000769.4:c.636G>A, (p.Trp212Ter)	*3	No function
	rs28399504	NM_000769.4:c.1A>G, (p.Met1Val)	*4	No function
	rs56337013	NM_000769.4:c.1297C>T, (p.Arg433Trp)	*5	No function
	rs72552267	NM_000769.4:c.395G>A, (p.Arg132Gln)	*6	No function
	rs41291556	NM_000769.4:c.358T>C, (p.Trp120Arg)	*8	No function
	rs6413438	NM_000769.4:c.680C>T, (p.Pro227Leu)	*10	Decreased function
	rs12248560	NM_000769.4:c.-806C>T, p.?	*17	Increased function
	rs118203757	NM_000769.4: c.1004G>A, (p.Arg335Gln)	*24	No function
SLCO1B1	rs4149056	NM_006446.5:c.521T>C, (:p.Val174Ala)	*5	No function
	rs2306283; rs11045819	NM_006446.5:c.388A>G, (p.Asn130Asp); NM_006446.5:c.463C>A, (p.Pro155Thr)	*14	Increased function (homozygotes only)
	rs2306283; rs4149056	NM_006446.5:c.388A>G, (:p.Asn130Asp); NM_006446.5:c.521T>C, (p.Val174Ala)	*15	No function
	rs59502379	NM_006446.5: c.1463G>C, (p.Gly488Ala)	*9	No function

Only core alleles are listed and as noted in PharmVar (https://www.pharmvar.org/genes). The above are recommended testing in CPIC/ClinPGx/FDA/AMP guidelines. Some normal function star alleles can be displayed on the report when they are associated with affected variants, such examples are SLCO1B1 *37 and CYPC19 *15.

Note: CPIC Guideline for CYP2C9, SLCO1B1co-allele detection can affect Fluvastatin dosage. Refer to PMID: 35152405 for classification of these dosing recommendations.

The amino acid change I331V (rs3758581), not listed above, is part of all CYP2C19 core alleles.