PharmCAT Report

pharmcat.example.hard-filtered

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PharmCAT Version 2.4.0
CPIC Version v1.25.0

Sections

Disclaimer: PharmCAT is only able to generate recommendations based on the information provided to the software. The gene and variant information for all reported sections are interpreted directly from user-supplied data. The user recognizes they are using PharmCAT at their own risk. For a detailed disclaimer see Section IV.

Section I: Genotype Summary

Genotypes called: 18 / 23

Drugs	Gene	Genotypes		
		Genotype	Allele Functionality	Phenotype
allopurinol rosuvastatin	ABCG2 ‡	rs2231142 reference (G)/rs2231142 reference (G)	Two Normal function alleles	Normal Function
desflurane enflurane halothane isoflurane methoxyflurane sevoflurane succinylcholine	CACNA1S † ‡	Reference/Reference	Two Normal function alleles	Uncertain Susceptibility
ivacaftor	CFTR ‡	No CPIC variants found	Two ivacaftor non-responsive alleles	ivacaftor non-responsive in CF patients
efavirenz sertraline	CYP2B6 † ‡	*1/*1	Two Normal function alleles	Normal Metabolizer
		Genotype based on missing variant input *.		
amitriptyline citalopram clomipramine clopidogrel dexlansoprazole doxepin escitalopram imipramine lansoprazole omeprazole pantoprazole sertraline trimipramine voriconazole	CYP2C19 † ‡	*38/*38	Two Normal function alleles	Normal Metabolizer
celecoxib flurbiprofen fluvastatin fosphenytoin	CYP2C9 † ‡	*1/*1	Two Normal function alleles	Normal Metabolizer

Drugs	Gene	Genotypes		
		Genotype	Allele Functionality	Phenotype
ibuprofen lornoxicam meloxicam phenytoin piroxicam siponimod tenoxicam warfarin				
quetiapine	CYP3A4 † ‡	*1/*1	Two Normal function alleles	Normal Metabolizer
tacrolimus	CYP3A5 † ‡	*1/*1	Two Normal function alleles	Normal Metabolizer
warfarin	CYP4F2 † ‡	*1/*1 N/A N/A		
capecitabine flucytosine fluorouracil tegafur	DPYD †	**Reference/Reference**		
		Reference	Normal function	See drug section
aminosalicylic acid aspirin chloramphenicol chloroquine ciprofloxacin dapsone dimercaprol doxorubicin furazolidone glyburide hydroxychloroquine mafenide methylene blue nalidixic acid nitrofurantoin norfloxacin ofloxacin pegloticase phenazopyridine primaquine quinine rasburicase sulfadiazine sulfadimidine sulfamethoxazole / trimethoprim sulfanilamide sulfasalazine sulfisoxazole tafenoquine tolbutamide toluidine blue vitamin c vitamin k	G6PD † ‡	B (reference)/B (reference)	Two IV/Normal alleles	Normal
peginterferon alfa-2a peginterferon alfa-2b	IFNL3/4 ‡	rs12979860 reference (C)/ rs12979860 reference (C)	N/A	N/A
azathioprine mercaptopurine thioguanine	NUDT15 † ‡	*1/*1	Two Normal function alleles	Normal Metabolizer

Drugs	Gene	Genotypes		
		Genotype	Allele Functionality	Phenotype
desflurane enflurane halothane isoflurane methoxyflurane sevoflurane succinylcholine	RYR1 † ‡	Reference/Reference	Two Normal function alleles	Uncertain Susceptibility
atorvastatin fluvastatin lovastatin pitavastatin pravastatin rosuvastatin simvastatin	SLCO1B1 † ‡	*1/*1	Two Normal function alleles	Normal Function
azathioprine mercaptopurine thioguanine	TPMT † ‡	*1/*1	Two Normal function alleles	Normal Metabolizer
atazanavir irinotecan	UGT1A1 † ‡	*1/*1	Two Normal function alleles	Normal Metabolizer
acenocoumarol phenprocoumon warfarin	VKORC1 ‡	rs9923231 reference (C)/rs9923231 reference (C)	N/A	N/A

* Some alleles were not considered for the genotype calls due to missing variant information. Please see Section III for details. Alleles that could not be considered due to missing input might change the metabolizer phenotype and possible recommendation.

† Check Section III for more details about this call.

‡ PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

For a full list of disclaimers and limitations see the Section IV.

Multiple DPWG versions used to generate gene and drug reports: [2023-04-05-11-38, v1.25.0].

Section II: Prescribing Recommendations

acenocoumarol

Guideline	Genes	Implications	Recommendation	Comments	Classification
PharmGKB-DPWG [1] Population: N/A	Genotype VKORC1: rs9923231 reference (C)/ rs9923231 reference (C)	VKORC1: The guideline does not provide a description of the impact of the -1639 GG genotype on acenocoumarol.	The guideline does not provide a recommendation for acenocoumarol in patients with the VKORC1 rs9923231 CC genotype (-1639 GG genotype).	N/A	N/A

Citations:

• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232

allopurinol

Guideline	Genes	Implications	Recommendation	Comments	Classification
PharmGKB-DPWG [1] Population: N/A	Genotype ABCG2: rs2231142 reference (G)/ rs2231142 reference (G)	ABCG2: The guideline does not provide a description of the impact of the ABCG2 rs2231142 GG genotype (c. 421CC; p.141QQ) on allopurinol.	The guideline does not provide a recommendation for allopurinol in patients with the the ABCG2 rs2231142 GG genotype (c.421CC; p. 141QQ)	N/A	N/A

Citations:

• Dutch pharmacogenetics working group guideline for the gene-drug interaction of ABCG2, HLA-B and Allopurinol, and MTHFR, folic acid and methotrexate. *European journal of human genetics : EJHG*. 2022. PMID:36056234

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aminosalicylic acid

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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amitriptyline

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C19:*38/*38; CYP2D6:Unknown/Unknown Phenotypes CYP2C19: Normal Metabolizer CYP2D6: No Result Activity Scores CYP2C19: N/A CYP2D6: No Result	• CYP2C19: Normal metabolism of tertiary amines • CYP2D6: N/A	Initiate therapy with recommended starting dose.	Patients may receive an initial low dose of a tricyclic, which is then increased over several days to the recommended steady-state dose. The starting dose in this guideline refers to the recommended steady-state dose.	Strong

Citations:

• Clinical Pharmacogenetics Implementation Consortium Guideline for CYP2D6 and CYP2C19 Genotypes and Dosing of Tricyclic Antidepressants. *Clinical pharmacology and therapeutics*. 2013. PMID:23486447
• Clinical Pharmacogenetics Implementation Consortium Guideline (CPIC®) for CYP2D6 and CYP2C19 Genotypes and Dosing of Tricyclic Antidepressants: 2016 Update. *Clinical pharmacology and therapeutics*. 2016. PMID:27997040

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aspirin

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Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: <= 1g per day	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status	aspirin ≤ 1 g/day	Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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atazanavir

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype UGT1A1:*1/*1 Phenotype Normal Metabolizer Activity Score N/A	UGT1A1: Reference UGT1A1 activity; very low likelihood of bilirubin-related discontinuation of atazanavir.	There is no need to avoid prescribing of atazanavir based on UGT1A1 genetic test result. Inform the patient that some patients stop atazanavir because of jaundice (yellow eyes and skin), but that this patient's genotype makes this unlikely (less than about a 1 in 20 chance of stopping atazanavir because of jaundice).	All studies correlating UGT1A1 genotypes with atazanavir adverse events have involved ritonavir boosting. However, concentration-time profiles are equivalent when boosted with either cobicistat or ritonavir (PMID 23532097), and bilirubin-related adverse events including discontinuation of atazanavir occur in a similar percentage of patients prescribed atazanavir with cobicistat or ritonavir (PMID 23532097). Associations between UGT1A1 genotype, bilirubin elevations, and atazanavir/r discontinuation therefore almost certainly translate to atazanavir/	Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
				cobicistat. "reference" function refers to the UGT1A1 allele to which other alleles are compared.	

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for UGT1A1 and Atazanavir Prescribing. *Clinical pharmacology and therapeutics*. 2015. PMID:26417955

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atorvastatin

The SLCO1B1 genotype (star nomenclature) will be displayed if determinable with the provided VCF based on the SLCO1B1 star allele definition published by CPIC and recommendations are provided based on the genotype.

In case no genotype can be determined, recommendations are based on the rs4149056 genotype alone as per guideline. The minor C allele at rs4149056 defines SLCO1B1*5.

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype SLCO1B1:*1/*1; rs4149056:T/T Phenotype Normal Function	SLCO1B1: Typical myopathy risk and statin exposure	Prescribe desired starting dose and adjust doses based on disease-specific guidelines.	The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.	Strong
PharmGKB-DPWG [1]	No annotation for SLCO1B1 *1/*1.				

Citations:

• The Clinical Pharmacogenomics Implementation Consortium: Guideline for SLCO1B1 and Simvastatin-Induced Myopathy. *Clinical pharmacology and therapeutics*. 2012. PMID:22617227
• The Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for SLCO1B1 and simvastatin-induced myopathy: 2014 update. *Clinical pharmacology and therapeutics*. 2014. PMID:24918167
• The Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for SLCO1B1, ABCG2, and CYP2C9 and statin-associated musculoskeletal symptoms. *Clinical pharmacology and therapeutics*. 2022. PMID:35152405

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azathioprine

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype NUDT15:*1/*1; TPMT:*1/*1 Phenotypes NUDT15: Normal Metabolizer TPMT: Normal Metabolizer Activity Scores	• NUDT15: Normal risk of thiopurine-related leukopenia, neutropenia, myelosuppression • TPMT: Lower concentrations of TGN metabolites, higher MeTIMP, this is the ‘normal’ pattern. Normal risk of thiopurine-related	Start with normal starting dose (e.g., 2-3 mg/kg/day) and adjust doses of azathioprine based on disease-specific guidelines. Allow 2 weeks to reach steady-state after each dose adjustment (PMID	Normal starting doses vary by race/ethnicity and treatment regimens. If standard dose is below normal recommended dose, dose reduction might not be recommended for	Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
	NUDT15: N/A TPMT: N/A	leukopenia, neutropenia, myelosuppression.	20354201, 11302950, 15606506).	intermediate metabolizers.	
PharmGKB-DPWG [1, 2] Population: N/A	Genotype NUDT15:*1/*1 Phenotype Normal Metabolizer	NUDT15: The guideline does not provide a description of the impact of a normal metabolizer phenotype on azathioprine.	The guideline does not provide a recommendation for azathioprine in normal metabolizers	N/A	N/A
PharmGKB-DPWG [1, 2] Population: N/A	Genotype TPMT:*1/*1 Phenotype Normal Metabolizer	TPMT: The guideline does not provide a description of the impact of a normal metabolizer phenotype on azathioprine.	The guideline does not provide a recommendation for azathioprine in normal metabolizers.	N/A	N/A

Citations:

• Clinical pharmacogenetics implementation consortium guidelines for thiopurine methyltransferase genotype and thiopurine dosing. *Clinical pharmacology and therapeutics*. 2011. PMID:21270794
• Clinical Pharmacogenetics Implementation Consortium Guidelines for Thiopurine Methyltransferase Genotype and Thiopurine Dosing: 2013 Update. *Clinical pharmacology and therapeutics*. 2013. PMID:23422873
• Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for thiopurine dosing based on TPMT and NUDT15 genotypes: 2018 update. *Clinical pharmacology and therapeutics*. 2018. PMID:30447069
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232

capecitabine

The two lowest activity values (variant activity scores, see CPIC guideline PMID: 29152729) are used for uphased data and the lowest activity value per allele are used for phased data to determine the gene activity score and phenotype type to retrieve prescribing recommendations.

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype DPYD:Reference/Reference Phenotype Normal Metabolizer Activity Score 2.0	DPYD: Normal DPD activity and "normal" risk for fluoropyrimidine toxicity	Based on genotype, there is no indication to change dose or therapy. Use label-recommended dosage and administration.	N/A	Strong
PharmGKB-DPWG [1] Population: N/A	Genotype DPYD:Reference/Reference Activity Score 2	DPYD: The guideline does not provide a description of the impact of a *DPYD* activity score of 2 on capecitabine.	The guideline does not provide a recommendation for capecitabine in patients with a *DPYD* activity score of 2.	N/A	N/A

Citations:

• Clinical Pharmacogenetics Implementation Consortium Guidelines for Dihydropyrimidine Dehydrogenase Genotype and Fluoropyrimidine Dosing. *Clinical pharmacology and therapeutics*. 2013. PMID:23988873
• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Dihydropyrimidine Dehydrogenase Genotype and Fluoropyrimidine Dosing: 2017 Update. *Clinical pharmacology and therapeutics*. 2017. PMID:29152729

• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232
• Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction of DPYD and fluoropyrimidines. *European journal of human genetics : EJHG*. 2019. PMID:31745289

celecoxib

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C9:*1/*1 Phenotype Normal Metabolizer Activity Score 2.0	CYP2C9: Normal metabolism	Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.	N/A	Strong

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C9 and Nonsteroidal Anti-inflammatory Drugs. *Clinical pharmacology and therapeutics*. 2020. PMID:32189324

chloramphenicol

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Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

chloroquine

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Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

ciprofloxacin

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Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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citalopram

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C19:*38/*38 Phenotype Normal Metabolizer Activity Score N/A	CYP2C19: Normal metabolism	Initiate therapy with recommended starting dose	N/A	Strong
PharmGKB-DPWG [1]	No annotation for CYP2C19 *38/*38.				

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6, CYP2C19, CYP2B6, SLC6A4, and HTR2A Genotypes and Serotonin Reuptake Inhibitor Antidepressants. *Clinical pharmacology and therapeutics*. 2023. PMID:37032427
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232
• Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction between CYP2C19 and CYP2D6 and SSRIs. *European journal of human genetics : EJHG*. 2022. PMID:34782755

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clomipramine

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C19:*38/*38; CYP2D6:Unknown/Unknown Phenotypes CYP2C19: Normal Metabolizer CYP2D6: No Result Activity Scores CYP2C19: N/A CYP2D6: No Result	• CYP2C19: Normal metabolism of tertiary amines • CYP2D6: N/A	Initiate therapy with recommended starting dose.	Patients may receive an initial low dose of a tricyclic, which is then increased over several days to the recommended steady-state dose. The starting dose in this guideline refers to the recommended	Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
				steady-state dose.	
PharmGKB-DPWG [1, 2]	No annotation for CYP2C19 *38/*38 and CYP2D6 Unknown/Unknown.				

Citations:

• Clinical Pharmacogenetics Implementation Consortium Guideline for CYP2D6 and CYP2C19 Genotypes and Dosing of Tricyclic Antidepressants. *Clinical pharmacology and therapeutics*. 2013. PMID:23486447
• Clinical Pharmacogenetics Implementation Consortium Guideline (CPIC®) for CYP2D6 and CYP2C19 Genotypes and Dosing of Tricyclic Antidepressants: 2016 Update. *Clinical pharmacology and therapeutics*. 2016. PMID:27997040
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232

clopidogrel

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: CVI ACS PCI	Genotype CYP2C19:*38/*38 Phenotype Normal Metabolizer	CYP2C19: Normal clopidogrel active metabolite formation; normal on-treatment platelet reactivity	If considering clopidogrel, use at standard dose (75 mg/day)	For cardiovascular indications of acute coronary syndrome (ACS) and/or percutaneous coronary intervention (PCI). ACS and/or PCI includes patients undergoing PCI for an ACS or non-ACS (elective) indication.	Strong
CPIC [1] Population: CVI non-ACS non-PCI	Genotype CYP2C19:*38/*38 Phenotype Normal Metabolizer	CYP2C19: Normal clopidogrel active metabolite formation; normal on-treatment platelet reactivity	If considering clopidogrel, use at standard dose (75 mg/day)	For non-acute coronary syndrome (non-ACS) and non-percutaneous coronary intervention (non-PCI) cardiovascular indications. Non-ACS, non-PCI cardiovascular indications include peripheral arterial disease and stable coronary artery disease following a recent myocardial infarction outside the setting of PCI.	Strong
CPIC [1] Population: NVI	Genotype CYP2C19:*38/*38 Phenotype Normal Metabolizer	CYP2C19: Normal clopidogrel active metabolite formation; normal on-treatment platelet reactivity	If considering clopidogrel, use at standard dose (75 mg/day)	For neurovascular indications. Neurovascular disease includes acute ischemic stroke or transient ischemic attack, secondary prevention of stroke, or prevention of thromboembolic events following neurointerventional procedures such as carotid artery stenting and stent-assisted coiling of	Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
				intracranial aneurysms.	
PharmGKB-DPWG [1]	No annotation for CYP2C19 *38/*38.				

Citations:

• Clinical Pharmacogenetics Implementation Consortium Guidelines for Cytochrome P450-2C19 (CYP2C19) Genotype and Clopidogrel Therapy. *Clinical pharmacology and therapeutics*. 2011. PMID:21716271
• Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for cytochrome P450-2C19 (CYP2C19) genotype and clopidogrel therapy: 2013 Update. *Clinical pharmacology and therapeutics*. 2013. PMID:23698643
• Clinical Pharmacogenetics Implementation Consortium Guideline for CYP2C19 Genotype and Clopidogrel Therapy: 2022 Update. *Clinical pharmacology and therapeutics*. 2022. PMID:35034351
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232

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dapsone

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Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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desflurane

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CACNA1S:Reference/ Reference; RYR1:Reference/ Reference Phenotypes CACNA1S: Uncertain Susceptibility RYR1: Uncertain Susceptibility Activity Scores CACNA1S: N/A RYR1: N/A	• CACNA1S: These results do not eliminate the chance that this patient is susceptible to malignant hyperthermia (MH). The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID 28902675). • RYR1: These results do not eliminate the chance that this patient is susceptible to malignant hyperthermia (MH). The genetic cause of	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.	N/A	Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
		about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID 28902675).			

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the use of potent volatile anesthetic agents and succinylcholine in the context of RYR1 or CACNA1S genotypes. *Clinical pharmacology and therapeutics*. 2018. PMID:30499100

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dexlansoprazole

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C19:*38/*38 Phenotype Normal Metabolizer Activity Score N/A	CYP2C19: Normal PPI metabolism; may be at increased risk of therapeutic failure compared to CYP2C19 IMs and PMs	Initiate standard starting daily dose. Consider increasing dose by 50-100% for the treatment of H. pylori infection and erosive esophagitis. Daily dose may be given in divided doses. Monitor for efficacy.	N/A	Optional

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C19 and Proton Pump Inhibitor Dosing. *Clinical pharmacology and therapeutics*. 2020. PMID:32770672

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dimercaprol

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Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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doxepin

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1]	Genotype CYP2C19:*38/*38; CYP2D6:Unknown/Unknown	• CYP2C19: Normal metabolism of		Patients may receive an initial low dose of a	Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
Population: general	Phenotypes CYP2C19: Normal Metabolizer CYP2D6: No Result Activity Scores CYP2C19: N/A CYP2D6: No Result	tertiary amines • CYP2D6: N/A	Initiate therapy with recommended starting dose.	tricyclic, which is then increased over several days to the recommended steady-state dose. The starting dose in this guideline refers to the recommended steady-state dose.	

Citations:

• Clinical Pharmacogenetics Implementation Consortium Guideline for CYP2D6 and CYP2C19 Genotypes and Dosing of Tricyclic Antidepressants. *Clinical pharmacology and therapeutics*. 2013. PMID:23486447
• Clinical Pharmacogenetics Implementation Consortium Guideline (CPIC®) for CYP2D6 and CYP2C19 Genotypes and Dosing of Tricyclic Antidepressants: 2016 Update. *Clinical pharmacology and therapeutics*. 2016. PMID:27997040

doxorubicin

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Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

efavirenz

Some position data used to define CYP2B6 alleles is missing which may change the matched genotype. See CYP2B6 in Section III for for more information.

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: child >40kg_adult	Genotype CYP2B6:*1/*1 Phenotype Normal Metabolizer Activity Score N/A	CYP2B6: Normal efavirenz metabolism	Initiate efavirenz with standard dosing (600 mg/day)	The ENCORE study showed that in treatment-naïve patients randomized to initiate efavirenz-based regimens (combined with tenofovir and emtricitabine), 400 mg/day was non-inferior to 600 mg/ day regardless of	Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
				CYP2B6 genotype (PMID 24522178).	
PharmGKB-DPWG [1] Population: N/A	Genotype CYP2B6:*1/*1 Phenotype Normal Metabolizer	CYP2B6: The guideline does not provide a description of the impact of a normal metabolizer phenotype on efavirenz.	The guideline does not provide a recommendation for efavirenz in normal metabolizers.	N/A	N/A

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2B6 and Efavirenz-containing Antiretroviral Therapy. *Clinical pharmacology and therapeutics*. 2019. PMID:31006110

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enflurane

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CACNA1S:Reference/Reference; RYR1:Reference/Reference Phenotypes CACNA1S: Uncertain Susceptibility RYR1: Uncertain Susceptibility Activity Scores CACNA1S: N/A RYR1: N/A	• CACNA1S: These results do not eliminate the chance that this patient is susceptible to malignant hyperthermia (MH). The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID 28902675). • RYR1: These results do not eliminate the chance that this patient is susceptible to malignant hyperthermia (MH). The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID 28902675).	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.	N/A	Strong

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the use of potent volatile anesthetic agents and succinylcholine in the context of RYR1 or CACNA1S genotypes. *Clinical pharmacology and therapeutics*. 2018. PMID:30499100

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escitalopram

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C19:*38/*38 Phenotype Normal Metabolizer Activity Score N/A	CYP2C19: Normal metabolism	Initiate therapy with recommended starting dose	N/A	Strong
PharmGKB-DPWG [1]	No annotation for CYP2C19 *38/*38.				

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6, CYP2C19, CYP2B6, SLC6A4, and HTR2A Genotypes and Serotonin Reuptake Inhibitor Antidepressants. *Clinical pharmacology and therapeutics*. 2023. PMID:37032427
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232
• Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction between CYP2C19 and CYP2D6 and SSRIs. *European journal of human genetics : EJHG*. 2022. PMID:34782755

flucytosine

The two lowest activity values (variant activity scores, see CPIC guideline PMID: 29152729) are used for uphased data and the lowest activity value per allele are used for phased data to determine the gene activity score and phenotype type to retrieve prescribing recommendations.

Guideline	Genes	Implications	Recommendation	Comments	Classification
PharmGKB-DPWG [1] Population: N/A	Genotype DPYD:Reference/Reference Activity Score 2	DPYD: The guideline does not provide a description of the impact of a DPYD activity score of 2 on flucytosine.	The guideline does not provide a recommendation for flucytosine in patients with a DPYD activity score of 2.	N/A	N/A

fluorouracil

The two lowest activity values (variant activity scores, see CPIC guideline PMID: 29152729) are used for uphased data and the lowest activity value per allele are used for phased data to determine the gene activity score and phenotype type to retrieve prescribing recommendations.

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype DPYD:Reference/Reference Phenotype Normal Metabolizer Activity Score 2.0	DPYD: Normal DPD activity and "normal" risk for fluoropyrimidine toxicity	Based on genotype, there is no indication to change dose or therapy. Use label-recommended dosage and administration.	N/A	Strong
PharmGKB-DPWG [1]	Genotype DPYD:Reference/Reference	DPYD: The guideline does not provide a description of the	The guideline does not provide a recommendation for	N/A	N/A

Guideline	Genes	Implications	Recommendation	Comments	Classification
Population: N/A	Activity Score 2	impact of a DPYD activity score of 2 on fluorouracil.	fluorouracil in patients with a DPYD activity score of 2.		

Citations:

• Clinical Pharmacogenetics Implementation Consortium Guidelines for Dihydropyrimidine Dehydrogenase Genotype and Fluoropyrimidine Dosing. *Clinical pharmacology and therapeutics*. 2013. PMID:23988873
• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Dihydropyrimidine Dehydrogenase Genotype and Fluoropyrimidine Dosing: 2017 Update. *Clinical pharmacology and therapeutics*. 2017. PMID:29152729
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232
• Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction of DPYD and fluoropyrimidines. *European journal of human genetics : EJHG*. 2019. PMID:31745289

flurbiprofen

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C9:*1/*1 Phenotype Normal Metabolizer Activity Score 2.0	CYP2C9: Normal metabolism	Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.	N/A	Strong

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C9 and Nonsteroidal Anti-inflammatory Drugs. *Clinical pharmacology and therapeutics*. 2020. PMID:32189324

fluvastatin

The SLCO1B1 genotype (star nomenclature) will be displayed if determinable with the provided VCF based on the SLCO1B1 star allele definition published by CPIC and recommendations are provided based on the genotype.

In case no genotype can be determined, recommendations are based on the rs4149056 genotype alone as per guideline. The minor C allele at rs4149056 defines SLCO1B1*5.

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C9:*1/*1; SLCO1B1:*1/*1; rs4149056:T/T Phenotypes CYP2C9: Normal Metabolizer SLCO1B1: Normal Function Activity Score 2.0	• CYP2C9: Normal exposure. • SLCO1B1: Typical myopathy risk and statin exposure.	Prescribe desired starting dose and adjust doses of fluvastatin based on disease-specific guidelines.	The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.	Strong

Citations:

• The Clinical Pharmacogenomics Implementation Consortium: Guideline for SLCO1B1 and Simvastatin-Induced Myopathy. *Clinical pharmacology and therapeutics*. 2012. PMID:22617227

• The Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for SLCO1B1 and simvastatin-induced myopathy: 2014 update. *Clinical pharmacology and therapeutics*. 2014. PMID:24918167
• The Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for SLCO1B1, ABCG2, and CYP2C9 and statin-associated musculoskeletal symptoms. *Clinical pharmacology and therapeutics*. 2022. PMID:35152405

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fosphenytoin

The displayed recommendation for CYP2C9 and phenytoin is ONLY valid for non-carriers of the HLA-B*15:02 high-risk allele. PharmCAT Named Allele Matcher does not determine HLA status. CPIC guidance: Fos-/Phenytoin is contraindicated in individuals with the HLA-B*15:02 variant allele ("HLA-B*15:02-positive") due to significantly increased risk of fos-/phenytoin-induced cutaneous adverse reactions of Stevens-Johnson syndrome (SJS) and toxic epidermal necrolysis (TEN). In HLA-B*15:02 carriers, carbamazepine should not be used as an alternative. Alternative medications such as oxcarbazepine, eslicarbazepine acetate, and lamotrigine have some evidence linking SJS/TEN with the HLA-B*15:02 allele, and thus caution should be used in choosing alternatives to phenytoin.

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1]	No annotation for CYP2C9 *1/*1 and HLA-B Unknown/Unknown.				

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guidelines for CYP2C9 and HLA-B Genotype and Phenytoin Dosing. *Clinical pharmacology and therapeutics*. 2014. PMID:25099164
• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C9 and HLA-B Genotypes and Phenytoin Dosing: 2020 Update. *Clinical pharmacology and therapeutics*. 2020. PMID:32779747

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furazolidone

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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glyburide

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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halothane

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CACNA1S:Reference/Reference; RYR1:Reference/Reference Phenotypes CACNA1S: Uncertain Susceptibility RYR1: Uncertain Susceptibility Activity Scores CACNA1S: N/A RYR1: N/A	• CACNA1S: These results do not eliminate the chance that this patient is susceptible to malignant hyperthermia (MH). The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID 28902675). • RYR1: These results do not eliminate the chance that this patient is susceptible to malignant hyperthermia (MH). The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID 28902675).	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.	N/A	Strong

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the use of potent volatile anesthetic agents and succinylcholine in the context of RYR1 or CACNA1S genotypes. *Clinical pharmacology and therapeutics*. 2018. PMID:30499100

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hydroxychloroquine

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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ibuprofen

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C9:*1/*1 Phenotype Normal Metabolizer Activity Score 2.0	CYP2C9: Normal metabolism	Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.	N/A	Strong

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C9 and Nonsteroidal Anti-inflammatory Drugs. *Clinical pharmacology and therapeutics*. 2020. PMID:32189324

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imipramine

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C19:*38/*38; CYP2D6:Unknown/Unknown Phenotypes CYP2C19: Normal Metabolizer CYP2D6: No Result Activity Scores CYP2C19: N/A CYP2D6: No Result	• CYP2C19: Normal metabolism of tertiary amines • CYP2D6: N/A	Initiate therapy with recommended starting dose.	Patients may receive an initial low dose of a tricyclic, which is then increased over several days to the recommended steady-state dose. The starting dose in this guideline refers to the recommended steady-state dose.	Strong
PharmGKB-DPWG [1, 2]	No annotation for CYP2C19 *38/*38 and CYP2D6 Unknown/Unknown.				

Citations:

• Clinical Pharmacogenetics Implementation Consortium Guideline for CYP2D6 and CYP2C19 Genotypes and Dosing of Tricyclic Antidepressants. *Clinical pharmacology and therapeutics*. 2013. PMID:23486447
• Clinical Pharmacogenetics Implementation Consortium Guideline (CPIC®) for CYP2D6 and CYP2C19 Genotypes and Dosing of Tricyclic Antidepressants: 2016 Update. *Clinical pharmacology and therapeutics*. 2016. PMID:27997040
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232

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irinotecan

Alleles determined based on the CPIC UGT1A1 allele definition file due to limited allele definition information in the DPWG UGT1A1 document

Guideline	Genes	Implications	Recommendation	Comments	Classification
PharmGKB-DPWG [1] Population: N/A	Genotype UGT1A1:*1/*1 Phenotype Normal Metabolizer	UGT1A1: The guideline does not provide a description of the impact of a normal metabolizer phenotype on irinotecan.	The guideline does not provide a recommendation for irinotecan in normal metabolizers	N/A	N/A

Citations:

• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232
• Dutch pharmacogenetics working group (DPWG) guideline for the gene-drug interaction between UGT1A1 and irinotecan. *European journal of human genetics : EJHG*. 2022. PMID:36443464

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isoflurane

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CACNA1S:Reference/Reference; RYR1:Reference/Reference Phenotypes CACNA1S: Uncertain Susceptibility RYR1: Uncertain Susceptibility Activity Scores CACNA1S: N/A RYR1: N/A	• CACNA1S: These results do not eliminate the chance that this patient is susceptible to malignant hyperthermia (MH). The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID 28902675). • RYR1: These results do not eliminate the chance that this patient is susceptible to malignant hyperthermia (MH). The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID 28902675).	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.	N/A	Strong

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the use of potent volatile anesthetic agents and succinylcholine in the context of RYR1 or CACNA1S genotypes. *Clinical pharmacology and therapeutics*. 2018. PMID:30499100

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ivacaftor

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CFTR: No CPIC variants found Phenotype ivacaftor non-responsive in CF patients Activity Score N/A	CFTR: An individual diagnosed with cystic fibrosis (CF) and negative for a CFTR variant listed in the FDA-approved drug label as being responsive to ivacaftor.	Ivacaftor is not recommended	N/A	Moderate

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guidelines for Ivacaftor Therapy in the Context of CFTR Genotype. *Clinical pharmacology and therapeutics*. 2014. PMID:24598717

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lansoprazole

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C19:*38/*38 Phenotype Normal Metabolizer Activity Score N/A	CYP2C19: Normal PPI metabolism; may be at increased risk of therapeutic failure compared to CYP2C19 IMs and PMs	Initiate standard starting daily dose. Consider increasing dose by 50-100% for the treatment of H. pylori infection and erosive esophagitis. Daily dose may be given in divided doses. Monitor for efficacy.	N/A	Moderate
PharmGKB-DPWG [1]	No annotation for CYP2C19 *38/*38.				

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C19 and Proton Pump Inhibitor Dosing. *Clinical pharmacology and therapeutics*. 2020. PMID:32770672
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232

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lornoxicam

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C9:*1/*1 Phenotype Normal Metabolizer Activity Score 2.0	CYP2C9: Normal metabolism	Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.	N/A	Strong

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C9 and Nonsteroidal Anti-inflammatory Drugs. *Clinical pharmacology and therapeutics*. 2020. PMID:32189324

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lovastatin

The SLCO1B1 genotype (star nomenclature) will be displayed if determinable with the provided VCF based on the SLCO1B1 star allele definition published by CPIC and recommendations are provided based on the genotype.

In case no genotype can be determined, recommendations are based on the rs4149056 genotype alone as per guideline. The minor C allele at rs4149056 defines SLCO1B1*5.

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype SLCO1B1:*1/*1; rs4149056:T/T Phenotype Normal Function	SLCO1B1: Typical myopathy risk and statin exposure	Prescribe desired starting dose and adjust doses based on disease-specific guidelines.	The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.	Strong

Citations:

• The Clinical Pharmacogenomics Implementation Consortium: Guideline for SLCO1B1 and Simvastatin-Induced Myopathy. *Clinical pharmacology and therapeutics*. 2012. PMID:22617227
• The Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for SLCO1B1 and simvastatin-induced myopathy: 2014 update. *Clinical pharmacology and therapeutics*. 2014. PMID:24918167
• The Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for SLCO1B1, ABCG2, and CYP2C9 and statin-associated musculoskeletal symptoms. *Clinical pharmacology and therapeutics*. 2022. PMID:35152405

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mafenide

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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meloxicam

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1]	Genotype CYP2C9:*1/*1	CYP2C9: Normal metabolism	Initiate therapy with recommended starting dose. In	N/A	Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
Population: general	Phenotype Normal Metabolizer Activity Score 2.0		accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.		

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C9 and Nonsteroidal Anti-inflammatory Drugs. *Clinical pharmacology and therapeutics*. 2020. PMID:32189324

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mercaptopurine

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype NUDT15:*1/*1; TPMT:*1/*1 Phenotypes NUDT15: Normal Metabolizer TPMT: Normal Metabolizer Activity Scores NUDT15: N/A TPMT: N/A	• NUDT15: Normal risk of thiopurine-related leukopenia, neutropenia, myelosuppression • TPMT: Lower concentrations of TGN metabolites, higher MeTIMP, this is the 'normal' pattern. Normal risk of thiopurine-related leukopenia, neutropenia, myelosuppression.	Start with normal starting dose (e.g., 75 mg/m2/day or 1.5 mg/kg/day) and adjust doses of mercaptopurine (and of any other myelosuppressive therapy) without any special emphasis on mercaptopurine compared to other agents. Allow at least 2 weeks to reach steady-state after each dose adjustment (PMID 20354201, 16401827, 11302950).	Normal starting doses vary by race/ethnicity and treatment regimens. If standard dose is below normal recommended dose, dose reduction might not be recommended for intermediate metabolizers.	Strong
PharmGKB-DPWG [1, 2] Population: N/A	Genotype NUDT15:*1/*1 Phenotype Normal Metabolizer	NUDT15: The guideline does not provide a description of the impact of a normal metabolizer phenotype on mercaptopurine.	The guideline does not provide a recommendation for mercaptopurine in normal metabolizers	N/A	N/A
PharmGKB-DPWG [1, 2] Population: N/A	Genotype TPMT:*1/*1 Phenotype Normal Metabolizer	TPMT: The guideline does not provide a description of the impact of a normal metabolizer phenotype on mercaptopurine.	The guideline does not provide a recommendation for mercaptopurine in normal metabolizers.	N/A	N/A

Citations:

• Clinical pharmacogenetics implementation consortium guidelines for thiopurine methyltransferase genotype and thiopurine dosing. *Clinical pharmacology and therapeutics*. 2011. PMID:21270794
• Clinical Pharmacogenetics Implementation Consortium Guidelines for Thiopurine Methyltransferase Genotype and Thiopurine Dosing: 2013 Update. *Clinical pharmacology and therapeutics*. 2013. PMID:23422873
• Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for thiopurine dosing based on TPMT and NUDT15 genotypes: 2018 update. *Clinical pharmacology and therapeutics*. 2018. PMID:30447069
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232

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methoxyflurane

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CACNA1S:Reference/ Reference; RYR1:Reference/ Reference Phenotypes CACNA1S: Uncertain Susceptibility RYR1: Uncertain Susceptibility Activity Scores CACNA1S: N/A RYR1: N/A	• CACNA1S: These results do not eliminate the chance that this patient is susceptible to malignant hyperthermia (MH). The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID 28902675). • RYR1: These results do not eliminate the chance that this patient is susceptible to malignant hyperthermia (MH). The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID 28902675).	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.	N/A	Strong

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the use of potent volatile anesthetic agents and succinylcholine in the context of RYR1 or CACNA1S genotypes. *Clinical pharmacology and therapeutics*. 2018. PMID:30499100

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methylene blue

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low risk of acute hemolytic anemia	No reason to avoid ased on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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nalidixic acid

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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nitrofurantoin

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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norfloxacin

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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ofloxacin

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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omeprazole

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C19:*38/*38 Phenotype Normal Metabolizer Activity Score N/A	CYP2C19: Normal PPI metabolism; may be at increased risk of therapeutic failure compared to CYP2C19 IMs and PMs	Initiate standard starting daily dose. Consider increasing dose by 50-100% for the treatment of H. pylori infection and erosive esophagitis. Daily dose may be given in divided doses. Monitor for efficacy.	N/A	Moderate
PharmGKB-DPWG [1]	No annotation for CYP2C19 *38/*38.				

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C19 and Proton Pump Inhibitor Dosing. *Clinical pharmacology and therapeutics*. 2020. PMID:32770672
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232

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pantoprazole

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C19:*38/*38 Phenotype Normal Metabolizer Activity Score N/A	CYP2C19: Normal PPI metabolism; may be at increased risk of therapeutic failure compared to CYP2C19 IMs and PMs	Initiate standard starting daily dose. Consider increasing dose by 50-100% for the treatment of H. pylori infection and erosive esophagitis. Daily dose may be given in divided doses. Monitor for efficacy.	N/A	Moderate
PharmGKB-DPWG [1]	No annotation for CYP2C19 *38/*38.				

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C19 and Proton Pump Inhibitor Dosing. *Clinical pharmacology and therapeutics*. 2020. PMID:32770672
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232

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peginterferon alfa-2a

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1]	No annotation for IFNL3/4 rs12979860 reference (C)/rs12979860 reference (C).				

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for IFNL3 (IL28B) genotype and peginterferon alpha based regimens. *Clinical pharmacology and therapeutics*. 2013. PMID:24096968

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peginterferon alfa-2b

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1]	No annotation for IFNL3/4 rs12979860 reference (C)/rs12979860 reference (C).				

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for IFNL3 (IL28B) genotype and peginterferon alpha based regimens. *Clinical pharmacology and therapeutics*. 2013. PMID:24096968

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pegloticase

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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phenazopyridine

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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phenprocoumon

Guideline	Genes	Implications	Recommendation	Comments	Classification
PharmGKB-DPWG [1] Population: N/A	Genotype VKORC1: rs9923231 reference (C)/ rs9923231 reference (C)	VKORC1: The guideline does not provide a description of the impact of the VKORC1 rs9923231 CC genotype (-1639 GG genotype) on phenprocoumon.	The guideline does not provide a recommendation for phenprocoumon in patients with the VKORC1 rs9923231 CC genotype (-1639 GG genotype).	N/A	N/A

Citations:

• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232

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phenytoin

The displayed recommendation for CYP2C9 and phenytoin is ONLY valid for non-carriers of the HLA-B*15:02 high-risk allele. PharmCAT Named Allele Matcher does not determine HLA status. CPIC guidance: Fos-/Phenytoin is contraindicated in individuals with the HLA-B*15:02 variant allele ("HLA-B*15:02-positive") due to significantly increased risk of fos-/phenytoin-induced cutaneous adverse reactions of Stevens-Johnson syndrome (SJS) and toxic epidermal necrolysis (TEN). In HLA-B*15:02 carriers, carbamazepine should not be used as an alternative. Alternative medications such as oxcarbazepine, eslicarbazepine acetate, and lamotrigine have some evidence linking SJS/TEN with the HLA-B*15:02 allele, and thus caution should be used in choosing alternatives to phenytoin.

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1]	No annotation for CYP2C9 *1/*1 and HLA-B Unknown/Unknown.				
PharmGKB-DPWG [1] Population: N/A	Genotype CYP2C9:*1/*1 Phenotype Normal Metabolizer	CYP2C9: The guideline does not provide a description of the impact of a normal metabolizer phenotype on phenytoin.	The guideline does not provide a recommendation for phenytoin in normal metabolizers.	N/A	N/A

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guidelines for CYP2C9 and HLA-B Genotype and Phenytoin Dosing. *Clinical pharmacology and therapeutics*. 2014. PMID:25099164
• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C9 and HLA-B Genotypes and Phenytoin Dosing: 2020 Update. *Clinical pharmacology and therapeutics*. 2020. PMID:32779747
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232

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piroxicam

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C9:*1/*1 Phenotype Normal Metabolizer Activity Score 2.0	CYP2C9: Normal metabolism	Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.	N/A	Strong

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C9 and Nonsteroidal Anti-inflammatory Drugs. *Clinical pharmacology and therapeutics*. 2020. PMID:32189324

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pitavastatin

The SLCO1B1 genotype (star nomenclature) will be displayed if determinable with the provided VCF based on the SLCO1B1 star allele definition published by CPIC and recommendations are provided based on the genotype.

In case no genotype can be determined, recommendations are based on the rs4149056 genotype alone as per guideline. The minor C allele at rs4149056 defines SLCO1B1*5.

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1]	Genotype SLCO1B1:*1/*1; rs4149056:T/T		Prescribe desired starting dose and adjust doses	The potential for drug-drug	Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
Population: general	Phenotype Normal Function	SLCO1B1: Typical myopathy risk and statin exposure	based on disease-specific guidelines.	interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.	

Citations:

• The Clinical Pharmacogenomics Implementation Consortium: Guideline for SLCO1B1 and Simvastatin-Induced Myopathy. *Clinical pharmacology and therapeutics*. 2012. PMID:22617227
• The Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for SLCO1B1 and simvastatin-induced myopathy: 2014 update. *Clinical pharmacology and therapeutics*. 2014. PMID:24918167
• The Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for SLCO1B1, ABCG2, and CYP2C9 and statin-associated musculoskeletal symptoms. *Clinical pharmacology and therapeutics*. 2022. PMID:35152405

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pravastatin

The SLCO1B1 genotype (star nomenclature) will be displayed if determinable with the provided VCF based on the SLCO1B1 star allele definition published by CPIC and recommendations are provided based on the genotype.

In case no genotype can be determined, recommendations are based on the rs4149056 genotype alone as per guideline. The minor C allele at rs4149056 defines SLCO1B1*5.

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype SLCO1B1:*1/*1; rs4149056:T/T Phenotype Normal Function	SLCO1B1: Typical myopathy risk and statin exposure	Prescribe desired starting dose and adjust doses based on disease-specific guidelines.	The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.	Strong

Citations:

• The Clinical Pharmacogenomics Implementation Consortium: Guideline for SLCO1B1 and Simvastatin-Induced Myopathy. *Clinical pharmacology and therapeutics*. 2012. PMID:22617227
• The Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for SLCO1B1 and simvastatin-induced myopathy: 2014 update. *Clinical pharmacology and therapeutics*. 2014. PMID:24918167
• The Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for SLCO1B1, ABCG2, and CYP2C9 and statin-associated musculoskeletal symptoms. *Clinical pharmacology and therapeutics*. 2022. PMID:35152405

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primaquine

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype	G6PD: Low risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
	Normal				

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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quetiapine

The CYP3A4 alleles are determined based on PharmVar CYP3A4 allele definitions. See PharmCAT disclaimer for further information.

Guideline	Genes	Implications	Recommendation	Comments	Classification
PharmGKB-DPWG [1] Population: N/A	Genotype CYP3A4:*1/*1 Phenotype Normal Metabolizer	CYP3A4: The guideline does not provide a description of the impact of a normal metabolizer phenotype on quetiapine.	The guideline does not provide a recommendation for quetiapine in normal metabolizers.	N/A	N/A

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quinine

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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rasburicase

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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rosuvastatin

The SLCO1B1 genotype (star nomenclature) will be displayed if determinable with the provided VCF based on the SLCO1B1 star allele definition published by CPIC and recommendations are provided based on the genotype.

In case no genotype can be determined, recommendations are based on the rs4149056 genotype alone as per guideline. The minor C allele at rs4149056 defines SLCO1B1*5.

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype ABCG2: rs2231142 reference (G)/ rs2231142 reference (G); SLCO1B1:*1/*1; rs4149056:T/T Phenotypes ABCG2: Normal Function SLCO1B1: Normal Function	• ABCG2: Typical myopathy risk and rosuvastatin exposure • SLCO1B1: Typical myopathy risk and statin exposure	Prescribe desired starting dose and adjust doses of rosuvastatin based on disease-specific and specific population guidelines.	The potential for drug-drug interactions and dose limits based on renal and hepatic function and Asian ancestry should be evaluated prior to initiating rosuvastatin.	Strong

Citations:

• The Clinical Pharmacogenomics Implementation Consortium: Guideline for SLCO1B1 and Simvastatin-Induced Myopathy. *Clinical pharmacology and therapeutics*. 2012. PMID:22617227
• The Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for SLCO1B1 and simvastatin-induced myopathy: 2014 update. *Clinical pharmacology and therapeutics*. 2014. PMID:24918167
• The Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for SLCO1B1, ABCG2, and CYP2C9 and statin-associated musculoskeletal symptoms. *Clinical pharmacology and therapeutics*. 2022. PMID:35152405

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sertraline

Some position data used to define CYP2B6 alleles is missing which may change the matched genotype. See CYP2B6 in Section III for for more information.

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2B6:*1/*1; CYP2C19:*38/*38 Phenotypes CYP2B6: Normal Metabolizer CYP2C19: Normal Metabolizer Activity Scores CYP2B6: N/A CYP2C19: N/A	• CYP2B6: Normal metabolism of sertraline to less active compounds. • CYP2C19: Normal metabolism	Initiate therapy with recommended starting dose.	N/A	Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
PharmGKB-DPWG [1]	No annotation for CYP2C19 *38/*38.				

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2D6, CYP2C19, CYP2B6, SLC6A4, and HTR2A Genotypes and Serotonin Reuptake Inhibitor Antidepressants. *Clinical pharmacology and therapeutics*. 2023. PMID:37032427
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232
• Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction between CYP2C19 and CYP2D6 and SSRIs. *European journal of human genetics : EJHG*. 2022. PMID:34782755

sevoflurane

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CACNA1S:Reference/Reference; RYR1:Reference/Reference Phenotypes CACNA1S: Uncertain Susceptibility RYR1: Uncertain Susceptibility Activity Scores CACNA1S: N/A RYR1: N/A	• CACNA1S: These results do not eliminate the chance that this patient is susceptible to malignant hyperthermia (MH). The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID 28902675). • RYR1: These results do not eliminate the chance that this patient is susceptible to malignant hyperthermia (MH). The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID 28902675).	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.	N/A	Strong

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the use of potent volatile anesthetic agents and succinylcholine in the context of RYR1 or CACNA1S genotypes. *Clinical pharmacology and therapeutics*. 2018. PMID:30499100

simvastatin

The SLCO1B1 genotype (star nomenclature) will be displayed if determinable with the provided VCF based on the SLCO1B1 star allele definition published by CPIC and recommendations are provided based on the genotype.

In case no genotype can be determined, recommendations are based on the rs4149056 genotype alone as per guideline. The minor C allele at rs4149056 defines SLCO1B1*5.

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype SLCO1B1:*1/*1; rs4149056:T/T Phenotype Normal Function	SLCO1B1: Typical myopathy risk and statin exposure	Prescribe desired starting dose and adjust doses based on disease-specific guidelines.	The potential for drug-drug interactions and dose limits based on renal and hepatic function and ancestry should be evaluated prior to initiating a statin.	Strong
PharmGKB-DPWG [1]	No annotation for SLCO1B1 *1/*1.				

Citations:

• The Clinical Pharmacogenomics Implementation Consortium: Guideline for SLCO1B1 and Simvastatin-Induced Myopathy. *Clinical pharmacology and therapeutics*. 2012. PMID:22617227
• The Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for SLCO1B1 and simvastatin-induced myopathy: 2014 update. *Clinical pharmacology and therapeutics*. 2014. PMID:24918167
• The Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for SLCO1B1, ABCG2, and CYP2C9 and statin-associated musculoskeletal symptoms. *Clinical pharmacology and therapeutics*. 2022. PMID:35152405

siponimod

Guideline	Genes	Implications	Recommendation	Comments	Classification
PharmGKB-DPWG [1] Population: N/A	Genotype CYP2C9:*1/*1 Phenotype Normal Metabolizer	CYP2C9: The guideline does not provide a description of the impact of a normal metabolizer phenotype on siponimod.	The guideline does not provide a recommendation for siponimod in normal metabolizers.	N/A	N/A

succinylcholine

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CACNA1S:Reference/Reference; RYR1:Reference/Reference Phenotypes CACNA1S: Uncertain Susceptibility RYR1: Uncertain Susceptibility Activity Scores CACNA1S: N/A RYR1: N/A	• CACNA1S: These results do not eliminate the chance that this patient is susceptible to malignant hyperthermia (MH). The genetic cause of about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID 28902675). • RYR1: These results do not eliminate the chance that this patient is susceptible to malignant hyperthermia (MH). The genetic cause of	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.	N/A	Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
		about half of all MH survivors, with MH susceptibility confirmed by contracture test, remains unknown (PMID 28902675).			

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the use of potent volatile anesthetic agents and succinylcholine in the context of RYR1 or CACNA1S genotypes. *Clinical pharmacology and therapeutics*. 2018. PMID:30499100

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sulfadiazine

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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sulfadimidine

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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sulfamethoxazole / trimethoprim

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1]					Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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sulfanilamide

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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sulfasalazine

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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sulfisoxazole

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

tacrolimus

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP3A5:*1/*1 Phenotype Normal Metabolizer Activity Score N/A	CYP3A5: Lower dose-adjusted trough concentrations of tacrolimus and decreased chance of achieving target tacrolimus concentrations.	Increase starting dose 1.5 to 2 times recommended starting dose. Total starting dose should not exceed 0.3 mg/kg/day. Use therapeutic drug monitoring to guide dose adjustments.	This recommendation includes the use of tacrolimus in kidney, heart, lung and hematopoietic stem cell transplant patients, and liver transplant patients where the donor and recipient genotypes are identical. Further dose adjustments or selection of alternative therapy may be necessary due to other clinical factors (e.g., medication interactions, or hepatic function). Typically with other CYP enzymes, a normal metabolizer would be classified as having normal metabolism, and therefore, the drug dose would not change based on the patient's genotype. However, in the case of CYP3A5 and tacrolimus, a CYP3A5 expresser (i.e., CYP3A5 normal metabolizer or intermediate metabolizer) would require a higher recommended starting dose, and the CYP3A5 non-expresser (i.e., poor metabolizer) would require the standard recommended starting dose.	Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
PharmGKB-DPWG [1] Population: N/A	Genotype CYP3A5:*1/*1	CYP3A5: An increase of the initial dose can result in an increased chance of reaching a tacrolimus concentration within the target range before the start of therapeutic drug monitoring. However, there is no direct evidence that this results in improved clinical results. The genetic variation results in an increased conversion of tacrolimus to inactive metabolites and therefore a higher required dose.	LIVER TRANSPLANTATION In addition to the patient's genotype, the metabolism of tacrolimus is also determined by the genotype of the transplanted liver. LIVER is also of the genotype HOMOZYGOUS EXPRESSOR: Use 2.5 times the normal initial dose. Adjustment of the dose should then be based on therapeutic drug monitoring. LIVER has a DIFFERENT genotype: There is insufficient evidence in the literature to support a dose recommendation. OTHER TRANSPLANTATION Use 2.5 times the initial dose that would yield the desired result in non-expressers. Adjustment of the dose should then be based on therapeutic drug monitoring. For example: One Dutch study found a median trough concentration for tacrolimus after three days of 9.4 ng/mL at an initial dose of 0.15 mg/kg twice daily for 5 homozygous kidney transplant patients. Their target value was 10 - 15 ng/mL.	N/A	N/A

Citations:

• Clinical pharmacogenetics implementation consortium (CPIC) guidelines for CYP3A5 genotype and tacrolimus dosing. *Clinical pharmacology and therapeutics*. 2015. PMID:25801146
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232

tafenoquine

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1]	Genotype G6PD:B (reference)/B (reference)	G6PD: Low risk of acute hemolytic anemia	No reason to avoid based on G6PD status	Tafenoquine's safety has been established for a	Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
Population: general	Phenotype Normal			G6PD enzyme activity ≥70% of normal. (Inclusion criteria for clinical trials involving tafenoquine included G6PD activity ≥70%.)	

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

tegafur

The two lowest activity values (variant activity scores, see CPIC guideline PMID: 29152729) are used for uphased data and the lowest activity value per allele are used for phased data to determine the gene activity score and phenotype type to retrieve prescribing recommendations.

Guideline	Genes	Implications	Recommendation	Comments	Classification
PharmGKB-DPWG [1] Population: N/A	Genotype DPYD:Reference/Reference Activity Score 2	DPYD: The guideline does not provide a description of the impact of a DPYD activity score of 2 on tegafur.	The guideline does not provide a recommendation for tegafur in patients with a DPYD activity score of 2.	N/A	N/A

Citations:

• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232
• Dutch Pharmacogenetics Working Group (DPWG) guideline for the gene-drug interaction of DPYD and fluoropyrimidines. *European journal of human genetics : EJHG*. 2019. PMID:31745289

tenoxicam

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C9:*1/*1 Phenotype Normal Metabolizer Activity Score 2.0	CYP2C9: Normal metabolism	Initiate therapy with recommended starting dose. In accordance with the prescribing information, use the lowest effective dosage for shortest duration consistent with individual patient treatment goals.	N/A	Strong

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2C9 and Nonsteroidal Anti-inflammatory Drugs. *Clinical pharmacology and therapeutics*. 2020. PMID:32189324

thioguanine

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1]					Strong

Guideline	Genes	Implications	Recommendation	Comments	Classification
Population: general	Genotype NUDT15:*1/*1; TPMT:*1/*1 Phenotypes NUDT15: Normal Metabolizer TPMT: Normal Metabolizer Activity Scores NUDT15: N/A TPMT: N/A	• NUDT15: Normal risk of thiopurine-related leukopenia, neutropenia, myelosuppression • TPMT: Lower concentrations of TGN metabolites, but note that TGN after thioguanine are 5-10X higher than TGN after mercaptopurine or azathioprine. Normal risk of thiopurine-related leukopenia, neutropenia, myelosuppression.	Start with normal starting dose (e.g., 40-60 mg/m2/day) and adjust doses of thioguanine and of other myelosuppressive therapy without any special emphasis on thioguanine. Allow 2 weeks to reach steady-state after each dose adjustment (PMID 20354201, 11037857).	Normal starting doses vary by race/ethnicity and treatment regimens. If standard dose is below normal recommended dose, dose reduction might not be recommended for intermediate metabolizers.	
PharmGKB-DPWG [1, 2] Population: N/A	Genotype NUDT15:*1/*1 Phenotype Normal Metabolizer	NUDT15: The guideline does not provide a description of the impact of a normal metabolizer phenotype on thioguanine.	The guideline does not provide a recommendation for thioguanine in normal metabolizers	N/A	N/A
PharmGKB-DPWG [1, 2] Population: N/A	Genotype TPMT:*1/*1 Phenotype Normal Metabolizer	TPMT: The guideline does not provide a description of the impact of a normal metabolizer phenotype on thioguanine.	The guideline does not provide a recommendation for thioguanine in normal metabolizers.	N/A	N/A

Citations:

• Clinical pharmacogenetics implementation consortium guidelines for thiopurine methyltransferase genotype and thiopurine dosing. *Clinical pharmacology and therapeutics*. 2011. PMID:21270794
• Clinical Pharmacogenetics Implementation Consortium Guidelines for Thiopurine Methyltransferase Genotype and Thiopurine Dosing: 2013 Update. *Clinical pharmacology and therapeutics*. 2013. PMID:23422873
• Clinical Pharmacogenetics Implementation Consortium (CPIC) guideline for thiopurine dosing based on TPMT and NUDT15 genotypes: 2018 update. *Clinical pharmacology and therapeutics*. 2018. PMID:30447069
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232

tolbutamide

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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toluidine blue

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low risk of acute hemolytic anemia	No reason to avoid ased on G6PD status	Toluidine blue classification strength is based on extrapolation from methylene blue data	Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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trimipramine

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype CYP2C19:*38/*38; CYP2D6:Unknown/Unknown Phenotypes CYP2C19: Normal Metabolizer CYP2D6: No Result Activity Scores CYP2C19: N/A CYP2D6: No Result	• CYP2C19: Normal metabolism of tertiary amines • CYP2D6: N/A	Initiate therapy with recommended starting dose.	Patients may receive an initial low dose of a tricyclic, which is then increased over several days to the recommended steady-state dose. The starting dose in this guideline refers to the recommended steady-state dose.	Strong

Citations:

• Clinical Pharmacogenetics Implementation Consortium Guideline for CYP2D6 and CYP2C19 Genotypes and Dosing of Tricyclic Antidepressants. *Clinical pharmacology and therapeutics*. 2013. PMID:23486447
• Clinical Pharmacogenetics Implementation Consortium Guideline (CPIC®) for CYP2D6 and CYP2C19 Genotypes and Dosing of Tricyclic Antidepressants: 2016 Update. *Clinical pharmacology and therapeutics*. 2016. PMID:27997040

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vitamin c

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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vitamin k

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: general	Genotype G6PD:B (reference)/ B (reference) Phenotype Normal	G6PD: Low-to-no risk of acute hemolytic anemia	No reason to avoid based on G6PD status		Strong

Citations:

• Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype. *Clinical pharmacology and therapeutics*. 2022. PMID:36049896

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voriconazole

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: adults	Genotype CYP2C19:*38/*38 Phenotype Normal Metabolizer	CYP2C19: Normal voriconazole metabolism	Initiate therapy with recommended standard of care dosing	Further dose adjustments or selection of alternative therapy may be necessary due to other clinical factors, such as drug interactions, hepatic function, renal function, species, site of	Strong

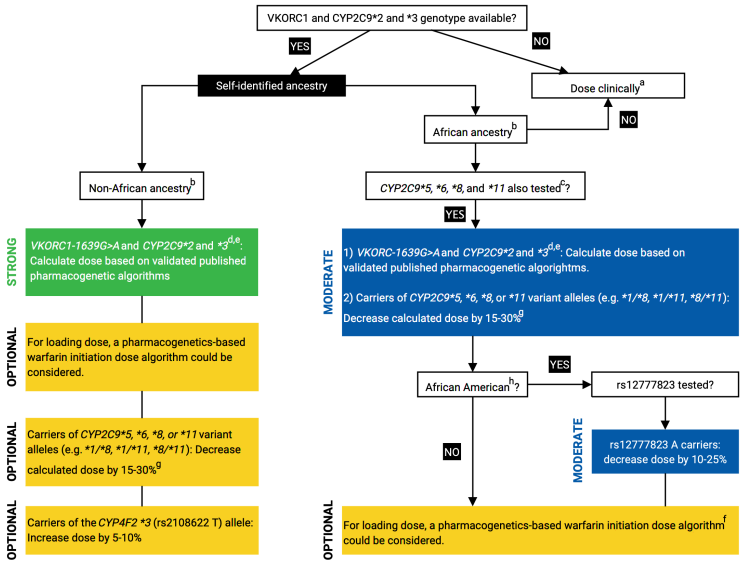
Guideline	Genes	Implications	Recommendation	Comments	Classification
				infection, therapeutic drug monitoring, and comorbidities.	
CPIC [1] Population: pediatrics	Genotype CYP2C19:*38/*38 Phenotype Normal Metabolizer	CYP2C19: Normal voriconazole metabolism	Initiate therapy with recommended standard of care dosing	Further dose adjustments or selection of alternative therapy may be necessary due to other clinical factors, such as drug interactions, hepatic function, renal function, species, site of infection, therapeutic drug monitoring, and comorbidities.	Strong
PharmGKB-DPWG [1]	No annotation for CYP2C19 *38/*38.				

Citations:

• Clinical Pharmacogenetics Implementation Consortium (CPIC®) Guideline for CYP2C19 and Voriconazole Therapy. *Clinical pharmacology and therapeutics*. 2016. PMID:27981572
• Pharmacogenetics: from bench to byte--an update of guidelines. *Clinical pharmacology and therapeutics*. 2011. PMID:21412232

warfarin

Guideline	Genes	Implications	Recommendation	Comments	Classification
CPIC [1] Population: N/A	Genotype CYP2C9:*1/*1; CYP4F2:*1/*1; VKORC1: rs9923231 reference (C)/ rs9923231 reference (C); rs12777823:G/G	Please follow the flow chart in figure 2 of the CPIC warfarin guideline to determine the appropriate dosing recommendation. The CPIC warfarin guideline only considers a single SNV in VKORC1 (rs9923231), which has varying frequency among different ancestral populations, and largely explains the differences in average dose requirements between people of European, African, and Asian descents. While other functional variants in VKORC1 have been associated with warfarin resistance (high dose requirements), there are currently no CPIC recommendations for how to use these other variants in warfarin dosing. An alternate name for rs9923231 is -1639G>A (note that VKORC1 is on the negative chromosomal strand, so displayed alleles are complemented).			

Guideline	Genes	Implications	Recommendation	Comments	Classification
		 **FIGURE 2: DOSING RECOMMENDATIONS FOR WARFARIN DOSING BSED ON GENOTYPE FOR ADULT PATIENTS** (a) "Dose clinically" means to dose without genetic information, which may include use of a clinical dosing algorithm or standard dose approach (b) Data strongest for European and East Asian ancestry populations and consistent in other populations. (c) 45-50% of individuals with self-reported African ancestry carry CYP2C9*5,*6,*8,*11, or rs12777823. IF CYP2C9*5, *6, *8, and *11 WERE NOT TESTED, DOSE WARFARIN CLINICALLY. Note: these data derive primarily from African Americans, who are largely from West Africa. It is unknown the same associations are present for those from other parts of Africa. (d) Most algorithms are developed for the target INR 2-3. (e) Consider an alternative agent in individuals with genotypes associated with CYP2C9 poor metabolism (e.g., CYP2C9*3/*3, *2/*3, *3/*3) or both increased sensitivity (VKORC1 A/G or A/A) and CYP2C9 poor metabolism. (f) See the EU-PACT trial for pharmacogenetics-based warfarin initiation (loading) dose algorithm (33) with the caveat that the loading dose PG algorithm has not been specifically tested or validated in populations of African ancestry. (g) Larger dose reduction might be needed in variant homozygotes (i.e. 20-40%). (h) African American refers to individuals mainly originating from West Africa.			
PharmGKB-DPWG [1, 2] Population: N/A	Genotype CYP2C9:*1/*1 Phenotype Normal Metabolizer	CYP2C9: The guideline does not provide a description of the impact of a normal metabolizer phenotype on warfarin.	The guideline does not provide a recommendation for warfarin in normal metabolizers.	N/A	N/A
PharmGKB-DPWG [1, 2] Population: N/A	Genotype VKORC1: rs9923231 reference (C)/ rs9923231 reference (C)	VKORC1: The guideline does not provide a description of the impact of the VKORC1 rs9923231 CC genotype (-1639 GG genotype) on warfarin.	The guideline does not provide a recommendation for warfarin in patients with the VKORC1 rs9923231 CC genotype (-1639 GG genotype).	N/A	N/A

Citations:

• Clinical Pharmacogenetics Implementation Consortium Guidelines for CYP2C9 and VKORC1 Genotypes and Warfarin Dosing. *Clinical pharmacology and therapeutics*. 2011. PMID:21900891
• Clinical pharmacogenetics implementation consortium (cpic) guideline for pharmacogenetics-guided warfarin dosing: 2017 update. *Clinical pharmacology and therapeutics*. 2017. PMID:28198005

Section III: Allele Matching Details

1. ABCG2 allele match data
2. CACNA1S allele match data
3. CFTR allele match data
4. CYP2B6 allele match data
5. CYP2C19 allele match data

6. CYP2C9 allele match data
7. CYP3A4 allele match data
8. CYP3A5 allele match data
9. CYP4F2 allele match data
10. DPYD allele match data
11. G6PD allele match data
12. IFNL3/4 allele match data
13. NUDT15 allele match data
14. RYR1 allele match data
15. SLCO1B1 allele match data
16. TPMT allele match data
17. UGT1A1 allele match data
18. VKORC1 allele match data

No data provided for CYP2D6, F5, HLA-A, HLA-B, MT-RNR1.

ABCG2 allele match data

Genotype Matched: rs2231142 reference (G)/rs2231142 reference (G)

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr4:88131171	rs2231142	G/G	G	rs2231142 variant (T)	

CACNA1S allele match data

Genotype Matched: Reference/Reference

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

The CACNA1S Reference allele assignment is characterized by the absence of variants at the positions that are included in the underlying allele definitions.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr1:201060815	rs1800559	C/C	C	c.3257G>A	
chr1:201091993	rs772226819	G/G	G	c.520C>T	

CFTR allele match data

Genotype Matched: No CPIC variants found

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr7:117509035	rs397508256	G/G	G	E56K	
chr7:117509069	rs368505753	C/C	C	P67L	
chr7:117509089	rs115545701	C/C	C	R74W	
chr7:117530953	rs113993958	G/G	G	D110H	
chr7:117530955	rs397508537	C/C	C	D110E	
chr7:117530974	rs77834169	C/C	C	R117C	
chr7:117530975	rs78655421	G/G	G	R117H	
chr7:117534318	rs80282562	G/G	G	G178R	
chr7:117534363	rs397508759	G/G	G	E193K	
chr7:117534368	rs397508761	A/A	A	711+3A->G	
chr7:117535285	rs121908752	T/T	T	L206W	
chr7:117540270	rs77932196	G/G	G	R347H	
chr7:117540285	rs121908753	G/G	G	R352Q	
chr7:117548795	rs74551128	C/C	C	A455E	
chr7:117587799	rs121908757	A/A	A	S549R(A>C)	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr7:117587800	rs121908755	G/G	G	S549N	
chr7:117587801	rs121909005	T/T	T	S549R(T>G)	
chr7:117587805	rs121909013	G/G	G	G551S	
chr7:117587806	rs75527207	G/G	G	G551D	
chr7:117590409	rs397508288	A/A	A	D579G	
chr7:117594930	rs397508387	G/G	G	E831X	
chr7:117602868	rs80224560	G/G	G	2789+5G->A	
chr7:117603708	rs397508442	C/C	C	S945L	
chr7:117606695	rs141033578	C/C	C	S977F	
chr7:117611555	rs76151804	A/A	A	3272-26A->G	
chr7:117611595	rs150212784	T/T	T	F1052V	
chr7:117611620	rs397508513	A/A	A	K1060T	
chr7:117611640	rs121909020	G/G	G	A1067T	
chr7:117611646	rs200321110	G/G	G	G1069R	
chr7:117611649	rs202179988	C/C	C	R1070W	
chr7:117611650	rs78769542	G/G	G	R1070Q	
chr7:117611663	rs186045772	T/T	T	F1074L	
chr7:117614699	rs75541969	G/G	G	D1152H	
chr7:117639961	rs75039782	C/C	C	3849+10kbC->T	
chr7:117642451	rs267606723	G/G	G	G1244E	
chr7:117642472	rs74503330	G/G	G	S1251N	
chr7:117642483	rs121909041	T/T	T	S1255P	
chr7:117642528	rs11971167	G/G	G	D1270N	
chr7:117664770	rs193922525	G/G	G	G1349D	

CYP2B6 allele match data

Genotype Matched: *1/*1

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In

	case of unphased data, additional genotypes might be possible and cannot be ruled out.
Alleles Not Considered:	The following alleles are not considered due to 12 missing positions of the total 47 positions: *44, *45, *46, *47, *48, *49 Carriage of these alleles might result in a different phenotype and different guideline recommendations.

The CYP2B6 *1 allele assignment is characterized by the absence of variants at the positions that are included in the underlying allele definitions either because the position is reference or missing.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:40991224	rs34223104	T/T	T	*22 *34 *35 *36	
chr19:40991367	rs34883432	A/A	A	*10	
chr19:40991369	rs8192709	C/C	C	*2 *10	
chr19:40991381	rs33973337	A/A	A	*17	
chr19:40991388	rs33980385	A/A	A	*17	
chr19:40991390	rs33926104	C/C	C	*17	
chr19:40991391	rs34284776	G/G	G	*17	
chr19:40991441	rs35303484	A/A	A	*11	
chr19:41004015	rs281864907	T/T	T	*38	
chr19:41004125	rs36060847	G/G	G	*12	
chr19:41004158	rs186335453	G/G	G	*35	
chr19:41004303	rs139801276	T/T	T	*35	
chr19:41004377	rs12721655	A/A	A	*8 *13	
chr19:41004381	rs35773040	G/G	G	*14	
chr19:41004406	rs145884402	G/G	G	*35	
chr19:41006919	rs3826711	C/C	C	*26	
chr19:41006923	rs36056539	C/C	C	*20	
chr19:41006936	rs3745274	G/G	G		

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
				*6 *7 *9 *13 *19 *20 *26 *34 *36 *37 *38 *39 *40 *41 *42 *43	
chr19:41006968	rs373489637	T/T	T	*37	
chr19:41007013	rs36079186	T/T	T	*27 *35	
chr19:41009350	rs45482602	C/C	C	*3	
chr19:41009358	rs2279343	A/A	A	*4 *6 *7 *13 *18 *19 *20 *26 *34 *36 *37 *38 *39 *40 *41 *42 *43	
chr19:41010006	rs139029625	G/G	G	*35	
chr19:41010088	rs34698757	C/C	C	*28	
chr19:41010108	rs193922917	C/C	C	*31	
chr19:41012316	rs28399499	T/T	T	*18	
chr19:41012339	rs34826503	C/C	C	*19	
chr19:41012465	rs34097093	C/C	C	*28	
chr19:41012693	rs35979566	T/T	T	*15	
chr19:41012740	rs193922918	G/G	G	*32	
chr19:41012803	rs35010098	C/C	C	*21	
chr19:41016726	rs3211369	A/A	A	*23	
chr19:41016778	rs564083989	G/G	G	*24	
chr19:41016805		A/A	A	*25	
chr19:41016810	rs3211371	C/C	C	*5 *7 *33 *34	
chr19:41004133	rs148009906	*Missing*	G	*44	
chr19:41004380	rs535039125	*Missing*	C	*39	
chr19:41006967	rs58871670	*Missing*	G	*45	
chr19:41009313		*Missing*	A	*46	
chr19:41012393	rs754621576	*Missing*	T	*47	
chr19:41012394	rs780991919	*Missing*	A	*47	
chr19:41012466	rs200458614	*Missing*	G	*40	
chr19:41012471	rs201500445	*Missing*	T	*41	
chr19:41012478	rs200238771	*Missing*	T	*48	
chr19:41016652	rs764288403	*Missing*	G	*49	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:41016679	rs374099483	*Missing*	G	*42	
chr19:41016741	rs117872433	*Missing*	G	*43	

CYP2C19 allele match data

Genotype Matched: *38/*38

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

The CYP2C19 *38 allele assignment is characterized by the absence of variants at the positions that are included in the underlying allele definitions.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr10:94761900	rs12248560	C/C	C	*1 *4 *17	
chr10:94762706	rs28399504	A/A	A	*1 *4	
chr10:94762712	rs367543002	C/C	C	*1 *34	
chr10:94762715	rs367543003	T/T	T	*1 *34	
chr10:94762755	rs55752064	T/T	T	*1 *14	
chr10:94762760	rs17882687	A/A	A	*1 *15 *28 *35 *39	
chr10:94762788	rs1564656981	A/A	A	*1 *29	
chr10:94762856	rs1564657013	A/A	A	*1 *19	
chr10:94775106	rs145328984	C/C	C	*1 *30	
chr10:94775121	rs1564660997	C/C	C	*1 *31	
chr10:94775160	rs118203756	G/G	G	*1 *23	
chr10:94775185	rs1288601658	A/A	A	*1 *32	
chr10:94775367	rs12769205	A/A	A	*1 *2 *35	
chr10:94775416	rs41291556	T/T	T	*1 *8	
chr10:94775423	rs17885179	A/A	A	*1 *39	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr10:94775453	rs72552267	G/G	G	*1 *6	
chr10:94775489	rs17884712	G/G	G	*1 *9	
chr10:94775507	rs58973490	G/G	G	*1 *2 *11	
chr10:94780574	rs140278421	G/G	G	*1 *22	
chr10:94780579	rs370803989	G/G	G	*1 *33	
chr10:94780653	rs4986893	G/G	G	*1 *3	
chr10:94781858	rs6413438	C/C	C	*1 *10	
chr10:94781859	rs4244285	G/G	G	*1 *2	
chr10:94781944	rs375781227	G/G	G	*1 *26	
chr10:94781999	rs72558186	T/T	T	*1 *7	
chr10:94842861	rs138142612	G/G	G	*1 *18	
chr10:94842866	rs3758581	A/A	A	*1 *2 *3 *4 *5 *6 *7 *8 *9 *10 *11 *12 *13 *14 *15 *17 *18 *19 *22 *23 *24 *25 *26 *28 *29 *31 *32 *33 *35 *39	
chr10:94842879	rs118203757	G/G	G	*1 *24	
chr10:94842995	rs113934938	G/G	G	*1 *28	
chr10:94849995	rs17879685	C/C	C	*1 *13	
chr10:94852738	rs56337013	C/C	C	*1 *5	
chr10:94852765	rs192154563	C/C	C	*1 *16	
chr10:94852785	rs118203759	C/C	C	*1 *25	
chr10:94852914	rs55640102	A/A	A	*1 *12	

CYP2C9 allele match data

Genotype Matched: *1/*1

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

The CYP2C9 *1 allele assignment is characterized by the absence of variants at the positions that are included in the underlying allele definitions.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr10:94938683	rs114071557	A/A	A	*36	
chr10:94938719		T/T	T	*80	
chr10:94938737	rs67807361	C/C	C	*7	
chr10:94938771	rs142240658	C/C	C	*21	
chr10:94938788		C/C	C	*83	
chr10:94938800	rs1364419386	G/G	G	*76	
chr10:94938803	rs2031308986	A/A	A	*22	
chr10:94938828	rs564813580	A/A	A	*37	
chr10:94941897	rs371055887	G/G	G	*20	
chr10:94941915		G/G	G	*23	
chr10:94941958	rs72558187	T/T	T	*13	
chr10:94941975		G/G	G	*77	
chr10:94941976		G/G	G	*38	
chr10:94941982	rs762239445	G/G	G	*39	
chr10:94942018		T/T	T	*40	
chr10:94942205	rs1304490498	CAATGGAAAGA/CAATGGAAAGA	CAATGGAAAGA	*25	
chr10:94942216	rs774607211	A/A	A	*41	
chr10:94942230	rs767576260	C/C	C	*43	
chr10:94942231	rs12414460	G/G	G	*42	
chr10:94942233	rs375805362	C/C	C	*62	
chr10:94942234	rs72558189	G/G	G	*14 *35	
chr10:94942243	rs1375956433	T/T	T	*78	
chr10:94942249	rs200965026	C/C	C	*26 *44	
chr10:94942254	rs199523631	C/C	C	*45	
chr10:94942255	rs200183364	G/G	G	*33	
chr10:94942290	rs1799853	C/C	C	*2 *35 *61	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr10:94942291	rs141489852	G/G	G	*63	
chr10:94942305	rs754487195	G/G	G	*46	
chr10:94942306	rs1289704600	C/C	C	*72	
chr10:94942308	rs17847037	C/C	C	*73	
chr10:94942309	rs7900194	G/G	G	*8 *27	
chr10:94947782	rs72558190	C/C	C	*15	
chr10:94947785	rs774550549	C/C	C	*47	
chr10:94947869		A/A	A	*69	
chr10:94947907		A/A	A	*57	
chr10:94947917	rs1326630788	T/T	T	*48	
chr10:94947938	rs2031531005	A/A	A	*28	
chr10:94947939	rs370100007	G/G	G	*74	
chr10:94949129		A/A	A	*49	
chr10:94949144		C/C	C	*50	
chr10:94949145	rs772782449	C/C	C	*82	
chr10:94949161		AT/AT	AT	*85	
chr10:94949217	rs2256871	A/A	A	*9	
chr10:94949280	rs9332130	A/A	A	*10 *71	
chr10:94949281	rs9332131	GA/GA	GA	*6	
chr10:94972119	rs182132442	C/C	C	*29	
chr10:94972123		C/C	C	*64	
chr10:94972134		A/A	A	*51	
chr10:94972179	rs72558192	A/A	A	*16	
chr10:94972180	rs988617574	C/C	C	*52	
chr10:94972183		A/A	A	*81	
chr10:94972233	rs1237225311	C/C	C	*53	
chr10:94981199		G/G	G	*65	
chr10:94981201	rs57505750	T/T	T	*31	
chr10:94981224	rs28371685	C/C	C	*11	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr10:94981225	rs367826293	G/G	G	*34	
chr10:94981230	rs1274535931	C/C	C	*58	
chr10:94981250	rs750820937	C/C	C	*54	
chr10:94981258	rs1297714792	C/C	C	*79	
chr10:94981281	rs749060448	G/G	G	*24	
chr10:94981296	rs1057910	A/A	A	*3 *18 *68	
chr10:94981297	rs56165452	T/T	T	*4	
chr10:94981301	rs28371686	C/C	C	*5	
chr10:94981302	rs1250577724	C/C	C	*55	
chr10:94981305	rs578144976	C/C	C	*66	
chr10:94981365		C/C	C	*17	
chr10:94981371	rs542577750	G/G	G	*68	
chr10:94986042	rs764211126	A/A	A	*56	
chr10:94986073	rs72558193	A/A	A	*18	
chr10:94986136	rs1254213342	A/A	A	*75	
chr10:94986174	rs1441296358	G/G	G	*84	
chr10:94988852	rs776908257	C/C	C	*67	
chr10:94988855		A/A	A	*59	
chr10:94988880		G/G	G	*70	
chr10:94988917	rs769942899	G/G	G	*19	
chr10:94988925	rs202201137	A/A	A	*61	
chr10:94988955	rs767284820	T/T	T	*60	
chr10:94988984	rs781583846	G/G	G	*30	
chr10:94989020	rs9332239	C/C	C	*12 *71	
chr10:94989023	rs868182778	G/G	G	*32	

Other Positions of Interest

Position in VCF	RSID	Call in VCF
chr10:94645745	rs12777823	G/G

CYP3A4 allele match data

Genotype Matched: *1/*1

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

The CYP3A4 alleles are determined based on PharmVar CYP3A4 allele definitions. See PharmCAT disclaimer for further information.

The CYP3A4 *1 allele assignment is characterized by the absence of variants at the positions that are included in the underlying allele definitions.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr7:99758183	rs67666821	G/G	G	*20	
chr7:99760836	rs4986913	G/G	G	*19	
chr7:99760901	rs4986910	A/A	A	*3 *37 *38	
chr7:99760956	rs774109750	T/T	T	*34	
chr7:99762047	rs4986909	G/G	G	*13	
chr7:99762177	rs12721629	G/G	G	*12	
chr7:99762186	rs756833413	C/C	C	*33	
chr7:99762206	rs67784355	G/G	G	*11 *38	
chr7:99763877	rs368296206	A/A	A	*32	
chr7:99763909	rs1303250043	G/G	G	*31	
chr7:99763925		T/T	T	*21	
chr7:99764003	rs28371759	A/A	A	*18	
chr7:99766411	rs4646438	G/G	G	*6	
chr7:99766440	rs138105638	G/G	G	*26	
chr7:99768360	rs55785340	A/A	A	*2	
chr7:99768371	rs55901263	G/G	G	*5	
chr7:99768424	rs113667357	T/T	T	*24	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr7:99768458	rs4987161	A/A	A	*17	
chr7:99768470	rs12721627	G/G	G	*16	
chr7:99768693	rs35599367	G/G	G	*22 *37	
chr7:99769769	rs4986908	C/C	C	*10	
chr7:99769781	rs72552798	C/C	C	*9	
chr7:99769804	rs4986907	C/C	C	*15	
chr7:99769805	rs57409622	G/G	G	*23	
chr7:99770165	rs72552799	C/C	C	*8	
chr7:99770166	rs778013004	G/G	G	*30	
chr7:99770202	rs55951658	T/T	T	*4	
chr7:99770217	rs1449865051	A/A	A	*29	
chr7:99778079	rs56324128	C/C	C	*7	
chr7:99784018	rs570051168	G/G	G	*28	
chr7:99784038	rs12721634	A/A	A	*14	
chr7:99784075	rs188389063	G/G	G	*35	

CYP3A5 allele match data

Genotype Matched: *1/*1

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

The CYP3A5 *1 allele assignment is characterized by the absence of variants at the positions that are included in the underlying allele definitions.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr7:99652770	rs41303343	T/T	T	*7	
chr7:99660516	rs28383479	C/C	C	*9	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr7:99665212	rs10264272	C/C	C	*6	
chr7:99672916	rs776746	T/T	T	*3	
chr7:99676198	rs55817950	G/G	G	*8	

CYP4F2 allele match data

Genotype Matched: *1/*1

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

The CYP4F2 *1 allele assignment is characterized by the absence of variants at the positions that are included in the underlying allele definitions.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:15878779	rs3093200	G/G	G	*5	
chr19:15879621	rs2108622	C/C	C	*3 *4	
chr19:15890405	rs3093153	C/C	C	*6	
chr19:15897566	rs114099324	C/C	C	*7	
chr19:15897578	rs3093105	A/A	A	*2 *4	

DPYD allele match data

Genotype Matched: Reference/Reference

Phasing Status: Unphased

The two lowest activity values (variant activity scores, see CPIC guideline PMID: 29152729) are used for uphased data and the lowest activity value per allele are used for phased data to determine the gene activity score and phenotype type to retrieve prescribing recommendations.

The DPYD Reference allele assignment is characterized by the absence of variants at the positions that are included in the underlying allele definitions.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr1:97078987	rs114096998	G/G	G	c.3067C>A	
chr1:97078993	rs148799944	C/C	C	c.3061G>C	
chr1:97079005	rs140114515	C/C	C	c.3049G>A	
chr1:97079071	rs1801268	C/C	C	c.2983G>T (*10)	
chr1:97079076	rs139459586	A/A	A	c.2978T>G	
chr1:97079077	rs202144771	G/G	G	c.2977C>T	
chr1:97079121	rs72547601	T/T	T	c.2933A>G	
chr1:97079133	rs72547602	T/T	T	c.2921A>T	
chr1:97079139	rs145529148	T/T	T	c.2915A>G	
chr1:97082365	rs141044036	T/T	T	c.2872A>G	
chr1:97082391	rs67376798	T/T	T	c.2846A>T	
chr1:97098598	rs1801267	C/C	C	c.2657G>A (*9B)	
chr1:97098599	rs147545709	G/G	G	c.2656C>T	
chr1:97098616	rs55674432	C/C	C	c.2639G>T	
chr1:97098632	rs201035051	T/T	T	c.2623A>C	
chr1:97193109	rs60139309	T/T	T	c.2582A>G	
chr1:97193209	rs200687447	C/C	C	c.2482G>A	
chr1:97234958	rs199634007	G/G	G	c.2336C>A	
chr1:97234991	rs56005131	G/G	G	c.2303C>A	
chr1:97305279	rs112766203	G/G	G	c.2279C>T	
chr1:97305363	rs60511679	A/A	A	c.2195T>G	
chr1:97305364	rs1801160	C/C	C	c.2194G>A (*6)	
chr1:97305372	rs146529561	G/G	G	c.2186C>T	
chr1:97306195	rs145548112	C/C	C	c.2161G>A	
chr1:97373598	rs137999090	C/C	C	c.2021G>A	
chr1:97373629	rs138545885	C/C	C	c.1990G>T	
chr1:97382461	rs55971861	T/T	T	c.1906A>C	
chr1:97450058	rs3918290	C/C	C	c.1905+1G>A (*2A)	
chr1:97450059	rs3918289	G/G	G	c.1905C>G	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr1:97450065	rs72549303	TG/TG	TG	c.1898delC (*3)	
chr1:97450068	rs17376848	A/A	A	c.1896T>C	
chr1:97450168	rs147601618	A/A	A	c.1796T>C	
chr1:97450187	rs145773863	C/C	C	c.1777G>A	
chr1:97450189	rs138616379	C/C	C	c.1775G>A	
chr1:97450190	rs59086055	G/G	G	c.1774C>T	
chr1:97515784	rs201615754	C/C	C	c.1682G>T	
chr1:97515787	rs55886062	A/A	A	c.1679T>G (*13)	
chr1:97515839	rs1801159	T/T	T	c.1627A>G (*5)	
chr1:97515851	rs142619737	C/C	C	c.1615G>A	
chr1:97515865	rs1801158	C/C	C	c.1601G>A (*4)	
chr1:97515889	rs190951787	G/G	G	c.1577C>G	
chr1:97515923	rs148994843	C/C	C	c.1543G>A	
chr1:97549565	rs138391898	C/C	C	c.1519G>A	
chr1:97549600	rs111858276	T/T	T	c.1484A>G	
chr1:97549609	rs72549304	G/G	G	c.1475C>T	
chr1:97549681	rs199549923	G/G	G	c.1403C>A	
chr1:97549713	rs57918000	G/G	G	c.1371C>T	
chr1:97549726	rs144395748	G/G	G	c.1358C>G	
chr1:97549735	rs72975710	G/G	G	c.1349C>T	
chr1:97573785	rs186169810	A/A	A	c.1314T>G	
chr1:97573805	rs142512579	C/C	C	c.1294G>A	
chr1:97573821	rs764666241	C/C	C	c.1278G>T	
chr1:97573839	rs200064537	A/A	A	c.1260T>A	
chr1:97573863	rs56038477	C/C	C	c.1129-5923C>G, c.1236G>A (HapB3)	
chr1:97573881	rs61622928	C/C	C	c.1218G>A	
chr1:97573918	rs143815742	C/C	C	c.1181G>T	
chr1:97573919	rs140602333	G/G	G	c.1180C>T	
chr1:97573943	rs78060119	C/C	C	c.1156G>T (*12)	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr1:97579893	rs75017182	G/G	G	c.1129-5923C>G, c.1236G>A (HapB3)	
chr1:97593238	rs72549305	T/T	T	c.1108A>G	
chr1:97593289	rs143154602	G/G	G	c.1057C>T	
chr1:97593322	rs183385770	C/C	C	c.1024G>A	
chr1:97593343	rs72549306	C/C	C	c.1003G>T (*11)	
chr1:97593379	rs201018345	C/C	C	c.967G>A	
chr1:97595083	rs145112791	G/G	G	c.934C>T	
chr1:97595088	rs150437414	A/A	A	c.929T>C	
chr1:97595149	rs146356975	T/T	T	c.868A>G	
chr1:97679170	rs45589337	T/T	T	c.775A>G	
chr1:97691776	rs1801266	G/G	G	c.703C>T (*8)	
chr1:97699399	rs72549307	T/T	T	c.632A>G	
chr1:97699430	rs72549308	T/T	T	c.601A>C	
chr1:97699474	rs115232898	T/T	T	c.557A>G	
chr1:97699506	rs6670886	C/C	C	c.525G>A	
chr1:97699533	rs139834141	C/C	C	c.498G>A	
chr1:97699535	rs2297595	T/T	T	c.496A>G	
chr1:97721542	rs200562975	T/T	T	c.451A>G	
chr1:97721650	rs141462178	T/T	T	c.343A>G	
chr1:97740400	rs150385342	C/C	C	c.313G>A	
chr1:97740410	rs72549309	GATGA/GATGA	GATGA	c.295_298delTCAT (*7)	
chr1:97883329	rs1801265	A/A	A	c.85T>C (*9A)	
chr1:97883352	rs80081766	C/C	C	c.62G>A	
chr1:97883353	rs72549310	G/G	G	c.61C>T	
chr1:97883368	rs150036960	G/G	G	c.46C>G	

G6PD allele match data

Genotype Matched: B (reference)/B (reference)

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

PharmCAT reports based on VCF file input and some VCF files do not specify hemizygous (one X chromosome) and instead represent samples as homozygous. See PharmCAT FAQs (https://pharmcat.org/faqs/).

The G6PD B (reference) allele assignment is characterized by the absence of variants at the positions that are included in the underlying allele definitions.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chrX:154532046		A/A	A	Bangkok Noi	
chrX:154532055		CTCT/CTCT	CTCT	Brighton	
chrX:154532082		G/G	G	Arakawa	
chrX:154532083		G/G	G	Buenos Aires	
chrX:154532085		C/C	C	Campinas	
chrX:154532086		C/C	C	Fukaya	
chrX:154532203	rs137852348	G/G	G	Split	
chrX:154532231		T/T	T	Laibin	
chrX:154532245	rs137852344	G/G	G	Neapolis	
chrX:154532257	rs72554664	C/C	C	Kaiping, Anant, Dhon, Sapporo-like, Wosera	
chrX:154532258		G/G	G	Flores Kamiube, Keelung	
chrX:154532264	rs782608284	C/C	C	Yunan	
chrX:154532265		C/C	C	Nice	
chrX:154532269	rs72554665	C/C	C	Bangkok Noi Canton, Taiwan-Hakka, Gifu-like, Agrigento-like Cosenza	
chrX:154532278		T/T	T	Amiens	
chrX:154532279		C/C	C	Figuera da Foz	
chrX:154532389	rs137852324	C/C	C	Andalus	
chrX:154532390	rs398123546	G/G	G		

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
				Hermoupolis Honiara Union,Maewo, Chinese-2, Kalo	
chrX:154532392		A/A	A	Harima	
chrX:154532403		C/C	C	Cassano Hermoupolis	
chrX:154532408		T/T	T	S. Antioco	
chrX:154532411	rs137852317	C/C	C	Santiago de Cuba, Morioka	
chrX:154532432		G/G	G	Telti/Kobe	
chrX:154532434	rs137852337	C/C	C	Pawnee	
chrX:154532458		A/A	A	Sumare	
chrX:154532459	rs782098548	C/C	C	Surabaya	
chrX:154532570		G/G	G	Georgia	
chrX:154532590		G/G	G	202G>A_376A>G_1264C>G	
chrX:154532608		C/C	C	Tokyo, Fukushima	
chrX:154532623		T/T	T	Munich	
chrX:154532625	rs137852336	C/C	C	Japan, Shinagawa Kawasaki	
chrX:154532626	rs137852323	C/C	C	Riverside	
chrX:154532628		G/G	G	Suwalki	
chrX:154532629		G/G	G	Utrecht	
chrX:154532634		T/T	T	Abeno	
chrX:154532639		C/C	C	Clinic	
chrX:154532649		G/G	G	Covao do Lobo	
chrX:154532661		T/T	T	Anadia	
chrX:154532662	rs137852325	C/C	C	Puerto Limon	
chrX:154532667		G/G	G	Bari	
chrX:154532674	rs137852335	C/C	C	Alhambra	
chrX:154532676	rs137852316	C/C	C	Nashville, Anaheim, Portici	
chrX:154532677		G/G	G	Wisconsin	
chrX:154532679		A/A	A	Krakow	
chrX:154532688		T/T	T	Praha	
chrX:154532692		T/T	T	Hartford	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chrX:154532694	rs137852321	C/C	C	Beverly Hills, Genova, Iwate, Niigata, Yamaguchi	
chrX:154532695	rs137852334	G/G	G	Guadalajara Mt Sinai	
chrX:154532698	rs137852320	T/T	T	Iowa, Walter Reed, Springfield	
chrX:154532699		G/G	G	Madrid	
chrX:154532700		C/C	C	Lynwood	
chrX:154532701	rs137852322	A/A	A	Tomah	
chrX:154532713		A/A	A	Olomouc	
chrX:154532715		A/A	A	Riley	
chrX:154532716		T/T	T	Calvo Mackenna	
chrX:154532722	rs371489738	C/C	C	Montpellier	
chrX:154532752		CGGCCTTGCGCTCGTTCAG/CGGCCTTGCGCTCGTTCAG	CGGCCTTGCGCTCGTTCAG	Tondela	
chrX:154532758		T/T	T	Tenri	
chrX:154532765	rs137852329	G/G	G	Aachen Loma Linda	
chrX:154532772	rs137852345	G/G	G	Serres	
chrX:154532773		C/C	C	Iwatsuki	
chrX:154532797	rs137852333	G/G	G	Ierapetra	
chrX:154532802		C/C	C	Partenope	
chrX:154532945	rs34193178	C/C	C	Mira d'Aire	
chrX:154532956	rs398123544	T/T	T	Cincinnati	
chrX:154532969	rs137852342	G/G	G	Chinese-5	
chrX:154532987		T/T	T	Torun	
chrX:154532989		G/G	G	Fushan	
chrX:154532990	rs5030869	C/C	C	Chatham	
chrX:154533004		C/C	C	Insuli	
chrX:154533012		CGTGGGGTCGTCCAGGTACCCTTTG/CGTGGGGTC	CGTGGGGTCGTCCAGGTACCCTTTG	Nara	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
		GTCCAGGTA CCCTTTG			
chrX:154533016		G/G	G	Farroupilha	
chrX:154533025	rs76723693	A/A	A	A- 968C_376G	
chrX:154533029	rs137852347	A/A	A	Rehevot	
chrX:154533031		C/C	C	Manhattan	
chrX:154533044	rs137852339	C/C	C	Kalyan-Kerala, Jamnaga, Rohini	
chrX:154533064		C/C	C	Ludhiana	
chrX:154533072		C/C	C	Omiya	
chrX:154533077		C/C	C	Seoul	
chrX:154533083		C/C	C	West Virginia	
chrX:154533122	rs137852327	C/C	C	Ananindeua Hechi Viangchan, Jammu	
chrX:154533586	rs74575103	C/C	C	Montalbano	
chrX:154533587		G/G	G	Osaka	
chrX:154533589		A/A	A	Piotrkow	
chrX:154533591		G/G	G	Papua	
chrX:154533592		T/T	T	Mizushima	
chrX:154533596	rs137852318	C/C	C	Bajo Maumere Seattle, Lodi, Modena, Ferrara II, Athens-like	
chrX:154533605		T/T	T	Chinese-1 Haikou	
chrX:154533607		G/G	G	Wexham	
chrX:154533608		A/A	A	La Jolla	
chrX:154533614		G/G	G	Sugao	
chrX:154533615		C/C	C	Bangkok	
chrX:154533619		T/T	T	Lille	
chrX:154533620		C/C	C	Cleveland Corum	
chrX:154533629		C/C	C	Roubaix	
chrX:154533634	rs137852346	C/C	C	Aveiro	
chrX:154534036		G/G	G	Wayne	
chrX:154534074			TCAGTGC	Stonybrook	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
		TCAGTGC/ TCAGTGC			
chrX:154534092		T/T	T	Durham	
chrX:154534102	rs782757170	G/G	G	Nanning	
chrX:154534110		C/C	C	Asahikawa	
chrX:154534116		ATGT/ATGT	ATGT	North Dallas	
chrX:154534125	rs137852328	C/C	C	A- 680T_376G Mexico City	
chrX:154534126		G/G	G	Radlowo	
chrX:154534157	rs137852319	A/A	A	Harilaou	
chrX:154534345	rs137852326	C/C	C	Cincinnati Minnesota, Marion, Gastonia, LeJeune	
chrX:154534348	rs782754619	T/T	T	Sibari	
chrX:154534387	rs781865768	T/T	T	Dagua	
chrX:154534389	rs137852332	C/C	C	Nilgiri Santiago	
chrX:154534390	rs137852330	G/G	G	Coimbra Shunde Vancouver	
chrX:154534409		G/G	G	Pedoplis-Ckaro	
chrX:154534414		GGGA/GGGA	GGGA	Tsukui	
chrX:154534419	rs5030868	G/G	G	Mediterranean, Dallas, Panama, Sassari, Cagliari, Birmingham	
chrX:154534438	rs267606836	G/G	G	Vancouver	
chrX:154534440	rs5030872	T/T	T	Malaga Santa Maria	
chrX:154534447		T/T	T	Chikugo	
chrX:154534455		T/T	T	Shinshu	
chrX:154534463		G/G	G	Miaoli	
chrX:154534465	rs137852343	A/A	A	Nankang	
chrX:154534468		G/G	G	Volendam	
chrX:154534485		C/C	C	Naone	
chrX:154534486		G/G	G	Toledo	
chrX:154534489	rs137852331	T/T	T	Taipei, Chinese-3	
chrX:154534494		C/C	C	Plymouth	
chrX:154534495	rs137852314	C/C	C	Mahidol	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chrX:154535176	rs370918918	C/C	C	Gond	
chrX:154535180	rs782487723	C/C	C	Shenzen	
chrX:154535187	rs137852313	C/C	C	Ilesha	
chrX:154535190		G/G	G	Acrokorinthos	
chrX:154535211		C/C	C	Liuzhou	
chrX:154535244		G/G	G	Belem	
chrX:154535247		G/G	G	Valladolid	
chrX:154535249	rs782322505	T/T	T	Cairo	
chrX:154535261		C/C	C	Quing Yan	
chrX:154535269		G/G	G	Crispim	
chrX:154535270	rs78365220	A/A	A	Crispim Salerno Pyrgos Vanua Lava	
chrX:154535274		C/C	C	Crispim	
chrX:154535277	rs1050829	T/T	T	202G>A_376A>G_1264C>G A A- 202A_376G A- 680T_376G A- 968C_376G Acrokorinthos Ananindeua Mt Sinai Santa Maria Sierra Leone	
chrX:154535278		C/C	C	Crispim	
chrX:154535301		A/A	A	Bao Loc	
chrX:154535316	rs5030870	C/C	C	Sao Borja	
chrX:154535330		A/A	A	Hammersmith	
chrX:154535336	rs267606835	G/G	G	Vancouver	
chrX:154535342	rs181277621	C/C	C	Sierra Leone	
chrX:154535367		GCTT/GCTT	GCTT	Urayasu	
chrX:154535379		G/G	G	Guangzhou	
chrX:154535962	rs782308266	C/C	C	Lagosanto	
chrX:154535963	rs138687036	G/G	G	Ube Konan	
chrX:154535980		A/A	A	Swansea	
chrX:154535995	rs782090947	T/T	T	Murcia Oristano	
chrX:154535996	rs137852349	A/A	A	Namouru	
chrX:154536002	rs1050828	C/C	C		

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
				202G>A_376A>G_1264C>G A- 202A_376G Asahi Hechi	
chrX:154536008		A/A	A	Songklanagarind	
chrX:154536019		G/G	G	Amazonia Musashino	
chrX:154536021		CAGA/CAGA	CAGA	Amsterdam	
chrX:154536025		A/A	A	Costanzo	
chrX:154536032	rs137852315	C/C	C	Metaponto	
chrX:154536034		C/C	C	Palestrina	
chrX:154536035		G/G	G	Kamogawa	
chrX:154536045		C/C	C	Kozukata	
chrX:154536151		G/G	G	Kambos	
chrX:154536156	rs76645461	A/A	A	Aures	
chrX:154536168	rs78478128	G/G	G	Orissa	
chrX:154536169		C/C	C	Rignano	
chrX:154546045	rs137852338	CATG/CATG	CATG	Sunderland	
chrX:154546046		A/A	A	Gidra	
chrX:154546057		T/T	T	Honiara	
chrX:154546061	rs137852340	T/T	T	Gaohe	
chrX:154546116		C/C	C	Lages	
chrX:154546122		C/C	C	Sinnai	
chrX:154546131		G/G	G	No name	

IFNL3/4 allele match data

Genotype Matched: rs12979860 reference (C)/ rs12979860 reference (C)

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:39248147	rs12979860	C/C	C	rs12979860 variant (T)	

NUDT15 allele match data

Genotype Matched: *1/*1

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

The NUDT15 *1 allele assignment is characterized by the absence of variants at the positions that are included in the underlying allele definitions.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr13:48037748	rs769369441	T/T	T	*10	
chr13:48037749		G/G	G	*19	
chr13:48037782	rs746071566	AGGAGTC/ AGGAGTC	AGGAGTC	*2 *6 *9	
chr13:48037798	rs186364861	G/G	G	*5	
chr13:48037825	rs777311140	C/C	C	*14	
chr13:48037834	rs1202487323	C/C	C	*16	
chr13:48037847	rs766023281	G/G	G	*7	
chr13:48037849		A/A	A	*8	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr13:48037885	rs1950545307	G/G	G	*11	
chr13:48037902	rs149436418	C/C	C	*12	
chr13:48040977	rs1457579126	GA/GA	GA	*18	
chr13:48041103	rs761191455	T/T	T	*13	
chr13:48041113	rs1368252918	G/G	G	*17	
chr13:48045690	rs768324690	C/C	C	*20	
chr13:48045719	rs116855232	C/C	C	*2 *3	
chr13:48045720	rs147390019	G/G	G	*4	
chr13:48045771	rs139551410	T/T	T	*15	

RYR1 allele match data

Genotype Matched: Reference/Reference

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

The RYR1 Reference allele assignment is characterized by the absence of variants at the positions that are included in the underlying allele definitions.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:38440802	rs193922747	T/T	T	c.103T>C	
chr19:38440829	rs193922748	C/C	C	c.130C>T	
chr19:38444211	rs118192161	C/C	C	c.487C>T	
chr19:38444212	rs193922753	G/G	G	c.488G>T	
chr19:38446710	rs1801086	G/G	G	c.742G>A c.742G>C	
chr19:38448673	rs193922762	C/C	C	c.982C>T	
chr19:38448712	rs121918592	G/G	G	c.1021G>A c.1021G>C	
chr19:38451842	rs193922764	C/C	C	c.1201C>T	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:38451850	rs118192116	C/C	C	c.1209C>G	
chr19:38455359	rs118192162	A/A	A	c.1565A>C	
chr19:38455463	rs111888148	G/G	G	c.1589G>A	
chr19:38455471	rs193922768	C/C	C	c.1597C>T	
chr19:38455472	rs144336148	G/G	G	c.1598G>A	
chr19:38455528	rs193922770	C/C	C	c.1654C>T	
chr19:38457545	rs118192172	C/C	C	c.1840C>T	
chr19:38457546	rs193922772	G/G	G	c.1841G>T	
chr19:38494564	rs118192175	C/C	C	c.6487C>T	
chr19:38494565	rs118192163	G/G	G	c.6488G>A	
chr19:38494579	rs118192176	G/G	G	c.6502G>A	
chr19:38496283	rs118192177	C/C	C	c.6617C>G c.6617C>T	
chr19:38499223	rs112563513	G/G	G	c.7007G>A	
chr19:38499644	rs121918596	TGGA/TGGA	TGGA	c.7042_7044delGAG	
chr19:38499655	rs193922802	G/G	G	c.7048G>A	
chr19:38499670	rs193922803	C/C	C	c.7063C>T	
chr19:38499731	rs193922807	G/G	G	c.7124G>C	
chr19:38499975	rs193922809	G/G	G	c.7282G>A	
chr19:38499993	rs121918593	G/G	G	c.7300G>A	
chr19:38499997	rs28933396	G/G	G	c.7304G>A	
chr19:38500636	rs118192124	C/C	C	c.7354C>T	
chr19:38500642	rs193922816	C/C	C	c.7360C>T	
chr19:38500643	rs118192122	G/G	G	c.7361G>A	
chr19:38500654	rs28933397	C/C	C	c.7372C>T	
chr19:38500655	rs121918594	G/G	G	c.7373G>A	
chr19:38500898	rs118192178	C/C	C	c.7522C>G c.7522C>T	
chr19:38500899	rs193922818	G/G	G	c.7523G>A	
chr19:38512321	rs193922832	G/G	G	c.9310G>A	
chr19:38543832	rs193922843	G/G	G	c.11969G>T	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr19:38580004	rs118192167	A/A	A	c.14387A>G	
chr19:38580094	rs121918595	C/C	C	c.14477C>T	
chr19:38580114	rs193922876	C/C	C	c.14497C>T	
chr19:38580370	rs193922878	C/C	C	c.14512C>G	
chr19:38580403	rs118192168	G/G	G	c.14545G>A	
chr19:38580440	rs63749869	G/G	G	c.14582G>A	
chr19:38584989	rs118192170	T/T	T	c.14693T>C	

SLCO1B1 allele match data

Genotype Matched: *1/*1

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

The SLCO1B1 genotype (star nomenclature) will be displayed if determinable with the provided VCF based on the SLCO1B1 star allele definition published by CPIC and recommendations are provided based on the genotype.

In case no genotype can be determined, recommendations are based on the rs4149056 genotype alone as per guideline. The minor C allele at rs4149056 defines SLCO1B1*5.

The SLCO1B1 *1 allele assignment is characterized by the absence of variants at the positions that are included in the underlying allele definitions.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr12:21172734	rs139257324	C/C	C	*33	
chr12:21172776	rs373327528	G/G	G	*23	
chr12:21172782	rs56101265	T/T	T	*2 *12	
chr12:21174595	rs56061388	T/T	T	*3 *13	
chr12:21176804	rs2306283	A/A	A	*14 *15 *20 *24 *25 *27 *28 *29 *30 *31 *32 *33 *37 *39 *42 *43 *44 *46 *47	
chr12:21176868	rs2306282	A/A	A	*16	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr12:21176871		G/G	G	*38	
chr12:21176879	rs11045819	C/C	C	*4 *14 *25 *32 *43	
chr12:21176883	rs72559745	A/A	A	*3 *13	
chr12:21176898	rs77271279	G/G	G	*41	
chr12:21178612	rs141467543	A/A	A	*42	
chr12:21178615	rs4149056	T/T	T	*5 *15 *40 *46 *47	
chr12:21178957	rs79135870	A/A	A	*30	
chr12:21196951	rs11045852	A/A	A	*24 *25 *28 *32 *33 *43 *44	
chr12:21196975	rs183501729	C/C	C	*39	
chr12:21196976	rs11045853	G/G	G	*25 *28 *33	
chr12:21200544	rs72559747	C/C	C	*47	
chr12:21200595	rs55901008	T/T	T	*6	
chr12:21202553	rs1228465562	T/T	T	*36	
chr12:21202555	rs59113707	C/C	C	*27	
chr12:21202649	rs56387224	A/A	A	*7	
chr12:21202664	rs142965323	G/G	G	*26	
chr12:21205921	rs72559748	A/A	A	*8	
chr12:21205999	rs59502379	G/G	G	*9 *31	
chr12:21206031	rs74064213	A/A	A	*43 *44	
chr12:21222355	rs71581941	C/C	C	*45 *46	
chr12:21239042	rs34671512	A/A	A	*19 *20 *40	
chr12:21239077	rs56199088	A/A	A	*10 *12	
chr12:21239113	rs55737008	A/A	A	*11 *13	
chr12:21239145	rs200995543	C/C	C	*34	
chr12:21239158	rs140790673	C/C	C	*29	

TPMT allele match data

Genotype Matched: *1/*1

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

The TPMT *1 allele assignment is characterized by the absence of variants at the positions that are included in the underlying allele definitions.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr6:18130687	rs1142345	T/T	T	*3A *3C *41	
chr6:18130694	rs150900439	T/T	T	*20	
chr6:18130725	rs72552736	A/A	A	*7	
chr6:18130729	rs139392616	C/C	C	*40	
chr6:18130758	rs398122996	A/A	A	*37	
chr6:18130762	rs56161402	C/C	C	*8	
chr6:18130772	rs377085266	A/A	A	*25	
chr6:18130781	rs1800584	C/C	C	*4	
chr6:18132136	rs72556347	A/A	A	*26	
chr6:18132147	rs79901429	A/A	A	*31	
chr6:18132163		C/C	C	*36	
chr6:18133845	rs75543815	T/T	T	*6	
chr6:18133847	rs6921269	C/C	C	*24	
chr6:18133870	rs772832951	A/A	A	*38	
chr6:18133884	rs74423290	G/G	G	*23	
chr6:18133887	rs201695576	T/T	T	*44	
chr6:18133890	rs9333570	C/C	C	*15	
chr6:18138969	rs144041067	C/C	C	*16 *22	
chr6:18138970	rs112339338	G/G	G	*33	
chr6:18138997	rs1800460	C/C	C	*3A *3B	
chr6:18139027	rs72552737	C/C	C	*10	
chr6:18139689	rs72552738	C/C	C	*11	

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr6:18139710	rs200220210	G/G	G	*12	
chr6:18143597		T/T	T	*19	
chr6:18143606	rs151149760	T/T	T	*9	
chr6:18143613		C/C	C	*28	
chr6:18143622	rs115106679	C/C	C	*32	
chr6:18143643		A/A	A	*27	
chr6:18143700	rs753545734	C/C	C	*43	
chr6:18143718	rs111901354	G/G	G	*34	
chr6:18143724	rs1800462	C/C	C	*2	
chr6:18143728	rs1256618794	C/C	C	*43	
chr6:18147838	rs281874771	G/G	G	*39	
chr6:18147845	rs777686348	C/C	C	*18	
chr6:18147851	rs200591577	G/G	G	*21	
chr6:18147856		A/A	A	*35	
chr6:18147910	rs72552740	A/A	A	*5	
chr6:18149004		G/G	G	*17	
chr6:18149022	rs750424422	C/C	C	*30	
chr6:18149032	rs759836180	C/C	C	*42	
chr6:18149045	rs72552742	T/T	T	*13	
chr6:18149126	rs267607275	A/A	A	*29	
chr6:18149127	rs9333569	T/T	T	*14	

UGT1A1 allele match data

Genotype Matched: *1/*1

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

The UGT1A1 *1 allele assignment is characterized by the absence of variants at the positions that are included in the underlying allele definitions.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr2:233759924	rs887829	C/C	C	*80 *80+*28 *80+*37	
chr2:233760233	rs3064744	CAT/CAT	CAT	*28 *36 *37 *80+*28 *80+*37	
chr2:233760498	rs4148323	G/G	G	*6	
chr2:233760973	rs35350960	C/C	C	*27	

VKORC1 allele match data

Genotype Matched: rs9923231 reference (C)/rs9923231 reference (C)

Phasing Status: Unphased

PharmCAT reports the genotype(s) that receive the highest score during the matcher process. In case of unphased data, additional genotypes might be possible and cannot be ruled out.

Calls at Positions

Position in VCF	RSID	Call in VCF	Reference	Related Alleles	Warnings
chr16:31096368	rs9923231	C/C	C	rs9923231 variant (T)	

Disclaimers and Other Information

Liability: PharmCAT assumes no responsibility for any injury to person or damage to persons or property arising out of, or related to any use of PharmCAT, or for any errors or omissions. The user recognizes that PharmCAT is a research tool and that they are using PharmCAT at their own risk.

A. Allele and Genotype Determination

1. PharmCAT uses gene allele definitions included in the CPIC database, with exceptions as noted in Gene Definition Exceptions document. For allele definitions and the positions used in PharmCAT, see the gene definition tables.
2. Genes with DPWG recommendations that are not included in CPIC are discussed in Section C.
3. PharmCAT results are dependent on the supplied VCF calls for the queried positions (for technical information about PharmCAT input formatting and requirements, please go to pharmcat.org). PharmCAT does not assume any reference calls for positions missing from the submitted VCF file; all missing queried positions are not considered in the allele determination process. See the *pharmcat_positions* file for which positions are queried in the VCF file. Missing positions might alter the assigned genotype and subsequent phenotype prediction and CPIC recommendation. If the supplied VCF file has missing positions, those positions will be noted in Section III: Allele Matching Details for each gene of this report. For the most reliable allele determination, reference calls as well as

variant calls in the VCF file for every queried position must be provided by the user. If an allele that includes a missing position is defined by an additional position(s) for which calls are provided, the allele will be considered in the matching process based on the available information. This might lead to the output of multiple possible genotypes that received the same highest matching score. In addition, alternate calls with a lower score is also possible. For instructions on getting PharmCAT to output all possible matching genotypes, consult the documentation.

4. For cytochrome P450 genes, TPMT, NUDT15, UGT1A1, and SLCO1B1, the *1 allele is defined by the absence of variation specified in the gene definition tables. This allele cannot be identified by variants; rather, *1 is assigned by default when no variation for the queried positions is reported in the submitted VCF file. The same is true for all other genes with multiple variant positions in the definition table (CACNA1S, CFTR, DPYD, RYR1): the reference sequence is the default result when variants are not reported in the VCF file. It is always possible un-interrogated variation can occur which could potentially affect allele function, but because it is undetected, the assignment would be defaulted to a *1 (or reference) allele and normal function.
5. For all genes, variation reported in the VCF file but NOT included in the gene definition table will not be considered during allele assignment. There is a possibility that any such variation results in a reduced or no activity allele which could lead to inaccurate phenotype and CPIC recommendation, similar to the situation in point 3, above.
6. Nucleotide base calls are displayed on the positive chromosomal strand regardless of the gene strand; further information is provided under Gene-specific warnings in Section III: Allele Matching Details.
7. PharmCAT matches variants to genotypes assuming unphased data (unless phased data is provided in the VCF file and noted as such, see pharmcat.org for details). The assumption is that defined alleles exist in trans configuration, i.e. on opposite chromosomes, with exceptions noted in Section III: Allele Matching Details under "Gene-specific warnings." However, in cases where an allele is defined by a combination of two or more variants, where each variant alone also defines an allele, the match is based on the longer allele. For example, TPMT*3B is defined by one SNP, *3C is defined by another SNP, and *3A is defined by the combination of those two SNPs. In the case of unphased data that is heterozygous for both SNPs, the *1/*3A genotype is returned though the possibility of *3B/*3C cannot be ruled out.

   Below cases are summarized for which two calls with different scores are possible when provided unphased data and heterozygous calls for the variants that define the two alleles. The genotype with the higher score (longer allele) will be used to determine allele functionality, phenotype, and recommendation but the genotype with the lower score cannot be ruled out.

Table 1: Cases for which there is an overlap in the allele definitions.

Gene	Genotype (Higher Score)	Phenotype	Genotype (Lower Score)	Phenotype
UGT1A1	*1/*80+*28	Intermediate	*28/*80	Indeterminate
UGT1A1	*1/*80+*37	Intermediate	*37/*80	Indeterminate
TPMT	*1/*3A	Intermediate	*3B/*3C	Poor
NUDT15	*1/*2	Intermediate	*3/*6	Possible Intermediate
CYP2C9	*1/*71	N/A	*10/*22	Indeterminate
CYP2B6	*1/*36	Intermediate	*6/*22	Intermediate
CYP2B6	*1/*34	Intermediate	*33/*36	Indeterminate
CYP2B6	*1/*6	Intermediate	*4/*9	Intermediate
CYP2B6	*1/*7	Intermediate	*5/*6	Intermediate
CYP2B6	*1/*13	Intermediate	*6/*8	Intermediate
SLCO1B1	*1/*46	Decreased function	*15/*45	Possible Decreased Function
SLCO1B1	*1/*20	Normal Function	*19/*37	Indeterminate

Gene	Genotype (Higher Score)	Phenotype	Genotype (Lower Score)	Phenotype
SLCO1B1	*1/*12	Indeterminate	*2/*10	Indeterminate
SLCO1B1	*1/*13	Indeterminate	*3/*11	Indeterminate
SLCO1B1	*1/*14	Normal Function	*4/*37	Indeterminate
SLCO1B1	*1/*15	Decreased function	*5/*37	Decreased function
SLCO1B1	*1/*25	Indeterminate	*4/*28	Indeterminate
SLCO1B1	*1/*31	Decreased function	*9/*37	Decreased Function
SLCO1B1	*1/*32	Indeterminate	*4/*24	Indeterminate
SLCO1B1	*1/*40	Indeterminate	*5/*19	Possible Decreased Function
SLCO1B1	*1/*43	Indeterminate	*4/*44	Indeterminate
CYP4F2	*1/*4	N/A	*2/*3	N/A
CYP3A4	*1/*37	N/A	*3/*22	N/A
CYP3A4	*1/*38	N/A	*3/*11	N/A
G6PD	A- 202A_376G/B (reference)	Variable	A/Asahi	Variable
G6PD	B (reference)/Mt Sinai	Variable	A/Guadalajara	Variable
G6PD	B (reference)/Santa Maria	Variable	A/Malaga	Variable
G6PD	Ananindeua/B (reference)	Variable	A/Viangchan, Jammu	Variable
G6PD	B (reference)/Hechi	Variable	Asahi/Viangchan, Jammu	Deficient
G6PD	B (reference)/ Hermoupolis	Variable	Cassano/ Union,Maewo, Chinese-2, Kalo	Deficient

Table 2: Cases for which there is an overlap in the allele definitions because the definition of the non-*1 allele in the genotype with the higher score allows for reference or variant at the position that defines the first allele listed in the genotype with the lower score. Both genotypes cannot be ruled out with unphased data if the position that overlaps between the respective alleles is heterozygous (0/1) in addition to heterozygous calls for the other variants that define the non-*1 allele in the genotype with the higher score.

Gene	Genotype (Higher Score)	Phenotype	Genotype (Lower Score)	Phenotype
CYP2C19	*1/*4	Intermediate	*17/*4	Intermediate
CYP2C19	*1/*2	Intermediate	*11/*2	Intermediate
CYP2C19	*1/*35	Intermediate	*15/*35	Intermediate

Gene	Genotype (Higher Score)	Phenotype	Genotype (Lower Score)	Phenotype
CYP2B6	*1/*18	Intermediate	*4/*18	Indeterminate

B. CPIC Allele Function, Phenotype and Recommendation

All content is sourced from the CPIC database.

C. DPWG Allele Function, Phenotype and Recommendation

1. PharmGKB annotates PGx-based drug dosing guidelines published by the Royal Dutch Association for the Advancement of Pharmacy - Pharmacogenetics Working Group (DPWG). PharmGKB curates allele function assignments and phenotype mappings from the DPWG to provide genotype specific DPWG guideline recommendations. Where possible, PharmGKB maps DPWG terms to CPIC terms, as outlined on PharmGKB.
2. CYP3A4 is currently not part of a CPIC guideline. Since the DPWG CYP3A4 documentation includes limit variant notations for the included alleles (only *16, *20, and *22 are specified) PharmCAT relies on PharmVar CYP3A4 allele definitions. However, the CYP3A4*16, *20 and *22 definitions are the same in both sources.
3. The CPIC UGT1A1 allele definition file includes *6, *27, *28, *36, *37, and *80. Since the DPWG UGT1A1 document does not include allele definitions besides for the UGT1A1 TA box promoter polymorphism, PharmCAT only includes the UGT1A1 positions from the CPIC UGT1A1 allele definition file. Other UGT1A1 alleles can be supplied as outside calls but not be determined from the VCF file by the Named Allele Matcher.
4. IMPORTANT: As of March 2022, gene information documents from the DPWG are no longer publicly available from the KNMP website. PharmGKB is currently providing PDF versions of these documents to users. These files were downloaded in February 2022. As such, it cannot be guaranteed that these documents match the mappings DPWG may use internally as there have been no publicly accessible updates since February 2022.

D. PharmCAT Exceptions to the CPIC Guideline Gene List

1. PharmCAT does not determine CYP2D6, MT-RNR1, HLA-A, or HLA-B genotypes from the VCF file, but genotypes for CYP2D6, MT-RNR1, HLA-A, or HLA-B can be provided to PharmCAT from an outside source and the corresponding phenotype prescribing recommendations will be included in the generated report. For the required format of the outside calls refer to PharmCAT documentation.
2. CPIC has assigned function to the following CYP2D6 CNV alleles: *1x2, *1x≥3, *2x2, *2x≥3, *3x2, *4x2, *4x≥3, *6x2, *9x2, *10x2, *17x2, *29x2, *35x2, *36x2, *41x2, *41x3, *43x2, *45x2, *146x2. These alleles are part of the CPIC diplotype to phenotype translation and can be connected to recommendations. Other CNV notations from outside calls need to be mapped accordingly.

E. CPIC Guideline Disclaimers and Caveats

1. A version of the following quoted disclaimer is part of each CPIC guideline and applies to the CPIC recommendations as used in PharmCAT. For the full description of potential benefits and risks, additional considerations (general and specific to gene-drug pairs), limitations, information about respective gene nomenclature systems, potential drug-drug interactions and clinical factors to consider, please see individual CPIC guidelines at (cpicpgx.org).
    1. "CPIC guidelines reflect expert consensus based on clinical evidence and peer-reviewed literature available at the time they are written and are intended only to assist clinicians in decision-making and to identify questions for further research. New evidence may have emerged since the time a guideline was submitted for publication. Guidelines are limited in scope and are not applicable to interventions or diseases not specifically identified. Guidelines do not account for all individual variations among patients and cannot be considered inclusive of all proper methods of care or exclusive of other treatments. It remains the responsibility of the health-care provider to determine the best course of treatment for a patient. Adherence to any guidelines is voluntary, with the ultimate determination regarding its application to be made solely by the clinician and the patient. CPIC assumes no responsibility for any injury to persons or damage to persons or property arising out of or related to any use of CPIC's guidelines, or for any errors or omissions." (PMID: 27997040)
    2. "Caveats: appropriate use and/or potential misuse of genetic tests. The application of genotype-based dosing is most appropriate when initiating therapy with a tricyclic. Obtaining a pharmacogenetic test after months of drug therapy may be less helpful in some instances, as the drug dose may have already been adjusted based on plasma concentrations, response, or side effects. Similar to all diagnostic tests, genetic

tests are one of several pieces of clinical information that should be considered before initiating drug therapy." (PMID: 27997040)

2. CPIC guidelines reflect the alleles/genotypes known and considered by the guideline authors for inclusion by the time of publication, however they may be updated online at cpicpgx.org and in the CPIC database in between publications. Additional alleles and/or more extensive allele definitions might exist by the representative gene nomenclatures for various genes.

3. CPIC is a registered service mark of the U.S. Department of Health & Human Services (HHS).

F. PharmGKB Disclaimers and Caveats

PharmGKB is a registered service mark of the U.S. Department of Health & Human Services (HHS).