



GeneDose Pharmacogenomic Drug-Gene Associations and Assays

Overview and Audience

This document describes the available drug-gene associations on Genedose system reports, as well as the linkage between those genes and the rsIDs that are offered for testing by PMCDx and part of Coriell's interpretation.

Drug-Gene Associations

CYP2C19

Anticonvulsants	Brivaracetam	Briviact
Anticonvulsants	Clobazam	Onfi
Antidepressants	Moclobemide	
Antifungals	Voriconazole	
Antiplatelet Agents	Prasugrel	Effient
Antiplatelet Agents	Clopidogrel	Plavix
Anxiolytics	Diazepam	Diastat, Valium
Muscle Relaxants	Carisoprodol	Soma
Proton Pump Inhibitors (PPIs)	Rabeprazole	Aciphex
Proton Pump Inhibitors (PPIs)	Pantoprazole	Protonix
Proton Pump Inhibitors (PPIs)	Esomeprazole	Nexium
Proton Pump Inhibitors (PPIs)	Lansoprazole	Prevacid
Proton Pump Inhibitors (PPIs)	Omeprazole	Prilosec, Zegerid
Proton Pump Inhibitors (PPIs)	Dexlansoprazole	Kapidex, Dexilant
Selective Serotonin Reuptake Inhibitors (SSRIs)	Sertraline	Zoloft
Selective Serotonin Reuptake Inhibitors (SSRIs)	Citalopram	Celexa
Selective Serotonin Reuptake Inhibitors (SSRIs)	Escitalopram	Lexapro

CYP2C9

Anticoagulants	Acenocoumarol	Sintrom, Acitrom
Anticonvulsants	Phenytoin	Dilantin
Antidiabetics	Glimepiride	
Antidiabetics	Gliclazide	
Antidiabetics	Glibenclamide	Glyburide
Antidiabetics	Tolbutamide	Orinase
Nonsteroidal Anti-Inflammatory Drugs (NSAIDs)	Piroxicam	Feldene
Nonsteroidal Anti-Inflammatory Drugs (NSAIDs)	Lornoxicam	Xefo
Nonsteroidal Anti-Inflammatory Drugs (NSAIDs)	Ibuprofen	Motrin, Advil
Nonsteroidal Anti-Inflammatory Drugs (NSAIDs)	Meloxicam	Mobic
Nonsteroidal Anti-Inflammatory Drugs (NSAIDs)	Flurbiprofen	Ocufen
Nonsteroidal Anti-Inflammatory Drugs (NSAIDs)	Celecoxib	Celebrex

CYP2D6

Alpha-1 Blockers	Tamsulosin	Flomax
Anti-ADHD Agents	Atomoxetine	Strattera
Antiarrhythmics	Flecainide	Tambocor
Antiarrhythmics	Propafenone	Rythmol

Anticholinergic Agents	Fesoterodine	Toviaz
Anticholinergic Agents	Tolterodine	Detrol
Antidementia Agents	Donepezil	Aricept
Antidepressants	Mirtazapine	
Antidepressants	Amitriptyline	Elavil
Antidepressants	Vortioxetine	Brintellix
Antidepressants	Venlafaxine	Effexor
Antidepressants	Clomipramine	Anafranil
Antidepressants	Desipramine	Norpramin
Antidepressants	Doxepin	Deptran
Antidepressants	Duloxetine	Cymbalta
Antidepressants	Protriptyline	Vivactil
Antidepressants	Flupenthixol	
Antidepressants	Nortriptyline	Pamelor
Antiemetics	Ondansetron	Zofran
Antiemetics	Tropisetron	
Antipsychotics	Thioridazine	
Antipsychotics	Olanzapine	Zalasta, Zyprexa
Antipsychotics	Iloperidone	Fanapt
Antipsychotics	Aripiprazole	Abilify
Antipsychotics	Pimozide	Orap
Antipsychotics	Risperidone	Risperdal
Antipsychotics	Perphenazine	Trilafon
Antipsychotics	Haloperidol	Haldol
Antipsychotics	Clozapine	
Antipsychotics	Brexipiprazole	Rexulti
Antipsychotics	Zuclopenthixol	
Beta-3 Adrenergic Agonists	Mirabegron	Myrbetriq
Beta Blockers	Timolol	Blocadren
Beta Blockers	Metoprolol	Lopressor
Beta Blockers	Propranolol	Inderal

Beta Blockers	Nebivolol	Bystolic
Beta Blockers	Carvedilol	Coreg
Central Monoamine-Depleting Agents	Tetrabenazine	Xenazine
Central Nervous System Agents	Dextromethorphan-Quinidine	Nuedexta
Cholinergic Agonists	Cevimeline	Evoxac
Cholinesterase Inhibitors	Galantamine	Razadyne, Razadyne ER, Nivalin, Lycoremine, Reminyl
EGFR Inhibitors	Gefitinib	Iressa
Endocrine-Metabolic Agents	Eliglustat	Cerdelga
Estrogen Agonists/Antagonists	Tamoxifen	Soltamox, Nolvadex
Opioids	Codeine	
Opioids	Tramadol	Ultracet, Ultram
Opioids	Hydrocodone	
Opioids	Oxycodone	Oxycontin
Selective Serotonin Reuptake Inhibitors (SSRIs)	Fluvoxamine	Luvox
Selective Serotonin Reuptake Inhibitors (SSRIs)	Paroxetine	Paxil
Selective Serotonin Reuptake Inhibitors (SSRIs)	Fluoxetine	Prozac

CYP3A4, CYP3A5

Anti-ADHD Agents	Guanfacine	Tenex, Intuniv
Antidepressants	Trazodone	Oleptro, Desyrel
Antidiabetics	Saxagliptin	Onglyza

Antifungals	Ketoconazole	Nizoral
Antiplatelet Agents	Ticagrelor	Brilinta
Antipsychotics	Quetiapine	Seroquel
Anxiolytics	Buspirone	Buspar
Anxiolytics	Alprazolam	Xanax, Niravam
Anxiolytics	Clonazepam	Klonopin
Hypnotics	Eszopiclone	Lunesta
Immunosuppressants	Cyclosporine	Gengraf, Neoral
Immunosuppressants	Sirolimus	Rapamune
Opioids	Fentanyl	Duragesic, Sublimaze
Opioids	Buprenorphine	Butrans, Buprenex
Statins	Atorvastatin	Lipitor, Caduet
Immunosuppressants	Tacrolimus	Prograf, Hecoria
Opioids	Oxycodone (CYP3A5)	Oxycontin

Multigenic

Antidepressants	Imipramine	CYP2C19,CYP2D6	Tofranil
Anticoagulants	Warfarin	CYP2C9,VKORC1,CYP4F2	Coumadin
Anticoagulants	Warfarin	VKORC1,CYP2C9	Coumadin

SLCO1B1

Statins	Simvastatin	Zocor
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TPMT

Immunosuppressants	Azathioprine	Imuran
Immunosuppressants	Mercaptopurine	Purinethol
Immunosuppressants	Thioguanine	6-TG, Tabloid, Lanvis

Other Genes

Drugs

ABCG2	Xanthine Oxidase Inhibitor	Allopurinol	Zyloprim
ADRA2A(C-1291G)	Anti-ADHD Agents	Methylphenidate (ADRA2A)	Concerta, Metadate, Ritalin, Ritalin LA, Quillivant, Daytrana, Methylin
ADRA2A(C-1291G)	Anti-ADHD Agents	Clonidine	Clonidine, Kapvay
ANKK1	Antidepressants	Bupropion	Wellbutrin
ATM	Antidiabetics	Metformin	Glucophage®
BCHE	Neuromuscular Depolarizing Agents	Succinylcholine	suxamethonium, Anectine
COMT(Val158Met)	Anti-ADHD Agents	Dextroamphetamine	Zenzedi, Dexedrine
COMT(Val158Met)	Anti-ADHD Agents	Methylphenidate (COMT)	Concerta, Metadate, Ritalin, Ritalin LA, Quillivant, Daytrana, Methylin
COMT(Val158Met)	Anti-ADHD Agents	Lisdexamfetamine	Vyvanse
COMT(Val158Met)	Anti-ADHD Agents	Dexmethylphenidate	Focalin
COMT(Val158Met)	Anti-ADHD Agents	Amphetamine	Adzenys, Evekeo
CYP2B6	Analgesics, Opioid	Methadone (CYP2B6)	Dolophine
CYP2B6	Anti-Retroviral Agents	Nevirapine	Viramune
CYP2B6	Anti-Retroviral Agents	Efavirenz	Sustiva
CYP2C9:rs1057910	Nonsteroidal Anti-Inflammatory Drugs (NSAIDs)	Diclofenac	Cataflam
F5	Contraceptives	Estrogen-containing oral contraceptives	
HLA-B*1502	Anticonvulsants	Carbamazepine	Tegretol
HLA-B*1502	Anticonvulsants	Oxcarbazepine	Trileptal

HLA-B*5701	Antivirals	Abacavir	
MTHFR	Antineoplastic Agents	Methotrexate	Rheumatrex, Trexall
OPRM1(A118G)	Opioids	Alfentanil	Rapifen, Alfenta
OPRM1(A118G)	Opioids	Sufentanil	Sufenta
OPRM1(A118G)	Opioids	Hydromorphone	Dilaudid
OPRM1(A118G)	Opioids	Morphine	MS-IR
OPRM1(A118G)	Opioids	Fentanyl (OPRM1)	Duragesic, Sublimaze
UGT2B15	Benzodiazepine	Oxazepam	Alepam
UGT2B15	Benzodiazepine	Lorazepam	Ativan

Gene rsID Associations

Gene	rsIDs
ABCB1	rs1128503, rs2032582, rs1045642
ABCG2	rs2231142
ANKK1	rs1800497
ApoE	rs429358, rs7412
ATM	rs11212617
BCHE	rs1799807, rs1803274, rs28933389, rs28933390
COMT(Val158Met)	rs4680
CYP1A2	rs2069514, rs35694136, rs2069526, rs12720461, rs762551

CYP2B6	rs34223104, rs3211371, rs3745274, rs28399499
CYP2C19	rs17884712, rs28399504, rs4986893, rs41291556, rs56337013, rs6413438, rs4244285, rs12248560, rs72558186, rs72552267
CYP2C8	rs17110453, rs1058930, rs11572103, rs10509681
CYP2C9	rs1057910, rs1799853, rs7900194, rs9332131, rs28371685, rs28371686, rs72558189, rs56165452
CYP2D6	rs1135840, rs59421388, rs28371725, rs5030867, rs16947, rs35742686, rs72549354, rs72549356, rs3892097, rs5030865, rs5030655, rs28371706, rs5030863, rs72549357, rs5030862, rs1065852, rs769258, rs5030656, rs774671100
CYP3A4	rs2740574, rs4986910, rs4987161, rs12721629, rs55785340, rs35599367, rs12721627
CYP3A5	rs15524, rs776746, rs10264272, rs28365083, rs28383468, rs28383479, rs41303343, rs55817950
CYP4F2	rs2108622
F2	rs1799963
F5	rs6025
GRIK4	rs1954787
HLA-B*1502	rs2844682, rs3909184

HLA-B*5701	rs2395029
HTR2A	rs7997012
MTHFR (A1298C)	rs1801131
MTHFR (C677T)	rs1801133
OPRM1(A118G)	rs1799971
SLCO1B1	rs4149056
TPMT	rs1800462, rs1142345, rs1800460
VKORC1	rs9923231

Appendix A: CYP2D6 Copy Number Variation

Copy number variation (CNV) is the result of a specific laboratory test, or multiple tests, related to detecting the number of copies present for CYP2D6 gene. Due to the clinical significance of CYP2D6 copy number variation, it is always necessary to provide Genedose with an integer CNV, in connection with CYP2D6 SNPs, in order to receive a PGx interpretation covering this gene.

In the event a PGx interpretation is requested for CYP2D6 with SNPs but without CNV, Genedose will report CYP2D6 as "Not Tested."

The CYP2D6 CNV value or values are most commonly extracted from the [CopyCaller](#) software's output, and are generally distinct from the SNP/genotyping data. It is the laboratory or integrator's responsibility to extract and use the correct integer value from any decimal representation of CNV, for example, by using the "Copy Number Predicted" integer value from CopyCaller, as well as to consider the results of multiple CNV assay replicates with associated confidence/quality scores.

In addition to the ("global") exon 9 CYP2D6 CNV test, a lab *may* also perform tests for the CYP2D7 pseudogene CNV.

The CNV values may be provided to the GeneDose API using the names below.

Concept	Allowable names
exon 9 / "global" CNV (<i>always required for CYP2D6</i>)	CYP2D6 CYP2D6_EXON9 or other variations matching wildcard pattern "CYP2D6*EX*9" Hs00010001_cn
intron 2 / "CYP2D7" / "*36" CNV	CYP2D6_INTRON2 or other variations matching wildcard pattern "CYP2D6*INT*2"
5' UTR (support added with GeneDose API v3.10.0)	CYP2D6_FIVEPRIME CYP2D6_5PRIME
exon 1 (support added with GeneDose API v3.10.0)	CYP2D6_EXON1
intron 4	CYP2D6_INTRON4

(support added with GeneDose API v3.10.0)	
intron 6 (support added with GeneDose API v3.10.0)	CYP2D6_INTRON6 HS04502391_cn
intron 7 (support added with GeneDose API v3.10.0)	CYP2D6_INTRON7