Prepared by the Association for Molecular Pathology Training and Education Committee
For More Educational Resources: *www.amp.org/AMPEducation*

Molecular In My Pocket™…

AMP Recommendations for Pharmacogenetic Testing

Pharmacogenetics Tier 1 Recommended Alleles				
Allele	**Allele Functional Status†**	**Core Variant(s)‖**	**HGVS cDNA Nomenclature**	**Multiethnic Allele Frequency**
CYP2C9 (NM_000771.3)				
*CYP2C9*2*	Decreased function	rs1799853	c.430C>T	0-12%
*CYP2C9*3*	Decreased function	rs1057910	c.1075A>C	1-11%
*CYP2C9*5*	Decreased function	rs28371686	c.1080C>A	0-1%
*CYP2C9*6*	No function	rs9332131	c.818delA	0-1%
*CYP2C9*8*	Possibly decreased function	rs7900194	c.449G>A	0-5%
*CYP2C9*11*	Possibly decreased function	rs28371685	c.1003C>T	0-2%
CYP2C19 (NM_000769.2)				
*CYP2C19*2*	No function	rs4244285	c.681G>A	12-54%
*CYP2C19*3*	No function	rs4986893	c.636G>A	0.3-15%
*CYP2C19*17*	Increased function	rs12248560	c.-806C>T	4-21%
CYP2D6 (NM_000106.6)				
*CYP2D6*2*	Normal function	rs16947, rs1135840	c.886C>T, c.1457G>C	3.9-29.5%
*CYP2D6*3*	No function	rs35742686	c.775del	<0.1-1.6%
*CYP2D6*4*	No function	rs3892097	c.506-1G>A	0.5-18.5%
*CYP2D6*5*	No function	*CYP2D6* full gene deletion		1.6-5.4%
*CYP2D6*6*	No function	rs5030655	c.454del	0-1.1%
*CYP2D6*9*	Decreased function	rs5030656	c.841_843del	0-2.8%
*CYP2D6*10*	Decreased function	rs1065852, rs1135840	c.100C>T, c.1457G>C	1.4-43.6%
*CYP2D6*17*	Decreased function	rs28371706, rs16947, rs1135840	c.320C>T, c.886C>T, c.1457G>C	<0.1-19.3%
*CYP2D6*29*	Decreased function	rs59421388, rs61736512+ rs1058164, rs16947, rs1135840	c.1012G>A, c.406_408delinsATC, c.886C>T, c.1457G>C	0-12.1%
*CYP2D6*41*	Decreased function	rs28371725, rs16947, rs1135840	c.985+39G>A, c.886C>T, c.1457G>C	0.8-15.4%
*CYP2D6*xN*	variable, depending the duplicated alleles	duplications	variable	variable
VKORC1 (NM_024006.5)				
VKORC1 c.-1639G>A	Decreased gene expression	rs9923231	c.-1639G>A	10-88%

Legend: †Citations for assignment of function can be found at https://www.pharmvar.org (last accessed 9/28/2021), HGVS nomenclature https://www.ncbi.nlm.nih.gov/snp (last accessed 9/28/2021), ‖core variant(s) can be found at https://www.pharmvar.org (last accessed 9/28/2021), the characteristic variant associated with altered function and corresponding HGVS nomenclature for each star allele are underlined.

Revised 12/21

Pharmacogenetics Tier 2 Recommended Alleles				
Allele	**Allele Functional Status†**	**Core Variant(s)‖**	**HGVS cDNA Nomenclature**	**Multiethnic Allele Frequency**
CYP2C cluster region				
2C Cluster	unknown; variant in linkage disequilibrium with warfarin effect in individuals of West African ancestry	rs12777823	N/A	0-30%
CYP2C9 (NM_000771.3)				
*CYP2C9*12*	Possibly decreased function	rs9332239	c.1465C>T	0-0.3%
*CYP2C9*13*	Possibly decreased function	rs72558187	c.269T>C	0-0.2%
*CYP2C9*15*	No function	rs72558190	c.485C>A	0-0.01%
CYP2C19 (NM_000769.2)				
*CYP2C19*4A*	No function	rs28399504	c.1A>G	0.1-0.3%
*CYP2C19*4B*	No function	rs28399504; rs12248560	c.[-806C>T; 1A>G]	0-0.2%
*CYP2C19*5*	No function	rs56337013	c.1297C>T	0%
*CYP2C19*6*	No function	rs72552267	c.395G>A	0-0.1%
*CYP2C19*7*	No function	rs72558186	c.819+2T>A	0%
*CYP2C19*8*	No function	rs41291556	c.358T>C	0.1-0.3%
*CYP2C19*9*	Decreased function	rs17884712	c.431G>A	0.1-4.2%
*CYP2C19*10*	Decreased function	rs6413438	c.680C>T	0.1-6%
*CYP2C19*35*	No function	rs12769205	c.332-23A>G	0.8-3.1%
CYP2D6 (NM_000106.6)				
*CYP2D6*7*	No function	rs5030867	c.971A>C	0-0.6%
*CYP2D6*8*	No function	rs5030865, rs16947, rs1135840	c.505G>T, c.886C>T, c.1457G>C	0-0.1%
*CYP2D6*12*	No function	rs5030862, rs16947, rs1135840	c.124G>A, c.886C>T, c.1457G>C	0-1.7%
*CYP2D6*14*	Decreased function	rs5030865, rs16947, rs1135840	c.505G>A, c.886C>T, c.1457G>C	0-0.3%
*CYP2D6*15*	No function	rs774671100	c.137dup	0-0.6%
*CYP2D6*21*	No function	rs72549352, rs16947, rs1135840	c.805dup, c.886C>T, c.1457G>C	0-0.4%
*CYP2D6*31*	No function	rs267608319, rs16947, rs1135840	c.1319G>A, c.886C>T, c.1457G>C	0-0.8%
*CYP2D6*40*	No function	rs72549356, rs28371706, rs16947, rs1135840	c.514_522TTTCGCCCC[3], c.320C>T, c.886C>T, c.1457G>C	0-1.3%
*CYP2D6*42*	No function	rs72549346, rs16947, rs1135840	c.1088_1089dup, c.886C>T, c.1457G>C	0-0.5%
*CYP2D6*49*	Decreased function	rs1135822, rs1065852, rs1135840	c.358T>A, c.100C>T, c.1457G>C	0-1.1%
*CYP2D6*56*	No function	rs72549347, rs1135840	c.1030C>T, c.1457G>C	0-0.2%
*CYP2D6*59*	Decreased function	rs79292917, rs16947, rs1135840	c.975G>A, c.886C>T, c.1457G>C	0-0.7%
CYP2D6 Hybrid genes	no function	variable	variable	variable
CYP4F2 (NM_001082.4)				
*CYP4F2*3*	Possibly decreased function	rs2108622	c.1297G>A	10-40%
VKORC1 (NM_024006.5)				
VKORC1	Warfarin resistant	rs72547529	c.196G>A	0-0.25%
VKORC1	Warfarin resistant	rs61742245	c.106G>T	0-3.8%

Legend: †Citations for assignment of function can be found at https://www.pharmvar.org (last accessed 9/28/2021), HGVS nomenclature https://www.ncbi.nlm.nih.gov/snp (last accessed 9/28/2021), ‖core variant(s) can be found at https://www.pharmvar.org (last accessed 9/28/2021), the characteristic variant associated with altered function and corresponding HGVS nomenclature for each star allele are underlined.