



Whole Exome and Whole Genome Sequencing

Secondary Findings | Pharmacogenomics

Gene	Variant	cDNA	Protein	dbSNP rsID	NM
CYP2C19	*2	c.681G>A	p.P227= splicing	rs4244285	NM_000769.2
	*3	c.636G>A	p.W212X	rs4986893	NM_000769.2
	*4	c.1A>G	p.M1?	rs28399504	NM_000769.2
	*6	c.395G>A	p.Arg132Gln	rs56337013	NM_000769.2
	*8	c.358T>C	p.W120R	rs72552267	NM_000769.2
	*17	c.-806C>T	-	rs72558186	NM_000769.2
	*35	c.332-23A>G		rs41291556	NM_000769.2
CYP2C9	*2	c.430C>T	p.Arg144Cys	rs1799853	NM_000771.3
	*3	c.1075A>C	p.Ile359Leu	rs1057910	NM_000771.3
	*5	c.1080C>G	p.Asp360Glu	rs28371686	NM_000771.3
	*6	c.818delA	-	rs9332131	NM_000771.3
	*8	c.449G>A	p.Arg150His	rs7900194	NM_000771.3
	*11	c.1003C>T	p.Arg335Trp	rs28371685	NM_000771.3
CYP2D6	*2	c.886C>T	p.Arg296Cys	rs16947	NM_000106.5
	*3	c.775del		rs35742686	NM_000106.5
	*4	c.506-1G>A		rs3892097	NM_000106.5
	*6	c.454del		rs5030655	NM_000106.5
	*7	c.971A>C	p.His324Pro	rs5030867	NM_000106.5
	*9	c.841_843delAAG		rs5030656	NM_000106.5
	*10	c.100C>T	p.Pro34Ser	rs1065852	NM_000106.5
	*17	c.320C>T	p.Thr107Ile	rs28371706	NM_000106.5
*41	c.985+39G>A		rs28371725	NM_000106.5	

Gene	Variant	cDNA	Protein	dbSNP rsID	NM
CYP3A5	*6	c.624G>A	p.K208=	rs10264272	NM_000777.4
	*7	c.1035dupT		rs41303343	NM_000777.4
SLCO1B1	*37	c.388A>G	p.Asn130Asp	rs2306283	NM_006446.4
	*5	c.521T>C	p.Val174Ala	rs4149056	NM_006446.4
	*15	c.388A>G	p.Asn130Asp	rs2306283	NM_006446.4
		c.521T>C	p.Val174Ala	rs4149056	NM_006446.4
TPMT	*2	c.238G>C	p.Ala80Pro	rs1800462	NM_000367.3
	*3A	c.460G>A	p.Ala154Thr	rs1800460	NM_000367.3
		c.719A>G	p.Tyr240Cys	rs1142345	NM_000367.3
	*3B	c.460G>A	p.Ala154Thr	rs1800460	NM_000367.3
	*3C	c.719A>G	p.Tyr240Cys	rs1142345	NM_000367.3
UGT1A1	*6	c.211G>A	p.Gly71Arg	rs4148323	NM_000463.2
	*28	c.-53_-52TA (7)		rs8175347	NM_000463.2
	*36	c.-53_-52TA (5)		rs8175347	NM_000463.2
	*37	c.-53_-52 (8)		rs8175347	NM_000463.2
NUDT15	*2	c.50_55dup	p.Gly17_Val18dup	rs61746486	NM_018283.4
		c.415C>T	p.Arg139Cys	rs116855232	NM_018283.4
	*3	c.415C>T	p.Arg139Cys	rs116855232	NM_018283.4
	*9	c.50_55del	p.Gly17_Val18del	rs746071566	NM_018283.4

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