

Laaja farmakogeneettinen testipaneeli (B- Farma-D, 12533) geenivariaatiot

Agena PGx Farmakogenetiikan paneeli sisältää seuraavat geenivariaatiot ja SNP:t. Abomics Pharmacogenetic (PGx) Interpretation Service-tulkintapalvelu antaa lausunnon **vahvennettuna** merkityistä geenivariaatioista. Uusia variantteja ja geenejä voidaan lisätä ym. paneeliin, kun uutta tietoa julkaistaan tai kun Agena päivittää tulkintapalveluaan.

Gene	SNP	Variantti	Nukleotidi
ABCB1	rs1045642		ABCB1 c.3435C>T
APOE	rs429358	C130R	APOE c.388T>C
APOE	rs7412	R176C	APOE c.526C>T
COMT	rs4680	472G>A	COMT c.322G>A or c.472G>A
CYP1A2	rs72547513	*11	CYP1A2*11 g.558C>A
CYP1A2	rs2069514	*1C	CYP1A2*1 g.-3860G>A
CYP1A2	rs762551	*1F	CYP1A2*1F g.-163C>A
CYP1A2	rs12720461	*1K	CYP1A2*1K g.-729C>T
CYP1A2	rs56107638	*7	CYP1A2*7 g.3533G>A
CYP2B6	rs28399499	*18	CYP2B6*16/*18 c.983T>C g.21011T>C
CYP2B6	rs3745274	*6	CYP2B6*6 c.516G>A
CYP2C19	rs12248560	*17	CYP2C19*17 g.-806C>T
CYP2C19	rs4244285	*2	CYP2C19*2 c.681G>A g.19154G>A
CYP2C19	rs4986893	*3	CYP2C19*3 c.636G>A g.17948G>A
CYP2C19	rs28399504	*4	CYP2C19*4 c.1A>G g. A>G
CYP2C19	rs56337013	*5	CYP2C19*5 c.1297C>T g.90033C>T
CYP2C19	rs72552267	*6	CYP2C19*6 c.395G>A g.12748G>A
CYP2C19	rs72558186	*7	CYP2C19*7 g.19294T>A
CYP2C19	rs41291556	*8	CYP2C19*8 c.358T>C g.12711T>C
CYP2C9	rs28371685	*11	CYP2C9*11 c.1003C>T g.42542C>T
CYP2C9	rs9332239	*12	CYP2C9*12 c.1465C>T

Gene	SNP	Variantti	Nukleotidi
CYP2C9	rs72558187	*13	CYP2C9*13 c.269T>C g.3276T>C
CYP2C9	rs72558190	*15	CYP2C9 *15 c.485C>A
CYP2C9	rs1799853	*2	CYP2C9*2 c.430C>T.g.3608C>T
CYP2C9	rs1057910	*3	CYP2C9*3/*18 c.1075A>C g.42614A>C
CYP2C9	rs56165452	*4	CYP2C9*4 c. 1076T>C g.42615T>C
CYP2C9	rs28371686	*5	CYP2C9*5 c.1080C>G g.42619C>G
CYP2C9	rs9332131	*6	CYP2C9*6 c.818delA g.10601delA
CYP2C9	rs7900194	*8	CYP2C9*8 c.449G>A g.3627G>A
CYP2D6	rs1065852	*10	CYP2D6*10 g.100C>T
	rs201377835 or		
CYP2D6	rs5030863	*11	CYP2D6*11 c.883G>C
CYP2D6	rs5030862	*12	CYP2D6*12 g.124G>A
CYP2D6	rs5030865	*14	CYP2D6*8 g.1758G>T
CYP2D6	rs72549357	*15	CYP2D6*15 g.137-138insT
CYP2D6	rs28371706	*17	CYP2D6*17 g.1023C>T
CYP2D6	dup4125_4133	*18	CYP2D6*18 c.dup4125-4133
CYP2D6	rs72549353	*19	CYP2D6*19 c.2539_2542delAACT
CYP2D6	rs16947	*2	CYP2D6*2 g.2850C>T
CYP2D6	rs1135840	*2	CYP2D6*2 g.4180G>C
CYP2D6	rs72549354	*20	CYP2D6*20 c.1973_1974insG
CYP2D6	rs59421388	*29	CYP2D6*29 g. 3183G>A
CYP2D6	rs35742686	*3	CYP2D6*3 g.2549delA
CYP2D6	rs3892097	*4	CYP2D6*4 g.1846G>A
CYP2D6	rs28371725	*41	CYP2D6*41 g.2988G>A
CYP2D6	rs28371735	*36	CYP2D6*41 g.4155C>T
CYP2D6	rs5030655	*6	CYP2D6*6 g.1707delT
CYP2D6	rs5030867	*7	CYP2D6*7 g.2935A>C
CYP2D6	rs5030865	*8	CYP2D6*8 g.1758G>T
			CYP2D6*9 g.2613_2615delAGA
CYP2D6	rs5030656	*9	(g.2615_2617delAAG)
CYP2D6	CNV	CNV	
CYP3A4	rs4987161	*17	CYP3A4*17 c.566T>C g.15615T>C
CYP3A4	rs55785340	*2	CYP3A4*2 c.664T>C g.15713T>C
CYP3A4	rs35599367	*22	CYP3A4*22 g.15389C>T
CYP3A5	rs28365083	*2	CYP3A5*2 g.27289C>A
CYP3A5	rs776746	*3	CYP3A5*3/*10 g.6986A>G
CYP3A5	rs10264272	*6	CYP3A5*6 c.624G>A

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CYP3A5	rs41303343	*7	CYP3A5*7 g.27131_27132insT
DRD2	rs1800497	Taq1A	DRD2 c.2137G>A
F2	rs1799963	G20210A	F2 c.*97G>A
F5	rs6025	R506Q	F5 c.1601G>A
GLP1R	rs1042044	A>C	GLP1R c.780A>C/T // Leu260Phe
GLP1R	rs6923761	A>G	GLP1R c.502G>A/C //Gly168Arg/Gly168Ser
GLP1R	rs2300615		GLP1R c.510-1135T>G
MTHFR	rs1801131	1286A>C	MTHFR A1298C Glu429Ala
MTHFR	rs1801133	665C>T	MTHFR C677T Ala222Val
OPRM1	rs1799971	118 A>G	OPRM1 A118G Asn40Asp
PNPLA5	rs5764010	C>T	PNPLA5 C>T
SLCO1B1	rs4149056	*5	SLCO1B1 g.37041T>C
SULT4A1	rs763120		SULT4A1 c.*1113A>G
VKORC1	rs9923231	1639 G>A	VKORC1 c.-1639G>A