

PHARMACOGENETIC PROFICIENCY TESTING 1

INFORME INDIVIDUAL

Código del laboratorio participante: L6

Técnicas empleadas en el análisis: PCR en tiempo real (Taqman)

Gen	rsID	C-3	Resultados L6 (C-3)		C-6	Resultados L6 (C-6)	
			Genotipo	Diplotipo		Genotipo	Diplotipo
<i>CYP2C19</i>	rs12248560	CC	CC	*1/*2	CT	CT	*2/*17
	rs4244285	GA	AG		GA	AG	
	rs4986893	GG	GG		GG	GG	
<i>CYP2C8</i>	rs10509681	CT	CT	*1/*3	TT	TT	*1/*2
	rs1058930	GG	GG		GG	GG	
	rs11572103	TT	TT		TA	AT	
<i>CYP2C9</i>	rs1057910	AA	AA	*1/*2	AA	AA	*1/*1
	rs1799853	CT	CT		CC	CC	
<i>CYP2D6</i>	rs16947	GG	GG	*3/*4	AA	AA	*17/*29
	rs35742686	A-	T-		AA	TT	
	rs3892097	AG	CT		GG	CC	
	rs1065852	CT	AG		CC	GG	
	rs28371725	GG	CC		GG	CC	
	rs5030655	TT	AA		TT	AA	
	rs5030656	AAG/AAG	TTC/TTC		AAG/AAG	TTC/TTC	
CNVs	2	2	2	2			
<i>CYP3A4</i>	rs2740574	TT	TT	*1A/*1A	CC	CC	*1B/*1B
	rs35599367	GG	GG		GG	GG	
<i>CYP3A5</i>	rs10264272	CC	CC	*3/*3	CT	CT	*1/*6
	rs776746	GG	CC		AA	TT	
<i>DPYD</i>	rs3918290	CC	CC		CC	CC	
	rs55886062	AA	AA		AA	AA	
	rs67376798	TT	TT		TT	TT	
<i>ABCB1</i>	rs2032582	GG	CC		GG	CC	
	rs1045642	AG	AG		GG	GG	
<i>MTHFR</i>	rs1801131	TG	GT		TT	TT	
	rs1801133	GA	GA		GG	GG	
<i>TPMT</i>	rs1800462	CC	CC	*1/*1	CC	CC	*1/*1
	rs1800584	CC	CC		CC	CC	
	rs1142345	TT	TT		TT	TT	
	rs1800460	CC	CC		CC	CC	

Gen	rsID	C-7	Resultados L6 (C-7)		C-9	Resultados L6 (C-9)	
			Genotipo	Diplotipo		Genotipo	Diplotipo
CYP2C19	rs12248560	TT	TT	*17/*17	CT	CT	*3/*17
	rs4244285	GG	GG		GG	GG	
	rs4986893	GG	GG		AG	AG	
CYP2C8	rs10509681	TT	TT	*2/*2	TT	TT	*1/*1
	rs1058930	GG	GG		GG	GG	
	rs11572103	AA	AA		TT	TT	
CYP2C9	rs1057910	AA	AA	*1/*1	AA	AA	*1/*1
	rs1799853	CC	CC		CC	CC	
CYP2D6	rs16947	AA	AA	*2A/*5	GG	GG	*10/*10X2
	rs35742686	AA	TT		AA	TT	
	rs3892097	GG	CC		GG	CC	
	rs1065852	CC	GG		TT	AA	
	rs28371725	GG	CC		GG	CC	
	rs5030655	TT	AA		TT	AA	
	rs5030656	AAG/AAG	TTC/TTC		AAG/AAG	TTC/TTC	
	CNVs	1	1		3	3	
CYP3A4	rs2740574	CC	CC	*1B/*1B	TT	TT	*1A/*1A
	rs35599367	GG	GG		GG	GG	
CYP3A5	rs10264272	CC	CC	*1/*7	CC	CC	*3/*3
	rs776746	AA	TT		GG	CC	
DPYD	rs3918290	CC	CC		CC	CC	
	rs55886062	AA	AA		AA	AA	
	rs67376798	TT	TT		TT	TT	
ABCB1	rs2032582	GG	CC		GT	CA	
	rs1045642	GG	GG		AG	AG	
MTHFR	rs1801131	TT	TT		TT	TT	
	rs1801133	GG	GG		GG	GG	
TPMT	rs1800462	CC	CC	*3C/*3C	CC	CC	*1/*1
	rs1800584	CC	CC		CC	CC	
	rs1142345	CC	CC		TT	TT	
	rs1800460	CC	CC		CC	CC	

*Se pueden identificar discrepancias al haber analizado una hebra diferente

Plantilla de Resultados por Diplotipos

Se muestra a continuación, a título informativo, la plantilla de resultados por diplotipos

Gen	rsID	Resultados			
		C-3	C-6	C-7	C-9
<i>CYP2C19</i>	rs12248560	*1/*2	*2/*17	*17/*17	*3/*17
	rs4244285				
	rs4986893				
<i>CYP2C8</i>	rs10509681	*1/*3	*1/*2	*2/*2	*1/*1
	rs1058930				
	rs11572103				
<i>CYP2C9</i>	rs1057910	*1/*2	*1/*1	*1/*1	*1/*1
	rs1799853				
<i>CYP2D6</i>	rs16947	*3/*4	(*17)/(*29)	*2/*5	*10/*10xN
	rs35742686				
	rs3892097				
	rs1065852				
	rs28371725				
	rs5030655				
	rs5030656				
CNVs					
<i>CYP3A4</i>	rs2740574	*1/*1	*1B/*1B	*1B/*1B	*1/*1
	rs35599367				
<i>CYP3A5</i>	rs10264272	*3/*3	*1/*6	*1/*7	*3/*3
	rs776746				
<i>DPYD</i>	rs3918290	*1/*4	*1/*9	*1/*1	*1/*1
	rs55886062				
	rs67376798				
<i>ABCB1</i>	rs2032582	GG	GG	GG	GT
	rs1045642	AG	GG	GG	AG
<i>MTHFR</i>	rs1801131	TG	TT	TT	TT
	rs1801133	GA	GG	GG	GG
<i>TPMT</i>	rs1800462	*1/*1	*1/*1	*3C/*3C	*1/*1
	rs1800584				
	rs1142345				
	rs1800460				