

## PHARMACOGENETIC PROFICIENCY TESTING 1

### INFORME INDIVIDUAL

Código del laboratorio participante: L11

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

Gen	rsID	C-3	Resultados L11 (C-3)		C-6	Resultados L11 (C-6)	
			Genotipo	Diplotipo		Genotipo	Diplotipo
<b>CYP2C19</b>	rs12248560	CC	CC	*2/*1	CT	CT	*2/*17
	rs4244285	GA	GA		GA	GA	
	rs4986893	GG			GG		
<b>CYP2C8</b>	rs10509681	CT			TT		
	rs1058930	GG			GG		
	rs11572103	TT			TA		
<b>CYP2C9</b>	rs1057910	AA	AA	*2/*1	AA	AA	*1/*1
	rs1799853	CT	CT		CC	CC	
<b>CYP2D6</b>	rs16947	GG		*3/*4	AA		*1/*1
	rs35742686	A-	T/-		AA	TT	
	rs3892097	AG	CT		GG	CC	
	rs1065852	CT	GA		CC	GG	
	rs28371725	GG	CC		GG	CC	
	rs5030655	TT	AA		TT	AA	
	rs5030656	AAG/AAG	CTT/CTT		AAG/AAG	CTT/CTT	
	CNVs	2	2		2	2	
<b>CYP3A4</b>	rs2740574	TT			CC		
	rs35599367	GG			GG		
<b>CYP3A5</b>	rs10264272	CC			CT		
	rs776746	GG			AA		
<b>DPYD</b>	rs3918290	CC			CC		
	rs55886062	AA			AA		
	rs67376798	TT			TT		
<b>ABCB1</b>	rs2032582	GG			GG		
	rs1045642	AG	GA		GG	GG	
<b>MTHFR</b>	rs1801131	TG			TT		
	rs1801133	GA			GG		
<b>TPMT</b>	rs1800462	CC			CC		
	rs1800584	CC			CC		
	rs1142345	TT			TT		
	rs1800460	CC			CC		

Gen	rsID	C-7	Resultados L11 (C-7)		C-9	Resultados L11 (C-9)	
			Genotipo	Diplotipo		Genotipo	Diplotipo
<b>CYP2C19</b>	rs12248560	TT	<b>TT</b>	<b>*17/*17</b>	CT	<b>CT</b>	<b>*1/*17</b>
	rs4244285	GG	<b>GG</b>		GG	<b>GG</b>	
	rs4986893	GG			AG		
<b>CYP2C8</b>	rs10509681	TT			TT		
	rs1058930	GG			GG		
	rs11572103	AA			TT		
<b>CYP2C9</b>	rs1057910	AA	<b>AA</b>	<b>*1/*1</b>	AA	<b>AA</b>	<b>*1/*1</b>
	rs1799853	CC	<b>CC</b>		CC	<b>CC</b>	
<b>CYP2D6</b>	rs16947	AA		<b>*1/*5</b>	GG		<b>*10/10X2 +híbrido</b>
	rs35742686	AA	<b>TT</b>		AA	<b>TT</b>	
	rs3892097	GG	<b>CC</b>		GG	<b>CC</b>	
	rs1065852	CC	<b>GG</b>		TT	<b>AA</b>	
	rs28371725	GG	<b>CC</b>		GG	<b>CC</b>	
	rs5030655	TT	<b>AA</b>		TT	<b>AA</b>	
	rs5030656	AAG/AAG	<b>CTT/CTT</b>		AAG/AAG	<b>CTT/CTT</b>	
	CNVs	1	<b>1</b>		3	<b>3 (EXON 9), 4 (INTRON2)</b>	
<b>CYP3A4</b>	rs2740574	CC			TT		
	rs35599367	GG			GG		
<b>CYP3A5</b>	rs10264272	CC			CC		
	rs776746	AA			GG		
<b>DPYD</b>	rs3918290	CC			CC		
	rs55886062	AA			AA		
	rs67376798	TT			TT		
<b>ABCB1</b>	rs2032582	GG			GT		
	rs1045642	GG	<b>GG</b>		AG	<b>GA</b>	
<b>MTHFR</b>	rs1801131	TT			TT		
	rs1801133	GG			GG		
<b>TPMT</b>	rs1800462	CC			CC		
	rs1800584	CC			CC		
	rs1142345	CC			TT		
	rs1800460	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	<b>*1/*2</b>	<b>*2/*17</b>	<b>*17/*17</b>	<b>*3/*17</b>
	rs4244285				
	rs4986893				
<i>CYP2C8</i>	rs10509681	<b>*1/*3</b>	<b>*1/*2</b>	<b>*2/*2</b>	<b>*1/*1</b>
	rs1058930				
	rs11572103				
<i>CYP2C9</i>	rs1057910	<b>*1/*2</b>	<b>*1/*1</b>	<b>*1/*1</b>	<b>*1/*1</b>
	rs1799853				
<i>CYP2D6</i>	rs16947	<b>*3/*4</b>	<b>(*17)/(*29)</b>	<b>*2/*5</b>	<b>*10/*10xN</b>
	rs35742686				
	rs3892097				
	rs1065852				
	rs28371725				
	rs5030655				
	rs5030656				
CNVs					
<i>CYP3A4</i>	rs2740574	<b>*1/*1</b>	<b>*1B/*1B</b>	<b>*1B/*1B</b>	<b>*1/*1</b>
	rs35599367				
<i>CYP3A5</i>	rs10264272	<b>*3/*3</b>	<b>*1/*6</b>	<b>*1/*7</b>	<b>*3/*3</b>
	rs776746				
<i>DPYD</i>	rs3918290	<b>*1/*4</b>	<b>*1/*9</b>	<b>*1/*1</b>	<b>*1/*1</b>
	rs55886062				
	rs67376798				
<i>ABCB1</i>	rs2032582	<b>GG</b>	<b>GG</b>	<b>GG</b>	<b>GT</b>
	rs1045642	<b>AG</b>	<b>GG</b>	<b>GG</b>	<b>AG</b>
<i>MTHFR</i>	rs1801131	<b>TG</b>	<b>TT</b>	<b>TT</b>	<b>TT</b>
	rs1801133	<b>GA</b>	<b>GG</b>	<b>GG</b>	<b>GG</b>
<i>TPMT</i>	rs1800462	<b>*1/*1</b>	<b>*1/*1</b>	<b>*3C/*3C</b>	<b>*1/*1</b>
	rs1800584				
	rs1142345				
	rs1800460				