

Supplementary information 2.2  
Pharmacogenomics genotype and  
diplotype calls

Patient ID	Diplotype	rs1799853	rs1057910	rs28371686	rs9332131	rs7900194	rs28371685	Notes
		10:94942290	10:94981296	10:94981301	10:94949282	10:94942309	10:94981224	
		g.94942290C>T p.Arg144Cys	g.94981296A>C p.Ile359Leu	g.94981301C>G p.Asp360Glu	g.94949282delA p.Lys273Argfs	g.94942309G>A p.Arg150His	g.94981224C>T p.Arg355Trp	
	*1	C	A	C	A	G	C	
	*2	T	A	C	A	G	C	
	*3	C	C	C	A	G	C	
	*5	C	A	G	A	G	C	
	*6	C	A	C	del A	G	C	
	*8	C	A	C	A	A	C	
	*11	C	A	C	A	G	T	
<b>BBS-001</b>	*1/*1	CC	AA	CC	AA	GG	CC	
<b>BBS-002</b>	*1/*2	CT	AA	CC	AA	GG	CC	
<b>BBS-003</b>	*1/*1	CC	AA	CC	AA	GG	CC	
<b>BBS-004</b>	*1/*2	CT	AA	CC	AA	GG	CC	
<b>BBS-005</b>	*1/*1	CC	AA	CC	AA	GG	CC	
<b>BBS-006</b>	*1/*2	CT	AA	CC	AA	GG	CC	
<b>BBS-007</b>	*1/*1	CC	AA	CC	AA	GG	CC	
<b>BBS-008</b>	*1/*3	CC	AC	CC	AA	GG	CC	
<b>BBS-009</b>	*1/*1	CC	AA	CC	AA	GG	CC	
<b>BBS-010</b>	*1/*1	CC	AA	CC	AA	GG	CC	
<b>BBS-011</b>	*1/*1	CC	AA	CC	AA	GG	CC	
<b>BBS-012</b>	*1/*1	CC	AA	CC	AA	GG	CC	
<b>BBS-013</b>	*1/*2	CT	AA	CC	AA	GG	CC	
<b>BBS-014</b>	*1/*2	CT	AA	CC	AA	GG	CC	
<b>BBS-015</b>	*1/*1	CC	AA	CC	AA	GG	CC	
<b>BBS-016</b>	*1/*3	CC	AC	CC	AA	GG	CC	
<b>BBS-017</b>	*1/*3	CC	AC	CC	AA	GG	CC	
<b>BBS-018</b>	*1/*1	CC	AA	CC	AA	GG	CC	
<b>IBD-001</b>	*1/*3	CC	AC	CC	AA	GG	CC	
<b>IBD-002</b>	*1/*1	CC	AA	CC	AA	GG	CC	
<b>IBD-003</b>	*3/*3	CC	CC	CC	AA	GG	CC	
<b>IBD-004</b>	*1/*1	CC	AA	CC	AA	GG	CC	
<b>IBD-005</b>	*1/*1	CC	AA	CC	AA	GG	CC	

Table A4.1: Diplotypes for *CYP2C9*

Patient ID	Diplotype	rs1799853	rs1057910	rs28371686	rs9332131	rs7900194	rs28371685	Notes
IBD-006	*1/*3	CC	AC	CC	AA	GG	CC	
IBD-007	*1/*2	CT	AA	CC	AA	GG	CC	
IBD-008	*1/*1	CC	AA	CC	AA	GG	CC	
IBD-009	*1/*1	CC	AA	CC	AA	GG	CC	
IBD-010	*1/*2	CT	AA	CC	AA	GG	CC	
IBD-011	*1/*3	CC	AC	CC	AA	GG	CC	
IBD-012	*1/*1	CC	AA	CC	AA	GG	CC	
IBD-013	*1/*1	CC	AA	CC	AA	GG	CC	
IBD-014	*1/*1	CC	AA	CC	AA	GG	CC	
IBD-015	*1/*3	CC	AC	CC	AA	GG	CC	
IBD-016	*1/*1	CC	AA	CC	AA	GG	CC	
IBD-017	*1/*1	CC	AA	CC	AA	GG	CC	
IBD-018	*1/*2	CT	AA	CC	AA	GG	CC	
IBD-019	*1/*1	CC	AA	CC	AA	GG	CC	
IBD-020	*1/*2	CT	AA	CC	AA	GG	CC	
JDM-001	*2/*3	CT	AC	CC	AA	GG	CC	
JDM-002	*1/*1	CC	AA	CC	AA	GG	CC	
JDM-003	*1/*3	CC	AC	CC	AA	GG	CC	
JDM-004	*1/*1	CC	AA	CC	AA	GG	CC	
JDM-005	*1/*3	CC	AC	CC	AA	GG	CC	
JDM-006	*1/*2	CT	AA	CC	AA	GG	CC	
JDM-007	*1/*1	CC	AA	CC	AA	GG	CC	
JDM-008	*1/*2	CT	AA	CC	AA	GG	CC	
JDM-009	*1/*3	CC	AC	CC	AA	GG	CC	
JDM-010	*1/*1	CC	AA	CC	AA	GG	CC	
JDM-011	*1/*11	CC	AA	CC	AA	GG	CT	
JDM-012	*1/*1	CC	AA	CC	AA	GG	CC	
JDM-013	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-001	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-002	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-003	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-004	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-005	*1/*1	CC	AA	CC	AA	GG	CC	

Table A4.1 continued: Diplotypes for *CYP2C9*

Patient ID	Diplotype	rs1799853	rs1057910	rs28371686	rs9332131	rs7900194	rs28371685	Notes
SRS-006	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-008	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-009	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-010	*1/*2	CT	AA	CC	AA	GG	CC	
SRS-011	*1/*12	CC	AA	CC	AA	GG	CC	Astrolabe
SRS-012	*1/*2	CT	AA	CC	AA	GG	CC	
SRS-013	*1/*2	CT	AA	CC	AA	GG	CC	
SRS-014	*1/*2	CT	AA	CC	AA	GG	CC	
SRS-015	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-016	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-017	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-018	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-019	*1/*2	CT	AA	CC	AA	GG	CC	
SRS-020	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-021	*1/*2	CT	AA	CC	AA	GG	CC	
SRS-022	*1/*3	CC	AC	CC	AA	GG	CC	
SRS-023	*1/*3	CC	AC	CC	AA	GG	CC	
SRS-024	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-025	*1/*3	CC	AC	CC	AA	GG	CC	
SRS-026	*1/*3	CC	AC	CC	AA	GG	CC	
SRS-027	*1/*1	CC	AA	CC	AA	GG	CC	
SRS-028	*1/*2	CT	AA	CC	AA	GG	CC	
SRS-029	*1/*2	CT	AA	CC	AA	GG	CC	
SRS-030	*1/*1	CC	AA	CC	AA	GG	CC	
USH-001	*1/*1	CC	AA	CC	AA	GG	CC	
USH-002	*1/*1	CC	AA	CC	AA	GG	CC	
USH-003	*1/*1	CC	AA	CC	AA	GG	CC	

Table A4.1 continued: Diplotypes for *CYP2C9*

Patient ID	Diplotype	rs4244285	rs4986893	rs28399504	rs56337013	rs72552267	rs72558186	rs41291556	rs12248560	Notes
		10:94781859	10:94780653	10:94762706	10:94852738	10:94775453	10:94781999	10:94775416	10:94761900	
		g.94781859 G>A p.Pro227=	g.9480653 G>A p.Trp212Ter	g.94762706 A>G p.Met1Leu	g.94852738 C>T p.Arg433Trp	g.94775453 G>A p.Arg132Gln	g.94781999 T>A	g.94775416 T>C p.Trp120Arg	g.94761900 C>T	
	*1	G	G	A	C	G	T	T	C	
	*2	A	G	A	C	G	T	T	C	
	*3	G	A	A	C	G	T	T	C	
	*4A	G	G	G	C	G	T	T	C	
	*4B	G	G	G	C	G	T	T	T	
	*5	G	G	A	T	G	T	T	C	
	*6	G	G	A	C	A	T	T	C	
	*7	G	G	A	C	G	A	T	C	
	*8	G	G	A	C	G	T	C	C	
	*17	G	G	A	C	G	T	T	T	
<b>BBS-001</b>	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
<b>BBS-002</b>	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
<b>BBS-003</b>	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
<b>BBS-004</b>	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
<b>BBS-005</b>	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
<b>BBS-006</b>	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
<b>BBS-007</b>	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
<b>BBS-008</b>	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
<b>BBS-009</b>	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
<b>BBS-010</b>	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
<b>BBS-011</b>	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
<b>BBS-012</b>	*17/*17	GG	GG	AA	CC	GG	TT	TT	TT	
<b>BBS-013</b>	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
<b>BBS-014</b>	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
<b>BBS-015</b>	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
<b>BBS-016</b>	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
<b>BBS-017</b>	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
<b>BBS-018</b>	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
<b>IBD-001</b>	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
<b>IBD-002</b>	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	

Table A4.2: Diplotypes for *CYP2C19*

Patient ID	Diplotype	rs4244285	rs4986893	rs28399504	rs56337013	rs72552267	rs72558186	rs41291556	rs12248560	Notes
IBD-003	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
IBD-004	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
IBD-005	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
IBD-006	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
IBD-007	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
IBD-008	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
IBD-009	*2/*2	AA	GG	AA	CC	GG	TT	TT	CC	
IBD-010	*1/*15	GG	GG	AA	CC	GG	TT	TT	CC	Astrolabe
IBD-011	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
IBD-012	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
IBD-013	*2/*17	GA	GG	AA	CC	GG	TT	TT	CT	
IBD-014	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
IBD-015	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
IBD-016	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
IBD-017	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
IBD-018	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
IBD-019	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
IBD-020	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
JDM-001	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
JDM-002	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
JDM-003	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
JDM-004	*17/*17	GG	GG	AA	CC	GG	TT	TT	TT	
JDM-005	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
JDM-006	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
JDM-007	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
JDM-008	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
JDM-009	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
JDM-010	*17/*17	GG	GG	AA	CC	GG	TT	TT	TT	
JDM-011	*2/*17	GA	GG	AA	CC	GG	TT	TT	CT	
JDM-012	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
JDM-013	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
SRS-001	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
SRS-002	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	

Table A4.2 continued: Diplotypes for *CYP2C19*

Patient ID	Diplotype	rs4244285	rs4986893	rs28399504	rs56337013	rs72552267	rs72558186	rs41291556	rs12248560	Notes
SRS-003	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
SRS-004	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
SRS-005	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
SRS-006	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
SRS-007	*2/*17	GA	GG	AA	CC	GG	TT	TT	CT	
SRS-008	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
SRS-009	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
SRS-010	*2/*17	GA	GG	AA	CC	GG	TT	TT	CT	
SRS-011	*2/*17	GA	GG	AA	CC	GG	TT	TT	CT	
SRS-012	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
SRS-013	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
SRS-014	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
SRS-015	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
SRS-016	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
SRS-017	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
SRS-018	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
SRS-019	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
SRS-020	*1/*17	GG	GG	AA	CC	GG	TT	C	CT	
SRS-021	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
SRS-022	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
SRS-023	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
SRS-024	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
SRS-025	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
SRS-026	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
SRS-027	*1/*2	GA	GG	AA	CC	GG	TT	TT	CC	
SRS-028	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
SRS-029	*1/*1	GG	GG	AA	CC	GG	TT	TT	CC	
SRS-030	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
USH-001	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
USH-002	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	
USH-003	*1/*17	GG	GG	AA	CC	GG	TT	TT	CT	

Table A4.2 continued: Diplotypes for *CYP2C19*

Patient ID	Diplotype	rs16947	rs1135840	rs35742686	rs1065852	rs3892097	rs5030655	rs5030656	rs28371706	rs28371725	rs769258	Notes
		22:42127941	22:42126611	22:42128242	22:42130692	22:42128945	22:42129084	22:42128174-42128176	22:42129770	22:42127803	22:42130761	
		g.42127941 G>A p.Arg296Cys	g.42126611 C>G p.Ser486Thr	g.42128242 delT p.Arg259Glyfs	g.42130692 G>A p.Pro34Ser	g.42128945 C>T	g.42129084 delA p.Trp152Glyfs	g.42128174_42128176 delCTT p.Lys281del	g.42129770 G>A p.Thr107Asn	g.42127803 C>T	g.42130761 C>T p.Val11Met	
*1		G	C	T	G	C	A	CTT	G	C	C	
*1xN												
*2		A	G									
*2xN		A	G									
*3				delT								
*3xN				delT								
*4			G		A	T						
*4xN			G		A	T						
*4J/*4P					A	T						
*4K		A	G		A	T						
*4M						T						
*5		delGene	delGene	delGene	delGene	delGene	delGene	delGene	delGene	delGene	delGene	
*6							delA					
*6xN							delA					
*6C			G				delA					
*9								delCTT				
*9x2								delCTT				
*10			G		A							
*10x2			G		A							
*17		A	G						A			
*17x2		A	G						A			
*35		A	G								T	
*41		A	G							T		
*41x2		A	G							T		
<b>BBS-001</b>	*1/*4	GG	GC	TT	GA	CT	AA	CTT/CTT	GG	CC	CC	

Table A4.3: Diplotypes for *CYP2D6*



Patient ID	Diplotype	rs16947	rs1135840	rs35742686	rs1065852	rs3892097	rs5030655	rs5030656	rs28371706	rs28371725	rs769258	Notes
BBS-002	*1/*41	GA	GC	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	
BBS-003	*1/*41	GA	GC	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	
BBS-004	*1/*1	GG	CC	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
BBS-005	*1/*1	GG	CC	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
BBS-006	*1/*9	GG	CC	TT	GG	CC	AA	delCTT (het)	GG	CC	CC	
BBS-007	*4/*4	GG	GG	TT	AA	TT	AA	CTT/CTT	GG	CC	CC	
BBS-008	*4/*4	GG	GG	TT	AA	TT	AA	CTT/CTT	GG	CC	CC	
BBS-009	*1/*41	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	
BBS-010	*1/*41	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	
BBS-011	*1/*41	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	
BBS-012	*1/*41	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	
BBS-013	*4/*4	GG	GG	TT	AA	TT	AA	CTT/CTT	GG	CC	CC	
BBS-014	*1/*2	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
BBS-015	*2/*35	AA	GG	TT	GG	CC	AA	CTT/CTT	GG	CC	CT	Astrolabe
BBS-016	*2/*2	AA	GG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
BBS-017	*2/*2	AA	GG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
BBS-018	*1/*4	GG	CG	TT	GA	CT	AA	CTT/CTT	GG	CC	CC	
IBD-001	*1/*1	GG	CC	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
IBD-002	*2/*2	AA	GG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
IBD-003	*1/*41	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	
IBD-004	*1/*1	GG	CC	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
IBD-005	*2/*4	GA	GG	TT	GA	CT	AA	CTT/CTT	GG	CC	CC	
IBD-006	*4/*4	GG	GG	TT	AA	TT	AA	CTT/CTT	GG	CC	CC	
IBD-007	*2/*41x2	AA	GG	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	Astrolabe
IBD-008	*1x2/*17	GA	CG	TT	GG	CC	AA	CTT/CTT	GA	CC	CC	Astrolabe
IBD-009	*1/*4	GG	CG	TT	GA	CT	AA	CTT/CTT	GG	CC	CC	
IBD-010	*1/*10	GG	CG	TT	GA	CC	AA	CTT/CTT	GG	CC	CC	
IBD-011	*1/*4	GG	CG	TT	GA	CT	AA	CTT/CTT	GG	CC	CC	
IBD-012	*1/*2	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
IBD-013	*1/*5	Del gene	Del gene	Del gene	Del gene	Del gene	Del gene	Del gene	Del gene	Del gene	Del gene	Astrolabe

Table A4.3 continued: Diplotypes for *CYP2D6*

Patient ID	Diplotype	rs16947	rs1135840	rs35742686	rs1065852	rs3892097	rs5030655	rs5030656	rs28371706	rs28371725	rs769258	Notes
IBD-014	*1/*2	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
IBD-015	*1/*2	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
IBD-016	*4/*9	GG	CG	TT	GA	CT	AA	DelCTT (het)	GG	CC	CC	
IBD-017	*4/*41	GA	GG	TT	GA	CT	AA	CTT/CTT	GG	CT	CC	
IBD-018	*9/*41	GA	CG	TT	GG	CC	AA	DelCTT (het)	GG	CT	CC	
IBD-019	*1/*2	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
IBD-020	*1/*9	GG	CC	TT	GG	CC	AA	DelCTT (het)	GG	CC	CC	
JDM-001	*2/*2	AA	GG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
JDM-002	*1/*1	GG	CC	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
JDM-003	*1/*1	GG	CC	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
JDM-004	*4/*41	GA	GG	TT	GA	CT	AA	CTT/CTT	GG	CT	CC	
JDM-005	*1/*41	GA	GC	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	
JDM-006	*1/*2	GA	GC	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
JDM-007	*1/*29	GA	GC	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	Astrolabe
JDM-008	*1/*1	GG	CC	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
JDM-009	*1/*3	GG	CC	T/delT	GG	CC	AA	CTT/CTT	GG	CC	CC	
JDM-010	*1/*1	GG	CC	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
JDM-011	*2/*9	GA	GC	TT	GG	CC	AA	delCTT	GG	CC	CC	Astrolabe
JDM-012	*2/*35	AA	GG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	Astrolabe
JDM-013	*2/*2	AA	GG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
SRS-001	*1/*2	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
SRS-002	*1x2/*2	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	3:1 ratio. Astrolabe
SRS-003	*1/*4	GG	CG	TT	GA	CT	AA	CTT/CTT	GG	CC	CC	
SRS-004	*1/*1	GG	CC	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
SRS-005	*1/*1	GG	CC	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
SRS-006	*1/*41	GA	CT	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	
SRS-007	*2/*41	AA	GG	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	

Table A4.3 continued: Diplotypes for *CYP2D6*

Patient ID	Diplotype	rs16947	rs1135840	rs35742686	rs1065852	rs3892097	rs5030655	rs5030656	rs28371706	rs28371725	rs769258	Notes
SRS-008	*41/*41	AA	GG	TT	GG	CC	AA	CTT/CTT	GG	TT	CC	
SRS-009	*1/*2	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
SRS-010	*2/*41	AA	GG	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	
SRS-011	*1/*2	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
SRS-012	*4/*41	GA	GG	TT	GA	CT	AA	CTT/CTT	GG	CT	CC	
SRS-013	*1x2/*1	GG	CC	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	Astrolabe
SRS-014	*1/*6	GG	CC	TT	GG	CC	Del A (het)	CTT/CTT	GG	CC	CC	
SRS-015	*1x2/*41	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	Astrolabe
SRS-016	*1/*41	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	
SRS-017	*1/*41	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	
SRS-018	*1/*41	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CT	CC	
SRS-019	*2/*4	GA	GG	TT	GA	CT	AA	CTT/CTT	GG	CC	CC	
SRS-020	*2/*4	GA	GG	TT	GA	CT	AA	CTT/CTT	GG	CC	CC	
SRS-021	*4/*4	GG	GG	TT	AA	TT	AA	CTT/CTT	GG	CC	CC	
SRS-022	*1/*2	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
SRS-023	*1/*2	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
SRS-024	*2/*4	GA	GG	TT	GA	CT	AA	CTT/CTT	GG	CC	CC	
SRS-025	*1/*2	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
SRS-026	*1/*2	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
SRS-027	*1/*1	GG	CC	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
SRS-028	*4/*4	GG	GG	TT	AA	TT	AA	CTT/CTT	GG	CC	CC	
SRS-029	*1/*4	GG	CG	TT	GA	CT	AA	CTT/CTT	GG	CC	CC	
SRS-030	*4/*41	GA	GG	TT	GA	CT	AA	CTT/CTT	GG	CT	CC	
USH-001	*1/*4	GG	CG	TT	GA	CT	AA	CTT/CTT	GG	CC	CC	
USH-002	*1/*2	GA	CG	TT	GG	CC	AA	CTT/CTT	GG	CC	CC	
USH-003	*10/*41	GA	GG	TT	GA	CC	AA	CTT/CTT	GG	CT	CC	

Table A4.3 continued: Diplotypes for *CYP2D6*

Patient ID	Diplotype	rs28365083	rs776746	rs56411402	rs55965422	rs10264272	rs41303343	rs55817950	rs28383479
		7:99652613	7:99672916	7:99665237	7:99666950	7:99665212	7:99652770-99652771	7:99676198	7:99660516
		g.99652613 G>T p.Thr398Asn	g.99672916 T>C	g.99665237 T>C p.Gln200Arg	g.99666950 A>G	g.99665212 C>T, p.Lys208=	g.99652770_ 99652771 ins A p.Thr346Tyrfs	g.99676198 G>A p.Arg28Cys	g.99660516 C>T p.Ala337Thr
	*1	G	T	T	A	C	-	G	C
	*2	T	T	T	A	C	-	G	C
	*3	G	C	T	A	C	-	G	C
	*4	G	T	C	A	C	-	G	C
	*5	G	T	T	G	C	-	G	C
	*6	G	T	T	A	T	-	G	C
	*7	G	T	T	A	C	insA	G	C
	*8	G	T	T	A	C	-	A	C
	*9	G	C	T	A	C	-	G	T
<b>BBS-001</b>	*3/*3	GG	CC	TT	AA	CC	└┘	GG	CC
<b>BBS-002</b>	*3/*3	GG	CC	TT	AA	CC	└┘	GG	CC
<b>BBS-003</b>	*3/*3	GG	CC	TT	AA	CC	└┘	GG	CC
<b>BBS-004</b>	*3/*3	GG	CC	TT	AA	CC	└┘	GG	CC
<b>BBS-005</b>	*3/*3	GG	CC	TT	AA	CC	└┘	GG	CC
<b>BBS-006</b>	*3/*3	GG	CC	TT	AA	CC	└┘	GG	CC
<b>BBS-007</b>	*3/*3	GG	CC	TT	AA	CC	└┘	GG	CC
<b>BBS-008</b>	*1/*3	GG	TC	TT	AA	CC	└┘	GG	CC
<b>BBS-009</b>	*3/*3	GG	CC	TT	AA	CC	└┘	GG	CC
<b>BBS-010</b>	*3/*3	GG	CC	TT	AA	CC	└┘	GG	CC
<b>BBS-011</b>	*3/*3	GG	CC	TT	AA	CC	└┘	GG	CC
<b>BBS-012</b>	*3/*3	GG	CC	TT	AA	CC	└┘	GG	CC
<b>BBS-013</b>	*3/*3	GG	CC	TT	AA	CC	└┘	GG	CC
<b>BBS-014</b>	*1/*3	GG	TC	TT	AA	CC	└┘	GG	CC
<b>BBS-015</b>	*3/*3	GG	CC	TT	AA	CC	└┘	GG	CC
<b>BBS-016</b>	*3/*3	GG	CC	TT	AA	CC	└┘	GG	CC

Table A4.4: Diplotypes for CYP3A5

Patient ID	Diplotype	rs28365083	rs776746	rs56411402	rs55965422	rs10264272	rs41303343	rs55817950	rs28383479
BBS-017	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
BBS-018	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-001	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-002	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-003	*1/*3	GG	TC	TT	AA	CC	┘┘	GG	CC
IBD-004	*1/*3	GG	TC	TT	AA	CC	┘┘	GG	CC
IBD-005	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-006	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-007	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-008	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-009	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-010	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-011	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-012	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-013	*1/*3	GG	TC	TT	AA	CC	┘┘	GG	CC
IBD-014	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-015	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-016	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-017	*1/*3	GG	TC	TT	AA	CC	┘┘	GG	CC
IBD-018	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
IBD-019	*1/*3	GG	TC	TT	AA	CC	┘┘	GG	CC
IBD-020	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
JDM-001	*3/*6	GG	TC	TT	AA	CT	┘┘	GG	CC
JDM-002	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
JDM-003	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
JDM-004	*1/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
JDM-005	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
JDM-006	*1/*3	GG	TC	TT	AA	CC	┘┘	GG	CC
JDM-007	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC
JDM-008	*3/*3	GG	CC	TT	AA	CC	┘┘	GG	CC

Table A4.4 continued: Diplotypes for *CYP3A5*

Patient ID	Diplotype	rs28365083	rs776746	rs56411402	rs55965422	rs10264272	rs41303343	rs55817950	rs28383479
JDM-009	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
JDM-010	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
JDM-011	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
JDM-012	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
JDM-013	*6/*6	GG	TT	TT	AA	TT	└┐	GG	CC
SRS-001	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-002	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-003	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-004	*1/*3	GG	TC	TT	AA	CC	└┐	GG	CC
SRS-005	*1/*3	GG	TC	TT	AA	CC	└┐	GG	CC
SRS-006	*3/*3	GG	CC	TT	AA	CC	└┐	GG	GG
SRS-007	*1/*3	GG	TC	TT	AA	CC	└┐	GG	CC
SRS-008	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-009	*1/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-010	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-011	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-012	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-013	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-014	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-015	*1/*3	GG	TC	TT	AA	CC	└┐	GG	CC
SRS-016	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-017	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-018	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-019	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-020	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-021	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-022	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-023	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-024	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC
SRS-025	*3/*3	GG	CC	TT	AA	CC	└┐	GG	CC

Table A4.4 continued: Diplotypes for *CYP3A5*

Patient ID	Diplotype	rs28365083	rs776746	rs56411402	rs55965422	rs10264272	rs41303343	rs55817950	rs28383479
SRS-026	*3/*3	GG	CC	TT	AA	CC	_/_	GG	CC
SRS-027	*3/*3	GG	CC	TT	AA	CC	_/_	GG	CC
SRS-028	*3/*3	GG	CC	TT	AA	CC	_/_	GG	CC
SRS-029	*3/*3	GG	CC	TT	AA	CC	_/_	GG	CC
SRS-030	*3/*3	GG	CC	TT	AA	CC	_/_	GG	CC
USH-001	*3/*3	GG	CC	TT	AA	CC	_	GG	CC
USH-002	*3/*3	GG	CC	TT	AA	CC	_	GG	CC
USH-003	*3/*3	GG	CC	TT	AA	CC	_	GG	CC

Table A4.4 continued: Diplotypes for *CYP3A5*

	<i>CYP2C</i>	<i>CYP4F2</i>	<i>VKORC1</i>	<i>VKORC1</i>
<b>Patient ID</b>	<b>rs12777823</b>	<b>rs2108622</b>	<b>rs9934438</b>	<b>rs9923231</b>
	10:94645745	19:15879621	16:31093557	16:31096368
	g.94645745 G>A	g.15879621 C>T p.Val433Met	g.31093557 G>A	g.31096368 C>T
<b>BBS-001</b>	GA	CT	GG	CC
<b>BBS-002</b>	GG	CT	GG	CC
<b>BBS-003</b>	GA	CC	GA	CT
<b>BBS-004</b>	GA	TT	GG	CC
<b>BBS-005</b>	GG	CC	AA	TT
<b>BBS-006</b>	GG	CT	GG	CC
<b>BBS-007</b>	GG	CT	GA	CT
<b>BBS-008</b>	GG	TT	GG	CC
<b>BBS-009</b>	GG	CT	AA	TT
<b>BBS-010</b>	GG	TT	GA	CT
<b>BBS-011</b>	GG	TT	GA	CT
<b>BBS-012</b>	GG	CC	GA	CT
<b>BBS-013</b>	GG	CT	GA	CT
<b>BBS-014</b>	GA	CC	GG	CC
<b>BBS-015</b>	GA	CC	GG	CC
<b>BBS-016</b>	GG	CC	GA	CT
<b>BBS-017</b>	GG	CC	GA	CT
<b>BBS-018</b>	GG	CC	GA	CT
<b>IBD-001</b>	GA	CC	GA	CT
<b>IBD-002</b>	GG	CC	GA	CT
<b>IBD-003</b>	GG	CT	GG	CC
<b>IBD-004</b>	GA	CC	GG	CC
<b>IBD-005</b>	GG	CC	AA	TT
<b>IBD-006</b>	GG	CC	GG	CC
<b>IBD-007</b>	GG	TT	GG	CC
<b>IBD-008</b>	GA	CC	GA	CT
<b>IBD-009</b>	AA	CT	GG	CC
<b>IBD-010</b>	GG	CT	GA	CT
<b>IBD-011</b>	GA	TT	GA	CT
<b>IBD-012</b>	GA	CC	GG	CC
<b>IBD-013</b>	GA	TT	GG	CC
<b>IBD-014</b>	GG	CC	GA	CT
<b>IBD-015</b>	GG	TT	GG	CC
<b>IBD-016</b>	GA	CC	GA	CT
<b>IBD-017</b>	GA	CC	GG	CC
<b>IBD-018</b>	GG	CC	GA	CT
<b>IBD-019</b>	GA	CT	GG	CC
<b>IBD-020</b>	GA	CT	GA	CT
<b>JDM-001</b>	GG	CT	GA	CT
<b>JDM-002</b>	GG	CC	GA	CT
<b>JDM-003</b>	GG	CT	GA	CT
<b>JDM-004</b>	GG	CC	GA	CT
<b>JDM-005</b>	GA	CT	AA	TT
<b>JDM-006</b>	GG	CC	GG	CC
<b>JDM-007</b>	GG	CC	GG	CC
<b>JDM-008</b>	GA	CT	GA	CT
<b>JDM-009</b>	GA	CC	GA	CT

Table A4.5: Genotypes for *CYP2C*, *CYP4F2* and *VKORC1*



Patient ID	rs12777823	rs2108622	rs9934438	rs9923231
JDM-010	GG	CC	AA	TT
JDM-011	GA	CC	GG	CC
JDM-012	GA	CC	GA	CT
JDM-013	GA	CC	GG	CC
SRS-001	GG	CT	GA	CT
SRS-002	GG	CT	GA	CT
SRS-003	GG	CC	GG	CC
SRS-004	GG	CC	GG	CC
SRS-005	GA	CC	GA	CT
SRS-006	GG	CT	GG	CC
SRS-007	GG	CT	GA	CT
SRS-008	GG	CT	GG	CC
SRS-009	GA	CT	AA	TT
SRS-010	GA	CC	GA	CT
SRS-011	GA	CC	GG	CC
SRS-012	GA	CT	AA	TT
SRS-013	GG	CC	GA	CT
SRS-014	GG	CC	AA	TT
SRS-015	GG	CT	GG	CC
SRS-016	GA	TT	GG	CC
SRS-017	GA	CT	GG	CC
SRS-018	GA	CC	GG	CC
SRS-019	GG	CT	GA	CT
SRS-020	GG	TT	GA	CT
SRS-021	GG	CT	GA	CT
SRS-022	GG	CC	GG	CC
SRS-023	GG	CT	GA	CT
SRS-024	GA	CC	GG	CC
SRS-025	GA	CC	GG	CC
SRS-026	GA	CC	GG	CC
SRS-027	GA	CC	GG	CC
SRS-028	GG	CC	GG	CC
SRS-029	GG	CC	GA	CT
SRS-030	GG	CT	GG	CC
USH-001	GG	CC	GA	CT
USH-002	GG	CC	GA	CT
USH-003	GG	CC	GA	CT

Table A4.5: Genotypes for *CYP2C*, *CYP4F2* and *VKORC1*

Patient ID	Diplotype	rs3918290	rs1801159	rs72549303	rs1801158	rs1801160	rs72549309	rs1801266	rs1801265
		1:97450058	1:97515839	1:97450066	1:97515865	1:97305364	1:97740415-97740418	1:97691776	1:97883329
		g.97450058 C>G	g.97515839 T>C p.Ile543Val	g.97450066 delG p.Pro633Glnfs	g.97515865 C>T p.Ser534Asn	g.97305364 C>T p.Val732Ile	g.97740415_97740418 delATGA p.Phe100Serfs	g.97691776 G>A p.Arg235Trp	g.97883329 A>G p.Cys29Arg
	*1	C	T	G	C	C	ATGA	G	A
	*2A	T	T	G	C	C	ATGA	G	A
	*2B	T	C	G	C	C	ATGA	G	A
	*3	C	T		C	C	ATGA	G	A
	*4	C	T	G	T	C	ATGA	G	A
	*5	C	C	G	C	C	ATGA	G	A
	*6	C	T	G	C	T	ATGA	G	A
	*7	C	T	G	C	C	///	G	A
	*8	C	T	G	C	C	ATGA	A	A
	*9A	C	T	G	C	C	ATGA	G	G
	*9B	C	T	G	C	C	ATGA	G	G
<b>BBS-001</b>	*1/*6	CC	TT	GG	CC	CT	ATGA/ATGA	GG	AA
<b>BBS-002</b>	*5/*9A	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AG
<b>BBS-003</b>	*6/*9A	CC	TT	GG	CC	CT	ATGA/ATGA	GG	AG
<b>BBS-004</b>	*1/*5	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AA
<b>BBS-005</b>	*1/*5	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AA
<b>BBS-006</b>	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
<b>BBS-007</b>	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
<b>BBS-008</b>	*5/*9A	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AG
<b>BBS-009</b>	*5/*9A	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AG
<b>BBS-010</b>	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
<b>BBS-011</b>	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
<b>BBS-012</b>	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
<b>BBS-013</b>	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
<b>BBS-014</b>	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
<b>BBS-015</b>	*5/*9A	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AG
<b>BBS-016</b>	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG

Table A4.6: Diploypes for *DPYD*. rs1801267, rs1801268, rs72539306, rs80081766, rs78060119 and rs55886062 are not shown. All patients were reference sequence at these SNPs. See chapter 5 for information on results in red

Patient ID	Diplotype	rs3918290	rs1801159	rs72549303	rs1801158	rs1801160	rs72549309	rs1801266	rs1801265
BBS-017	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
BBS-018	*1/*5	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AA
IBD-001	*9A/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	GG
IBD-002	*1/*5	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AA
IBD-003	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
IBD-004	*5/*9A	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AG
IBD-005	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
IBD-006	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
IBD-007	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
IBD-008	*1/*5	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AG
IBD-009	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
IBD-010	*5/*6	CC	TC	GG	CC	CT	ATGA/ATGA	GG	AA
IBD-011	*1/*5	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AA
IBD-012	*1/*5	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AA
IBD-013	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
IBD-014	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
IBD-015	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
IBD-016	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
IBD-017	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
IBD-018	*9A/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	GG
IBD-019	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
IBD-020	Uninterpretable	CC	TT	GG	CC	CT	ATGA/ATGA	GG	GG
JDM-001	*1/*5	CC	CT	GG	CC	CC	ATGA/ATGA	GG	AA
JDM-002	*1/*5	CC	CT	GG	CC	CC	ATGA/ATGA	GG	AA
JDM-003	*1/*5	CC	CT	GG	CC	CC	ATGA/ATGA	GG	AA
JDM-004	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
JDM-005	*1/*5	CC	CT	GG	CC	CC	ATGA/ATGA	GG	AA
JDM-006	*1/*5	CC	CT	GG	CC	CC	ATGA/ATGA	GG	AA
JDM-007	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
JDM-008	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	GA
JDM-009	*5/*9A	CC	CT	GG	CC	CC	ATGA/ATGA	GG	GA
JDM-010	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	GA

Table A4.6 continued: Diplotypes for *DPYD*. rs1801267, rs1801268, rs72539306, rs80081766, rs78060119 and rs55886062 are not shown. All patients were reference sequence at these SNPs. See chapter 5 for information on results in red

Patient ID	Diplotype	rs3918290	rs1801159	rs72549303	rs1801158	rs1801160	rs72549309	rs1801266	rs1801265
JDM-011	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
JDM-012	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
JDM-013	*1/*5	CC	CT	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-001	*5/*9A	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AG
SRS-002	Uninterpretable	CC	CC	GG	CC	CC	ATGA/ATGA	GG	AG
SRS-003	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-004	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
SRS-005	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
SRS-006	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
SRS-007	*5/*6	CC	TC	GG	CC	CT	ATGA/ATGA	GG	AA
SRS-008	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-009	Uninterpretable	CC	CC	GG	CC	CT	ATGA/ATGA	GG	AA
SRS-010	*1/*5	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-011	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-012	*1/*5	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-013	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-014	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-015	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-016	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
SRS-017	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
SRS-018	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
SRS-019	*1/*5	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-020	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-021	*4/*5	CC	TC	GG	CT	CC	ATGA/ATGA	GG	AA
SRS-022	*5/*9A	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AG
SRS-023	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
SRS-024	*1/*5	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-025	*1/*5	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-026	*1/*5	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-027	*5/*5	CC	CC	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-028	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA
SRS-029	*1/*1	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AA

Table A4.6 continued: Diplotypes for *DPYD*. rs1801267, rs1801268, rs72539306, rs80081766, rs78060119 and rs55886062 are not shown. All patients were reference sequence at these SNPs. See chapter 5 for information on results in red

Patient ID	Diplotype	rs3918290	rs1801159	rs72549303	rs1801158	rs1801160	rs72549309	rs1801266	rs1801265
<b>SRS-030</b>	*1/*5	CC	TC	GG	CC	CC	ATGA/ATGA	GG	AA
<b>USH-001</b>	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
<b>USH-002</b>	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG
<b>USH-003</b>	*1/*9A	CC	TT	GG	CC	CC	ATGA/ATGA	GG	AG

Table A4.6 continued: Diplotypes for *DPYD*. rs1801267, rs1801268, rs72539306, rs80081766, rs78060119 and rs55886062 are not shown. All patients were reference sequence at these SNPs. See chapter 5 for information on results in red

Patient ID	rs398123546	rs1050828	rs1050829	rs72554665	rs5030868	rs137852327	rs137852339	rs5030869	n/a	n/a	rs5030870
	X:154532390	X:154536002	X:154535277	X:154532269	X:154534419	X:154533122	X:154533044	X:154532990	X:154533615	X:154532992	X:154535316
	Orissa	A-(1)	A-(2)	Canton	Mediterranean	Viagchan	Kerala	Chatham	Bankok	Villeurbanne	Sao Boria
	g.154532390 G>A p.Arg484Cys	g.154536002 C>T p.Val98Met	g.154535277 T>A p.Asn156Asp	g.154532269 C>A p.Arg489Leu	g.154534419 G>A p.Ser218Phe	g.154533122 C>T p.Val321Met	g.154533044 C>T p.Glu347Lys	g.154532990 C>T p.Ala365Thr	g.154533615 C>G p.Leu305Asn	g.154532992 TGGdel p.Thr364del	g.154535316 C>T p.Asp143Asn
<b>BBS-001</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-002</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-003</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-004</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-005</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-006</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-007</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-008</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-009</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-010</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-011</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-012</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-013</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-014</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-015</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-016</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-017</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>BBS-018</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>IBD-001</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>IBD-002</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>IBD-003</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>IBD-004</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>IBD-005</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>IBD-006</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
<b>IBD-007</b>	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC

Table A4.7: Genotypes for *G6PD*

Patient ID	rs398123546	rs1050828	rs1050829	rs72554665	rs5030868	rs137852327	rs137852339	rs5030869	n/a	n/a	rs5030870
IBD-008	GG	CC	CT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
IBD-009	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
IBD-010	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
IBD-011	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
IBD-012	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
IBD-013	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
IBD-014	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
IBD-015	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
IBD-016	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
IBD-017	GG	CC	TT	CC	GA	CC	CC	CC	CC	TGG/TGG	CC
IBD-018	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
IBD-019	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
IBD-020	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
JDM-001	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
JDM-002	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
JDM-003	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
JDM-004	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
JDM-005	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
JDM-006	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
JDM-007	GG	CC	CC	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
JDM-008	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
JDM-009	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
JDM-010	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
JDM-011	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
JDM-012	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
JDM-013	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-001	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-002	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-003	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-004	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-005	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-006	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-007	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC

Table A4.7 continued: Genotypes for *G6PD*

Patient ID	rs398123546	rs1050828	rs1050829	rs72554665	rs5030868	rs137852327	rs137852339	rs5030869	n/a	n/a	rs5030870
SRS-008	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-009	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-010	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-011	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-012	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-013	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-014	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-015	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-016	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-017	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-018	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-019	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-020	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-021	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-022	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-023	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-024	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-025	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-026	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-027	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-028	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-029	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
SRS-030	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
USH-001	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
USH-002	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC
USH-003	GG	CC	TT	CC	GG	CC	CC	CC	CC	TGG/TGG	CC

Table A4.7 continued: Genotypes for *G6PD*



Patient ID	57*01	*15:02	*15:02	*15:02	*15:02
	rs2395029	rs2395148	rs10484555	rs144012689	rs2524160
	6:31464003	6:32353777	6:31313688	6:31355003	6:31292077
	g.31464003 T>G	g.32353777 G>T	g.31313688 T>C	g.31355003 T>A	g.31292077 G>A
BBS-001	TT	GG	TT	TT	GG
BBS-002	TT	GG	TT	TT	GG
BBS-003	TT	GG	TT	TT	GG
BBS-004	TT	GG	TT	TT	GG
BBS-005	TT	GG	TT	TT	GG
BBS-006	TT	GG	TT	TT	GG
BBS-007	TT	GG	TT	TT	GG
BBS-008	TT	GG	TT	TT	GA
BBS-009	TT	GG	TT	TT	GG
BBS-010	TT	GG	TT	TT	GG
BBS-011	TT	GG	TT	TT	GG
BBS-012	TG	GG	TT	TT	GG
BBS-013	TG	GG	TT	TT	GA
BBS-014	TG	GG	TT	TT	GG
BBS-015	TT	GG	TT	TT	GG
BBS-016	TT	GG	TT	TT	GA
BBS-017	TT	GG	TT	TT	GA
BBS-018	TT	GG	TT	TT	GG
IBD-001	TT	GG	TT	TT	GG
IBD-002	TT	GG	TT	TT	GG
IBD-003	TT	GG	TT	TT	GG
IBD-004	TT	GG	TT	TT	GA
IBD-005	TT	GG	TT	TT	GG
IBD-006	TT	GG	TT	TT	GG
IBD-007	TT	GG	TT	TT	GG
IBD-008	TT	GG	TT	TT	GG
IBD-009	TT	GG	TT	TT	GG
IBD-010	TT	GG	TT	TT	GG
IBD-011	TT	GG	TT	TT	GG
IBD-012	TT	GG	TT	TT	GG
IBD-013	TT	GG	TT	TT	GG
IBD-014	TT	GG	TT	TT	GG
IBD-015	TT	GG	TT	TT	GG
IBD-016	TT	GG	TT	TT	GA
IBD-017	TT	GG	TT	TT	GG
IBD-018	TT	GG	TT	TT	GG
IBD-019	GG	GG	TT	TT	GG
IBD-020	TT	GG	TT	TT	GG
SRS-001	TT	GG	TT	TT	GG
SRS-002	TT	GG	TT	TT	GA
SRS-003	TT	GG	TT	TT	GG
SRS-004	TT	GG	TT	TT	GG
SRS-005	TT	GG	TT	TT	GG
SRS-006	TT	GG	TT	TT	GG
SRS-007	TT	GG	TT	TT	GG
SRS-008	TG	GG	TT	TT	GG
SRS-009	TT	GG	TT	TT	GG

Table A4.8: Genotypes for *HLA-B* \*57:01 and *HLA-B* \*15:02

Patient ID	57*01	*15:02	*15:02	*15:02	*15:02
	rs2395029	rs2395148	rs10484555	rs144012689	rs2524160
SRS-010	TT	GG	TT	TT	GA
SRS-011	TT	GG	TT	TT	GA
SRS-012	TT	GG	TT	TT	GA
SRS-013	TT	GG	TT	TT	GG
SRS-014	TT	GA	TT	TT	GG
SRS-015	TT	GG	TT	TT	GG
SRS-016	TT	GG	TT	TT	GG
SRS-017	TT	GG	TT	TT	GG
SRS-018	TT	GG	TT	TT	GG
SRS-019	TT	GG	TT	TT	GG
SRS-020	TT	GG	TT	TT	GG
SRS-021	TT	GG	TT	TT	GG
SRS-022	TT	GG	TT	TT	GG
SRS-023	TT	GG	TT	TT	GG
SRS-024	TT	GG	TT	TT	GG
SRS-025	TT	GG	TT	TT	GG
SRS-026	TT	GG	TT	TT	GG
SRS-027	TT	GG	TT	TT	GG
SRS-028	TT	GG	TT	TT	GG
SRS-029	TT	GG	TT	TT	GG
SRS-030	TT	GG	TT	TT	GG
USH-001	TT	GG	TT	TT	GG
USH-002	TT	GG	TT	TT	GG
USH-003	TG	GG	TT	TT	GG

Table A4.8 continued: Genotypes for *HLA-B*\*57:01 and *HLA-B*\*15:02

Patient ID	Diplotype	rs1800462	rs1800460	rs1142345	rs1800584	rs72552740	rs75543815	rs72552736	rs56161402	rs151149760	rs72552737
		6:18143724	6:18138997	6:18130687	6:18130781	6:18147910	6:18133845	6:18130725	6:18130762	6:18143606	6:18139027
		g.18143724 C>G p.Ala80Pro	g.18138997 C>T p.Ala154Thr	g.18130687 T>C p.Tyr240Cys	g.18130781 C>T	g.1z8147910 A>G p.Leu49Ser	g.18133845 T>A p.Tyr180Phe	g.18130725 A>C p.His227Gln	g.18130762 C>T p.Arg215His	g.18143606 T>G p.Lys119Thr	g.18139027 C>G p.Gly114Arg
	*1	C	C	T	C	A	T	A	C	T	C
	*2	G	C	T	C	A	T	A	C	T	C
	*3A	C	T	C	C	A	T	A	C	T	C
	*3B	C	T	T	C	A	T	A	C	T	C
	*3C	C	C	C	C	A	T	A	C	T	C
	*4	C	C	T	T	A	T	A	C	T	C
	*5	C	C	T	C	G	T	A	C	T	C
	*6	C	C	T	C	A	A	A	C	T	C
	*7	C	C	T	C	A	T	C	C	T	C
	*8	C	C	T	C	A	T	A	T	T	C
	*9	C	C	T	C	A	T	A	C	G	C
	*10	C	C	T	C	A	T	A	C	T	G
<b>BBS-001</b>	*1/*3A	CC	CT	TC	CC	AA	TT	AA	CC	TT	CC
<b>BBS-002</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-003</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-004</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-005</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-006</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-007</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-008</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-009</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-010</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-011</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-012</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-013</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-014</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-015</b>	*1/*3A	CC	CT	TC	CC	AA	TT	AA	CC	TT	CC
<b>BBS-016</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-017</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>BBS-018</b>	*1/*3A	CC	CT	TC	CC	AA	TT	AA	CC	TT	CC
<b>IBD-001</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>IBD-002</b>	*1/*3A	CC	CT	TC	CC	AA	TT	AA	CC	TT	CC
<b>IBD-003</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
<b>IBD-004</b>	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC

Table A4.9: Diplotypes for *TMPT*. Results for rs72552738, rs20022021, rs72552742, rs9333569, rs144041067, 6:18149004 and rs777686348 (haplotypes \*11-\*18) not shown. All patients reference sequence at these SNPs

Patient ID	Diplotype	rs1800462	rs1800460	rs1142345	rs1800584	rs72552740	rs75543815	rs72552736	rs56161402	rs151149760	rs72552737
IBD-005	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-006	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-007	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-008	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-009	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-010	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-011	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-012	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-013	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-014	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-015	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-016	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-017	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-018	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-019	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
IBD-020	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
JDM-001	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
JDM-002	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
JDM-003	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
JDM-004	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
JDM-005	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
JDM-006	*1/*3A	CC	CT	TC	CC	AA	TT	AA	CC	TT	CC
JDM-007	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
JDM-008	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
JDM-009	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
JDM-010	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
JDM-011	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
JDM-012	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
JDM-013	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-001	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-002	*1/*3A	CC	CT	CT	CC	AA	TT	AA	CC	TT	CC
SRS-003	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-004	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-005	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-006	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-007	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-008	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-009	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC

Table A4.9: Diplotypes for *TMPT*. Results for rs72552738, rs20022021, rs72552742, rs9333569, rs144041067, 6:18149004 and rs777686348 (haplotypes \*11-\*18) not shown. All patients reference sequence at these SNPs

Patient ID	Diplotype	rs1800462	rs1800460	rs1142345	rs1800584	rs72552740	rs75543815	rs72552736	rs56161402	rs151149760	rs72552737
SRS-010	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-011	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-012	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-013	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-014	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-015	*1/*3A	CC	CT	CT	CC	AA	TT	AA	CC	TT	CC
SRS-016	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-017	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-018	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-019	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-020	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-021	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-022	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-023	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-024	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-025	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-026	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-027	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-028	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-029	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
SRS-030	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
USH-001	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
USH-002	*1/*1	CC	CC	TT	CC	AA	TT	AA	CC	TT	CC
USH-003	*1/*3A	CC	CT	TC	CC	AA	TT	AA	CC	TT	CC

Table A4.9: Diplotypes for *TMPT*. Results for rs72552738, rs20022021, rs72552742, rs9333569, rs144041067, 6:18149004 and rs777686348 (haplotypes \*11-\*18) not shown. All patients reference sequence at these SNPs

Patient ID	Diplotype	rs8175347	rs887829	rs4148323
		2:233760235-233760236 [TA]	2:233759924	2:233760498
		Number of repeats	g.233759924 C>T	g.233760498 G>A p.Gly71Arg
	*1	[TA]6	2 reference or increased function alleles- CC	GG
	*6	[TA]6	1 reference or increased function allele with one decreased function allele- CT	GA
	*28	[TA]7	2 decreased function alleles- TT	GG
	*36	[TA]8	2 reference or increased function alleles- CC	GG
	*37	[TA]5	2 decreased function alleles- TT	GG
<b>BBS-001</b>	*1/*1	[TA]6/[TA]6	CC	GG
<b>BBS-002</b>	*1/*28	[TA]6/[TA]7	CT	GG
<b>BBS-003</b>	*1/*1	[TA]6/[TA]6	CC	GG
<b>BBS-004</b>	*1/*28	[TA]6/[TA]7	CT	GG
<b>BBS-005</b>	*28/*28	[TA]7/[TA]7	TT	GG
<b>BBS-006</b>	*1/*28	[TA]6/[TA]7	CT	GG
<b>BBS-007</b>	*1/*28	[TA]6/[TA]7	CT	GG
<b>BBS-008</b>	*1/*1	[TA]6/[TA]6	CC	GG
<b>BBS-009</b>	*1/*1	[TA]6/[TA]6	CC	GG
<b>BBS-010</b>	*28/*28	[TA]7/[TA]7	TT	GG
<b>BBS-011</b>	*28/*28	[TA]7/[TA]7	TT	GG
<b>BBS-012</b>	*1/*1	[TA]6/[TA]6	CC	GG
<b>BBS-013</b>	*1/*1	[TA]6/[TA]6	CC	GG
<b>BBS-014</b>	*1/*1	[TA]6/[TA]6	CC	GG
<b>BBS-015</b>	*1/*1	[TA]6/[TA]6	CC	GG
<b>BBS-016</b>	*1/*28	[TA]6/[TA]7	CT	GG
<b>BBS-017</b>	*1/*28	[TA]6/[TA]7	CT	GG
<b>BBS-018</b>	*28/*28	[TA]7/[TA]7	TT	GG
<b>IBD-001</b>	*1/*1	[TA]6/[TA]6	CC	GG
<b>IBD-002</b>	*28/*28	[TA]7/[TA]7	TT	GG
<b>IBD-003</b>	*1/*28	[TA]6/[TA]7	CT	GG
<b>IBD-004</b>	*1/*36	[TA]6/[TA]8	CC	GG
<b>IBD-005</b>	*1/*28	[TA]6/[TA]7	CT	GG
<b>IBD-006</b>	*1/*1	[TA]6/[TA]6	CC	GG
<b>IBD-007</b>	*1/*28	[TA]6/[TA]7	CT	GG
<b>IBD-008</b>	*28/*28	[TA]7/[TA]7	TT	GG
<b>IBD-009</b>	*1/*28	[TA]6/[TA]7	CT	GG
<b>IBD-010</b>	*1/*28	[TA]6/[TA]7	CT	GG
<b>IBD-011</b>	*1/*1	[TA]6/[TA]6	CC	GG
<b>IBD-012</b>	*1/*28	[TA]6/[TA]7	CT	GG
<b>IBD-013</b>	*1/*28	[TA]6/[TA]7	CT	GG
<b>IBD-014</b>	*1/*1	[TA]6/[TA]6	CC	GG
<b>IBD-015</b>	*1/*1	[TA]6/[TA]6	CC	GG
<b>IBD-016</b>	*1/*28	[TA]6/[TA]7	CT	GG

Table A4.10: Diplotypes for *UGT1A1*

Patient ID	Diplotype	rs8175347	rs887829	rs4148323
IBD-017	*1/*28	[TA]6/[TA]7	CT	GG
IBD-018	*28/*28	[TA]7/[TA]7	TT	GG
IBD-019	*1/*1	[TA]6/[TA]6	CC	GG
IBD-020	*1/*28	[TA]6/[TA]7	CT	GG
JDM-001	*1/*1	[TA]6/[TA]6	CC	GG
JDM-002	*1/*28	[TA]6/[TA]7	CT	GG
JDM-003	*1/*1	[TA]6/[TA]6	CC	GG
JDM-004	*1/*1	[TA]6/[TA]6	CC	GG
JDM-005	*1/*28	[TA]6/[TA]7	CT	GG
JDM-006	*1/*1	[TA]6/[TA]6	CC	GG
JDM-007	*1/*28	[TA]6/[TA]7	CT	GG
JDM-008	*1/*1	[TA]6/[TA]6	CC	GG
JDM-009	*1/*28	[TA]6/[TA]7	CT	GG
JDM-010	*1/*28	[TA]6/[TA]7	CT	GG
JDM-011	*1/*1	[TA]6/[TA]6	CC	GG
JDM-012	*1/*1	[TA]6/[TA]6	CC	GG
JDM-013	*28/*28	[TA]7/[TA]7	TT	GG
SRS-001	*28/*28	[TA]7/[TA]7	TT	GG
SRS-002	*1/*28	[TA]6/[TA]7	CT	GG
SRS-003	*1/*28	[TA]6/[TA]7	CT	GG
SRS-004	*28/*28	[TA]7/[TA]7	TT	GG
SRS-005	*1/*28	[TA]6/[TA]7	CT	GG
SRS-006	*28/*28	[TA]7/[TA]7	TT	GG
SRS-007	*28/*28	[TA]7/[TA]7	TT	GG
SRS-008	*1/*28	[TA]6/[TA]7	CT	GG
SRS-009	*28/*28	[TA]7/[TA]7	TT	GG
SRS-010	*1/*1	[TA]6/[TA]6	CC	GG
SRS-011	*1/*1	[TA]6/[TA]6	CC	GG
SRS-012	*1/*1	[TA]6/[TA]6	CC	GG
SRS-013	*1/*28	[TA]6/[TA]7	CT	GG
SRS-014	*1/*28	[TA]6/[TA]7	CT	GG
SRS-015	*1/*1	[TA]6/[TA]6	CC	GG
SRS-016	*1/*1	[TA]6/[TA]6	CC	GG
SRS-017	*1/*28	[TA]6/[TA]7	CT	GG
SRS-018	*1/*1	[TA]6/[TA]6	CC	GG
SRS-019	*1/*28	[TA]6/[TA]7	CT	GG
SRS-020	*28/*28	[TA]7/[TA]7	TT	GG
SRS-021	*1/*1	[TA]6/[TA]6	CC	GG
SRS-022	*1/*28	[TA]6/[TA]7	CT	GG
SRS-023	*1/*1	[TA]6/[TA]6	CC	GG
SRS-024	*1/*28	[TA]6/[TA]7	CT	GG
SRS-025	*1/*1	[TA]6/[TA]6	CC	GG
SRS-026	*1/*1	[TA]6/[TA]6	CC	GG
SRS-027	*1/*1	[TA]6/[TA]6	CC	GG
SRS-028	*1/*1	[TA]6/[TA]6	CC	GG
SRS-029	*1/*1	[TA]6/[TA]6	CC	GG
SRS-030	*1/*1	[TA]6/[TA]6	CC	GG
USH-001	*1/*28	[TA]6/[TA]7	CT	GG
USH-002	*1/*28	[TA]6/[TA]7	CT	GG
USH-003	*1/*28	[TA]6/[TA]7	CT	GG

Table A4.10 continued: Diplotypes for *UGT1A1*

Patient ID	<i>F5</i>	<i>HLA-A *31:01</i>	<i>IFNL3</i>	<i>RARG</i>	<i>SLC28A3</i>	<i>SLCO1B1</i>	<i>UGT1A6</i>
	rs6025	rs1061235	rs12979860	rs2229774	rs7853758	rs4149056	rs17863783
	1:169549811	6:29945521	19:39248147	12:53211761	9:84286011	12:21178615	2:233693631
	g.169549811 C>T p.Arg534Gln	g.29945521 A>T	g.39248147 C>T	g.53211761 G>A p.Ser427Leu	g.84286011 G>A p.Leu461=	g.21178615 T>C p.Val174Ala	g.233693631 G>T p.Val209=
<b>BBS-001</b>	CC	AA	CT	GG	GG	TT	GG
<b>BBS-002</b>	CC	AA	CT	GG	GG	TT	GG
<b>BBS-003</b>	CC	AA	CC	GG	GG	TT	GG
<b>BBS-004</b>	CC	AA	CC	GG	GG	TC	GG
<b>BBS-005</b>	CC	AA	CC	GG	GG	TT	GG
<b>BBS-006</b>	CC	AA	CC	GG	AA	TT	GG
<b>BBS-007</b>	CC	AA	CC	GG	GG	TT	GG
<b>BBS-008</b>	CC	AA	CC	GG	GA	TT	GG
<b>BBS-009</b>	CC	AA	CC	GG	GG	TT	GT
<b>BBS-010</b>	CC	AA	CT	GG	GA	TT	GG
<b>BBS-011</b>	CC	AA	CT	GG	GA	TT	GG
<b>BBS-012</b>	CC	AA	CC	GG	GA	TT	GG
<b>BBS-013</b>	CC	AA	TT	GG	GG	TT	GG
<b>BBS-014</b>	CC	AA	CC	GG	GG	TT	GT
<b>BBS-015</b>	CC	AA	CT	GG	GG	TT	GG
<b>BBS-016</b>	CC	AA	TT	GG	GA	TT	GT
<b>BBS-017</b>	CC	AA	TT	GG	GA	TT	GT
<b>BBS-018</b>	CC	AA	CC	GG	GG	TC	GG
<b>IBD-001</b>	CC	AA	CT	GG	GG	TT	GG
<b>IBD-002</b>	CC	AA	CT	GG	GG	TC	GG
<b>IBD-003</b>	CC	AA	CC	GA	GG	TT	GG
<b>IBD-004</b>	CC	AA	TT	GA	AA	TT	GG
<b>IBD-005</b>	CC	AA	CC	GG	GG	TT	GT

Table 4.11: Genotypes for *F5*, *HLA-A \*31:01*, *IFNL3*, *RARG*, *SLC28A3*, *SLCO1B1* and *UGT1A6*



Patient ID	<i>F5</i>	<i>HLA-A *31:01</i>	<i>IFNL3</i>	<i>RARG</i>	<i>SLC28A3</i>	<i>SLCO1B1</i>	<i>UGT1A6</i>
	rs6025	rs1061235	rs12979860	rs2229774	rs7853758	rs4149056	rs17863783
<b>IBD-006</b>	CC	AA	CC	GG	GG	TT	GG
<b>IBD-007</b>	CC	AA	CC	GG	GG	TT	GG
<b>IBD-008</b>	CC	AA	TT	GG	GA	TT	GT
<b>IBD-009</b>	CC	AA	CC	GG	GG	TT	GG
<b>IBD-010</b>	CC	AA	CT	GG	GA	TC	GG
<b>IBD-011</b>	CC	AA	CC	GG	GA	TT	GG
<b>IBD-012</b>	CT	AA	CC	GG	GG	TT	GG
<b>IBD-013</b>	CC	AA	CT	GG	GG	TT	GG
<b>IBD-014</b>	CC	AA	CC	GG	GA	TT	GG
<b>IBD-015</b>	CC	TT	CC	GG	GG	TT	GG
<b>IBD-016</b>	CC	AA	CT	GG	GG	TC	GG
<b>IBD-017</b>	CC	AA	CC	GA	GA	TT	GG
<b>IBD-018</b>	CC	AA	CT	GG	GG	TT	GG
<b>IBD-019</b>	CC	AA	CC	GG	GG	TT	GG
<b>IBD-020</b>	CC	AA	CC	GA	GG	TT	GG
<b>JDM-001</b>	CC	AA	CT	GG	GG	TT	GG
<b>JDM-002</b>	CC	AA	CC	GG	GG	TC	GG
<b>JDM-003</b>	CC	AA	CT	GA	GA	TT	GG
<b>JDM-004</b>	CC	AA	CT	GA	GA	TT	GG
<b>JDM-005</b>	CC	AA	CT	GG	GA	TT	GG
<b>JDM-006</b>	CC	AA	CT	GG	GG	TC	GG
<b>JDM-007</b>	CC	AT	TT	GG	GA	TT	GG
<b>JDM-008</b>	CT	AA	CT	GG	GA	TT	GG
<b>JDM-009</b>	CC	AT	CT	GA	GA	TT	GG
<b>JDM-010</b>	CC	AA	CT	GG	GG	TC	GG
<b>JDM-011</b>	CC	AA	CC	GG	GG	TC	GG

Table 4.11 continued: Genotypes for *F5*, *HLA-A \*31:01*, *IFNL3*, *RARG*, *SLC28A3*, *SLCO1B1* and *UGT1A6*

Patient ID	<i>F5</i>	<i>HLA-A *31:01</i>	<i>IFNL3</i>	<i>RARG</i>	<i>SLC28A3</i>	<i>SLCO1B1</i>	<i>UGT1A6</i>
	rs6025	rs1061235	rs12979860	rs2229774	rs7853758	rs4149056	rs17863783
<b>JDM-012</b>	CC	AA	CT	GA	GG	TT	GG
<b>JDM-013</b>	CC	AA	TT	GG	GG	TT	GT
<b>SRS-001</b>	CC	AA	CT	GG	GG	TT	GG
<b>SRS-002</b>	CC	AA	CC	GA	GG	TT	GG
<b>SRS-003</b>	CC	AA	CT	GG	GA	TT	GG
<b>SRS-004</b>	CC	AA	CT	GG	GG	TT	GG
<b>SRS-005</b>	CC	AA	TT	GG	GG	TT	GG
<b>SRS-006</b>	CC	AA	CT	GG	GA	TT	GG
<b>SRS-007</b>	CC	AA	CC	GG	AA	TC	GT
<b>SRS-008</b>	CC	AA	CC	GG	AA	TC	GG
<b>SRS-009</b>	CC	AA	CC	GG	GA	TT	GT
<b>SRS-010</b>	CC	AT	CT	GG	GG	TT	GG
<b>SRS-011</b>	CC	AT	CC	GG	GG	TT	GG
<b>SRS-012</b>	CC	AA	CT	GG	GA	TT	GG
<b>SRS-013</b>	CC	AA	CT	GG	GG	TT	TT
<b>SRS-014</b>	CC	AA	CC	GG	GG	TT	GT
<b>SRS-015</b>	CC	AA	CT	GG	GG	TC	GT
<b>SRS-016</b>	CC	AT	CC	GG	GA	TT	GG
<b>SRS-017</b>	CC	AT	CC	GG	GG	TT	GG
<b>SRS-018</b>	CC	AT	CT	GA	GA	TT	GG
<b>SRS-019</b>	CC	AA	CC	GG	GG	TT	GG
<b>SRS-020</b>	CC	AA	CC	GG	GG	TT	GG
<b>SRS-021</b>	CC	AA	CC	GG	GG	TT	GG
<b>SRS-022</b>	CC	AA	CT	GG	GG	TC	GG
<b>SRS-023</b>	CC	AA	CT	GG	GG	CC	GG
<b>SRS-024</b>	CC	AA	CC	GG	GG	TT	GG

Table 4.11 continued: Genotypes for *F5*, *HLA-A \*31:01*, *IFNL3*, *RARG*, *SLC28A3*, *SLCO1B1* and *UGT1A6*

Patient ID	<i>F5</i>	<i>HLA-A *31:01</i>	<i>IFNL3</i>	<i>RARG</i>	<i>SLC28A3</i>	<i>SLCO1B1</i>	<i>UGT1A6</i>
	rs6025	rs1061235	rs12979860	rs2229774	rs7853758	rs4149056	rs17863783
<b>SRS-025</b>	CC	AA	CC	GG	GG	TT	GG
<b>SRS-026</b>	CC	AA	CC	GG	GG	TT	GG
<b>SRS-027</b>	CC	AA	CC	GG	GG	TC	GG
<b>SRS-028</b>	CC	AA	CC	GG	GG	TC	GG
<b>SRS-029</b>	CC	AA	CT	GG	GG	TT	GG
<b>SRS-030</b>	CC	AA	CT	GG	GG	TC	GG
<b>USH-001</b>	CT	AA	TT	GG	GG	TT	GG
<b>USH-002</b>	CC	AA	CT	GG	GG	TC	GG
<b>USH-003</b>	CC	AA	CT	GG	GG	TT	GT

Table 4.11 continued: Genotypes for *F5*, *HLA-A \*31:01*, *IFNL3*, *RARG*, *SLC28A3*, *SLCO1B1* and *UGT1A6*