Table S4. Coverage of 203 CPIC/PharmGKB variant positions

Gene	Genomic position (GRCh37)	mRNA	CPIC/PharmGKB variant	GS HQ GT cts (N=5)	ES HQ GT cts (N=973)	Chip HQ GT cts (N=5)
MTHFR	1:11856378	NM_005957.4:c.665C>T	p.Ala222Val	5	953	0
DPYD	1:97547947	NM_000110.3:c.2846A>T	p.Asp949Val	5	972	0
DPYD	1:97770920	NM_000110.3:c.2194G>A	p.Val732Ile	5	973	0
DPYD	1:97915614	NM_000110.3:c.1905+1G>A	IVS14+1G>A	5	923	5
DPYD	1:97981343	NM_000110.3:c.1679T>A	p.Ile560Asn	5	973	5
DPYD	1:97981395	NM_000110.3:c.1627A>G	p.Ile543Val	5	973	0
DPYD	1:97981421	NM_000110.3:c.1601G>A	p.Ser534Asn	5	971	5
DPYD	1:98348885	NM_000110.3:c.85T>C	p.Cys29Arg	5	973	5
XPC	3:14187449	NM_004628.4:c.2815C>A	p.Gln939Lys	5	544	0
TPMT	6:18130918	NM_000367.3:c.719A>G	p.Tyr240Cys	5	969	5
TPMT	6:18131012	NM_000367.3:c.626-1G>A	IVS8-1G>A	5	965	5
TPMT	6:18139214	NM_000367.3:c.474C>T	p.Ile158Ile	5	973	5
TPMT	6:18139228	NM_000367.3:c.460G>A	p.Ala154Thr	5	973	5
TPMT	6:18143955	NM_000367.3:c.238G>C	p.Ala80Pro	5	967	5
TPMT	6:18155397	NA	Promoter	0	0	0
CFTR	7:117174372	NM_000492.3:c.532G>A	p.Gly178Arg	5	971	0
CFTR	7:117199645_117199647	NM_000492.3:c.1520_1522del	p.Phe508del	5	918	0
CFTR	7:117199646_117199648	NM_000492.3:c.1521_1523del	p.Phe508del	5	897	0
CFTR	7:117227853	NM_000492.3:c.1645A>C	p.Ser549Arg	5	968	0
CFTR	7:117227854	NM_000492.3:c.1646G>A	p.Ser549Asn	5	967	0
CFTR	7:117227855	NM_000492.3:c.1647T>G	p.Ser549Arg	5	968	0
CFTR	7:117227859	NM_000492.3:c.1651G>A	p.Gly551Ser	5	962	0
CFTR	7:117227860	NM_000492.3:c.1652G>A	p.Gly551Asp	5	966	0
CFTR	7:117282505	NM_000492.3:c.3731G>A	p.Gly1244Glu	5	969	0
CFTR	7:117282526	NM_000492.3:c.3752G>A	p.Ser1251Asn	5	967	0
CFTR	7:117282537	NM_000492.3:c.3763T>C	p.Ser1255Pro	5	967	0
CFTR	7:117304824	NM_000492.3:c.4046G>A	p.Gly1349Asp	5	973	0
EGFR	7:55259515	NM_005228.3:c.2573T>G	p.Leu858Arg	5	943	0
CYP3A5	7:99270539	NM_000777.3:c.219-237G>A	IVS3-1G>A	4	59	5
CYP2C19	10:96519061	NA	Intergenic	5	0	0
CYP2C19	10:96521574	NM_000769.2:c.-889T>G	Promoter	5	0	0

CYP2C19	10:96521657	NM_000769.2:c.-806C>T	Promoter	5	0	5
CYP2C19	10:96522365	NM_000769.2:c.-98T>C	Promoter	5	661	0
CYP2C19	10:96522463	NM_000769.1:c.1A>G	p.Met1Val	5	959	5
CYP2C19	10:96522561	NM_000769.2:c.99C>T	p.Pro33Pro	5	967	0
CYP2C19	10:96534475	NM_000769.2:c.169-340T>G	IVS1-340T>G	5	0	0
CYP2C19	10:96534584	NM_000769.2:c.169-231G>A	IVS1-231G>A	5	66	0
CYP2C19	10:96534768	NM_000769.2:c.169-47G>A	IVS1-47G>A	5	889	0
CYP2C19	10:96534922	NM_000769.2:c.276G>C	p.Glu92Glu	5	973	5
CYP2C19	10:96535124	NM_000769.2:c.332-23A>G	IVS2-23A>G	5	968	0
CYP2C19	10:96535173	NM_000769.1:c.358T>C	p.Trp120Arg	5	973	5
CYP2C19	10:96535210	NM_000769.1:c.395G>A	p.Arg132Gln	5	972	5
CYP2C19	10:96535222	NM_000769.1:c.407T>A	p.Met136Lys	5	971	0
CYP2C19	10:96535296	NM_000769.2:c.481G>C	p.Ala161Pro	5	958	0
CYP2C19	10:96535378	NM_000769.2:c.481+332T>C	IVS3+332T>C	5	715	0
CYP2C19	10:96540410	NM_000769.1:c.636G>A	p.Trp212X	5	886	5
CYP2C19	10:96541373	NM_000769.2:c.643-205A>G	IVS4-205A>G	5	4	0
CYP2C19	10:96541616	NM_000769.1:c.681G>A	p.Pro227Pro	5	774	5
CYP2C19	10:96541748	NM_000769.1:c.813G>A	p.Met271Ile	5	907	0
CYP2C19	10:96541756	NM_000769.1:c.819+2T>A	IVS5+2T>A	5	884	5
CYP2C19	10:96541982	NM_000769.2:c.819+228A>G	IVS5+228A>G	5	24	0
CYP2C19	10:96580202	NM_000769.2:c.820-51C>G	IVS4-51C>G	4	912	0
CYP2C19	10:96602398	NM_000769.2:c.962-196T>A	IVS6-196T>A	5	69	0
CYP2C19	10:96602622	NM_000769.2:c.990C>T	p.Val330Val	5	887	0
CYP2C19	10:96602623	NM_000769.2:c.991A>G	p.Ile331Val	5	880	5
CYP2C19	10:96602653	NM_000769.1:c.1021G>A	p.Asp341Asn	5	912	0
CYP2C19	10:96602710	NM_000769.2:c.1078G>A	p.Asp360Asn	5	962	0
CYP2C19	10:96602711	NM_000769.1:c.1079A>T	p.Asp360Val	5	960	0
CYP2C19	10:96609710	NM_000769.1:c.1186C>G	p.His396Asp	5	972	0
CYP2C19	10:96609737	NM_000769.1:c.1213G>A	p.Glu405Lys	5	970	0
CYP2C19	10:96609775	NM_000769.2:c.1251A>C	p.Gly417Gly	5	970	0
CYP2C19	10:96609785	NM_000769.1:c.1261A>C	p.Lys421Gln	5	967	0
CYP2C19	10:96612495	NM_000769.1:c.1297C>T	p.Arg433Trp	5	973	5
CYP2C9	10:96702047	NM_000771.3:c.430C>T	p.Arg144Cys	4	973	5
CYP2C9	10:96741053	NM_000771.3:c.1075A>C	p.Ile359Leu	5	958	5
ANKK1	11:113270828	NM_178510.1:c.2137G>A	p.Glu713Lys	4	546	0
GRIK4	11:120663363	NM_014619.3:c.83-10039T>C	IVS3-10039T>C	5	0	0

SLCO1B1	12:21283322	NM_006446.4:c.-910G>A	Promoter	5	0	5
SLCO1B1	12:21329738	NM_006446.4:c.388A>G	p.Asn130Asp	5	962	5
SLCO1B1	12:21331549	NM_006446.4:c.521T>C	p.Val174Ala	5	973	5
VKORC1	16:31102321	NM_024006.4:c.*134G>A	3'UTR	5	335	5
VKORC1	16:31104878	NM_024006.4:c.174-136C>T	IVS1-136C>T	4	249	5
VKORC1	16:31107689	NA	Promoter	5	0	5
CYP4F2	19:15990431	NM_001082.3:c.1297G>A	p.Val433Met	5	973	5
IFNL3, IFNL4	19:39738787	NM_001276254.2:c.151-152G>A	IVS1-152G>A	3	3	0
IFNL3, IFNL4	19:39743165	NA	Intergenic	5	0	0
CYP2D6	22:42522312	NM_000106.5:c.*75+189A>G	Intergenic	4	76	0
CYP2D6	22:42522392	NM_000106.5:c.*75+109C>T	Intergenic	4	253	0
CYP2D6	22:42522613	NM_000106.5:c.1457G>C	p.Ser486Thr	4	824	5
CYP2D6	22:42522660_42522668	NM_000106.5:c.1402_1410dup	p.Val468_Thr470dup	4	447	1
CYP2D6	22:42522678	NM_000106.5:c.1392C>T	p.Phe464Phe	4	335	0
CYP2D6	22:42522749	NM_000106.5:c.1321C>T	p.Arg441Cys	0	254	0
CYP2D6	22:42522751	NM_000106.5:c.1319G>A	p.Arg440His	0	255	0
CYP2D6	22:42522906	NM_000106.5:c.1262T>C	p.Leu421Pro	4	794	0
CYP2D6	22:42522916	NM_000106.5:c.1252G>C	p.Glu418Gln	4	842	0
CYP2D6	22:42522940	NM_000106.5:c.1228C>T	p.Glu410Lys	4	913	0
CYP2D6	22:42522958	NM_000106.5:c.1210A>C	p.Lys404Gln	4	880	0
CYP2D6	22:42522965	NM_000106.5:c.1203G>A	p.Ser401Ser	4	862	0
CYP2D6	22:42523003	NM_000106.5:c.1174-9C>T	IVS7-9C>T	3	407	0
CYP2D6	22:42523209	NM_000106.5:c.1174-215A>G	IVS7-215A>G	4	250	0
CYP2D6	22:42523211	NM_000106.5:c.1174-217A>G	IVS7-217A>G	4	207	0
CYP2D6	22:42523302	NM_000106.5:c.1173+147G>A	IVS7+147G>A	4	257	0
CYP2D6	22:42523358	NM_000106.5:c.1173+91C>A	IVS7+91C>A	4	381	0
CYP2D6	22:42523409	NM_000106.5:c.1173+40A>C	IVS7+40A>C	5	870	0
CYP2D6	22:42523475	NM_000106.5:c.1147G>A	p.Glu383Lys	4	966	0
CYP2D6	22:42523505	NM_000106.5:c.1117G>A	p.Gly373Ser	4	832	0
CYP2D6	22:42523533_42523534	NM_000106.5:c.1088_1089dup	p.Gln364CysfsX12	4	970	5
CYP2D6	22:42523539	NM_000106.5:c.1083T>C	p.His310His	4	897	0
CYP2D6	22:42523592	NM_000106.5:c.1030C>T	p.Arg344X	4	962	5
CYP2D6	22:42523610	NM_000106.5:c.1012G>A	p.Val338Met	4	964	5
CYP2D6	22:42523621	NM_000106.5:c.1001A>C	p.Glu334Ala	4	964	0
CYP2D6	22:42523763	NM_000106.5:c.985+81G>A	IVS6+81G>A	1	544	0
CYP2D6	22:42523805	NM_000106.5:c.985+39G>A	IVS6+39G>A	4	874	5

CYP2D6	22:42523843	NM_000106.5:c.985+1G>C	IVS6+1G>C	4	935	5
CYP2D6	22:42523854	NM_000106.5:c.975G>A	p.Pro325Pro	4	941	0
CYP2D6	22:42523855	NM_000106.5:c.974C>T	p.Pro325Leu	4	945	0
CYP2D6	22:42523858	NM_000106.5:c.971A>C	p.His324Pro	4	949	5
CYP2D6	22:42523943	NM_000106.5:c.886C>T	p.Arg296Cys	4	954	5
CYP2D6	22:42523944	NM_000106.5:c.885G>A	p.Leu295Leu	4	933	0
CYP2D6	22:42524132	NM_000106.5:c.843+44G>A	IVS5+44G>A	3	343	0
CYP2D6	22:42524176_42524178	NM_000106.5:c.841_843del	p.Lys281del	4	619	5
CYP2D6	22:42524203_42524206	NM_000106.5:c.813_816del	p.Thr272ArgfsX25	4	727	5
CYP2D6	22:42524217	NM_000106.5:c.802C>T	p.Pro268Ser	3	799	0
CYP2D6	22:42524218	NM_000106.5:c.801C>A	p.Pro267Pro	3	776	0
CYP2D6	22:42524219_42524220	NM_000106.5:c.799_800insG	p.Pro267ArgfsX7	3	796	0
CYP2D6	22:42524237	NM_000106.5:c.782C>T	p.Thr261Ile	3	892	0
CYP2D6	22:42524244	NM_000106.5:c.775del	p.Arg259GlyfsX2	3	907	5
CYP2D6	22:42524251_42524254	NM_000106.5:c.765_768del	p.Thr256SerfsX4	4	951	5
CYP2D6	22:42524310	NM_000106.5:c.709G>T	p.Ala237Ser	5	969	0
CYP2D6	22:42524313	NM_000106.5:c.706C>T	p.Leu236Leu	5	968	0
CYP2D6	22:42524323	NM_000106.5:c.696T>C	p.Trp262Trp	5	959	0
CYP2D6	22:42524490	NM_000106.5:c.667-138C>T	IVS4-138C>T	4	249	0
CYP2D6	22:42524502	NM_000106.5:c.667-150G>A	IVS4-150G>A	4	223	0
CYP2D6	22:42524664	NM_000106.5:c.666+122A>C	IVS4+122A>C	4	258	0
CYP2D6	22:42524696	NM_000106.5:c.666+90A>G	IVS4+90A>G	5	400	0
CYP2D6	22:42524814	NM_000106.5:c.638T>C	p.Leu213Pro	4	843	0
CYP2D6	22:42524815	NM_000106.5:c.637C>T	p.Leu213Leu	4	837	0
CYP2D6	22:42524817	NM_000106.5:c.635G>A	p.Gly212Glu	4	837	0
CYP2D6	22:42524819_42524820	NM_000106.5:c.632_633insC	p.Glu211AspfsX43	4	830	5
CYP2D6	22:42524906_42524907	NM_000106.5:c.545_546insTA	p.Ser183ArgfsX4	4	361	0
CYP2D6	22:42524924	NM_000106.5:c.528T>C	p.Gly126Gly	3	350	0
CYP2D6	22:42524929_42524930	NM_000106.5:c.522_523insTTTCGCCCCTTTCGCCCC	p.Pro174_Asn175insPheArgProPheArgPro	4	333	5
CYP2D6	22:42524935	NM_000106.5:c.517C>T	p.Arg173Cys	4	329	0
CYP2D6	22:42524947	NM_000106.5:c.506-1G>A	IVS3-1G>A	3	445	5
CYP2D6	22:42525035	NM_000106.5:c.505G>T, G>A	p.Gly169X, p.Gly169Arg	4	500	5
CYP2D6	22:42525036	NM_000106.5:c.504C>T	p.Ser168Ser	4	471	0
CYP2D6	22:42525044	NM_000106.5:c.496A>G	p.Asn166Asp	3	472	0
CYP2D6	22:42525069	NM_000106.5:c.471C>T	p.Ala157Ala	4	495	0

CYP2D6	22:42525073	NM_000106.5:c.467A>C	p.Glu156Ala	4	497	0
CYP2D6	22:42525077	NM_000106.5:c.463G>A	p.Glu155Lys	4	505	0
CYP2D6	22:42525086	NM_000106.5:c.454del	p.Trp152GlyfsX2	4	517	5
CYP2D6	22:42525132	NM_000106.5:c.408G>C	p.Val136Val	4	563	5
CYP2D6	22:42525134	NM_000106.5:c.406G>A	p.Val136Met	4	543	4
CYP2D6	22:42525176	NM_000106.5:c.364G>T	p.Ala122Ser	3	512	0
CYP2D6	22:42525182	NM_000106.5:c.358T>A	p.Phe120Ile	3	497	0
CYP2D6	22:42525195	NM_000106.5:c.353-8A>G	IVS2-8A>G	3	463	0
CYP2D6	22:42525280	NM_000106.5:c.353-93C>T	IVS2-93C>T	4	254	0
CYP2D6	22:42525733	NM_000106.5:c.352+7A>G	IVS2+7T>C	0	681	0
CYP2D6	22:42525754	NM_000106.5:c.338G>A	p.Gly113Glu	1	608	0
CYP2D6	22:42525772	NM_000106.5:c.320C>T	p.Thr107Ile	1	494	5
CYP2D6	22:42525796	NM_000106.5:c.296C>G	p.Ala99Gly	0	191	0
CYP2D6	22:42525811	NM_000106.5:c.281A>G	p.His94Arg	1	324	0
CYP2D6	22:42525819	NM_000106.5:c.273G>T	p.Leu91Leu	1	185	0
CYP2D6	22:42525821	NM_000106.5:c.271C>A	p.Leu91Met	1	283	0
CYP2D6	22:42525823	NM_000106.5:c.269C>T	p.Ala90Val	1	182	0
CYP2D6	22:42525906	NM_000106.5:c.186G>A	p.Arg62Arg	0	6	0
CYP2D6	22:42525912	NM_000106.5:c.181-1G>C	IVS1-1G>C	0	6	5
CYP2D6	22:42525950	NM_000106.5:c.181-39A>C	IVS1-39T>G	0	8	0
CYP2D6	22:42526048	NM_000106.5:c.181-137G>C	Intronic	0	1	0
CYP2D6	22:42526192	NM_000106.5:c.181-281del	IVS1-281del	2	0	0
CYP2D6	22:42526484	NM_000106.5:c.180+130T>G	IVS1+130G>T	4	153	0
CYP2D6	22:42526567	NM_000106.5:c.180+47A>G	IVS1+47T>C	4	476	0
CYP2D6	22:42526571	NM_000106.5:c.180+43G>C	IVS1+43C>G	4	498	0
CYP2D6	22:42526573	NM_000106.5:c.180+41A>C	IVS1+41C>A	4	512	0
CYP2D6	22:42526580	NM_000106.5:c.180+34C>G	IVS1+34C>G	4	535	0
CYP2D6	22:42526656_42526657	NM_000106.5:c.137dup	p.Leu47ThrfsX207	5	614	5
CYP2D6	22:42526670	NM_000106.5:c.124G>A	p.Gly42Arg	4	594	5
CYP2D6	22:42526694	NM_000106.5:c.100C>T	p.Pro34Ser	4	563	5
CYP2D6	22:42526712	NM_000106.5:c.82C>T	p.Arg28Cys	3	439	0
CYP2D6	22:42526717	NM_000106.5:c.77G>A	P.Arg26His	4	448	0
CYP2D6	22:42526721	NM_000106.5:c.73C>T	p.Arg25Trp	3	483	0
CYP2D6	22:42526763	NM_000106.5:c.31G>A	p.Val11Met	4	529	0
CYP2D6	22:42527422	NM_000106.5:c.-629T>C	Promoter	4	0	0
CYP2D6	22:42527471	NM_000106.5:c.-678C>T	Promoter	4	0	0

CYP2D6	22:42527485_42527488	NM_000106.5:c.-695_-692del	Promoter	4	0	0
CYP2D6	22:42527533	NM_000106.5:c.-740C>T	Promoter	5	0	0
CYP2D6	22:42527542_42527543	NM_000106.5:c.-750_-749del	Promoter	4	0	0
CYP2D6	22:42527793	NM_000106.5:c.-1001C>T	Promoter	5	1	0
CYP2D6	22:42527804	NM_000106.5:c.-1011T>C	Promoter	5	0	0
CYP2D6	22:42527886_42527887	NM_000106.5:c.-1094_-1093insT	Promoter	4	0	0
CYP2D6	22:42528028	NM_000106.5:c.-1235G>A	Promoter	2	0	0
CYP2D6	22:42528030_42528031	NM_000106.5:c.-1238_-1237insAA	Promoter	0	0	0
CYP2D6	22:42528051_42528052	NM_000106.5:c.-1259_-1258insTTTTT	Promoter	1	0	0
CYP2D6	22:42528096	NM_000106.5:c.-1299C>T	Promoter	4	0	0
CYP2D6	22:42528224	NM_000106.5:c.-1427G>A	Promoter	5	0	0
CYP2D6	22:42528336	NM_000106.5:c.-1543C>T	Promoter	4	0	0
CYP2D6	22:42528377	NM_000106.5:c.-1584G>C	Promoter	5	0	0
CYP2D6	22:42528393_42528394	NM_000106.5:c.-1601_-1600del	Promoter	5	0	0
CYP2D6	22:42528568	NM_000106.5:c.-1772C>T	Promoter	4	3	5
G6PD	X:153760484	NM_001042351.1:c.1376G>C	p.Arg459Pro or p.Arg459Leu	0	787	5
G6PD	X:153760654	NM_001042351.2:c.1311C>T	p.Tyr437Tyr	1	849	0
G6PD	X:153761205	NM_001042351.2:c.1003G>A	p.Ala335Thr	1	595	5
G6PD	X:153761206_153761208	NM_001042351.1:c.1000_1002del	p.Thr334del	2	614	0
G6PD	X:153761240	NM_001042351.1:c.968T>C	p.Leu323Pro	3	718	0
G6PD	X:153761259	NM_001042351.1:c.949G>A	p.Glu317Lys	4	795	0
G6PD	X:153761337	NM_001042351.2:c.871G>A	p.Val291Met	2	803	0
G6PD	X:153761830	NM_001042351.1:c.825G>C	p.Lys275Asn	0	944	0
G6PD	X:153762340	NM_001042351.1:c.680G>T or G>A	p.Arg227Leu or p.Arg227Gln	3	925	0
G6PD	X:153762634	NM_001042351.2:c.563C>T	p.Ser188Phe	3	930	5
G6PD	X:153763492	NM_001042351.1:c.376A>G	p.Asn126Asp	2	753	5
G6PD	X:153763531	NM_001042351.1:c.337G>A	p.Asp113Asn	0	560	5
G6PD	X:153764217	NM_001042351.2:c.202G>A	p.Val68Met	0	887	0
G6PD	X:153764383	NM_001042351.1:c.131C>G	p.Ala44Gly	2	750	0

Chip, Affymetrix DMET Plus (Drug Metabolizing Enzymes and Transporters array); CPIC, Clinical Pharmacogenetics Implementation Consortium; GRCh37, Genome Reference Consortium human genome (build 37); HQ GT cts, high quality genotype counts (genotype quality score ≥50 for exomes and genomes, genotype confidence score <0.05 for chip); N, number of individuals tested per platform; PharmGKB, Pharmacogenomics Knowledgebase; SEM, standard error of the mean.

Coverage of 203 phamacogenetic positions selected from CPIC and PharmGKB were assessed in 973 exomes, 5 genomes, and 5 chip data. Coverage of the 203 variant positions from 3 platforms was assessed in coding and noncoding regions.

Summary statistics

Platform/location	Mean	SEM	N
Agilent38Mb/coding	91.38	7.46	393
Agilent50Mb/coding	109.17	4.87	318
TruSeqv1/coding	118.01	2.31	147
TruSeqv2/coding	122.11	1.78	115
Genome/coding	100.60	0.65	5
Chip/coding	44.80	0.33	5

Platform/location	Mean	SEM	N
Agilent38Mb/noncoding	13.66	6.63	393
Agilent50Mb/noncoding	18.36	6.73	318
TruSeqv1/noncoding	26.78	4.93	147
TruSeqv2/noncoding	27.37	4.59	115
Genome/noncoding	54.6	0.66	5
Chip/noncoding	14	0.9	5