

Table S3. SNVs/indels detected by PGRNseq data that were within 3 kb from either end of *CYP2D6*

No.	Position*	rs ID	Reference Allele	Alternative Allele	Function**	Amino Acids	Protein Position	CADD Score	Allele Count	Within Haplotype Definition Region***	In Translation Table****
1	42521442	rs535293691	G	A	Downstream-gene	-	-	4.086	2	No	No
2	42521489	rs145458535	C	T	Downstream-gene	-	-	1.976	79	No	No
3	42521577	rs201551607	G	A	Downstream-gene	-	-	0.172	2	No	No
4	42521625	rs72395204	GGGGTGGGGAA GGGTGGGGAA	G	Downstream-gene	-	-	-	52	No	No
5	42521625	rs72395204	GGGGTGGGGAA GGGTGGGGAA	GGGGTGGGGAA	Downstream-gene	-	-	-	98	No	No
6	42521625	rs72395204	GGGGTGGGGAA GGGTGGGGAA	GGGGTGGGGAA GGGTGGGGAA GGGTGGGGAA	Downstream-gene	-	-	-	7	No	No
7	42521676	-	G	A	Downstream-gene	-	-	0.257	1	No	No
8	42521799	-	GGCCTGTGGGCCCT	G	Downstream-gene	-	-	-	1	No	No
9	42521807	rs561101513	G	T	Downstream-gene	-	-	2.048	1	No	No
10	42521811	-	C	G	Downstream-gene	-	-	4.42	1	No	No
11	42521865	rs112866416	C	T	Downstream-gene	-	-	2.139	4	No	No
12	42521918	rs532182046	T	C	Downstream-gene	-	-	3.416	6	No	No
13	42521919	rs550576546	G	A	Downstream-gene	-	-	7.514	6	No	No
14	42521920	rs562602203	T	G	Downstream-gene	-	-	5.554	5	No	No
15	42521929	rs34385013	G	A	Downstream-gene	-	-	2.417	261	No	No
16	42521969	-	G	A	Downstream-gene	-	-	3.234	1	No	No
17	42521985	rs4078247	T	C	Downstream-gene	-	-	0.999	145	No	No
18	42522027	rs4078248	C	T	Downstream-gene	-	-	8.703	4	No	No
19	42522074	rs35028622	C	A	Downstream-gene	-	-	1.735	259	No	Yes
20	42522079	rs77827855	G	A	Downstream-gene	-	-	0.286	2	No	No
21	42522084	rs4078249	C	T	Downstream-gene	-	-	3.795	4	No	No
22	42522101	rs866408541	C	T	Downstream-gene	-	-	5.332	1	No	No
23	42522137	rs71184866	C	CTGT	Downstream-gene	-	-	-	405	No	No
24	42522141	-	G	T	Downstream-gene	-	-	3.167	2	No	No
25	42522142	-	A	G	Downstream-gene	-	-	3.045	2	No	No
26	42522312	rs116390392	T	C	Downstream-gene	-	-	2.511	413	Yes	Yes
27	42522349	rs148648640	T	C	Downstream-gene	-	-	0.894	1	Yes	Yes
28	42522392	rs28371738	G	A	Downstream-gene	-	-	1.136	145	Yes	Yes
29	42522464	rs77845838	G	A	Downstream-gene	-	-	5.327	7	Yes	Yes
30	42522550	rs201759814	G	A	3-prime-UTR	-	-	4.413	41	Yes	Yes
31	42522613	rs1135840	G	C	Missense	THR,SER	486/498	3.127	261	Yes	Yes
32†	42522649	rs141756339	C	T	Missense	ARG,GLN	474/498	11.57	1	Yes	No
33‡	42522724	rs79392742	G	T	Missense	ALA,ASP	449/498	32	1	Yes	No
34	42522765	rs548264542	G	T	Intron	-	-	6.939	1	Yes	No
35	42522774	rs772203297	G	A	Intron	-	-	1.665	3	Yes	No
36	42522846	-	C	T	Intron-near-splice	-	-	5.595	1	Yes	No
37	42522940	rs769157652	C	T	Missense	GLU,LYS	410/498	15.05	1	Yes	Yes
38	42522965	rs28371732	C	T	Synonymous	SER	401/498	0.891	2	Yes	Yes
39	42523003	rs116917064	A	G	Intron	-	-	3.767	368	Yes	Yes
40	42523070	rs1009883497	GAC	G	Intron	-	-	-	1	Yes	No

41	42523209	rs28371730	T	C	Intron	-	-	1.232	408	Yes	Yes
42	42523211	rs2004511	T	C	Intron	-	-	0.848	146	Yes	Yes
43	42523228	-	C	T	Intron	-	-	1.957	1	Yes	No
44	42523247	rs1269631565	C	CT	Intron	-	-	-	1	Yes	Yes
45	42523309	rs867985262	C	T	Intron	-	-	3.3	120	Yes	Yes
46	42523315	rs79596243	T	C	Intron	-	-	2.375	126	Yes	Yes
47	42523358	rs28371729	G	T	Intron	-	-	5.316	17	Yes	Yes
48	42523400	rs28578778	A	G	Intron	-	-	0.507	7	Yes	Yes
49	42523409	rs1985842	G	T	Intron	-	-	0.698	201	Yes	Yes
50	42523505	rs150552908	C	T	Missense	GLY,SER	373/498	11.82	68	Yes	Yes
51	42523514	rs61745683	C	T	Missense	VAL,ILE	370/498	6.769	65	Yes	Yes
52	42523528	rs1058172	C	T	Missense	ARG,HIS	365/498	34	124	Yes	Yes
53	42523539	rs28371726	A	G	Synonymous	HIS	361/498	7.593	36	Yes	Yes
54	42523558	rs202102799	T	C	Missense	TYR,CYS	355/498	15.61	19	Yes	Yes
55	42523567	rs61736517	T	C	Missense	HIS,ARG	352/498	0.008	1	Yes	Yes
56	42523609	rs771811053	ACGT	A	Coding	-	-	-	1	Yes	No
57	42523610	rs59421388	C	T	Missense	VAL,MET	338/498	22.9	1	Yes	Yes
58	42523636	rs3915951	C	A	Missense-near-splice	ARG,LEU	329/498	14.89	63	Yes	No
59	42523650	rs370010370	C	T	Intron	-	-	4.449	1	Yes	No
60	42523695	rs768806497	CCTT	C	Intron	-	-	-	1	Yes	No
61	42523721	rs867265808	G	A	Intron	-	-	7.023	1	Yes	Yes
62	42523805	rs28371725	C	T	Intron	-	-	6.324	75	Yes	Yes
63	42523813	rs143276168	G	A	Intron	-	-	1.046	2	Yes	Yes
64	42523820	rs772490933	G	A	Intron	-	-	5.597	1	Yes	No
65	42523854	rs79292917	C	T	Synonymous	PRO	325/498	4.125	5	Yes	Yes
66	42523943	rs16947	A	G	Missense	CYS,ARG	296/498	0.545	406	Yes	Yes
67	42524073	rs187203531	C	G	Intron	-	-	3.596	7	Yes	No
68	42524130	rs28371722	C	T	Intron	-	-	2.929	1	Yes	No
69	42524132	rs76015180	C	T	Intron	-	-	6.039	2	Yes	Yes
70	42524175	rs5030656	CCTT	C	Coding-near-splice	-	-	-	22	Yes	Yes
71	42524191	rs28371719	C	A	Synonymous	LEU	276/498	5.956	1	Yes	Yes
72	42524218	rs28371718	G	T	Synonymous	PRO	267/498	4.991	3	Yes	Yes
73	42524219	rs148769737	G	A	Missense	PRO,LEU	267/498	15.39	1	Yes	No
74	42524243	rs35742686	CT	C	Frameshift	-	-	-	8	Yes	Yes
75	42524310	rs28371717	C	A	Missense	ALA,SER	237/498	11.43	5	Yes	Yes
76	42524323	rs17002852	A	G	Synonymous	HIS	232/498	4.784	4	Yes	Yes
77	42524501	rs75203276	C	T	Intron	-	-	1.877	1	Yes	Yes
78	42524502	rs267608300	C	T	Intron	-	-	1.687	5	Yes	Yes
79	42524578	rs80262685	T	C	Intron	-	-	1.351	1	Yes	Yes
80	42524670	rs76327133	G	A	Intron	-	-	2.853	1	Yes	Yes
81	42524696	rs58440431	T	C	Intron	-	-	5.504	142	Yes	Yes
82	42524708	rs111564371	T	C	Intron	-	-	1.56	208	Yes	No
83	42524713	rs112568578	C	A	Intron	-	-	0.492	1	Yes	No
84	42524713	rs112568578	C	G	Intron	-	-	0.492	207	Yes	No
85	42524733	-	G	A	Intron	-	-	2.875	2	Yes	No
86	42524743	rs113889384	G	A	Intron	-	-	1.308	199	Yes	No
87	42524759	-	G	C	Intron	-	-	0.675	1	Yes	No
88	42524776	rs368129875	C	T	Intron	-	-	1.347	1	Yes	No
89	42524795	rs28371713	A	G	Synonymous	PHE	219/498	4.015	210	Yes	No
90	42524814	rs199535154	A	G	Missense	LEU,PRO	213/498	14.3	3	Yes	Yes
91	42524815	rs150163869	G	A	Synonymous	LEU	213/498	3.7	4	Yes	Yes

92	42524817	rs5030866	C	T	Missense	GLY,GLU	212/498	3.567	6	Yes	Yes
93	42524820	rs3831704	T	TC	Frameshift	-	-	-	3	Yes	Yes
94	42524860	rs763284150	C	T	Missense	ASP,ASN	198/498	8.097	1	Yes	No
95	42524924	rs111606937	A	G	Synonymous	GLY	176/498	6.286	126	Yes	Yes
96	42524947	rs3892097	C	T	Splice-acceptor	-	-	14.92	126	Yes	Yes
97	42524975	rs200720666	C	T	Intron	-	-	6.896	2	Yes	No
98	42525038	rs1135826	A	C	Missense	SER,ALA	168/498	0.028	1	Yes	Yes
99	42525039	rs1135825	G	T	Missense	HIS,GLN	167/498	0.021	1	Yes	Yes
100	42525044	rs1135824	T	C	Missense	ASN,ASP	166/498	0.003	1	Yes	Yes
101	42525045	rs768668762	G	A	Synonymous	ALA	165/498	0.032	1	Yes	Yes
102	42525085	rs5030655	CA	C	Frameshift	-	-	-	6	Yes	Yes
103	42525089	rs78482768	G	C	Missense	GLN,GLU	151/498	0.009	1	Yes	Yes
104	42525132	rs1058164	G	C	Synonymous	VAL	136/498	0.087	261	Yes	Yes
105	42525134	rs61736512	C	T	Missense	VAL,ILE	136/498	7.841	1	Yes	Yes
106	42525180	rs61736507	G	A	Synonymous	PHE	120/498	4.546	3	Yes	Yes
107	42525305	rs142302759	T	G	Intron	-	-	2.797	1	Yes	No
108	42525390	rs557722765	G	A	Intron	-	-	6.221	2	Yes	Yes
109	42525416	rs143170489	G	C	Intron	-	-	0.508	3	Yes	No
110	42525438	rs184086520	A	C	Intron	-	-	4.17	1	Yes	No
111	42525463	rs1046791114	G	A	Intron	-	-	1.17	3	Yes	No
112	42525500	rs189736703	C	T	Intron	-	-	1.057	5	Yes	No
113	42525547	rs267608277	G	A	Intron	-	-	4.796	5	Yes	Yes
114	42525625	rs1081004	C	T	Intron	-	-	4.952	34	Yes	Yes
115	42525628	-	T	A	Intron	-	-	4.379	1	Yes	No
116	42525728	rs78854695	A	C	Intron	-	-	5.434	1	Yes	Yes
117	42525733	rs267608289	T	C	Intron-near-splice	-	-	3.956	1	Yes	Yes
118	42525756	rs1081003	G	A	Synonymous	PHE	112/498	3.186	25	Yes	Yes
119	42525772	rs28371706	G	A	Missense	THR,ILE	107/498	5.817	4	Yes	Yes
120	42525798	rs28371705	G	C	Synonymous	THR	98/498	3.782	119	Yes	Yes
121	42525811	rs28371704	T	C	Missense	HIS,ARG	94/498	0.424	115	Yes	Yes
122	42525821	rs28371703	G	T	Missense	LEU,MET	91/498	14.36	115	Yes	Yes
123	42525952	rs71328650	C	A	Intron	-	-	2.593	259	Yes	Yes
124	42526049	rs147296446	C	G	Intron	-	-	3.125	275	Yes	Yes
125	42526421	rs926658668	A	G	Intron	-	-	3.423	1	Yes	No
126	42526477	rs34291018	T	C	Intron	-	-	4.764	1	Yes	Yes
127	42526484	rs28371699	A	C	Intron	-	-	6.725	258	Yes	Yes
128	42526524	rs76527171	G	A	Intron	-	-	6.971	24	Yes	Yes
129	42526549	rs56011157	C	T	Intron	-	-	6.694	408	Yes	Yes
130	42526561	rs28695233	G	T	Intron	-	-	2.422	407	Yes	Yes
131	42526562	rs75276289	G	C	Intron	-	-	1.702	407	Yes	Yes
132	42526567	rs1080998	G	A	Intron	-	-	0.469	407	Yes	Yes
133	42526571	rs1080997	C	G	Intron	-	-	1.522	407	Yes	Yes
134	42526573	rs1080996	T	G	Intron	-	-	2.388	407	Yes	Yes
135	42526580	rs1080995	G	C	Intron	-	-	3.908	407	Yes	Yes
136	42526694	rs1065852	G	A	Missense	PRO,SER	34/498	18.34	145	Yes	Yes
137	42526717	rs28371696	C	T	Missense	ARG,HIS	26/498	14.41	2	Yes	Yes
138	42526763	rs769258	C	T	Missense	VAL,MET	11/498	3.418	39	Yes	Yes
139	42526772	rs1346961531	G	A	Missense	PRO,SER	8/498	1.759	1	Yes	No
140	42526775	rs72549358	C	T	Missense	VAL,MET	7/498	1.817	1	Yes	Yes
141	42526811	rs376446555	AT	A	5-prime-UTR	-	-	-	3	Yes	No
142	42526816	rs756687222	C	G	5-prime-UTR	-	-	5.87	3	Yes	No

143	42526836	rs75085559	A	AC	5-prime-UTR	-	-	-	5	Yes	Yes
144	42526969	rs1080993	C	T	Upstream-gene	-	-	4.618	1	Yes	Yes
145	42527109	-	C	T	Upstream-gene	-	-	3.693	1	Yes	No
146	42527116	rs572914357	A	G	Upstream-gene	-	-	2.938	2	Yes	No
147	42527158	rs1080992	C	T	Upstream-gene	-	-	2.402	4	Yes	Yes
148	42527191	rs374672076	C	G	Upstream-gene	-	-	1.49	116	Yes	No
149	42527224	rs566383351	G	A	Upstream-gene	-	-	1.184	1	Yes	Yes
150	42527381	rs769257	C	T	Upstream-gene	-	-	0.566	1	Yes	Yes
151	42527471	rs28633410	T	C	Upstream-gene	-	-	0.843	401	Yes	Yes
152	42527530	-	T	C	Upstream-gene	-	-	0.539	1	Yes	No
153	42527533	rs28624811	A	G	Upstream-gene	-	-	0.79	405	Yes	Yes
154	42527580	rs949806652	C	T	Upstream-gene	-	-	4.046	1	Yes	No
155	42527684	rs114155186	C	A	Upstream-gene	-	-	0.902	1	Yes	No
156	42527741	rs200937699	G	T	Upstream-gene	-	-	0.61	1	Yes	No
157	42527793	rs1080989	C	T	Upstream-gene	-	-	0.265	145	Yes	Yes
158	42527886	rs375413467	A	AT	Upstream-gene	-	-	-	62	Yes	Yes
159	42527929	rs528127834	C	A	Upstream-gene	-	-	0.791	2	Yes	No
160	42527989	rs757622584	TC	T	Upstream-gene	-	-	-	1	Yes	No
161	42528028	rs28735595	C	T	Upstream-gene	-	-	0.256	164	Yes	Yes
162	42528096	rs59099247	C	T	Upstream-gene	-	-	0.489	1	Yes	Yes
163	42528224	rs28588594	G	A	Upstream-gene	-	-	0.835	41	Yes	Yes
164	42528382	rs1080985	C	G	Upstream-gene	-	-	0.174	486	Yes	Yes
165	42528392	rs1080984	A	G	Upstream-gene	-	-	1.42	1	Yes	Yes
166	42528477	rs775251692	C	T	Upstream-gene	-	-	5.077	3	No	No
167	42528538	rs58188898	G	A	Upstream-gene	-	-	10.95	3	No	No
168	42528568	rs1080983	T	C	Upstream-gene	-	-	0.709	414	No	Yes
169	42528682	rs551293469	G	A	Upstream-gene	-	-	4.149	4	No	No
170	42528851	rs28680494	A	C	Upstream-gene	-	-	1.804	260	No	Yes
171	42528858	rs28439297	C	T	Upstream-gene	-	-	3.928	260	No	Yes
172	42528976	rs28360521	C	T	Upstream-gene	-	-	2.547	146	No	No
173	42529156	rs73887946	C	T	Upstream-gene	-	-	14.44	1	No	No
174	42529176	rs138419008	T	C	Upstream-gene	-	-	12.69	2	No	No
175	42529219	rs28568508	A	G	Upstream-gene	-	-	4.072	148	No	No
176	42529253	-	T	A	Upstream-gene	-	-	4.734	1	No	No
177	42529321	rs28566059	C	T	Upstream-gene	-	-	6.32	104	No	No
178	42529366	rs118017929	C	G	Upstream-gene	-	-	6.933	5	No	No
179	42529407	rs28542726	G	T	Upstream-gene	-	-	7.75	35	No	Yes
180	42529540	rs28369142	C	A	Upstream-gene	-	-	0.099	8	No	Yes

* Human Genome version 19 was used.

** SeattleSeq Annotation was used to provide functional annotation for the variants.

*** The region contains variants positioned between -1619 and 4482 of the NG_008376.3 RefSeq.

**** PharmVar's translation table for Human Genome version 19 was used.

† The haplotype of the allele containing the R474Q change has been determined and designated *CYP2D6*138.001* by PharmVar.

‡ The haplotype of the allele containing the A449D change has been determined and designated *CYP2D6*4.028* by PharmVar.

§ The haplotype of the allele containing the R365H change has been determined and designated *CYP2D6*139.001* by PharmVar.