

Table S2

Homologous coding missense variants identified by deep sequencing of chromosomes 2 and der(2)

\*Allele frequencies

variant no	GeneName	rsid	chr	start	end	REF	ALT	genotype	freq	depth	1000G*	NHLBI6500*	GERP++	EXONIC info	Impact	SIFT	PP2
1	TAF1B	rs396190	chr2	10043024	10043024	G	A	hom	4.3	43	0.95	0.930801	3.27	nonsynonymous	TAF1B.NM_005890.e	0.38	0.970
2	RAD51AP2	rs7695414	chr2	17695773	17695773	G	A	hom	179	45	0.58	0.511569	2.29	nonsynonymous	RAD51AP2.NM_001144	0.53	0
3	GEN1	rs1812152	chr2	17942775	17942775	T	A	hom	177	47	0.79	0.671229	n	nonsynonymous	GEN1.NM_0013000	0.47	0
4	GEN1	rs300175	chr2	17964027	17964027	G	A	hom	32	45	0.99	0.963546	n	nonsynonymous	GEN1.NM_0013000	0.92	0
5	GEN1	rs300169	chr2	17962518	17962518	C	T	hom	153	44	0.91	0.617414	n	nonsynonymous	GEN1.NM_0013000	0.92	0
6	MATN3	rs28939676	chr2	20202930	20202930	G	A	hom	205	29	0.01	0.011459	5.5	nonsynonymous	MATN3.NM_002381.x	0	n
7	POB	rs584542	chr2	21232803	21232803	T	C	hom	209	43	0.97	0.981624	n	nonsynonymous	POB.NM_000384.e	0.77	0.009
8	POB	rs584133	chr2	21234575	21234575	T	C	hom	157	38	1	0.999692	n	nonsynonymous	POB.NM_000384.e	0.85	0
9	GPR113	rs7698929	chr2	26534901	26534901	G	A	hom	341	22	0.07	0.084915	5.18	nonsynonymous	GPR113.NM_001145	0.04	0.065
10	GPR113	rs2052937	chr2	26536694	26536694	C	T	hom	194	26	0.71	0.689835	3.47	nonsynonymous	GPR113.NM_001145	0.42	0
11	ABHD1	rs6715286	chr2	27361948	27361948	T	G	hom	118	26	0.99	0.983469	n	nonsynonymous	ABHD1.NM_032604.x	1	0
12	FAM179A	rs1127202	chr2	29240080	29240080	A	G	hom	23	25	0.21	0.233414	n	nonsynonymous	FAM179A.NM_19928	1	n
13	ALK	rs1881420	chr2	29416481	29416481	T	C	hom	529	21	0.2	0.223538	n	nonsynonymous	ALK.NM_004304.exo	0.31	0.123
14	ALK	rs1670283	chr2	29416572	29416572	T	C	hom	218	29	0.99	0.990312	n	nonsynonymous	ALK.NM_004304.exo	1	0
15	BIRC6	rs60197015	chr2	32687162	32687162	G	C	hom	163	43	1	n	5.64	nonsynonymous	BIRC6.NM_016252.e	1	n
16	BIRC6	rs2398984	chr2	32711708	32711708	A	T	hom	95	41	0.58	0.658968	4.48	nonsynonymous	BIRC6.NM_016252.e	0.97	n
17	LTPB1	rs4422143	chr2	33585796	33585796	T	C	hom	136	37	1	0.999923	4.89	nonsynonymous	LTPB1.NM_00116622	1	0
18	SULT1B1	rs1025833	chr2	37406880	37406880	C	G	hom	543	37	0.41	0.249731	n	nonsynonymous	SULT1B1.NM_0011003	1	0
19	CEBP2	rs2098368	chr2	37456032	37456032	C	T	hom	39	40	0.93	0.879499	n	nonsynonymous	CEBP2.NM_005760.x	1	0.127063
20	RMDN2	rs4670800	chr2	38179134	38179134	G	A	hom	59.5	51	1	n	1.879438	nonsynonymous	RMDN2.NM_144713	0.77	0
21	CYP11B1	rs1800440	chr2	38208139	38208139	T	C	hom	36	39	0.1	0.134998	5.95	nonsynonymous	CYP11B1.NM_000104	0.01	0.903
22	CYP11B1	rs1058396	chr2	38298203	38298203	C	G	hom	157	39	0.61	0.445564	5.95	nonsynonymous	CYP11B1.NM_000104	0.34	0
23	DHX57	rs7698922	chr2	39363244	39363244	T	C	hom	87	49	0.6	0.635907	n	nonsynonymous	DHX57.NM_196963.x	0.49	0
24	EML4	rs1020624	chr2	42515388	42515388	C	G	hom	140	46	0.85	0.917333	5.67	nonsynonymous	EML4.NM_00114507	0.85	0
25	HAAO	rs3816183	chr2	43015719	43015719	T	C	hom	222	22	0.62	0.793634	3.66	nonsynonymous	HAAO.NM_012205.e	1	0.067406
26	ABO3B	rs5844718	chr2	44104925	44104925	T	C	hom	58.3	35	0.89	0.837229	n	nonsynonymous	ABO3B.NM_022437.x	1	0
27	FSHR	rs6165	chr2	49191041	49191041	C	T	hom	20	42	0.51	0.454636	n	nonsynonymous	FSHR.NM_181446.e	0.42	0.07985
28	PSME4	rs805408	chr2	54120025	54120025	A	T	hom	38	45	0.3	0.271106	5.41	nonsynonymous	PSME4.NM_014614.x	0.75	0.709923
29	CD207F3	rs9257586	chr2	54587586	54587586	G	A	hom	4.81	39	0.33	0.761328	3.18	nonsynonymous	CD207F3.NM_0011002	1	0
30	CLCH1	rs14026	chr2	55407734	55407734	G	A	hom	1.5	1.5	0.51	0.511304	n	nonsynonymous	CLCH1.NM_001193	1	n
31	CLCH1	rs6716006	chr2	55407754	55407754	C	T	hom	172	44	0.97	0.982854	3.59	nonsynonymous	CLCH1.NM_0011355	1	0.438892
32	MTF2	rs11337	chr2	55470115	55470115	C	T	hom	153	35	0.68	0.690278	2.14	nonsynonymous	MTF2.NM_0010053e	1	0
33	SMK2	rs2043704	chr2	55854540	55854540	G	A	hom	189	33	0.9	0.919172	n	nonsynonymous	SMK2.NM_0011229	1	0
34	PNPT1	rs762572	chr2	55912120	55912120	T	C	hom	21.8	29	0.38	0.446871	n	nonsynonymous	PNPT1.NM_033109e	0.19	0
35	KIAA1841	n	chr2	61381327	61381327	G	C	hom	214	49	n	n	n	frameshift deletion	KIAA1841.NM_032520	1	0
36	CD207F3	rs1729674	chr2	61389737	61389737	T	C	hom	15	21	0.43	0.431137	n	nonsynonymous	CD207F3.NM_0011437	0	n
37	USP34	rs6722430	chr2	61575308	61575308	A	G	hom	179	35	0.99	0.985651	3.83	nonsynonymous	USP34.NM_014709e	0	n
38	ETAA1	rs3770657	chr2	67630980	67630980	G	A	hom	49	45	0.72	0.761538	3.4	nonsynonymous	ETAA1.NM_019002.e	1	0
39	ETAA1	rs3770655	chr2	67632125	67632125	C	G	hom	54.3	47	0.73	0.775958	n	nonsynonymous	ETAA1.NM_019002.e	0.82	0
40	FILG1	rs7619234	chr2	70112724	70112724	C	T	hom	127	21	0.97	0.992455	5.24	nonsynonymous	FILG1.NM_00100431	1	0
41	CD207	rs714326	chr2	71058835	71058835	A	G	hom	163	27	0.54	0.437219	n	nonsynonymous	CD207.NM_015717.e	0.75	0.2
42	ALMS1	rs2037814	chr2	73675669	73675669	T	G	hom	50	32	0.86	0.880566	n	nonsynonymous	ALMS1.NM_015120.x	0.14	0
43	ALMS1	rs2037814	chr2	73675669	73675669	T	G	hom	17	35	0.86	0.880566	n	nonsynonymous	ALMS1.NM_015120.x	1	0.001
44	NAT8	rs4852974	chr2	73927931	73927931	A	G	hom	26.8	32	0.74	0.749227	3.18	unknown	UNKNOWN	n	n
45	NAT8	rs2010490	chr2	73928008	73928008	C	G	hom	31	29	0.58	0.561126	2.4	unknown	UNKNOWN	n	n
46	POLR1A	rs4832242	chr2	86317037	86317037	G	C	hom	37	44	0.76	0.787664	n	nonsynonymous	POLR1A.NM_015425	0.62	0.001
47	IMMT	rs6408024	chr2	86403924	86403924	T	C	hom	39.8	23	0.98	0.990025	5.13	nonsynonymous	IMMT.NM_00110018	0.43	0
48	KDM3A	rs2030259	chr2	86683642	86683642	A	G	hom	138	27	0.85	0.801784	3.51	nonsynonymous	KDM3A.NM_0011466	0.95	0
49	ASTL	rs794958	chr2	96795857	96795857	T	C	hom	64.3	24	0.89	0.860531	n	nonsynonymous	ASTL.NM_00102030	1	0
50	ANKRD36	rs20049616	chr2	97623853	97623853	G	A	hom	18	21	0.9	0.91972	n	nonsynonymous	ANKRD36.NM_001161	1	0
51	AF3	rs4851223	chr2	10034357	10034357	C	T	hom	44	53	1	n	5.26	nonsynonymous	AF3.NM_00102510	1	0
52	RF38	rs4851436	chr2	10203849	10203849	T	A	hom	218	46	0.89	0.947657	4.74	nonsynonymous	RF38.NM_00114569	0.29	n
53	TMEM182	rs987987	chr2	103431404	103431404	T	C	hom	222	38	0.99	0.975012	2.93	nonsynonymous	TMEM182.NM_14446	1	0
54	CC22	rs9718928	chr2	103909572	103909572	T	C	hom	15	1	0.99	0.990025	5.13	nonsynonymous	CC22.NM_181453	0.66	0.071
55	CKAP2L	rs6713825	chr2	113513825	113513825	T	C	hom	210	34	0.97	0.940181	n	nonsynonymous	CKAP2L.NM_152515	0.36	0.071
56	IL36A	rs895497	chr2	113763575	113763575	A	G	hom	43.5	43	0.82	0.725041	n	nonsynonymous	IL36A.NM_014440.ex	0.36	0
57	DPF1B	rs1429265	chr2	115252960	115252960	G	A	hom	127	21	0.97	0.992455	5.25	nonsynonymous	DPF1B.NM_020865	1	0
58	CD207F6	rs1052500	chr2	120060082	120060082	T	C	hom	40.5	55	0.66	0.724126	n	nonsynonymous	CD207F6.NM_001017	0.6	0
59	PCDP1	rs2273058	chr2	120388412	120388412	G	A	hom	55.3	47	0.98	0.977241	n	nonsynonymous	PCDP1.NM_0012170	0.77	0.002
60	TMEM177	rs1863496	chr2	120439220	120439220	C	G	hom	27	22	0.78	0.771106	n	nonsynonymous	TMEM177.NM_00110	0.81	0
61	TUSK2	rs3849411	chr2	120949411	120949411	G	A	hom	76.3	24	0.9	0.947769	2.64	nonsynonymous	TUSK2.NM_002712	0	n
62	POTE1	rs489872	chr2	131212170	131212170	C	T	hom	29.8	22	n	n	n	nonsynonymous	POTE1.NM_0012744	0	n
63	RSHD1	rs961360	chr2	136393658	136393658	A	G	hom	22.8	58	0.31	0.183377	5.58	nonsynonymous	RSHD1.NM_015136	0.36	0.578
64	LCT	rs3675479	chr2	13657554	13657554	C	T	hom	25.8	23	0.98	0.979395	5.53	nonsynonymous	LCT.NM_002299.exo	1	0
65	LCT	rs3754689	chr2	136590746	136590746	A	T	hom	160	29	0.34	0.27026	6.21	nonsynonymous	LCT.NM_002299.exo	1	0.024
66	NEB	rs6															