

Supplemental Table 3. A list of variants that are present in group 1 and absent from groups 2+3.

Gene	Location	Type	Substitution	dbSNP	II-2	III-5	III-7
<Non-synonymous variants>							
RETNLB	3q13.13	nonsynonymous SNV	NM_032579:c.C59T;p.P20L	rs11708527	Hetero	Hetero	Hetero
BOC	3q13.2	nonsynonymous SNV	NM_033254:c.C664T;p.R222C		Hetero	Hetero	Hetero
WDR52	3q13.2	nonsynonymous SNV	NM_001164496:c.A3861C;p.K1287N	rs6768642	Hetero	Hetero	Hetero
WDR52	3q13.2	nonsynonymous SNV	NM_001164496:c.C2234T;p.P745L	rs58988763	Hetero	Hetero	Hetero
WDR52	3q13.2	nonsynonymous SNV	NM_001164496:c.G852T;p.K284N	rs16845107	Hetero	Hetero	Hetero
SPICE1	3q13.2	nonsynonymous SNV	NM_144718:c.C823G;p.L275V	rs16861032	Hetero	Hetero	Hetero
POLQ	3q13.33	nonsynonymous SNV	NM_199420:c.A3602G;p.H1201R	rs3218651	Hetero	Hetero	Hetero
POLQ	3q13.33	nonsynonymous SNV	NM_199420:c.A2884G;p.S962G	rs3218636	Hetero	Hetero	Hetero
IQCB1	3q13.33	nonsynonymous SNV	NM_001023571:c.G902A;p.C301Y	rs17849995	Hetero	Hetero	Hetero
PARP15	3q21.1	nonsynonymous SNV	NM_001113523:c.G124A;p.V42M	rs1875272	Hetero	Hetero	Hetero
SEMA5B	3q21.1	nonsynonymous SNV	NM_001031702:c.C1148G;p.A383G	rs78693594	Hetero	Hetero	Hetero
TNK2	3q29	nonsynonymous SNV	NM_005781:c.G2630A;p.R877H	rs112384084	Hetero	Hetero	Hetero
FYTTD1	3q29	nonsynonymous SNV	NM_001011537:c.C7G;p.P3A	rs7624719	Homo	Hetero	Hetero
LRCH3	3q29	nonsynonymous SNV	NM_032773:c.C1565T;p.P522L	rs36078463	Hetero	Hetero	Hetero
IQCG	3q29	nonsynonymous SNV	NM_001134435:c.G529C;p.D177H	rs67877771	Hetero	Hetero	Hetero
IQCG	3q29	nonsynonymous SNV	NM_001134435:c.C335A;p.A112D	rs9880989	Hetero	Hetero	Hetero
CASP8AP2	6q15	nonsynonymous SNV	NM_001137667:c.C4975T;p.P1659S	rs3799896	Hetero	Hetero	Hetero
KLHL32	6q16.1	nonsynonymous SNV	NM_052904:c.A386G;p.N129S	rs2294763	Hetero	Hetero	Hetero
PRDM1	6q21	nonsynonymous SNV	NM_001198:c.G220A;p.G74S	rs2185379	Hetero	Hetero	Hetero
ARMC2	6q21	nonsynonymous SNV	NM_032131:c.T497C;p.M166T	rs9386758	Hetero	Hetero	Hetero
AKD1	6q21	nonsynonymous SNV	NM_001145128:c.C2443T;p.P815S	rs10499052	Hetero	Hetero	Hetero
MARCKS	6q21	nonsynonymous SNV	NM_002356:c.C821T;p.A274V	rs3734458	Hetero	Hetero	Hetero
C6orf170	6q22.31	nonsynonymous SNV	NM_152730:c.A1795G;p.I599V	rs7745023	Hetero	Hetero	Hetero
NCOA7	6q22.32	nonsynonymous SNV	NM_001199622:c.T657G;p.D219E	rs1567	Hetero	Hetero	Hetero
LAMA2	6q22.33	nonsynonymous SNV	NM_000426:c.G1856A;p.R619H	rs3816665	Hetero	Hetero	Hetero
TAAR2	6q23.2	stopgain SNV	NM_014626:c.G368A;p.W123X	rs8192646	Hetero	Hetero	Hetero
VNN1	6q23.2	nonsynonymous SNV	NM_004666:c.G436A;p.D146N	rs45624336	Hetero	Hetero	Hetero
AHI1	6q23.3	nonsynonymous SNV	NM_001134830:c.A3257G;p.E1086G	rs148000791	Hetero	Hetero	Hetero
MAP3K5	6q23.3	nonsynonymous SNV	NM_005923:c.A3748G;p.I1250V	rs35551087	Hetero	Hetero	Hetero
CCDC28A	6q24.1	nonsynonymous SNV	NM_015439:c.C245T;p.P82L	rs2273510	Hetero	Hetero	Hetero
PHACTR2	6q24.2	nonsynonymous SNV	NM_001100165:c.G862A;p.V288I	rs186432598	Hetero	Hetero	Hetero
LRRTM3	10q21.3	nonsynonymous SNV	NM_178011:c.T1589G;p.I530R	rs75363119	Hetero	Hetero	Hetero
NEUROG3	10q22.1	nonsynonymous SNV	NM_020999:c.T596C;p.F199S	rs4536103	Hetero	Hetero	Hetero
ADAMTS14	10q22.1	nonsynonymous SNV	NM_080722:c.C2809A;p.L937M	rs12774070	Hetero	Hetero	Hetero
ADAMTS14	10q22.1	nonsynonymous SNV	NM_080722:c.C3322T;p.P1108S	rs61573157	Hetero	Hetero	Hetero
EP400	12q24.33	nonsynonymous SNV	NM_015409:c.C2494G;p.R832G		Hetero	Hetero	Hetero
ZNF268	12q24.33	nonsynonymous SNV	NM_001165887:c.A220T;p.M74L	rs61960670	Hetero	Hetero	Hetero
ZNF268	12q24.33	nonsynonymous SNV	NM_001165881:c.G2579C;p.R860T	rs62644541	Hetero	Hetero	Hetero
LOC647589	12q24.33	nonsynonymous SNV	NM_001191054:c.C689G;p.S230C	rs36146434	Hetero	Hetero	Hetero
CNTNAP4	16q23.1	nonsynonymous SNV	NM_138994:c.T3246G;p.D1082E	rs7202925	Hetero	Hetero	Hetero
WWOX	16q23.1	nonsynonymous SNV	NM_130791:c.A544G;p.K182E	rs77067228	Hetero	Hetero	Hetero
WWOX	16q23.1	nonsynonymous SNV	NM_016373:c.G535A;p.A179T	rs12918952	Hetero	Hetero	Hetero
GP1BA	17p13.2	frameshift deletion	NM_000173:c.I285delG;p.E429fs		Homo	Homo	Homo
CCDC165	18p11.22	nonsynonymous SNV	NM_015210:c.A1613G;p.D538G	rs3744979	Hetero	Hetero	Hetero
PIEZO2	18p11.22	nonsynonymous SNV	NM_022068:c.G3443T;p.C1148F	rs35033671	Hetero	Hetero	Hetero
CEP76	18p11.21	nonsynonymous SNV	NM_024899:c.G1327T;p.V443F		Hetero	Hetero	Hetero
LAMA3	18q11.2	nonsynonymous SNV	NM_001127718:c.C3450A;p.N1150K	rs1154232	Hetero	Hetero	Hetero
COL5A3	19p13.2	nonsynonymous SNV	NM_015719:c.G4462C;p.A1488P	rs3745584	Hetero	Hetero	Hetero
NWD1	19p13.11	nonsynonymous SNV	NM_001007525:c.C163T;p.R55W	rs139873139	Hetero	Hetero	Hetero
NCAN	19p13.11	nonsynonymous SNV	NM_004386:c.G2513A;p.S838N	rs10426537	Hetero	Hetero	Hetero
UR1I	19q12	nonsynonymous SNV	NM_003796:c.C82G;p.P28A		Hetero	Hetero	Hetero
OGFR	20q13.33	nonsynonymous SNV	NM_007346:c.G1670C;p.S557T	rs78981100	Hetero	Hetero	Hetero
FAM116B	22q13.33	nonsynonymous SNV	NM_001001794:c.G1207A;p.V403M	rs68178377	Hetero	Hetero	Hetero
SPANXD,SPANXE	Xq27.2	nonsynonymous SNV	NM_032417:c.G202C;p.V68L	rs5953618	Hetero	Hetero	Hetero
<Synonymous variants>							
ARMC9	2q37.1	synonymous SNV	NM_025139:c.C1833T;p.L611L	rs1669083	Hetero	Hetero	Hetero
ARMC9	2q37.1	synonymous SNV	NM_025139:c.G1866A;p.T622T	rs1669084	Hetero	Hetero	Hetero
DGKD	2q37.1	synonymous SNV	NM_003648:c.C1800T;p.F600F	rs146366997	Hetero	Hetero	Hetero
HJURP	2q37.1	synonymous SNV	NM_018410:c.C2157T;p.P719P	rs3771334	Hetero	Hetero	Hetero
TRPM8	2q37.1	synonymous SNV	NM_024080:c.A747T;p.P249P	rs28901637	Hetero	Hetero	Hetero
MORC1	3q13.13	synonymous SNV	NM_014429:c.G2340C;p.P780P	rs75923723	Hetero	Hetero	Hetero
MORC1	3q13.13	synonymous SNV	NM_014429:c.G2337C;p.V779V	rs76539963	Hetero	Hetero	Hetero
BOC	3q13.2	synonymous SNV	NM_033254:c.C903T;p.A301A	rs3814399	Hetero	Hetero	Hetero
BOC	3q13.2	synonymous SNV	NM_033254:c.C2985T;p.D995D	rs34600669	Hetero	Hetero	Hetero
WDR52	3q13.2	synonymous SNV	NM_001164496:c.A4176G;p.L1392L	rs7650771	Hetero	Hetero	Hetero
WDR52	3q13.2	synonymous SNV	NM_001164496:c.C156T;p.T52T	rs58474121	Hetero	Hetero	Hetero
SPICE1	3q13.2	synonymous SNV	NM_144718:c.G2301A;p.Q767Q	rs11920725	Hetero	Hetero	Hetero
SIDT1	3q13.2	synonymous SNV	NM_017699:c.C174T;p.L58L	rs3732799	Hetero	Hetero	Hetero

SIDT1	3q13.2	synonymous SNV	NM_017699:c.G1368T:p.A456A		Hetero	Hetero	Hetero
TNK2	3q29	synonymous SNV	NM_005781:c.C1719T:p.F573F	rs1056726	Hetero	Hetero	Hetero
LRRC33	3q29	synonymous SNV	NM_198565:c.G1893A:p.T631T	rs116984491	Hetero	Hetero	Hetero
BACH2	6q15	synonymous SNV	NM_001170794:c.C1071T:p.P357P	rs2236182	Hetero	Hetero	Hetero
ARMC2	6q21	synonymous SNV	NM_032131:c.T1314C:p.G438G	rs11153136	Hetero	Hetero	Hetero
VGLL2	6q22.1	synonymous SNV	NM_182645:c.T441C:p.N147N	rs13194610	Hetero	Hetero	Hetero
ROS1	6q22.1	synonymous SNV	NM_002944:c.A6247C:p.R2083R	rs552183	Hetero	Hetero	Hetero
GRM1	6q24.3	synonymous SNV	NM_000838:c.T3213G:p.P1071P	rs1047006	Hetero	Hetero	Hetero
PBLD	10q21.3	synonymous SNV	NM_001033083:c.G516A:p.S172S		Hetero	Hetero	Hetero
AIFM2	10q22.1	synonymous SNV	NM_001198696:c.C729T:p.T243T	rs118135233	Hetero	Hetero	Hetero
NPFPR1	10q22.1	synonymous SNV	NM_022146:c.C201T:p.I67I	rs3812696	Hetero	Hetero	Hetero
ADAMTS14	10q22.1	synonymous SNV	NM_080722:c.C2856T:p.C952C	rs2587475	Hetero	Hetero	Hetero
SGPL1	10q22.1	synonymous SNV	NM_003901:c.A1305G:p.E435E	rs114461640	Hetero	Hetero	Hetero
ZC3H12C	11q22.3	synonymous SNV	NM_033390:c.A2445G:p.Q815Q	rs74548991	Hetero	Hetero	Hetero
GOLGA3	12q24.33	synonymous SNV	NM_001172557:c.A414G:p.T138T	rs7134809	Hetero	Hetero	Hetero
ZNF140	12q24.33	synonymous SNV	NM_003440:c.A792G:p.Q264Q	rs710945	Hetero	Hetero	Hetero
LOC647589	12q24.33	synonymous SNV	NM_001191054:c.G351A:p.P117P	rs147259575	Hetero	Hetero	Hetero
LOC647589	12q24.33	synonymous SNV	NM_001191054:c.C240G:p.R80R	rs141475940	Hetero	Hetero	Hetero
HNRNPA1L2	13q14.3	synonymous SNV	NM_001011725:c.C408A:p.I136I	rs78145640	Homo	Hetero	Hetero
CNTNAP4	16q23.1	synonymous SNV	NM_138994:c.T3375C:p.T1125T	rs7184958	Hetero	Hetero	Hetero
KRTAP4-8	17q21.2	synonymous SNV	NM_031960:c.C522T:p.C174C	rs77163010	Homo	Hetero	Hetero
LAMA3	18q11.2	synonymous SNV	NM_001127718:c.C1152G:p.A384A	rs1154226	Hetero	Hetero	Hetero
LAMA3	18q11.2	synonymous SNV	NM_001127718:c.C3736T:p.L1246L	rs1131521	Hetero	Hetero	Hetero
TMEM38A	19p13.11	synonymous SNV	NM_024074:c.C189T:p.F63F	rs730120	Hetero	Hetero	Hetero
MAST3	19p13.11	synonymous SNV	NM_015016:c.C648T:p.H216H		Hetero	Hetero	Hetero
KIAA1683	19p13.11	synonymous SNV	NM_001145304:c.C2787T:p.P929P	rs12608744	Hetero	Hetero	Hetero
KIAA1683	19p13.11	synonymous SNV	NM_001145304:c.G2691A:p.L897L	rs12608843	Hetero	Hetero	Hetero
NCAN	19p13.11	synonymous SNV	NM_004386:c.G2856A:p.P952P	rs10406347	Hetero	Hetero	Hetero
VSTM2B	19q12	synonymous SNV	NM_001146339:c.G666C:p.S222S	rs115230064	Hetero	Hetero	Hetero
ALG12	22q13.33	synonymous SNV	NM_024105:c.A885G:p.A295A	rs8135963	Hetero	Hetero	Hetero
CRELD2	22q13.33	synonymous SNV	NM_001135101:c.T273C:p.N91N	rs9616388	Hetero	Hetero	Hetero
CRELD2	22q13.33	synonymous SNV	NM_001135101:c.C600T:p.T200T	rs7410276	Hetero	Hetero	Hetero
CRELD2	22q13.33	synonymous SNV	NM_024324:c.C1050T:p.R350R	rs1608	Hetero	Hetero	Hetero
FAM116B	22q13.33	synonymous SNV	NM_001001794:c.C1356G:p.P452P	rs67248662	Hetero	Hetero	Hetero
KLHDC7B	22q13.33	synonymous SNV	NM_138433:c.C309A:p.G103G		Hetero	Hetero	Hetero

