

Table S4. Complete List of 710 variants in 399 genes from 59 Trios with AVSD Displaying Rare Inheritance

Proband	Gene	Inheritance	Residual Variation Intoleranc e Score	Exonic Function	Amino Acid Change	Minor Allele Frequency EXAC	dbSNP138	PolyPhen2 Score	PolyPhen2 Prediction	GERP Score	Chromosome	Position	Ref	Alt	Quality Score	Paternal Genotype	Maternal Genotype	Proband Genotype
17	MARCH7	compound heterozygous	86.299835	nonsynonymous SNV	uc010for.3.c.C599T.p.T200M	0.02	rs76774368	0	B	1.22	chr2	160604514	C	T	1935.4	0/0	0/1	0/1
17	MARCH7	compound heterozygous	86.299835	nonsynonymous SNV	uc010for.3.c.G853A.p.A285T	0.02	rs141659304	0	A	-3.38	chr2	160604768	C	T	710.2	0/1	0/1	0/1
2036	ABCA3	compound heterozygous	3.7096013	nonsynonymous SNV	uc010bpb.1.c.A3610G.p.S1204G	8.54E-04	rs35089233	0.836	P	4.89	chr16	2334358	T	C	1064.9	0/0	0/1	0/1
2036	ABCA3	compound heterozygous	3.7096013	nonsynonymous SNV	uc002ypl.1.c.G863A.p.R288Q	5.96E-03	rs117603931	0	B	-8.84	chr16	2369592	C	T	622.6	0/1	0/1	0/1
60	ABCA4	compound heterozygous	86.305732	nonsynonymous SNV	uc010dqb.1.c.G505C.p.V169I	2.73E-03	rs41292677	0.953	D	5.08	chr1	94467548	C	G	588	0/1	0/1	0/1
60	ABCA4	compound heterozygous	86.305732	nonsynonymous SNV	uc010dqb.3.c.A3845G.p.Y1262C	2.62E-03	rs145625174	0.92	D	3.63	chr1	94526174	C	G	499	0/1	0/1	0/1
2812	ABCA4	compound heterozygous	86.305732	nonsynonymous SNV	uc001dqb.3.c.G6529A.p.D2177N	0.01	rs18005529	0.004	B	5.03	chr1	94463617	C	T	483.2	0/0	0/1	0/1
313	ABHD14A.ACY1	homozygous rare	NA	nonsynonymous SNV	uc001dqb.3.c.A466G.p.I156V	1.30E-03	rs62646863	0	B	-1.46	chr1	94568675	T	C	2879.7	0/1	0/1	0/1
155	ABO	homozygous rare	NA	nonframeshift substitution	uc011bea.2.c.G182A.p.R61H	8.20E-03	rs41292350				chr3	52012320	G	A	935.1	0/1	0/1	0/1
2020	ACACB	compound heterozygous	1.7751828	nonsynonymous SNV	ABO:uc004oda.1.exon4.c.188_189GC						chr9	136135237	AT	GC	3700.4	0/1	0/1	0/1
2020	ACACB	compound heterozygous	1.7751828	nonsynonymous SNV	uc0010cc.3.c.A2503G.p.M835V	0.02	rs17848802	0.558	P	4.01	chr12	109634834	A	G	1300.1	0/1	0/1	0/1
2020	ACACB	compound heterozygous	1.7751828	nonsynonymous SNV	uc010xsm.2.c.G1984A.p.V662M	5.07E-04	rs138562469	0.995	D	5.16	chr12	109690904	A	G	3649.8	0/1	0/1	0/1
2812	ACACB	compound heterozygous	1.7751828	nonsynonymous SNV	uc0010cc.3.c.A2503G.p.M835V	0.02	rs17848802	0.558	P	4.01	chr12	109634834	A	G	891.8	0/1	0/1	0/1
2812	ACACB	compound heterozygous	1.7751828	nonsynonymous SNV	uc0010cc.3.c.A3845G.p.Y1262C	6.59E-04	rs149793040	0.92	D	4.1	chr12	109661672	A	G	2718.8	0/0	0/1	0/1
3761	ACACB	compound heterozygous	1.7751828	nonsynonymous SNV	uc0010cc.3.c.A2503G.p.M835V	0.02	rs17848802	0.558	P	4.01	chr12	109634834	A	G	763.3	0/1	0/1	0/1
3761	ACACB	compound heterozygous	1.7751828	nonsynonymous SNV	uc010xsm.2.c.C1109T.p.A370V	0.036	rs10980303	0.036	B	4.32	chr12	109680303	C	T	1039.7	0/0	0/1	0/1
313	ACTN2	homozygous rare	2.4652041	frameshift deletion	uc001hyg.2.c.95del.C.P32fs						chr1	236899903	CC	C	728.4	0/1	0/1	0/1
3721	ADAM17	compound heterozygous	82.778957	nonsynonymous SNV	uc010ewz.3.c.T290G.p.L97R	0.003	rs19290520	0.003	B	1.61	chr2	9630520	A	C	2144.5	0/0	0/1	0/1
3721	ADAM17	compound heterozygous	82.778957	nonsynonymous SNV	uc010ewz.3.c.C269T.p.S90L	0.01	rs57596712	0.003	B	1.66	chr2	9630541	G	A	2781.7	0/1	0/1	0/1
2035	ADAM28	compound heterozygous	86.854211	nonsynonymous SNV	uc010lba.3.c.G122A.p.C41Y	2.11E-04	rs145612420	1	D	5.4	chr8	24187586	G	A	1867.6	0/1	0/1	0/1
2035	ADAM28	compound heterozygous	86.854211	nonsynonymous SNV	uc010lba.3.c.T1166C.p.L389P	1.33E-03	rs147942437	0	B	-8.9	chr8	24208750	T	C	2277.9	0/1	0/1	0/1
2037	ADAMTS15	homozygous rare	84.754659	nonsynonymous SNV	uc010hsc.2.c.G597C.p.E133Q	2.70E-03	rs18526910	0.988	D	3.08	chr11	130319265	G	C	976.5	0/1	0/1	0/1
2028	AFAP12	homozygous rare	87.296532	nonsynonymous SNV	uc001lba.3.c.G281A.p.DC1T	0.24E-02	rs45726190	0.026	B	4.87	chr1	6529319	A	G	6293.9	0/1	0/1	0/1
313	AFAP12	homozygous rare	87.296532	nonsynonymous SNV	uc001lbn.3.c.G46A.p.D16N	9.77E-03	rs41300231	0.243	P	4.63	chr10	116100461	C	T	949.1	0/1	0/1	0/1
3731	AFM1	homozygous rare	79.464496	nonsynonymous SNV	uc003hbh.3.c.G1184A.p.R395H	0.02	rs41265665	0.013	B	4.28	chr4	74361142	A	G	5345.4	0/1	0/1	0/1
7952	AFP	compound heterozygous	71.567587	nonsynonymous SNV	uc003hgz.1.c.A559C.p.K187Q	0.01	rs37565619	0.977	D	3.81	chr4	74308089	A	C	2227.4	0/1	0/1	0/1
7952	AFP	compound heterozygous	71.567587	nonsynonymous SNV	uc011cbp.1.c.C80G.p.T27S	1.87E-04	rs139295532	0.235	P	5.09	chr4	74310754	C	G	3774.8	0/0	0/1	0/1
3781	AGA	compound heterozygous	86.712668	nonsynonymous SNV	uc003iua.2.c.C313A.p.L105I	9.56E-03	rs76491548	0.282	P	5.58	chr4	178360811	C	T	2118.6	0/1	0/1	0/1
3781	AGA	compound heterozygous	86.712668	nonsynonymous SNV	uc003iua.2.c.G34T.p.V12L	0.01	rs74626221	0	B	-9.86	chr4	178363496	C	A	383.4	0/1	0/1	0/1
152	AH11	homozygous rare	78.066761	nonsynonymous SNV	uc003agq.3.c.C838T.p.R280W	0.02	rs15312995				chr6	135751024	A	G	2032.4	0/1	0/1	0/1
23	AIF1L	homozygous rare	81.216089	nonsynonymous SNV	uc011fmc.1.c.A388G.p.M130V	0.02	rs11284413	0.705	P	4.19	chr9	133956529	A	G	3259.5	0/1	0/1	0/1
3761	AIF1L	compound heterozygous	81.216089	nonsynonymous SNV	uc011fmc.1.c.T185C.p.T477I	2.16E-03	rs146896674	0.85	P	4.57	chr9	133989793	G	T	2251.9	0/0	0/1	0/1
3761	AIF1L	compound heterozygous	81.216089	nonsynonymous SNV	uc011fmc.1.c.A388G.p.M130V	0.02	rs11284413	0.705	P	4.19	chr9	133956529	A	G	926.1	0/0	0/1	0/1
2037	AKAP4	homozygous rare	84.613116	nonsynonymous SNV	uc004dou.1.c.G860A.p.G072B	5.04E-03	rs141513690	0.072	B	2.46	chrX	49958477	C	T	1123.9	0/1	0/1	0/1
2002	AKAP6	compound heterozygous	71.650153	nonsynonymous SNV	uc001wq.3.c.A1672G.p.N588D	6.82E-03	rs35210906	0.463	P	-6.88	chr14	33015531	A	G	643.1	0/1	0/1	0/1
2002	AKAP6	compound heterozygous	71.650153	nonsynonymous SNV	uc001wq.3.c.G4564A.p.V152I	0.02	rs34711402	0.001	B	2.84	chr14	33291583	A	G	1124.9	0/0	0/1	0/1
152	AKAP9	compound heterozygous	68.447747	nonsynonymous SNV	uc003iud.4.c.C139T.p.H47Y	8.09E-03	rs35669569	0	B	0.0934	chr7	91603115	C	T	1165	0/0	0/1	0/1
152	AKAP9	compound heterozygous	68.447747	nonsynonymous SNV	uc003iud.3.c.G2467C.p.E823Q	1.63E-05	rs41265665	0.884	D	3.25	chr7	91643622	G	C	644.4	0/1	0/1	0/1
2013	AKNA1D	homozygous rare	89.110403	nonsynonymous SNV	uc001dwa.3.c.C311T.p.A104V	0.02	rs17621411	0.068	B	-1.16	chr1	109394976	A	G	3744.6	0/1	0/1	0/1
3761	ALDH1A1	compound heterozygous	67.521821	nonsynonymous SNV	uc001lba.3.c.C1430A.p.T477I	3.91E-03	rs41340361	0.911	D	4.47	chr1	6529319	A	G	792.8	0/1	0/1	0/1
3781	ALDH1A1	compound heterozygous	67.521821	nonsynonymous SNV	uc001lbb.3.c.K126I.p.C421R	1.62E-03	rs149414160	0.959	D	4.53	chr1	19202886	A	G	476	0/0	0/1	0/1
17	ALG8	compound heterozygous	79.464496	nonsynonymous SNV	uc0010yz.1.c.T1316C.p.I439T	0.02	rs17825668	0.002	B	3.06	chr11	77815059	A	G	674.3	0/0	0/1	0/1
17	ALG8	compound heterozygous	79.464496	nonsynonymous SNV	uc0010yz.1.c.G803A.p.R268Q	0.01	rs61995925	0.998	D	5.22	chr11	77823791	T	G	242.7	0/1	0/1	0/1
151	AMPD1	compound heterozygous	59.170795	nonsynonymous SNV	uc001eff.2.c.A188T.p.L627F	1.55E-04	rs200717164	0.997	D	3.32	chr1	115217379	T	A	312.9	0/1	0/1	0/1
151	AMPD1	compound heterozygous	59.170795	nonsynonymous SNV	uc001eff.2.c.A947T.p.K316I	0.02	rs34526199	0.993	D	5.58	chr1	115222237	T	A	526.1	0/1	0/1	0/1
146	AMZ1	compound heterozygous	69.839585	nonsynonymous SNV	uc003smr.1.c.T680C.p.V227A	1.97E-03	rs150799683	0.021	B	-4.8	chr7	2748787	T	C	646.1	0/1	0/1	0/1
146	AMZ1	compound heterozygous	69.839585	nonsynonymous SNV	uc011jwa.1.c.C815A.p.S205R	3.05E-03	rs148177744	0.002	B	-4.11	chr7	2752383	C	T	682.9	0/1	0/1	0/1
2022	ANKRD11	compound heterozygous	60.093619	nonsynonymous SNV	uc002fmc.1.c.C7519A.p.G2057K	4.86E-05	rs41265665	0.98	D	3.94	chr6	86341551	G	C	791.9	0/0	0/1	0/1
2022	ANKRD11	compound heterozygous	60.093619	nonsynonymous SNV	uc002fmc.1.c.C3848T.p.T283M	6.91E-04	rs144673419	0.33	P	5.16	chr16	99348973	G	C	375.2	0/0	0/1	0/1
749	ANKRD12	compound heterozygous	45.364473	nonsynonymous SNV	uc010dxc.1.c.C713G.p.T238S	0.02	rs7243088	0	B	4.5	chr18	9254857	A	G	789.6	0/0	0/1	0/1
749	ANKRD12	compound heterozygous	45.364473	nonframeshift deletion	uc010dxc.1.c.1668_1670del.p.556_557del						chr18	9255811	ATGA	A	928.1	0/1	0/1	0/1
313	ANKRD31	homozygous rare	NA	nonframeshift deletion	uc003kdc.2.c.752_754del.p.251_252del						chr5	74491718	ATCA	A	1475.1	0/1	0/1	0/1
1346	AOC2	homozygous rare	44.102383	nonsynonymous SNV	uc002bit.3.c.G818A.p.R273Q	7.37E-03	rs35508987	0.042	B	-0.175	chr17	40997461	G	A	3650.7	0/1	0/1	0/1
3781	APBA3	compound heterozygous	89.266336	nonsynonymous SNV	uc002ypl.1.c.C236T.p.A79V	6.04E-03	rs11556574	0.005	B	-1.69	chr19	3760027	G	A	731.2	0/1	0/1	0/1
3781	APBA3	compound heterozygous	89.266336	nonsynonymous SNV	uc002ypl.1.c.G48A.p.M16I	2.12E-04	rs200523433	0.04	B	4.5	chr19	3760215	T	C	535.3	0/0	0/1	0/1
2036	APOBEC3H	homozygous rare	85.527247	nonframeshift substitution	uc021wps.1.c.45_53GCGCTAPOBEC3H						chr22	39496328	CAAGCGCCG	CGCGCCT	65.0	0/1	0/1	0/1
123	APOBEC3H	homozygous rare	85.527247	nonframeshift substitution	uc021wps.1.c.45_53GCGCTAPOBEC3H						chr22	39496328	CAAGCGCCG	CGCGCCT	4561.5	0/1	0	

Proband	Gene	Inheritance	Residual Variation Intoleranc e Score	Exonic Function	Amino Acid Change	Minor Allele Frequency EXAC	dbSNP138	PolyPhen2 Score	PolyPhen2 Prediction	GERP Score	Chromosome	Position	Ref	Alt	Quality Score	Paternal Genotype	Maternal Genotype	Proband Genotype
135	C6orf132	compound heterozygous	NA	nonsynonymous SNV	uc0030rw.2:c.22480T>P.R827L						chr8	42071370	G	A	420.8	0/1	0/1	1/0
135	C6orf132	compound heterozygous	NA	nonsynonymous SNV	uc0030rw.2:c.T778G>P.S260A	0.02	rs11758539				chr8	42074872	A	C	486.7	0/1	0/1	0/1
2008	C6orf186	homozygous rare	NA	nonsynonymous SNV	uc0030pb.2:c.A143G>P.H48R	0.02	rs41288584	0.977	D	1.91	chr6	110620177	T	C	334.1	0/1	0/1	1/1
1131	C8orf46	homozygous rare	38.281434	nonsynonymous SNV	uc0030qk.3:c.G235T>P.A79S	0.02	rs61736270	0.015	B	0.125	chr8	67417718	T	C	834	0/1	0/1	1/1
10	C8orf58	compound heterozygous	87.137297	nonsynonymous SNV	uc0030kc.3:c.C661T>P.P221S	7.55E-03	rs147403979	0.479	P	0.876	chr8	22459488	C	T	481.8	0/1	0/1	0/1
10	C8orf58	compound heterozygous	87.137297	nonsynonymous SNV	uc0030kc.3:c.C931T>P.R311W	0.02	rs117289616	0.003	B	0.395	chr8	22460101	T	T	70.14	0/1	0/1	0/1
2026	C9orf131	compound heterozygous	82.944091	nonsynonymous SNV	uc0032vw.3:c.G45A>P.M15I	0.02	rs61742471	0.08	B	-3.07	chr9	35042296	C	A	2025.5	0/1	0/1	0/1
151	C9orf47	homozygous rare	NA	nonsynonymous SNV	uc0032zv.3:c.T155G>P.I518T	0.01	rs34368062	0.065	B	-3.09	chr9	35044398	T	C	1265.4	0/1	0/1	0/1
2010	CACNA1C	homozygous rare	1.5746638	nonsynonymous SNV	uc0034aq.2:c.G44A>P.S144A	0.01	rs34421923	0.012	B	1.52	chr9	91606912	G	A	314.4	0/1	0/1	1/1
60	CACNA2D4	homozygous rare	89.661477	nonsynonymous SNV	uc001nqv.1:c.G168A>P.V55A1						chr12	2714881	G	A	1338.3	0/1	0/1	0/1
4405	CAPN2	homozygous rare	85.828025	nonsynonymous SNV	uc001nqv.1:c.C266T>P.P69L	7.28E-03	rs61741336				chr12	1906632	G	A	1304.6	0/1	0/1	0/1
3765	CAPS2	homozygous rare	67.303609	nonsynonymous SNV	uc001thc.3:c.C34T>P.P12S	6.03E-03	rs114318533				chr1	223946945	C	T	1531.2	0/1	0/1	1/1
23	CCDC159	homozygous rare	53.509082	frameshift substitution	uc001sxi.4:c.G418A>P.D140N	0.0023	rs117753158	0.005	B	0.356	chr12	75687039	C	T	7175.2	0/1	0/1	1/1
3721	CCDC165	compound heterozygous	NA	nonsynonymous SNV	CCDC159:uc010xw.1:exon2:c.66_68AGCA						chr19	11456667	GCG	AGCA	2777.1	0/1	0/1	1/1
3721	CCDC165	compound heterozygous	NA	nonsynonymous SNV	uc002kns.2:c.A5G>P.E2G	8.13E-06		0.995	D	5.47	chr18	8796234	A	C	1579.2	0/1	0/1	0/1
3721	CCDC165	compound heterozygous	NA	nonsynonymous SNV	uc002kns.2:c.C2676G>P.H89ZQ	8.72E-04	rs146312067	0.995	D	-8.4	chr18	8826164	C	G	999.8	0/1	0/1	0/1
151	CCDC169	compound heterozygous	NA	nonsynonymous SNV	uc001vpm.3:c.8872_8874del>P.N3597S						chr13	103392257	C	G	428.7	0/1	0/1	0/1
151	CCDC169	compound heterozygous	NA	nonframeshift deletion	uc001vpm.3:c.8872_8874del>P.N256_2958del						chr13	103394172	T	CTTC	356.6	0/1	0/1	0/1
7952	CCDC48	compound heterozygous	NA	nonsynonymous SNV	uc011bkt.2:c.G1381T>P.A461S	1.22E-04	rs377018804	0.006	B	0.177	chr3	128753104	G	T	1108.5	0/1	0/1	0/1
7952	CCDC48	compound heterozygous	NA	nonsynonymous SNV	uc011bkt.2:c.G1662C>P.E545D	0.01	rs75140996	0.004	B	4.044	chr3	128758556	G	C	413.9	0/1	0/1	0/1
289	CCDC88B	compound heterozygous	71.083982	nonsynonymous SNV	uc001nzz.1:c.G1328A>P.R443Q	8.35E-03	rs143386417	0.695	P	2.15	chr11	64112394	G	A	714.3	0/1	0/1	0/1
289	CCDC88B	compound heterozygous	71.083982	nonsynonymous SNV	uc001oab.1:c.C502T>P.R168C	0.0023	rs143386417	0.999	D	2.82	chr11	64121552	G	C	379.9	0/1	0/1	0/1
3761	CCDC88B	compound heterozygous	71.083982	nonsynonymous SNV	uc001oaa.3:c.C284T>P.T95I	0.01	rs144037797	0.053	B	-7.49	chr11	64117106	C	C	630.1	0/1	0/1	0/1
3761	CCDC88B	compound heterozygous	71.083982	nonsynonymous SNV	uc001oac.3:c.G317C>P.R106P	3.90E-04	rs202018582				chr11	64124205	G	C	315	0/1	0/1	0/1
123	CNCL2	compound heterozygous	53.986892	nonsynonymous SNV	uc001afi.2:c.C212G>P.T71S	0.02	rs17851391	0.016	B	1.39	chr1	1334475	C	C	314.9	0/1	0/1	0/1
2013	CNNT1	compound heterozygous	52.093654	nonsynonymous SNV	uc009kz.2:c.C168G>P.D66E	0.01	rs114112990	0.001	B	-1.82	chr1	1331919	G	T	1484.9	0/1	0/1	0/1
2013	CNNT1	compound heterozygous	52.093654	nonsynonymous SNV	uc009kz.2:c.G86A>P.R287H	5.53E-04	rs61751602	0.991	D	4.82	chr12	49087282	C	T	397.4	0/1	0/1	0/1
123	CNNT1	compound heterozygous	52.093654	nonsynonymous SNV	uc009kz.2:c.A817G>P.T273A	0.02	rs61751603	0.001	B	-1.09	chr12	49087325	T	C	526.1	0/1	0/1	0/1
2013	CD1B	homozygous rare	72.59967	nonsynonymous SNV	uc001fhw.3:c.G409A>P.G137R	0.01	rs35841099	0.001	B	-7.49	chr1	158299840	C	T	2832.5	0/1	0/1	1/1
3731	CD1E	homozygous rare	60.56853	nonsynonymous SNV	uc001ffy.3:c.A305G>P.H102R	5.13E-03	rs2873587	0.005	B	0.583	chr1	158324413	G	A	1620.8	0/1	0/1	1/1
50	CD2BP2	compound heterozygous	58.740269	nonsynonymous SNV	uc002zhr.3:c.C785T>P.T262I	0.01	rs34391305	0.913	D	3.62	chr16	30364712	A	G	1680.7	0/1	0/1	0/1
50	CD2BP2	compound heterozygous	58.740269	nonsynonymous SNV	uc002zhr.3:c.A262T>P.T88S	4.53E-03	rs139111996	0.995	D	4.93	chr16	30365335	T	A	1092	0/1	0/1	0/1
2007	CD300LD	compound heterozygous	72.22812	nonsynonymous SNV	uc002jzk.2:c.A325G>P.T109A	2.28E-04	rs180816540	0.002	B	3.83	chr17	72584704	T	C	3519.3	0/1	0/1	0/1
2007	CD300LD	compound heterozygous	72.22812	nonsynonymous SNV	uc002jzk.2:c.T142A>P.Y429I	6.35E-04	rs142719069	0.897	D	-8.42	chr17	72584967	A	A	2.136	0/1	0/1	0/1
2023	CD7	homozygous rare	81.481481	nonsynonymous SNV	uc002kxi.1:c.G86A>P.R973H	0.02	rs113733263	0.973	D	-3.84	chr17	40273234	T	T	1937.7	0/1	0/1	1/1
14	CDH5	homozygous rare	7.5784383	nonframeshift substitution	CDH5:uc002eom.4:exon10:c.1550_1551CT						chr16	66432423	TC	CT	2902	0/1	0/1	1/1
2001	CDK5RAP2	homozygous rare	1.8895447	nonsynonymous SNV	uc0040ke.3:c.G920A>P.G307E	0.02	rs34523498	0.981	D	-0.892	chr9	123205981	C	T	26859.6	0/1	0/1	1/1
50	CDKL5	homozygous rare	15.864591	nonsynonymous SNV	uc0022bn.1:c.C1303T>P.R435C	1.79E-04	rs61753977	0.999	D	4.0	chrX	18622374	C	T	1862.374	0/1	0/1	1/1
155	CELSR2	compound heterozygous	2.1762208	nonsynonymous SNV	uc001dwx.4:c.A3800G>P.H1267R	7.39E-03	rs138543788	0.998	D	4.4	chr1	109801543	A	G	1520.5	0/1	0/1	0/1
155	CELSR2	compound heterozygous	2.1762208	nonframeshift substitution	CELSR2:uc001dwx.4:exon16:c.6044_6045AA						chr1	109810200	GG	AA	852	0/1	0/1	0/1
2027	CHD6	compound heterozygous	8.9112998	nonsynonymous SNV	uc002kxa.1:c.C7165T>P.R239R	0.01	rs61752057	0.999	D	4.81	chr20	40040870	G	A	942.3	0/1	0/1	0/1
2027	CHD6	compound heterozygous	8.9112998	nonsynonymous SNV	uc002kxa.1:c.G430A>P.R1434K	6.91E-04	rs140327030	0.245	P	4.59	chr20	40053863	C	T	324.63	0/1	0/1	0/1
7952	CHRD	compound heterozygous	15.326324	nonsynonymous SNV	uc011br.2:c.C44C>P.S251I	5.29E-04	rs15204264	0.951	P	3.28	chr3	70254170	T	T	7025.417	0/1	0/1	0/1
7952	CHRD	compound heterozygous	15.326323	nonsynonymous SNV	uc011br.2:c.C1085T>P.T362I	2.36E-04	rs138569844	0.992	D	3.88	chr3	184105726	T	T	479.8	0/1	0/1	0/1
60	CLDN16	homozygous rare	43.571597	frameshift substitution	uc003fsi.3:c.165_166C>CLDN16						chr3	190106073	GG	C	3897.6	0/1	0/1	1/1
3731	CLECA6	homozygous rare	89.956358	nonsynonymous SNV	uc001qum.1:c.G223C>P.A75P	1.30E-03	rs150852294	0.086	B	2.37	chr12	8612294	C	C	4063.9	0/1	0/1	1/1
2023	CLECL1	homozygous rare	83.138712	frameshift insertion	uc001qwi.3:c.153_154insAACTT>L51fs						chr12	9885708	T	TAAGTT	6907.5	0/1	0/1	1/1
2037	CNOT4	homozygous rare	57.313046	nonsynonymous SNV	uc003vss.3:c.C20G>P.A7G	0.02	rs17480616	0.001	B	0.995	chr7	135123060	G	C	6067.2	0/1	0/1	1/1
10	CNR2	homozygous rare	60.474168	nonframeshift substitution	uc021oij.1:c.188_189GG>CNR2						chr1	24201919	TT	CC	975.2	0/1	0/1	1/1
151	CNR2	homozygous rare	60.474168	nonframeshift substitution	uc021oij.1:c.188_189GG>CNR2						chr1	24201919	TT	CC	1185.1	0/1	0/1	1/1
17	COL8	homozygous rare	81.552253	nonframeshift deletion	uc003jfp.4:c.G4_6del>P.C22_22del						chr7	51085923	CTTC	C	1580.9	0/1	0/1	1/1
2007	COL4A5	homozygous rare	6.3163482	nonsynonymous SNV	uc0040eb.1:c.G1039G>P.F347A	1.10E-03	rs104886164	0.745693	NA	1.75	chrX	107846262	CT	A	1113.7	0/1	0/1	0/1
2812	COL5A1	compound heterozygous	0.5248879	nonsynonymous SNV	uc004dfe.3:c.G574A>P.D192N	0.02	rs138579182	0.32044	NA	3.29	chr9	137593099	G	A	1520.3	0/1	0/1	0/1
2812	COL5A1	compound heterozygous	0.5248879	nonsynonymous SNV	uc004dfe.3:c.G4322A>P.G144E	0.02	rs478282	NA	4.49	chr9	137710593	G	A	588.9	0/1	0/1	0/1	
2034	COL6A2	homozygous rare	8.2212786	nonsynonymous SNV	uc0020zh.1:c.C1552T>P.P518S	8.83E-03	rs141166141	0.998	D	4.23	chr21	47542052	C	T	3854.2	0/1	0/1	1/1
2007	COL6A3	compound heterozygous	0.8610521	nonsynonymous SNV	uc0020wv.2:c.G1291A>P.A431T	1.04E-03	rs114596320	0.391154	C	2.32	chr2	238243350	C	T	912.4	0/1	0/1	0/1
2007	COL6A3	compound heterozygous	0.8610521	nonsynonymous SNV	uc002wvr.3:c.T257C>P.V86A	1.27E-03	rs116794756	0.697623	NA	5.15	chr2	238289977	A	C	939.8	0/1	0/1	0/1
2033	COL6A6	homozygous rare	NA	nonsynonymous SNV	uc010hw.1:c.T167C>P.M56T	0.02	rs113396273											

Proband	Gene	Inheritance	Residual Variation Intoleranc e Score	Exonic Function	Amino Acid Change	Minor Allele Frequency EXAC	dbSNP138	PolyPhen2 Score	PolyPhen2 Prediction	GERP Score	Chromosome	Position	Ref	Alt	Quality Score	Paternal Genotype	Maternal Genotype	Proband Genotype
23	DC583205	homozygous rare	NA	nonsynonymous SNV	uc010yph.1.c.C7527.p.P251L	0.02	rs1754918				chr16	89300242	C	T	882.5	0/1	0/1	1/1
2022	DYNC2H1	compound heterozygous	29.564756	nonsynonymous SNV	uc001phn.1.c.G8881A.p.A2961T	1.79E-03	rs19958537				chr11	103090699	G	A	2453.3	0/1	0/1	0/1
2022	DYNC2H1	compound heterozygous	29.564756	nonsynonymous SNV	uc009yx.1.c.G2704C.p.G902R	0.01	rs144717489				chr11	103349922	C	G	3145.1	0/1	0/1	0/1
749	DYNC2H1	compound heterozygous	29.564756	nonsynonymous SNV	uc001phn.1.c.C4728C.p.N1576K	0.01	rs72989738				chr11	103036743	C	C	1108.4	0/1	0/1	0/1
749	DYNC2H1	compound heterozygous	29.564756	nonsynonymous SNV	uc009yx.1.c.G2704C.p.G902R	0.01	rs144717489				chr11	103349922	C	C	2137.8	0/1	0/1	0/1
3781	E2F1	compound heterozygous	72.381458	nonsynonymous SNV	uc002wzu.4.c.G598A.p.G200S	0.02	rs3385772	0	B	-7.32	chr20	32266134	T	C	332.4	0/0	0/1	0/1
3781	E2F1	compound heterozygous	72.381458	nonsynonymous SNV	uc002wzu.4.c.G304A.p.A102T	4.66E-03	rs145741678	0.28	P	3.3	chr20	32268180	C	T	1411.8	0/1	0/1	0/1
8522	EHF1	compound heterozygous	47.222222	nonsynonymous SNV	uc002bcb.3.c.T802C.p.F268L	1.71E-04	rs140051312	0.687	P	4.23	chr2	63091910	T	C	3630.6	0/1	0/1	0/1
8522	EHF1	compound heterozygous	47.222222	nonsynonymous SNV	uc002bcb.3.c.C2749G.p.L517V	9.84E-04	rs150215775	0.006	B	5.57	chr2	63226080	G	A	3235.6	0/1	0/1	0/1
2028	EHMT1	deNovo	3.1316346	nonsynonymous SNV	uc004cbb.1.c.G779A.p.R260Q	0.999	D	5.07	D	5.07	chr3	140637871	A	A	1571.7	0/0	0/1	0/1
313	E1F5A	homozygous rare	49.386648	nonsynonymous SNV	uc010yfs.1.c.G367A.p.V123I	0.02	rs78916808				chr17	7213352	G	A	6532.8	0/1	0/1	0/1
123	ELOVL5	compound heterozygous	59.760557	nonsynonymous SNV	uc003pbs.2.c.G746A.p.C249Y	2.28E-04	rs146646780	0	B	5.61	chr6	53133954	C	T	696.1	0/1	0/1	0/1
123	ELOVL5	compound heterozygous	59.760557	stopgain SNV	uc003pbs.2.c.G736T.p.G246X	0.02	rs41273878				chr6	53133964	C	A	1043.3	0/1	0/1	0/1
8522	ENOSF1	homozygous rare	82.254069	nonsynonymous SNV	uc002kku.4.c.C93A.p.D31E	0.01	rs34724061	0.036	B	0.557	chr18	706570	G	T	2893.8	0/1	0/1	1/1
2023	EPB41L3	homozygous rare	89.325313	nonsynonymous SNV	uc0101dr.2.c.G230A.p.R77Q	4.25E-03	rs117538203	0.996	D	5.57	chr18	5419723	C	T	4532.4	0/1	0/1	1/1
2028	EPG5	compound heterozygous	28.314461	nonsynonymous SNV	uc002bfn.2.c.G1722C.p.E574D	0.02	rs16734402	0.855	D	2.22	chr18	43467728	C	G	2713.8	0/1	0/1	0/1
2028	EPG5	compound heterozygous	28.314461	nonsynonymous SNV	uc002bfn.3.c.A1766G.p.Q589R	0.01	rs61734402	0.855	D	4.83	chr18	43522897	T	A	4263.5	0/1	0/1	0/1
152	ERC1	homozygous rare	10.539042	nonsynonymous SNV	uc010qf.2.c.A1486G.p.S50Q	0.02	rs35037408	0	B	-3.85	chr12	1137217	A	A	1442.1	0/1	0/1	1/1
66	ERCC2	compound heterozygous	1.8872376	nonsynonymous SNV	uc002pbk.2.c.A470C.p.K157T	3.25E-05	0.961	D	3.33	chr19	45888148	T	G	1284.7	0/1	0/1	0/1	
66	ERCC2	compound heterozygous	1.8872376	nonsynonymous SNV	uc002pbk.2.c.T52G.p.F18V	0.01	rs11562095				chr19	45885046	A	C	748.9	0/1	0/1	0/1
2025	FAM56C	homozygous rare	NA	nonsynonymous SNV	uc003dvn.3.c.T1269G.p.H423Q	0.02	rs35589292	0.165	P	-2.72	chr3	101543087	G	G	4893.7	0/1	0/1	1/1
2023	FAM58BP	homozygous rare	NA	nonframeshift deletion	uc009wzi.1.c.13_36del.p.5_12del	0.02	rs141962771				chr1	200182703	G	G	5751.1	0/1	0/1	1/1
2020	FAM70B	compound heterozygous	84.123614	nonsynonymous SNV	uc001vuh.3.c.C347T.p.P116L	2.03E-03	rs114871504	0.918	D	3.29	chr13	114502316	C	C	4281.7	0/1	0/1	0/1
2020	FAM70B	compound heterozygous	84.123614	nonsynonymous SNV	uc001vuh.3.c.G547A.p.V183I	0.02	rs41284482	0.995	D	4.34	chr13	114504663	G	A	226.6	0/1	0/1	0/1
2020	FBLN2	compound heterozygous	6.9868766	nonsynonymous SNV	uc011auz.2.c.G1009A.p.A337T	3.12E-03	rs113899508				chr3	13612786	G	A	423.3	0/1	0/1	0/1
2020	FBLN2	compound heterozygous	6.9868766	nonsynonymous SNV	uc011auz.2.c.C221A.p.V712I	4.65E-04	rs136814144				chr3	136814144	G	A	382.1	0/1	0/1	0/1
2020	FBN1	compound heterozygous	0.6428403	nonsynonymous SNV	uc001zww.2.c.T766T.p.R2554W	1.63E-05	rs369294972	0.995	D	4.48	chr15	48713794	A	A	385.0	0/1	0/1	0/1
2020	FBN1	compound heterozygous	0.6428403	nonsynonymous SNV	uc001zww.2.c.T986C.p.I329T	2.28E-03	rs12324002	0.003	B	5.68	chr15	48818329	A	G	1595.2	0/1	0/1	0/1
2026	FBXL18	homozygous rare	12.485256	nonsynonymous SNV	uc0030sn.4.c.T1656G.p.N552K	0.01	rs33941092	0.047	B	2.46	chr7	5540244	A	G	1056.9	0/1	0/1	0/1
135	FBXL6	compound heterozygous	24.534088	nonsynonymous SNV	uc003bzb.3.c.T338C.p.L113P	5.96E-03	rs72695499	0.989	D	4.21	chr8	145580028	A	G	727.2	0/1	0/1	0/1
135	FBXL6	compound heterozygous	24.534088	nonsynonymous SNV	uc010mfx.3.c.C135C.p.L12P	0.02	rs11762319	0.852	D	4.43	chr8	145580669	A	G	588.8	0/1	0/1	0/1
2026	FBXO38	compound heterozygous	10.503657	nonsynonymous SNV	uc003jpf.1.c.T1002A.p.D334E	0.99	D	2.93	chr5	147790237	T	A	4208.4	0/1	0/1	0/1		
2026	FBXO38	compound heterozygous	10.503657	nonsynonymous SNV	uc003jpf.1.c.C2341T.p.P781S	1.06E-03	rs116266000	0.001	B	0.934	chr5	147807198	C	T	628.7	0/1	0/1	0/1
2023	FBXO40	compound heterozygous	12.57372	nonsynonymous SNV	uc003seg.2.c.G307A.p.P23K	3.42E-04	rs12144020	0.854	D	4.96	chr3	12144020	G	A	2562.7	0/1	0/1	0/1
2023	FBXO40	compound heterozygous	12.57372	nonsynonymous SNV	uc003seg.2.c.G1540A.p.S14T	1.71E-04	rs139583536	0.88	D	4.6	chr3	12341816	A	G	1266.9	0/1	0/1	0/1
3761	FCHSD1	compound heterozygous	68.536211	nonsynonymous SNV	uc010jgg.3.c.A1031C.p.D344A	0.02	rs116772194				chr5	141021294	T	G	1584.1	0/1	0/1	0/1
3761	FCHSD1	compound heterozygous	68.536211	nonsynonymous SNV	uc003llk.3.c.C1032A.p.N344K	0.02	rs3749760				chr5	141026182	G	T	865.0	0/1	0/1	0/1
3761	FCN2	deNovo	82.295353	nonsynonymous SNV	uc004cfh.1.c.G666A.p.M222I	0.02	rs13779099	0.006	B	-3.05	chr9	137779099	G	A	547.6	0/1	0/1	0/1
7952	FGFBP3	compound heterozygous	NA	nonsynonymous SNV	uc021pwg.1.c.G676C.p.D226H	0.01	rs80269321	0.871	D	0.648	chr10	93688051	C	C	754.4	0/1	0/1	0/1
7952	FGFBP3	compound heterozygous	NA	nonsynonymous SNV	uc021pwg.1.c.A587G.p.K196R	0.01	rs11186740	0.024	B	1.25	chr10	93688140	T	G	1746.1	0/1	0/1	0/1
1346	FMN1	homozygous rare	88.635291	nonsynonymous SNV	uc001zmf.4.c.C2048T.p.P683L	0.02	rs117804335				chr15	32611185	A	A	1691.5	0/1	0/1	1/1
3737	FMN1	deNovo	88.635291	nonsynonymous SNV	uc001zmf.4.c.C1439T.p.A380L	0.02	rs1148251675				chr15	33357211	G	A	2091.7	0/1	0/1	1/1
45	FNIP2	compound heterozygous	45.647558	nonsynonymous SNV	uc0030iq.3.c.T891C.p.S301I	0.02	rs19790252				chr4	15978941	G	A	979.2	0/1	0/1	0/1
120	FNIP2	compound heterozygous	45.647558	nonsynonymous SNV	uc0030iq.4.c.C1653A.p.S551R	0.02	rs62001914				chr4	159789411	A	A	1073.8	0/1	0/1	0/1
120	FNIP2	compound heterozygous	45.647558	nonsynonymous SNV	uc0030iq.4.c.G1426T.p.A476S	0.01	rs62001915				chr4	159782889	G	A	544.3	0/1	0/1	0/1
120	FNIP2	compound heterozygous	45.647558	nonsynonymous SNV	uc0030iq.4.c.C1653A.p.S551R	0.02	rs62001914				chr4	159789411	C	A	421.6	0/1	0/1	0/1
14	FREM2	homozygous rare	16.772824	nonframeshift substitution	uc001uww.3.c.T16_117GC:FREM2						chr13	39424253	CT	GC	1177.2	0/1	0/1	0/1
28	FSCN2	compound heterozygous	NA	nonsynonymous SNV	uc010wuo.2.c.G829T.p.V277F		rs181420326				chr17	79502080	G	T	497.7	0/1	0/1	0/1
28	FSCN2	compound heterozygous	NA	nonsynonymous SNV	uc010wuo.2.c.G967A.p.A323T	0.01	rs18636879				chr17	79502218	G	A	977.1	0/1	0/1	0/1
2035	G2E3	homozygous rare	75.294881	nonsynonymous SNV	uc001wql.1.c.G442A.p.A148T	5.73E-03	rs45475495	0.002	B	3.13	chr14	31085525	A	A	3273.8	0/1	0/1	1/1
2033	G4A	homozygous rare	6.9891964	nonsynonymous SNV	uc002jxp.3.c.G271A.p.D91M	0.02	rs1800299	0.999	D	4.7	chr17	78078566	A	A	984.4	0/1	0/1	0/1
2007	GAB3	homozygous rare	50.011795	nonsynonymous SNV	uc004fml.1.c.G4186C.p.S139T	7.42E-03	rs184809317	0.123	B	3.4	chr1	15308497	G	A	2937.4	0/1	0/1	0/1
3731	GAL3ST1	compound heterozygous	85.208776	nonsynonymous SNV	uc0030ah.1.c.G119C.p.G40A	1.63E-05	rs200175330	0.016	B	3.68	chr22	30953261	C	G	709.1	0/1	0/1	0/1
3731	GAL3ST1	compound heterozygous	85.208776	nonsynonymous SNV	uc0030ah.1.c.G100A.p.V34M	0.02	rs55674628	0.065	B	3.67	chr22	30953280	C	G	878.1	0/1	0/1	0/1
3731	GHTM	homozygous rare	45.126858	nonsynonymous SNV	uc010mqb.1.c.C353T.p.P118L	5.49E-03	rs11550184	0.012	B	4.76	chr10	85908565	C	T	6801.1	0/1	0/1	1/1
2027	GLI3	homozygous rare	52.860344	nonsynonymous SNV	uc003zhy.1.c.C870A.p.D290E	0.02	rs148199056	0	B	2.56	chr9	4117942	T	T	2111.4	0/1	0/1	1/1
2034	GMPPB	compound heterozygous	30.555556	nonsynonymous SNV	uc003cck.1.c.C869T.p.T290M	4.07E-05	rs139668958	0.28	P	4.52	chr3	49759480	A	A	568.2	0/1	0/1	0/1
2034	GMPPB	compound heterozygous	30.555556	nonsynonymous SNV	uc003cck.1.c.C376G.p.H126D	0.01	rs34345884	0.001	B	4.59	chr3	49760431	G	C	1313.1	0/1	0/1	0/1
749	GOLGB1	homozygous rare	37.768342	nonsynonymous SNV	uc011bjm.1.c.C2200T.p.L734F	8.65E-03	rs114420009	0.45	P	4.84	chr4	12141613	A	A	4347.4	0/1	0/1	1/1
3721	GPNMB	compound heterozygous	89.289927	nonsynonymous SNV	uc001bnc.3.c.A1463G.p.S992D	8.13E-06	rs19409392	0.992	D	4.68	chr2	2721781	G	A	1723.1	0/1	0/1	1/1
54	GPNMB	compound heterozygous	89.289927	nonsynonymous SNV	uc011jyz.2.c.C2347A.p.N98K	1.73E-03	rs17147995	0.926	D	4.34	chr7	32299648	G	G	350.3	0/1	0/1	0/1
54	GPNMB	compound heterozygous	89.289927	stopgain SNV	uc011jyz.2.c.G1366T.p.E456X	9.08E-03	rs11537976	0.403943	NA	0.185	chr7	23313823	T	T	3031.4	0/1	0/1	0/1
28	GPR111	homozygous rare	NA	nonframeshift substitution	uc010jzj.1.c.1278_1279TG:GPR111						chr6	47649573	CA	TG	1376.1	0/1	0/1	0/1
289	GPR111	homozygous rare	NA	nonframeshift substitution	uc010jzj.1.c.1278_1279TG:GPR111						chr6	47649573	CA	TG	1548.0	0/1	0/1	0/1
2020	GPR123	compound heterozygous	43.772116	nonsynonymous SNV	uc001llw.3.c.G592A.p.A198T	0.02	rs111011757											

Proband	Gene	Inheritance	Residual Variation Intoleranc	Exonic Function	Amino Acid Change	Minor Allele Frequency EXAC	dbSNP138	PolyPhen2 Score	PolyPhen2 Prediction	GERP Score	Chromosome	Position	Ref	Alt	Quality Score	Paternal Genotype	Maternal Genotype	Proband Genotype
4405	KIAA100	compound heterozygous	73.425336	nonsynonymous SNV	uc002bhu.3.c.G2852A.p.R951H	3.80E-03	rs61732688	0.998	D	5.55	chr17	26961753	C	T	1356	0/0	0/1	0/1
4405	KIAA100	compound heterozygous	73.425336	nonsynonymous SNV	uc010crr.2.c.G321C.p.K107N			0.001	B	-4.36	chr17	26968923	C	G	916.8	0/0	0/0	1/0
2034	KIAA0556	homozygous rare	88.133994	nonsynonymous SNV	uc002dow.3.c.G4276A.p.E1426K	0.02	rs117316062	0.98	D	4.42	chr16	27784497	G	A	10634.5	0/1	0/1	0/1
32	KIAA0564	compound heterozygous	10.509554	nonsynonymous SNV	uc001uyj.3.c.G4690A.p.V1564M	4.57E-03	rs73464952	0.875	D	5.73	chr13	42189142	G	T	1487.6	0/1	0/1	0/1
32	KIAA0564	compound heterozygous	10.509554	nonsynonymous SNV	uc001uyj.3.c.G2693A.p.R989K	8.67E-03	rs41288291	0.727	P	5.25	chr13	42301395	C	C	1848.6	0/1	0/0	1/0
3721	KIAA0564	compound heterozygous	10.509554	nonsynonymous SNV	uc001uyj.3.c.A2795G.p.K932R	3.25E-05	rs4295675	0.349	P	5.27	chr13	42295675	T	C	222.6	0/1	0/0	1/0
3721	KIAA0564	compound heterozygous	10.509554	nonsynonymous SNV	uc001uyj.3.c.T2783C.p.V928A	3.33E-04	rs138978193	0.305	P	5.27	chr13	42295687	A	G	2278.6	0/1	0/0	1/0
155	KIAA0664	compound heterozygous	1.3328615	nonsynonymous SNV	uc002fux.1.c.G2233A.p.G745S	5.72E-05	rs370270067		C	4.77	chr17	25995688	C	T	168.4	0/0	0/1	0/1
146	KIAA0754	compound heterozygous	NA	nonsynonymous SNV	uc002fux.1.c.G1567A.p.A523T	6.92E-03	rs145919520		T	260	chr17	2601226	T	G	461.9	0/1	0/0	1/0
146	KIAA0754	compound heterozygous	NA	nonsynonymous SNV	uc0030wv.1.c.A536C.p.L121F	9.01E-03	rs74607331		C	4.1	chr1	39876573	A	C	2048.1	0/0	0/1	0/1
2013	KIAA0889	compound heterozygous	NA	nonsynonymous SNV	uc009v1t.1.c.A4082C.p.E1361A	4.09E-03	rs78342613		A	3.8	chr1	39800219	A	C	414.7	0/1	0/0	1/0
2013	KIAA0889	compound heterozygous	NA	nonsynonymous SNV	uc002ygd.1.c.C1625T.p.P542L	3.82E-03	rs145513467		G	4.1	chr20	35441225	G	A	1140.1	0/1	0/0	1/0
2013	KIAA0889	compound heterozygous	NA	nonsynonymous SNV	uc002ygd.1.c.C1641T.p.V481L	2.44E-05			C	3.0	chr20	35443690	C	G	703.5	0/0	0/1	0/1
2022	KIAA1797	compound heterozygous	NA	nonsynonymous SNV	uc003zoh.1.c.C644T.p.T215I	1.28E-03	rs147376982	0.254	P	0.566	chr9	20881888	C	T	1203.9	0/0	0/1	0/1
2022	KIAA1797	compound heterozygous	NA	nonsynonymous SNV	uc003zoh.1.c.G3355A.p.A119T	7.97E-03	rs150147497		B	4.4	chr9	20990164	G	A	2268.9	0/1	0/0	1/0
3721	KIAA1797	compound heterozygous	NA	nonsynonymous SNV	uc003zoh.1.c.A122C.p.D41A	8.14E-06			D	5	chr9	20823008	A	C	2080.7	0/0	0/1	0/1
3721	KIAA1797	compound heterozygous	NA	nonsynonymous SNV	uc003zoh.1.c.A554T.p.D185V			0.008	B	4.7	chr9	20874735	A	T	2572.6	0/1	0/0	1/0
2008	KIF13A	compound heterozygous	46.496815	nonsynonymous SNV	uc003nce.2.c.C516G.p.D172E	6.66E-03	rs41267712		G	3.8	chr6	17764896	G	C	2247.4	0/0	0/0	1/0
2008	KIF13A	compound heterozygous	46.496815	nonsynonymous SNV	uc003nce.2.c.A1G.p.M1V	0.02	rs17689215		T	3.8	chr6	17773790	T	C	3984.1	0/0	0/1	0/1
2013	KIF20A	compound heterozygous	12.532437	nonsynonymous SNV	uc003lqj.3.c.C131T.p.S44F	2.86E-03	rs150704301	0.995	D	4.05	chr5	137515500	C	T	1234.7	0/1	0/0	1/0
2013	KIF20A	compound heterozygous	12.532437	nonsynonymous SNV	uc003lqj.3.c.C2516T.p.P89L	0.01	rs3172747	0.303	P	5.48	chr5	137522945	T	C	4046.7	0/0	0/1	0/1
45	KLHL38	homozygous rare	80.372729	nonframeshift substitution	KLHL38:uc003yqs.1.exon1.c.293_294GC				AT	GC	2252	0/1	0/1	1/1				
123	KLHL38	homozygous rare	80.372729	nonframeshift substitution	KLHL38:uc003yqs.1.exon1.c.293_294GC				AT	GC	1309.5	0/1	0/1	1/1				
2026	KLK3	homozygous rare	67.359688	nonsynonymous SNV	uc002pts.1.c.G304A.p.D102N	0.02	rs61752561	0	B	-2.85	chr19	5161382	G	A	5170.5	0/1	0/1	1/1
17	KLK3	homozygous rare	67.359688	nonsynonymous SNV	uc002pts.1.c.G304A.p.D102N	0.02	rs61752561	0	B	-2.85	chr19	5161382	G	A	2478.1	0/1	0/1	1/1
2010	KRT72	compound heterozygous	67.585515	nonsynonymous SNV	uc002hwh.1.c.A170G.p.R106A	2.44E-05	rs10789156	0.769	P	5.68	chr7	38941004	A	T	5094	0/0	0/1	1/0
2010	KRT72	compound heterozygous	67.585515	nonsynonymous SNV	uc002hwh.1.c.G565C.p.V189L	3.42E-03	rs139807956	0.01	B	3.26	chr17	38954162	C	G	3384.3	0/0	0/1	0/1
152	KRT6B	homozygous rare	32.248172	nonsynonymous SNV	uc001sak.3.c.G1495A.p.G499S	0.02	rs61746355	0.174644	NA	-0.274	chr12	52841174	C	T	3452.3	0/1	0/1	1/1
3731	KRTAP17-1	homozygous rare	67.716443	nonsynonymous SNV	uc002hwj.3.c.G1340A.p.G47D	1.34E-03	rs74252500	0.414147	NA	-0.3	chr17	39471763	T	G	735.9	0/1	0/1	1/1
2007	KRTAP19-6	homozygous rare	80.455296	frameshift substitution	KRTAP19-6:uc002yok.1.exon1.c.153_171TTATGGATTCTCTGGATT				chr21	31913982	GAATCCAGAGAATCCAT	AATCCAGAGAATCCAT	5664.2	0/1	0/1	1/1		
17	KRTAP19-6	homozygous rare	80.455296	frameshift substitution	KRTAP19-6:uc002yok.1.exon1.c.153_171TTATGGATTCTCTGGATT				chr21	31913982	GAATCCAGAGAATCCAT	AATCCAGAGAATCCAT	6149.8	0/1	0/1	1/1		
54	KRTAP19-6	homozygous rare	80.455296	frameshift substitution	KRTAP19-6:uc002yok.1.exon1.c.153_171TTATGGATTCTCTGGATT				chr21	31913982	GAATCCAGAGAATCCAT	AATCCAGAGAATCCAT	5995	0/1	0/1	1/1		
2001	LAI2	homozygous rare	75.12385	stopgain SNV	uc002zgc.3.c.C226T.p.R76X	0.02	rs61737751	0.534453	NA	0.692	chr19	95019261	T	T	15219.2	0/1	0/1	1/1
3731	LAMA1	homozygous rare	66.217268	nonsynonymous SNV	uc010wzj.2.c.C599A.p.L1967M	4.71E-03	rs60009220	0.001	B	3.1	chr16	9861680	G	T	2599.2	0/0	0/1	1/1
155	LAMA2	compound heterozygous	79.641425	nonsynonymous SNV	uc003qbn.3.c.G222A.p.E308K	1.15E-03	rs14662599	0.620829	NA	5.57	chr6	129470136	A	A	794.2	0/1	0/0	1/0
155	LAMA2	compound heterozygous	79.641425	nonsynonymous SNV	uc003qbn.3.c.G4750A.p.G1584S	0.02	rs11781224	0.348762	NA	3.7	chr6	129687396	G	A	980.7	0/1	0/0	1/0
2812	LAMB1	compound heterozygous	8.5456476	nonsynonymous SNV	uc003evz.2.c.G2941A.p.D981N	0.01	rs61751041	0	B	2.75	chr7	107594185	C	T	1171.2	0/1	0/0	1/0
2812	LAMB1	compound heterozygous	8.5456476	nonsynonymous SNV	uc003evz.2.c.C2455G.p.R819G	0.01	rs80095409	0.999	D	4.96	chr7	107600211	G	C	382.3	0/0	0/1	0/1
53	LARS	homozygous rare	24.675631	nonframeshift substitution	uc003nwh.1.c.147_148GG:LARS				chr5	145508636	TA	CC	1338.3	0/1	0/1	0/1	1/1	
146	LEPRE1	compound heterozygous	74.681529	nonsynonymous SNV	uc001hxx.4.c.G2248A.p.G750R	5.05E-03	rs116577636	0.276323	NA	-1.8	chr12	43212750	C	T	126.5	0/1	0/1	0/1
246	LEPRE1	compound heterozygous	74.681529	nonsynonymous SNV	uc001hxx.4.c.G2248A.p.G750R				chr12	43212750	C	T	351.7	0/1	0/0	1/0		
18	LMT1	homozygous rare	81.086341	nonsynonymous SNV	uc003gdv.3.c.G1496A.p.R496H	0.02	rs28681468	0.005	B	-1.51	chr4	1824020	C	T	91.9	0/1	0/1	1/1
123	LMT2	homozygous rare	63.851144	nonframeshift substitution	uc003nwh.1.c.139_140GC:LMT2				chr9	1398199	336	163	15	216	15	0/1	0/1	1/1
2033	LMTK2	compound heterozygous	1.0497759	nonsynonymous SNV	uc003upd.2.c.G2584A.p.A862T	3.74E-03	rs34005293	0	B	-6.27	chr7	97822361	G	A	1495.8	0/0	0/1	1/0
2033	LMTK2	compound heterozygous	1.0497759	nonsynonymous SNV	uc003upd.2.c.C4022G.p.A1341G	5.07E-03	rs56343792	0	B	1.01	chr7	97823799	C	G	845.7	0/0	0/1	0/1
7760	LOC285033	homozygous rare	NA	nonsynonymous SNV	uc002yup.1.c.G298C.p.V100L	1.17E-03	rs116858801		chr2	9690359	G	C	1278	0/1	0/1	1/1		
2027	LOC286238	homozygous rare	NA	nonframeshift substitution	LOC286238:uc010mq1.1.exon2.c.149_150AG				chr9	91262493	AG	CT	5610.8	0/1	0/1	1/1		
66	LOC646508	homozygous rare	NA	nonsynonymous SNV	uc0021uz.1.c.G341A.p.S114N	0.01	rs201240639		chr19	53786079	G	A	2457.4	0/1	0/1	1/1		
66	LOC646508	homozygous rare	NA	nonframeshift substitution	LOC646508:uc021uze.1.exon3.c.343_344AG				chr19	53786081	GA	AG	2457.4	0/1	0/1	1/1		
146	LOC646508	homozygous rare	NA	nonsynonymous SNV	uc0021uze.1.c.G341A.p.S114N	0.01	rs201240639		chr19	53786079	G	A	1522.9	0/1	0/1	1/1		
146	LOC646508	homozygous rare	NA	nonframeshift substitution	LOC646508:uc021uze.1.exon3.c.343_344AG				chr19	53786081	GA	AG	1522.9	0/1	0/1	1/1		
45	LRRRC16B	compound heterozygous	16.153574	nonsynonymous SNV	uc001wjk.2.c.G287A.p.R98H	0.02	rs117092113	0.515007	NA	4.81	chr1	24533474	A	A	580.9	0/1	0/0	1/0
45	LRRRC16B	compound heterozygous	16.153574	nonsynonymous SNV	uc001wjk.2.c.G391A.p.V131M	7.34E-05	rs10304735	0.304735	NA	-5.33	chr14	24534189	G	A	2317.5	0/0	0/1	0/1
50	LRRRC3C	homozygous rare	NA	nonsynonymous SNV	uc021hw1.1.c.C514G.p.L172V	0.02	rs72832968		C	3.8	chr17	38100673	C	G	2685.8	0/1	0/1	1/1
17	LRRK1	compound heterozygous	2.9723992	nonsynonymous SNV	uc002bwr.3.c.C1246A.p.L416M	8.54E-03	rs55739947	0.724424	NA	1.16	chr15	101551007	C	A	316.4	0/1	0/0	1/0
17	LRRK1	compound heterozygous	2.9723992	nonframeshift substitution	LRRK1:uc002bwr.3.exon33.c.S813_5814AA				chr15	101606889	GC	AA	379.1	0/1	0/0	1/1		
69	LRRK1	homozygous rare	2.9723992	nonframeshift substitution	LRRK1:uc002bwr.3.exon33.c.S813_5814AA				chr15	101606889	GC	AA	2232	0/1	0/1	1/1		
3781	LRRK1	homozygous rare	2.9723992	nonframeshift substitution	LRRK1:uc002bwr.3.exon33.c.S813_5814AA				chr15	101606889	GC	AA	5947.2	0/1	0/1	1/1		
151	LSG1	homozygous rare	77.311866	nonframeshift substitution	LSG1:uc0													

Proband	Gene	Inheritance	Residual Variation Intoleranc	Exonic Function	Amino Acid Change	Minor Allele Frequency EXAC	dbSNP138	PolyPhen2 Score	PolyPhen2 Prediction	GERP Score	Chromosome	Position	Ref	Alt	Quality Score	Paternal Genotype	Maternal Genotype	Proband Genotype	
2037	MYO6	compound heterozygous	10.92525	nonsynonymous SNV	uc010eak.3:c.16155C>P.V2052A	0.02	rs117297085				chr19	17322800	T	C	476.4	0/0	0/1	0/1	
3731	MYO9T	compound heterozygous	32.05948	nonsynonymous SNV	uc003blb.3:c.A1496G>Q50R	7.73E-03	rs34711730	0.578	P	5.67	chr7	157206489	A	G	1654.2	0/1	0/1	0/1	
146	NALCN	compound heterozygous	2.9841944	nonsynonymous SNV	uc001vox.1:c.C2305T>H769Y	0.02	rs76776920	0.984	D	5.09	chr13	101763029	G	A	1525.9	0/1	0/1	0/1	
1131	NAPA	compound heterozygous	51.65723	nonsynonymous SNV	uc002pha.2:c.G25A>P.E89K	9.43E-04	rs141536777	0.98	D	5.99	chr19	48018173	C	T	1077.4	0/1	0/0	1/0	
1131	NAPA	compound heterozygous	51.65723	nonsynonymous SNV	uc010eaf.2:c.C134T>P.S45F						chr19	47996327	G	A	1984.9	0/0	0/1	0/1	
2812	NCAPD2	compound heterozygous	48.950224	nonsynonymous SNV	uc001qoo.2:c.G67T>P.V23L	0.02	rs61753197	0.16	P	4.29	chr12	6604331	G	T	4827.2	0/1	0/1	1/1	
50	NCOR1	compound heterozygous	1.4979948	frameshift deletion	NCOR1:uc010c0y.3:A942C>P.L314F	8.90E-03	rs16753150	0.834	P	0.65	chr17	15973774	T	C	575.9	0/1	0/0	1/0	
50	NCOR1	compound heterozygous	1.4979948	frameshift deletion	NCOR1:uc010c0y.3:WholeGene.	0.02	rs147795041				chr7	15953843	CAT	G	835.4	0/0	0/1	0/1	
7952	NDE1	compound heterozygous	11.765747	nonsynonymous SNV	uc002ddu.1:c.C393T>P.L29I	8.73E-03	rs113493697	0.042	B	5.07	chr16	157850949	C	G	319.1	0/1	0/0	1/0	
7952	NDE1	compound heterozygous	11.765747	nonsynonymous SNV	uc002ddu.1:c.G556T>P.A165S	4.07E-05	rs147233260	0.855	D	4.75	chr16	157851663	T	C	1350.2	0/0	0/1	0/1	
3731	NDUFA7	compound heterozygous	81.888417	nonsynonymous SNV	uc002mjm.2:c.A158G>P.Y53C	1.63E-05	rs0992	0.999	D	5.39	chr19	8381473	T	C	1087.5	0/0	0/1	0/1	
3731	NDUFA7	compound heterozygous	81.888417	nonsynonymous SNV	uc002mjm.2:c.A152G>P.N51S	5.47E-04	rs146105751	0	B	3.31	chr19	8381479	T	C	1087.5	0/1	0/0	1/0	
2035	NDUFA7	compound heterozygous	82.165605	nonsynonymous SNV	uc0012mx.3:c.G526A>P.E176K	0.02	rs35227875	0.088	B	3.34	chr15	41688732	C	T	1082.1	0/1	0/0	1/0	
2035	NDUFA7	compound heterozygous	82.165605	nonsynonymous SNV	uc0012mx.3:c.T249A>P.D83E	3.17E-04	rs146540015	0.1	B	-1.31	chr15	41689009	A	T	1537.1	0/0	0/0	1/0	
17	NDUFS7	compound heterozygous	39.95046	nonsynonymous SNV	uc002lse.4:c.C613G>P.R205G	2.49E-05	rs14680508	0.387	P	2.19	chr19	1395458	C	G	535.5	0/1	0/0	1/0	
17	NDUFS7	compound heterozygous	39.95046	nonsynonymous SNV	uc002lsg.2:c.C77T>P.A26V	1.51E-04	rs1388765				chr19	1388765	C	C	184.1	0/0	0/1	0/1	
3731	NEK10	compound heterozygous	89.189667	nonsynonymous SNV	uc003cfd.2:c.A1202G>P.N401S	8.89E-03	rs150013785	0.001	B	2.41	chr14	75533765	T	C	2392.5	0/1	0/1	0/1	
2001	NEK9	compound heterozygous	25.825666	nonsynonymous SNV	uc001wjl.3:c.A430C>P.T144A	1.87E-04	rs112467144	0.549	P	4.12	chr14	75558163	G	A	1240.2	0/1	0/0	1/0	
2001	NEK9	compound heterozygous	25.825666	nonsynonymous SNV	uc001wjl.3:c.C752T>P.P251L	0.01	rs112467144	0.549	P	4.12	chr14	75558163	G	A	1240.2	0/1	0/0	1/0	
151	NIAP2	compound heterozygous	51.916726	frameshift deletion	uc001yva.3:c.1025_1400del>p.342_342del		rs368460716				chr15	23006218	TTCTT	C	T	1174.5	0/1	0/1	0/1
2027	NLRP7	compound heterozygous	73.11866	nonsynonymous SNV	uc002oig.4:c.A1532G>P.K511R	0.01	rs61743949	0.071	B	-4.69	chr19	55450655	T	C	1443.1	0/1	0/0	1/0	
2027	NLRP7	compound heterozygous	73.11866	nonsynonymous SNV	uc002oig.4:c.A574C>P.M192L	1.09E-03	rs104895529	0.938	D	-0.67	chr19	55451613	T	G	1847.9	0/1	0/0	1/0	
2033	NOTCH1	compound heterozygous	0.3302666	nonsynonymous SNV	uc004chz.3:c.G6853A>P.V2285I	0.02	rs61751489	0	B	1.38	chr9	139391338	C	C	621.8	0/1	0/0	1/0	
2033	NOTCH1	compound heterozygous	0.3302666	nonsynonymous SNV	uc004cia.1:c.G1526A>P.R509H	0.01	rs61751543	0.874	D	3.49	chr9	139401233	C	C	541.2	0/1	0/0	1/0	
155	NOTCH2	compound heterozygous	2.1467327	nonsynonymous SNV	uc0016ek.3:c.T723A>P.L240H	1.77E-03	rs35586704	0.688	P	5	chr1	120458122	A	T	1173.5	0/0	0/1	0/1	
155	NOTCH2	compound heterozygous	2.1467327	nonsynonymous SNV	uc0016ek.3:c.C398G>P.L067	8.13E-06	rs61752454	0.067	B	5.47	chr1	120458122	A	T	1695.2	0/1	0/1	0/1	
749	NPHP4	compound heterozygous	83.581033	nonsynonymous SNV	uc0010nv.2:c.G367A>P.L238H	3.90E-03	rs79119325	0.98	D	5.63	chr11	108032614	T	C	1504.5	0/1	0/1	1/1	
2026	NPHP4	compound heterozygous	81.788158	nonsynonymous SNV	uc001a1q.2:c.C2542T>P.R848H	0.01	rs17472401				chr1	5940243	G	A	2536.9	0/1	0/0	1/0	
2026	NPHP4	compound heterozygous	81.788158	nonsynonymous SNV	uc001a1q.2:c.G1852A>P.E618K	0.01	rs571655				chr1	5965455	C	T	1266.6	0/1	0/0	1/0	
152	NPHP4	compound heterozygous	81.788158	nonsynonymous SNV	uc001a1q.2:c.G2219A>P.R740H	0.02	rs34248917				chr1	5951013	C	T	185.7	0/0	0/1	0/1	
152	NPHP4	compound heterozygous	81.788158	nonsynonymous SNV	uc001a1q.2:c.G1852A>P.E618K	0.01	rs571655				chr1	5965455	C	T	610	0/1	0/0	1/0	
14	NPVF	compound heterozygous	72.009908	frameshift substitution	NPVF:uc003oxo.3:exon3:c.547_553AG		rs7	25264779			chr7	25264779	TTGAATAG	CT	CT	1189.5	0/1	0/0	1/0
14	NPVF	compound heterozygous	72.009908	nonsynonymous SNV	uc003oxo.3:c.C168G>P.S56R	7.32E-04	rs147644964	0.991	D	2.23	chr7	25266616	G	C	258.9	0/0	0/1	0/1	
2035	NPFD2	compound heterozygous	50.56853	nonsynonymous SNV	uc003oxo.2:c.C523T>P.R179I	1	rs147644964	0.991	D	2.23	chr7	25266616	G	C	258.9	0/0	0/1	0/1	
155	NSD1	compound heterozygous	3.2377919	nonsynonymous SNV	uc003mfs.1:c.G279T>C.A833P	0.02	rs28932179	0.004	B	-8.89	chr3	24303473	G	C	1555.7	0/0	0/1	0/1	
155	NSD1	compound heterozygous	3.2377919	nonsynonymous SNV	uc021vlp.1:c.A1083T>P.R361S	1.63E-05	rs377546578	0.405	P	3.13	chr5	176721980	A	T	236.6	0/1	0/0	1/0	
2036	NTRK1	compound heterozygous	48.908841	nonsynonymous SNV	uc001fgh.1:c.G482A>P.R161H	3.01E-04	rs150271893	0.869	D	-0.246	chr1	156837949	G	A	1766.5	0/1	0/0	1/0	
2036	NTRK1	compound heterozygous	48.908841	nonsynonymous SNV	uc001fgh.1:c.G1456A>P.E486K	5.20E-04	rs144901788	0.98	D	-4.29	chr1	156845431	G	A	2537.5	0/1	0/0	1/0	
2002	NUP210	compound heterozygous	1.7162067	nonsynonymous SNV	uc003bxv.1:c.G3655A>P.V1219I	3.25E-05	rs201994997	0	B	0.887	chr3	13378316	C	T	5511.9	0/1	0/0	1/0	
2002	NUP210	compound heterozygous	1.7162067	nonsynonymous SNV	uc003bxv.1:c.G3154A>P.G1052S	2.74E-03	rs151008831	0.98	D	4.3	chr3	13383322	C	T	1000.2	0/1	0/0	1/0	
28	ORC1	compound heterozygous	82.932295	nonsynonymous SNV	uc001ctt.3:c.C1115T>P.A372V	0.01	rs3087476	0.004	B	2.94	chr1	52854961	A	G	1283.7	0/1	0/0	1/0	
28	ORC1	compound heterozygous	82.932295	nonsynonymous SNV	uc001ctt.3:c.G573C>P.R195H	4.15E-03	rs3087476	0.656	P	3.2	chr1	52867839	C	G	1684.2	0/1	0/0	1/0	
2001	OSBP1_A	compound heterozygous	NA	nonsynonymous SNV	uc002kvs.3:c.C825G>P.E218E	8.13E-06	rs34907319	0.001	B	-4.2	chr18	21742348	G	A	1224.8	0/1	0/0	1/0	
2001	OSBP1_A	compound heterozygous	NA	nonsynonymous SNV	uc002kvs.3:c.T495A>P.N165K	4.03E-03	rs34907319	0.001	B	1.98	chr18	21913036	A	T	8862.2	0/1	0/0	1/0	
2034	OSBP1_5	compound heterozygous	20.960132	nonsynonymous SNV	uc001lqj.2:c.G401A>P.R134Q	0.02	rs35733103	0.725	P	-0.354	chr11	3114190	C	T	1085.8	0/1	0/1	0/1	
2034	OSBP1_5	compound heterozygous	20.960132	nonsynonymous SNV	uc009ydx.3:c.C55T>P.P19S	4.48E-05	rs377237273				chr11	3150396	A	C	1347.1	0/1	0/0	1/0	
2033	OTOF	compound heterozygous	45.576787	nonsynonymous SNV	uc002zhi.3:c.T1681G>P.C561G	0.02	rs41288773	0.001	B	-1.8	chr2	26695500	A	C	765.4	0/1	0/0	1/0	
2033	OTOF	compound heterozygous	45.576787	nonsynonymous SNV	uc002zhi.3:c.G311A>P.R104H	8.97E-04	rs80356592	0.939	D	4.09	chr2	26700309	C	T	1310.9	0/1	0/0	1/0	
45	OTOF	compound heterozygous	45.576787	nonsynonymous SNV	uc002zhi.3:c.G1723A>P.V57M	0.01	rs55676840	0.07	B	3.78	chr2	26703734	C	T	1384.1	0/1	0/0	1/1	
151	PBRX5	compound heterozygous	83.474876	frameshift deletion	uc002zhi.3:c.329del>C.T110fs		rs329949				chr17	3594280	GG	G	884.6	0/1	0/1	1/1	
17	PALLD	compound heterozygous	63.670677	nonsynonymous SNV	uc0030ru.3:c.A625C>P.N299H	1.56E-03	rs146018183	0.374	P	5.2	chr4	169433260	A	C	773.8	0/0	0/1	1/0	
17	PALLD	compound heterozygous	63.670677	nonsynonymous SNV	uc0030ru.3:c.G784A>P.R255H		rs146018183	0.374	P	0.176	chr4	169433419	G	A	289	0/0	0/1	0/1	
24	PALLD	compound heterozygous	63.670677	nonframeshift substitution	uc0030ru.3:c.671_672CA>PALLD		rs146018183	0.374	P	0.176	chr4	169433326	TG	CA	1921.4	0/1	0/1	1/1	
3737	PCDH18	compound heterozygous	5.3078556	nonsynonymous SNV	uc0030he.4:c.G3154A>P.G1052R	0.99	D	4.3	chr4	138442437	C	T	937.6	0/1	0/0	1/1			
3737	PCDH18	compound heterozygous	5.3078556	nonsynonymous SNV	uc0030he.4:c.T2807C>P.L936R	0.956	D	4.06	chr4	138442784	A	G	1391.7	0/1	0/0	1/0			
151	PCDH8	compound heterozygous	87.715263	nonframeshift substitution	PCDH8:uc0030ip.1:exon1:c.763_764TT		rs146018183	0.374	P	0.176	chr4	140502343	CC	TT	2331.9	0/1	0/1	1/1	
3731	PCDHGA1	compound heterozygous	62.190375	nonsynonymous SNV	uc0030jj.2:c.C2335T>P.L79F	0.01	rs11575968	0.556785	NA	2.59	chr5	140712586	C	C	3919.3	0/1	0/1	1/1	

Proband	Gene	Inheritance	Residual Variation Intoleranc	Exonic Function	Amino Acid Change	Minor Allele Frequency	dbSNP138	PolyPhen2 Score	PolyPhen2 Prediction	GERP Score	Chromosome	Position	Ref	Alt	Quality Score	Paternal Genotype	Maternal Genotype	Proband Genotype	
151	PTPRC	compound heterozygous	68.51597	nonsynonymous SNV	uc009wze.1.c.G169C.p.D57H	0.01	rs11269905	0.992	D	2.71	chr1	19868761	G	C	1741	0/0	0/1	0/1	
151	PTPRC	compound heterozygous	68.51597	nonsynonymous SNV	uc009wze.1.c.G169C.p.D57H	0.01	rs116161234				chr1	198661414	T	C	434.7	0/1	0/1	0/1	
8522	PTPRG	compound heterozygous	2.4298184	nonsynonymous SNV	uc003dlb.3.c.G623T.p.P208L	1.63E-04	rs61757808	0.07	B	4.47	chr3	62118283	C	T	3852.9	0/1	0/1	0/1	
8522	PTPRG	compound heterozygous	2.4298184	nonsynonymous SNV	uc003dlb.3.c.A644G.p.D215G	7.32E-05	rs140916981	0.984	D	5.45	chr3	62118304	A	G	4102.6	0/1	0/1	0/1	
3731	PTPRJ	compound heterozygous	86.417787	nonsynonymous SNV	uc001nng.4.c.G1114A.p.V372I	8.80E-03	rs2229703	0.014	B	-10.8	chr11	48149352	G	A	632.1	0/1	0/1	0/1	
3731	PTPRJ	compound heterozygous	86.417787	nonsynonymous SNV	uc010hr.1.c.G517A.p.E173K	4.07E-04	rs181029182	0.881	D	0.0462	chr11	48161067	G	A	1410	0/1	0/1	0/1	
3721	PTPRN	compound heterozygous	89.195565	nonsynonymous SNV	uc003wmp.3.c.T1112A.p.L371H	1.31E-03	rs7456452	0.03	B	-7.81	chr7	157929357	A	T	1490.3	0/1	0/1	0/1	
3721	PTPRN	compound heterozygous	89.195565	nonsynonymous SNV	uc003wmp.3.c.G1099A.p.D367N	9.56E-03	rs5645575	0.023	B	0.387	chr7	157929370	T	T	1122.3	0/1	0/1	0/1	
114	PTPRU	deNovo	2.2823779	nonsynonymous SNV	uc001brw.3.c.C2722T.p.R69W	0.02	rs29613149			2.49	chr7	29613149	C	T	147.7	0/1	0/1	0/1	
3731	PTPRZ1	homozygous rare	1.350544	nonframeshift substitution	PTPRZ1:uc003v9r.3.exon2.c.4290_4298CGAGAGA	4.07E-05	rs150833997	1	D	2.49	chr7	121663390	G	A	2434.2	0/1	0/1	0/1	
749	RAG2	homozygous rare	55.608634	nonsynonymous SNV	uc021qpe.1.c.G229A.p.V8I	3.13E-03	rs150762709	0.024	B	4.48	chr11	36615697	G	A	2265.6	0/1	0/1	0/1	
146	RALGDS	homozygous rare	4.6414249	nonsynonymous SNV	uc004cvc.1.c.C2017T.p.P673S	0.02	rs641999				chr9	135976826	G	A	3732.9	0/1	0/1	0/1	
2035	RBM10	homozygous rare	15.764331	nonsynonymous SNV	uc004dgg.3.c.G1657C.p.A563P	0.02	rs79693964	0.121	B	4.3	chrX	47041666	G	C	1681.9	0/1	0/1	0/1	
7952	RCBTB1	compound heterozygous	18.783911	nonsynonymous SNV	uc001vde.1.c.A421G.p.M141V	2.03E-04	rs147650879	0.048	B	3.72	chr13	50134077	T	C	1368.7	0/1	0/1	0/1	
7952	RCBTB1	compound heterozygous	18.783911	nonsynonymous SNV	uc001vde.1.c.A207C.p.E69D	5.40E-03	rs138303890	0.004	B	4.49	chr13	50140824	T	C	3767.4	0/1	0/1	0/1	
3721	RECOL	compound heterozygous	8.8995046	nonsynonymous SNV	uc001rey.3.c.G1483C.p.D495H	3.88E-03	rs6499	0.003	B	5.09	chr12	21624546	C	C	1381.8	0/1	0/1	0/1	
3721	RECOL	compound heterozygous	8.8995046	nonsynonymous SNV	uc001rey.3.c.G138I.p.R454H	1.63E-05				1	D	4.75	chr12	21626571	C	T	1970	0/1	0/1
2001	RELN	compound heterozygous	1.4626091	nonsynonymous SNV	uc0101z.3.c.A1886C.p.S630R	0.02	rs115734214	0.949	D	5.27	chr7	103292112	G	G	1998.2	0/1	0/1	0/1	
2001	RELN	compound heterozygous	1.4626091	nonsynonymous SNV	uc0101z.3.c.C1799T.p.S60F	0.01	rs78008536	0.975	D	5.45	chr7	103292201	G	A	2944.8	0/1	0/1	0/1	
2037	RHBG	homozygous rare	NA	nonsynonymous SNV	uc010hph.2.c.C243G.p.F81L	0.01	rs150963900				chr1	15634717	C	G	4179.3	0/1	0/1	0/1	
23	RHBG	homozygous rare	NA	frameshift deletion	uc009wrz.3.c.1169delC.p.S390fs		rs11303415				chr1	15635437	T	T	1516	0/1	0/1	0/1	
50	RTKN2	homozygous rare	48.779193	nonframeshift deletion	uc009xpf.1.c.321_326del.p.107_109del		rs59044276				chr10	63976913	A	A	6522.1	0/1	0/1	0/1	
114	RTKN2	homozygous rare	48.779193	nonframeshift deletion	uc009xpf.1.c.321_326del.p.107_109del		rs59044276				chr10	63976913	A	A	2430.8	0/1	0/1	0/1	
1131	RTL1	compound heterozygous	73.649446	nonsynonymous SNV	uc010mj.1.c.G332A.p.R1108Q	0.02	rs117476938				chr14	101347803	C	T	248.5	0/1	0/1	0/1	
1131	RTL1	compound heterozygous	73.649446	nonsynonymous SNV	uc010mj.1.c.C1672.p.F55T	0.01	rs41266580				chr14	10134944	G	G	1248.4	0/1	0/1	0/1	
1131	RYR1	compound heterozygous	0.0058976	nonsynonymous SNV	uc0001z.3.c.A207I.p.S38R	2.37E-04	rs143771708	0.158	P	4.41	chr1	8891269	C	T	189.8	0/1	0/1	0/1	
2025	SAM03	compound heterozygous	29.429111	nonsynonymous SNV	uc0020iu.3.c.G13498C.p.D4500H	3.01E-03	rs150336398	0.45689	P	3.76	chr19	30957626	G	C	1245.1	0/1	0/1	0/1	
2025	SAM03	compound heterozygous	29.429111	nonsynonymous SNV	uc003aby.3.c.G541A.p.D181N	2.22E-03	rs150968705	0.99	D	5.31	chr6	130505611	C	T	2037.7	0/1	0/1	0/1	
2025	SAM03	compound heterozygous	29.429111	nonsynonymous SNV	uc003aby.3.c.G303T.p.R101S	5.69E-04	rs138130162	0	B	-4.5	chr6	130530720	C	A	1812.8	0/1	0/1	0/1	
4405	SCARF2	compound heterozygous	NA	nonsynonymous SNV	uc002zsj.2.c.C1982T.p.P661L	0.02	rs9680797	0.563889	NA	2.78	chr22	20780296	G	A	424.7	0/1	0/1	0/1	
4405	SCARF2	compound heterozygous	NA	nonsynonymous SNV	uc002zsj.2.c.C1661T.p.S554L	2.83E-03	rs150853613	0.564209	NA	4.36	chr22	20781732	G	A	1787.6	0/1	0/1	0/1	
4405	SCML4	compound heterozygous	50.224109	nonsynonymous SNV	uc003pry.4.c.C283T.p.R95W	0.02	rs56215636				chr6	108029180	A	T	661.2	0/1	0/1	0/1	
4405	SCML4	compound heterozygous	50.224109	nonsynonymous SNV	uc003pry.4.c.G142A.p.A48T	1.90E-03	rs143639662	0	B	0.998	chr6	108080864	C	T	680.3	0/1	0/1	0/1	
2001	SCN5A	compound heterozygous	73.625855	nonsynonymous SNV	uc010pl.3.c.T4612C.p.S38R	2.07E-03	rs202064411				chr2	167069594	G	A	17500.7	0/1	0/1	0/1	
2001	SCN5A	compound heterozygous	73.625855	nonsynonymous SNV	uc010pl.3.c.G3329A.p.R1110Q	0.02	rs74401238				chr2	167108385	G	T	10215.2	0/1	0/1	0/1	
135	SCRN3	homozygous rare	53.851144	frameshift deletion	uc002uis.3.c.477_489del.p.158_163del						chr2	175292598	T	A	2873.8	0/1	0/1	0/1	
3731	SCUBE1	homozygous rare	3.9278132	nonsynonymous SNV	uc003bdt.2.c.C674T.p.T225M	9.38E-03	rs73420094	0	B	0.643	chr22	43654278	G	A	1903.7	0/1	0/1	0/1	
2002	SEL1L2	compound heterozygous	77.229299	nonsynonymous SNV	uc010gdf.3.c.C1162T.p.P388S	1.80E-04	rs201283948	0.999	D	2.84	chr20	13850792	G	A	3324.8	0/1	0/1	0/1	
2002	SEL1L2	compound heterozygous	77.229299	nonsynonymous SNV	uc010gcf.3.c.A577G.p.I193V	0.02	rs141957868	0.588	P	5.24	chr20	13869131	T	T	678.4	0/1	0/1	0/1	
2035	SERINC2	compound heterozygous	66.165369	nonsynonymous SNV	uc001bst.3.c.G259A.p.D87N	5.64E-03	rs112561697	0.024	B	3.95	chr1	31897587	G	A	2388.4	0/1	0/1	0/1	
2035	SERINC2	compound heterozygous	66.165369	nonsynonymous SNV	uc001bst.3.c.G563C.p.R181P	4.07E-05	rs113753753	0.988	D	-4.65	chr1	31898713	G	C	1074.6	0/1	0/1	0/1	
2812	SEZ1L2	homozygous rare	6.5758434	nonsynonymous SNV	uc002duz.4.c.G1210A.p.D44D	0.02	rs113753753	0.988	D	5.35	chr16	29861206	C	T	2553.3	0/1	0/1	0/1	
2028	SID1	deNovo	51.281332	nonframeshift substitution	SGK223:uc002vz3.1.exon2.c.1701T>A						chr3	8231718	G	T	715.1	0/1	0/1	0/1	
2010	SID1	deNovo	9.7782496	nonsynonymous SNV	uc010blj.2.c.G818A.p.R273Q	3.25E-05	rs147948601	0.001	B	2.24	chr3	113346530	G	A	1272.2	0/1	0/1	0/1	
2010	SLC22A31	homozygous rare	NA	nonframeshift substitution	SLC22A31:uc021fmr.1.exon4.c.233_234AT						chr16	98265100	GC	AT	2242.8	0/1	0/1	0/1	
2035	SLC26A7	compound heterozygous	62.096013	nonsynonymous SNV	uc003yez.3.c.A643G.p.I215V	0.02	rs16912250	0.004	B	4.49	chr8	92346523	A	T	4861.5	0/1	0/1	0/1	
2035	SLC26A7	compound heterozygous	62.096013	nonsynonymous SNV	uc003yez.3.c.A1454T.p.E485V	3.22E-03	rs140569478	0.01	B	5.33	chr8	92375732	A	G	2366.5	0/1	0/1	0/1	
2023	SLC27A4	homozygous rare	7.1950932	nonsynonymous SNV	uc004but.3.c.A1052G.p.N351S	0.02	rs111417655	0	B	-1.94	chr9	131115368	A	A	2414.4	0/1	0/1	0/1	
2034	SLC6A17	homozygous rare	18.901864	nonframeshift substitution	SLC6A17:uc009wfg.3.exon2.c.168_169CA						chr1	110709719	TG	CA	4217.5	0/1	0/1	0/1	
2035	SLC6A17	homozygous rare	18.901864	nonframeshift substitution	SLC6A17:uc009wfg.3.exon2.c.168_169CA						chr1	110709719	TG	CA	3111.2	0/1	0/1	0/1	
10	SLC6A17	compound heterozygous	80.337344	nonsynonymous SNV	SLC6A17:uc009wfg.3.exon2.c.168_169CA						chr1	110709719	TG	CA	914.3	0/1	0/1	0/1	
10	SLC6A17	compound heterozygous	80.337344	nonsynonymous SNV	uc0030yk.3.c.G1124A.p.R375Q	4.23E-03	rs149382749	1	D	4.94	chr8	70667793	G	T	1006.9	0/1	0/1	0/1	
152	SMARCAD1	compound heterozygous	70.582685	nonsynonymous SNV	uc0030yk.3.c.G685A.p.A229T	8.78E-04	rs111622961	0.003	B	4.12	chr8	70744224	C	T	1497.5	0/1	0/1	0/1	
54	SMARCAD1	compound heterozygous	70.582685	nonsynonymous SNV	uc011cdw.2.c.T37C.p.W13R	8.95E-04	rs200569328	0.998	D	5.35	chr4	95185903	C	T	1380.2	0/1	0/1	0/1	
54	SMARCAD1	compound heterozygous	70.582685	nonsynonymous SNV	uc011cdw.2.c.C637T.p.H213Y	8.95E-04	rs151125076	0.958	D	5.07	chr4	95197608	C	T	568.2	0/1	0/1	0/1	
69	SNRPC	homozygous rare	51.037981	nonframeshift substitution	SNRPC:uc021yyw.1.exon1.c.41_41delinsATCT						chr6	34725658	G	T	6487.5	0/1	0/1	0/1	
69	SON	compound heterozygous	1.9933947	nonsynonymous SNV	uc002ysg.3.c.T517G.p.L173V	4.07E-05	rs15170616	0.813	P	4.04	chr21	34925081	T	G	529.5	0/1	0/1	0/1	
69	SON	compound heterozygous	1.9933947	nonsynonymous SNV	uc002ysg.3.c.G1063A.p.A355T	1.65E-05	rs140276173	0	B	2.16	chr21	34925627</							

Proband	Gene	Inheritance	Residual Variation Intoleranc e Score	Exonic Function	Amino Acid Change	Minor Allele Frequency EXAC	dbSNP138	PolyPhen2 Score	PolyPhen2 Prediction	GERP Score	Chromosome	Position	Ref	Alt	Quality Score	Paternal Genotype	Maternal Genotype	Proband Genotype
289	THSD7B	compound heterozygous		nonsynonymous SNV	uc002bvb.3.c.G635A.p.R212H	0.02	rs61741426				chr2	137825550	G	A	375.7	0/0	0/1	0/1
289	THSD7B	compound heterozygous		nonsynonymous SNV	uc002bvb.3.c.C1400T.p.T467M	3.76E-04	rs61741154				chr2	137988713	C	T	389.9	0/1	0/0	0/1
152	TIAM1	homozygous rare	9.8431234	nonframeshift substitution	uc011adk.1.c.739_740AT:TIAM1						chr21	32638549	CC	AT	2434	0/1	0/1	0/1
749	TIAM1	compound heterozygous	9.8431234	nonsynonymous SNV	uc011adk.1.c.A4226G.p.K1409R	8.42E-03	rs111536576	0.178	P	0.538	chr21	32493056	T	C	1193	0/1	0/1	0/1
749	TIAM1	compound heterozygous	9.8431234	nonsynonymous SNV	uc011adk.1.c.G92A.p.R31H	8.38E-03	rs141720377	0.999	D	3.67	chr21	32639197	C	T	1146	0/0	0/1	0/1
3781	TMIP1	homozygous rare	58.262562	nonsynonymous SNV	uc011mr.1.c.C32T.p.P11L	0.01	rs1043428				chrX	4744663	T	T	1592.9	0/1	0/1	0/1
7952	TJAP1	homozygous rare	69.509318	nonsynonymous SNV	uc0101yp.2.c.G100A.p.D34N	6.42E-03	rs146174516	0.941	D	4.47	chr7	43469358	G	A	1403.2	0/1	0/1	0/1
3721	TMEM184A	homozygous rare	45.570889	nonframeshift substitution	uc003skt.4.c.1105_1105delinsGGCC:TMEM184A						chr7	1586662	G	T	3157.2	0/1	0/1	0/1
89	TMIE	homozygous rare	41.247336	nonframeshift deletion	uc010h9.1.c.391_393del:p.131_131del						chr9	48751099	G	G	1574.5	0/1	0/1	0/1
1346	TMFR3S13	homozygous rare	84.642604	nonsynonymous SNV	uc0022ou.3.c.G1304T.p.R435L	0.02	rs80264810				chr11	117779305	G	G	641.6	0/1	0/1	0/1
151	TMPT	homozygous rare	87.402689	nonsynonymous SNV	uc003nmc.3.c.G460A.p.A154T	0.02	rs1800460	0.848	P	-0.936	chr6	18139228	C	C	2051.1	0/1	0/1	0/1
7760	TRAF4	deNovo	26.533381	stopgain SNV	uc002hcs.3.c.C622T.p.Q208X						chr17	27075439	C	T	377.1	0/0	0/0	0/1
66	TRGV3	homozygous rare	NA	nonsynonymous SNV	uc003gr.2.c.A332C.p.Y111S	0.01	rs150804886				chr7	38398135	T	G	1692.7	0/1	0/1	0/1
2028	TRMT11	homozygous rare	40.44586	nonsynonymous SNV	uc003qam.3.c.A916G.p.I306V	0.02	rs116913944	0.026	B	3.95	chr6	126332554	A	G	2469.7	0/1	0/1	0/1
23	TRPM3	compound heterozygous	6.982779	nonsynonymous SNV	uc004ah.3.c.G4012A.p.G1338R	3.46E-03	rs148192709	0.988	D	5.25	chr9	73131387	C	T	303.5	0/1	0/1	0/1
23	TRPM3	compound heterozygous	6.982779	nonsynonymous SNV	uc004ahv.3.c.C2105G.p.S702C	1.63E-05	rs138882109	0.961	D	5.24	chr9	73218319	G	C	425.5	0/0	0/1	0/1
2020	TSC2	compound heterozygous	0.8728474	nonsynonymous SNV	uc0022op.3.c.G500A.p.R167Q	0.01	rs1800725	0.999	D	4.66	chr16	2110795	G	G	1870.9	0/1	0/1	0/1
62	TSC2	compound heterozygous	0.8728474	nonsynonymous SNV	uc0022op.3.c.G500A.p.R167Q	0.01	rs1800725	0.999	D	4.66	chr16	2110795	G	G	941.9	0/0	0/1	0/1
62	TSC2	compound heterozygous	0.8728474	nonsynonymous SNV	uc0022op.3.c.G500A.p.R167Q	0.01	rs1800725	0.999	D	4.66	chr16	2110795	G	G	763.6	0/1	0/0	0/1
28	TSC22D1	compound heterozygous	5.3432413	nonsynonymous SNV	uc0022ocq.3.c.A149G.p.H50R	2.17E-04	rs184611539				chr16	2133636	A	G	529.7	0/1	0/1	0/1
28	TSC22D1	compound heterozygous	5.3432413	nonsynonymous SNV	uc001uzo.2.c.A1667G.p.Y556C	0.01	rs41288339				chr13	45008147	T	C	1662.3	0/0	0/1	0/1
28	TSC22D1	compound heterozygous	5.3432413	nonframeshift deletion	uc001luzn.4.c.1343_1345del:p.448_449del	0.01	rs201688574				chr13	45148865	T	T	1810.7	0/1	0/1	0/1
3765	TSHR	deNovo	54.098844	nonsynonymous SNV	uc001xvb.1.c.G100A.p.E34K	1.87E-04	rs45499704	0.023	B	2.24	chr14	81422124	G	A	301.9	0/0	0/1	0/1
749	TSH21	compound heterozygous	5.0070771	nonsynonymous SNV	uc021uh.1.c.G1402A.p.A468T	0.02	rs33930274	0	B	-9.83	chr18	72998989	G	A	781	0/0	0/1	0/1
749	TSH21	compound heterozygous	5.0070771	nonsynonymous SNV	uc021uh.1.c.G2591A.p.R864Q	1.29E-03	rs150160967	0.977	D	4.83	chr18	73000088	A	A	736.3	0/1	0/0	0/1
2033	TTC25	homozygous rare	NA	nonsynonymous SNV	uc0022ia.4.c.C10T.p.P4S	0.02	rs45156586				chr17	40093686	T	T	3814.8	0/1	0/1	0/1
53	TTC40	compound heterozygous	NA	nonsynonymous SNV	uc021qbc.1.c.G5520A.p.M1840I	7.48E-04	rs144154094	0.410823	NA	0.0344	chr10	134671148	G	T	1843.6	0/1	0/0	0/1
53	TTC40	compound heterozygous	NA	nonsynonymous SNV	uc021qbc.1.c.T5260C.p.S1754P	7.48E-04	rs144154094	0.410823	NA	0.0344	chr10	134672690	A	G	563.8	0/0	0/1	0/1
313	TTL6	homozygous rare	44.031611	frameshift deletion	uc021zlm.1.c.154_170del:p.52_57del	0.02					chr17	46882286	G	G	1155.6	0/1	0/1	0/1
151	TXNDC16	homozygous rare	88.110403	nonsynonymous SNV	uc001wzs.3.c.A1691G.p.D564G	0.02	rs61744423	0.831	P	5	chr14	52923820	T	C	776.9	0/1	0/1	0/1
2035	UBR1	compound heterozygous	16.814107	nonsynonymous SNV	uc001zqz.3.c.A4834C.p.R1612G	5.45E-03	rs78948790	0.951	D	3.26	chr15	43250212	C	C	2869.1	0/1	0/0	0/1
2035	UBR1	compound heterozygous	16.814107	nonsynonymous SNV	uc001zqz.3.c.A2695G.p.I899V	0.02	rs35069201	0	B	-3	chr15	43317071	T	T	5287.2	0/1	0/1	0/1
313	UBT1L1	homozygous rare	NA	nonsynonymous SNV	uc010rb.2.c.C346T.p.P116S						chr11	89819463	C	C	1361.4	0/1	0/1	0/1
45	UGT2B7	homozygous rare	40.563812	nonframeshift substitution	uc003heg.4.c.801_802TC:UGT2B7						chr4	69694337	AT	TC	5163.8	0/1	0/1	0/1
89	UGT2B7	homozygous rare	40.563812	nonframeshift substitution	uc003heg.4.c.801_802TC:UGT2B7						chr4	69694337	AT	TC	1983.7	0/1	0/1	0/1
313	UGT2B7	homozygous rare	40.563812	nonframeshift substitution	uc003heg.4.c.801_802TC:UGT2B7						chr4	69694337	AT	TC	2844.3	0/1	0/1	0/1
2001	UIMC1	compound heterozygous	59.105921	nonsynonymous SNV	uc003mf.2.c.T580C.p.Y194H	2.14E-03	rs115224789	0.99	D	5.56	chr5	176335682	A	G	6584.8	0/1	0/1	0/1
2001	UIMC1	compound heterozygous	59.105921	nonsynonymous SNV	uc021yil.1.c.T1058C.p.M353T	5.20E-04	rs143282828	0.009	B	-0.432	chr5	176395698	A	G	11345.9	0/1	0/1	0/1
3737	VAV2	compound heterozygous	2.5536683	nonsynonymous SNV	uc004cer.3.c.G2396A.p.R799Q	2.85E-04	rs138753594	0.997	D	4.63	chr9	136633640	C	T	360.1	0/1	0/0	0/1
3737	VAV2	compound heterozygous	2.5536683	nonsynonymous SNV	uc004cer.3.c.C2337G.p.I779M	0.01	rs61751477	0.031	B	0.87	chr9	136633699	G	C	584.9	0/1	0/1	0/1
54	VCAN	compound heterozygous	19.957537	nonsynonymous SNV	uc003kl.3.c.G241A.p.V81I	8.13E-06	rs1410000	0.877	D	3.44	chr5	82833071	A	G	830.1	0/1	0/0	0/1
54	VCAN	compound heterozygous	19.957537	nonsynonymous SNV	uc003kl.3.c.A596G.p.E199G	3.00E-03	rs61749614	0.01	B	1.67	chr5	82833426	A	A	399.5	0/1	0/0	0/1
7952	VPS13A	compound heterozygous	2.034678	nonsynonymous SNV	uc004ap.4.c.T293C.p.V99R	0.02	rs78048112	0.006	B	2.43	chr9	5918155	T	A	5918.5	0/1	0/1	0/1
7952	VPS13A	compound heterozygous	2.034678	nonsynonymous SNV	uc004aks.3.c.T7340C.p.I2447T	1.55E-03	rs141138349	0.307	P	4.87	chr9	79968362	T	C	3403.8	0/0	0/1	0/1
3781	VPS13B	compound heterozygous	17.504128	nonsynonymous SNV	uc003yv.3.c.G6337A.p.D2113N	0.03	rs141138349	0.307	P	4.87	chr9	79968362	T	C	3403.8	0/0	0/1	0/1
3781	VPS13B	compound heterozygous	17.504128	nonsynonymous SNV	uc003yv.3.c.G6337A.p.D2113N	0.03	rs141138349	0.307	P	4.87	chr9	79968362	T	C	3403.8	0/0	0/1	0/1
2812	VPS39	deNovo	11.276244	nonsynonymous SNV	uc003yv.3.c.C8645T.p.P2882L	9.43E-04	rs145890213	0.121	B	4.45	chr8	100831065	C	T	1702.2	0/1	0/0	0/1
2034	VTN	compound heterozygous	21.650153	nonsynonymous SNV	uc001zpc.3.c.G574A.p.G192R	0.02	rs145890213	0.121	B	4.45	chr8	100831065	C	T	1134	0/0	0/1	0/1
2034	VTN	compound heterozygous	21.650153	nonsynonymous SNV	uc002hbc.3.c.A831C.p.K277N	2.44E-05	rs201356175	0.001	B	-9.54	chr17	26695690	T	G	2457.8	0/1	0/1	0/1
2034	VTN	compound heterozygous	21.650153	nonsynonymous SNV	uc002hbc.3.c.A307T.p.I126V	4.82E-03	rs112887300	0	B	1.03	chr17	26696680	T	A	599.4	0/1	0/1	0/1
1131	WNR2	compound heterozygous	NA	nonsynonymous SNV	uc004at.3.c.C2975T.p.S992L		rs41296061				chr9	96051100	C	T	430.6	0/1	0/1	0/1
1131	WNR2	compound heterozygous	NA	nonsynonymous SNV	uc004at.1.c.G118T.p.A49S	1.63E-05	rs41296061				chr9	96051369	G	T	303.6	0/1	0/1	0/1
17	WKC3	homozygous rare	85.851616	nonsynonymous SNV	uc004csx.4.c.G11A.p.E51K	5.15E-03	rs61748714	0.956	D	4.96	chrX	10036461	A	A	1042.1	0/1	0/1	0/1
3721	XPO4	compound heterozygous	12.974758	nonsynonymous SNV	uc001unq.4.c.A2695T.p.I899L	0.01	rs117470697	0	B	-1.86	chr13	21370371	A	A	2858.7	0/1	0/1	0/1
3721	XPO4	compound heterozygous	12.974758	nonsynonymous SNV	uc001unq.4.c.A446G.p.N149S	0.02	rs17320607	0.024	B	3.41	chr13	21429776	T	C	2956.7	0/1	0/1	0/1
2023	ZFXH3	compound heterozygous	0.1179523	nonsynonymous SNV	uc002fd.3.c.G247T.p.A83S	0.01	rs2213978	0.733599	NA	4.77	chr16	72984595	C	A	1927	0/0	0/1	0/1
2023	ZFXH3	compound heterozygous	0.1179523	nonsynonymous SNV	uc002fd.3.c.G247T.p.A83S	0.01	rs2213978	0.733599	NA	4.77	chr16	72984595	C	A	1927	0/0	0/1	0/1
24	ZFXH4	compound heterozygous	2.435716	nonsynonymous SNV	uc003yav.1.c.G196C.p.V66L	0.02	rs56261025				chr8	77616519	G	C	767.1	0/1	0/1	0/1
24	ZFXH4	compound heterozygous	2.435716	nonsynonymous SNV	uc003yav.1.c.C7271T.p.P2424L	5.78E												