Patient	Gene	cDNA	Amino acid	ACMG	origin	ExAC	SIFT	Polyphen2	Mutationtaster
1	*HECW2*	c.3665G>T	p.R1222I	likely pathogenic	mother	0	deleterious	probably damaging	disease-causing
1	*KIF1A*	c.2748_2750del	p.E917_E917del	US	father	0	--	--	--
1	*MYRF*	c.1129T>A	p.Y377N	US	mother	0	deleterious	probably damaging	disease-causing
1	*NOTCH3*	c.4348G>A	p.A1450T	US	father	0.0002	tolerated	possibly damaging	disease-causing
1	*SHANK3*	c.1775C>T	p.A592V	US	father	0	--	--	--
1	*SLC2A1*	c.668G>A	p.R223Q	likely pathogenic	father	1.648E-05	tolerated	benign	disease-causing
2	*ADGRL2*	c.3496G>A	p.D1166N	US	mother	0	deleterious	probably damaging	disease-causing
2	*AUTS2*	c.2553C>G	p.H851Q	US	mother	0.0002	deleterious	probably damaging	disease-causing
2	*DYRK1A*	c.976T>G	p.F326V	US	father	0	deleterious	probably damaging	disease-causing
2	*HDDC2*	c.84+1G>C	-	US	mother	0	--	--	disease-causing
2	*SLC44A1*	c.766A>C	p.T256P	US	mother	8.238E-06	deleterious	probably damaging	disease-causing
2	*TSC2*	c.586G>A	p.A196T	US	father	4.942E-05	tolerated	probably damaging	disease-causing
2	*UBE3D*	c.976C>T	p.L326F	US	mother	0.0002	deleterious	probably damaging	disease-causing
3	*CACNA1A*	c.6881G>C	p.R2294P	US	father	0	deleterious	probably damaging	disease-causing
3	*CACNB3*	c.449G>A	p.R150Q	US	mother	0.0001	tolerated	possibly damaging	disease-causing
3	*KIF1A*	c.2748_2750del	p.E917_E917del	US	mother	0	--	--	--
4	*CACNA1A*	c.6881G>C	p.R2294P	US	father	0	deleterious	probably damaging	disease-causing
4	*CHD2*	c.1774C>T	p.L592F	US	father	0	deleterious	probably damaging	disease-causing
4	*EPB41L1*	c.67G>A	p.E23K	US	mother	0	tolerated	benign	disease-causing
4	*GJB2*	c.571T>C	p.F191L	likely pathogenic	mother	0.0001	deleterious	probably damaging	disease-causing
4	*IRF2BPL*	c.366_371del	p.Q126_Q127del	US	mother	0	--	--	--
4	*SHANK3*	c.2215G>A	p.E739K	US	mother	0	--	--	--
4	*TNRC6A*	c.1516C>T	p.H506Y	US	mother	0	deleterious	benign	disease-causing
5	*GRIN2C*	c.1297T>G	p.C433G	US	mother	0	deleterious	probably damaging	disease-causing
5	*NID2*	c.2041A>G	p.S681G	US	father	0.0002	deleterious	possibly damaging	polymorphism
5	*PRRT2*	c.649dupC	p.R217Pfs*8	pathogenic	mother	0.0063	--	--	--
5	*RBFOX3*	c.271G>A	p.D91N	US	father	0.0001	--	benign	disease-causing
5	*TLK2*	c.731A>G	p.D244G	US	mother	0	deleterious	probably damaging	disease-causing
6	*ATN1*	c.2546T>C	p.V849A	US	mother	0	tolerated	possibly damaging	polymorphism

7	*CACNA1A*	c.6985_6986insCAG	p.G2330delins	US	mother	0	--	--	--
7	*CHD2*	c.5232G>A	p.M1744I	US	father	3.316E-05	tolerated	benign	disease-causing
7	*NEURL4*	c.4303G>A	p.G1435R	US	mother	0	deleterious	possibly damaging	disease-causing
7	*SMARCA2*	c.670_672del	p.Q238_Q238del	US	mother	0	--	--	--
8	*DIAPH1*	c.3097G>A	p.D1033N	US	father	0	deleterious	probably damaging	disease-causing
8	*NSF*	c.1900C>T	p.P634S	US	father	0	deleterious	benign	disease-causing
8	*PDE8B*	c.1258G>A	p.V420M	US	mother	0.0000165	deleterious	possibly damaging	disease-causing
8	*YWHAE*	c.143C>G	p.A48G	US	father	0	tolerated	benign	disease-causing
9	*ADGRV1*	c.12586C>T	p.P4196S	US	father	0.0001	tolerated	benign	disease-causing
9	*CDON*	c.3574G>A	p.D1192N	US	father	0.0001	deleterious	benign	polymorphism
9	*SHANK3*	c.2956G>T	p.G986C	US	mother	0	--	--	--
9	*TGM6*	c.7+1G>T	-	US	father	0.0002	--	--	disease-causing
10	*AUTS2*	c.1322T>A	p.L441Q	US	father	0	deleterious	possibly damaging	disease-causing
10	*CHI3L1*	c.727T>G	p.Y243D	US	father	0	deleterious	probably damaging	disease-causing
10	*CUX2*	c.1891C>T	p.R631Trp	US	mother	9.133E-06	deleterious	probably damaging	disease-causing
10	*ERMARD*	c.1680G>T	p.Q560H	US	mother	0	tolerated	benign	disease-causing
10	*PAFAH1B1*	c.182T>C	p.L61S	US	mother	0	deleterious	probably damaging	disease-causing
10	*RELN*	c.10357C>T	p.R3453*	US	father	4.127E-05	--	--	--
10	*TRIP12*	c.3920A>C	p.K1307T	US	mother	0	deleterious	probably damaging	disease-causing
11	*PIGA*	c.328C>G	p.L110V	US	mother	0.00015	tolerated	probably damaging	disease-causing
11	*PREPL*	c.1282_c.1285delTTTG	p.F428Rfs*18	likely pathogenic	father	0.00001	--	--	--
11	*RELN*	c.449C>T	p.A150V	US	father	0.00001	deleterious	probably damaging	disease-causing
11	*SNIP1*	c.422A>C	p.D141A	US	father	0	tolerated	possibly damaging	disease-causing
11	*SYNGAP1*	c.3963_c.3964delAG	p.P1321Pfs*41	US	mother	0	--	--	--
12	*CLCN2*	c.1325A>C	p.K442T	US	mother	0	tolerated	probably damaging	disease-causing
12	*KCNB1*	c.2506C>A	p.H836N	US	father	0.00023	deleterious	probably damaging	disease-causing
13	*ADGRV1*	c.15743(exon74)C>T	p.T5248I	US	mother	0.00058	tolerated	benign	disease-causing

13	*INTS1*	c.3346G>A	p.V1116M	US	mother	0.00012	deleterious	probably damaging	disease-causing
13	*IQSEC3*	c.3283C>G	p.Q1095E	US	mother	0.00017	deleterious	possibly damaging	disease-causing
13	*PIK3R2*	c.286C>G	p.L96V	US	mother	0.00002	deleterious	probably damaging	disease-causing
13	*SPTBN2*	c.3557G>A	p.G1186D	US	mother	0.00059	deleterious	probably damaging	disease-causing
13	*TSC2*	c.5050_c.5051insCCCTGCAGTGCAGGAAAGGTAGGGCCGGGTGGGG	p.S1684Sfs*33	pathogenic	de novo	0	--	--	--
14	*ATP7A*	c.3112G>A	p.V1038I	US	mother	3.422E-05	tolerated	benign	disease-causing
14	*EHMT1*	c.2047G>A	p.D683N	US	father	8.274E-06	tolerated	benign	polymorphism
14	*FIG4*	c.788A>G	p.Y263C	US	mother	0	deleterious	probably damaging	disease-causing
14	*NRXN3*	c.2758G>A	p.A920T	likely pathogenic	mother	0	deleterious	probably damaging	disease-causing
15	*CACNA1A*	c.7178G>A	p.G2393E	US	father	0	tolerated	probably damaging	polymorphism
15	*HECW2*	c.4366A>G	p.I1456V	US	mother	0	tolerated	probably damaging	disease-causing
15	*INTS1*	c.3584C>T	p.P1195L	US	father	0	deleterious	probably damaging	disease-causing
15	*VPS9D1*	c.1144G>T	p.D382Y	US	father	0.00019	deleterious	probably damaging	disease-causing
16	*KCNQ2*	c.2602G>C	p.A868P	US	father	0	tolerated	benign	disease-causing
16	*KIDINS220*	c.883A>C	p.I295L	US	father	0	deleterious	probably damaging	disease-causing
16	*SETBP1*	c.814A>G	p.N272D	US	mother	0	deleterious	benign	polymorphism
17	*CNTNAP2*	c.3517C>A	p.P1173T	US	mother	0.00035	tolerated	probably damaging	disease-causing
17	*KCNT1*	c.3683A>C	p.E1228A	US	mother	0.0002	deleterious	possibly damaging	disease-causing
17	*NPRL3*	c.1567C>T	p.R523C	US	de novo	0.00069	deleterious	probably damaging	disease-causing
17	*POLG*	c.2890C>T	p.R964C	likely pathogenic	father	0.0093	deleterious	probably damaging	disease-causing
17	*TSC2*	c.4469A>G	p.E1490G	US	mother	0.00022	deleterious	probably damaging	disease-causing
18	*ATP1A2*	c.1262G>A	p.R421Q	US	father	0.00037	tolerated	possibly damaging	disease-causing
18	*KAT6A*	c.2986A>G	p.S996G	US	father	0.00081	deleterious	possibly damaging	polymorphism
18	*SCN5A*	c.3067C>T	p.R1023C	likely pathogenic	father	0.00012	tolerated	probably damaging	disease-causing
18	*SLC6A7*	c.1724C>T	p.P575L	US	mother	0.00012	deleterious	probably damaging	disease-causing
19	*DISC1*	c.1228delT	p.F410Ffs*26	US	mother	0	--	--	--
19	*TAS2R19*	c.542_c.543insA	p.T181Tfs*24	US	father	0	--	--	--

20	*CACNA1B*	c.347A>C	p.Q116P	US	mother	0.00081	deleterious	probably damaging	disease-causing
20	*SLC6A1*	c.589A>G	p.M197V	US	mother	0	tolerated	benign	disease-causing
20	*SPG7*	c.679C>T	p.R227X,569	likely pathogenic	father	0.00006	--	--	--
21	*ATXN2*	c.537_c.538insG	p.Q180Afs*70	US	father	0.00032	--	--	--
21	*BSN*	c.5902G>A	p.E1968K	US	mother	0	deleterious	possibly damaging	disease-causing
21	*CHD8*	c.6893T>G	p.V2298G	US	mother	0	deleterious	probably damaging	disease-causing
21	*GABRD*	c.1145G>A	p.R382H	US	father	0.00007	tolerated	benign	disease-causing
22	*BSN*	c.7687C>T	p.R2563W	US	mother	0.00009	deleterious	probably damaging	disease-causing
22	*CACNA1H*	c.1961C>T	p.P654L	likely pathogenic	de novo	0	deleterious	benign	polymorphism
22	*CLCN2*	c.1492G>T	p.G498C	US	father	0	deleterious	probably damaging	disease-causing
22	*HECW2*	c.2668G>A	p.D890N	US	mother	0	tolerated	possibly damaging	disease-causing
23	*CACNA1D*	c.3914C>T	p.A1305V	US	father	0.00051	tolerated	benign	disease-causing
23	*GJB2*	c.253T>C	p.S85P	likely pathogenic	mother	0	deleterious	probably damaging	disease-causing
23	*SMARCA4*	c.979C>T	p.P327S	US	father	0	tolerated	probably damaging	disease-causing
23	*TAS2R19*	c.542_c.543insA	p.T181Tfs*24	US	mother	0	--	--	--
23	*TBP*	c.216_c.217insG	p.Q73Afs*105	US	father	0.00079	--	--	--
23	*TRIO*	c.6876_c.6877insGGC	p.G2292_G2293insG	US	de novo	0.0066	--	--	--
24	*PRRT2*	c.236C>T	p.S79L	US	father	0.0002	deleterious	benign	polymorphism

US: Uncertain significance.