Supplementary table 1. Summary of the phenotypic features of the 80 patients in this study.

Pt	Gene	Current age, Sex	Seizure onset age	Seizure semiology	Other Phenotype	Developmental delay	Epilepsy syndrome	EEG	MRI	ASMs tried	Effective ASMs	Seizure outcome
1	*SCN2A*	10y, F	3d	FS, GTCS, T	No	Severe → Severe	DEE	Frequent → Infrequent	N → N	B6 TPM LEV VPA LTG OXC PB KD	No	Uncontrolled
2	*SCN2A*	10y, F	5m	FS, GTCS, T, SE	No	Severe → Severe	DEE	BS → Frequent → Infrequent	N → Atrophy	TPM VPA LTG OXC LEV PB Mexitil KD VNS	No	Uncontrolled
3	*SCN2A*	5y, M	4m	S	No	Severe →	WS	BS →	N →	VPA MP	MP KD	Controlled Free for 3y

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						Moderate		H → Infrequent	N	KD		
4	*SCN3A*	8y, M	6m	S	No	Severe → Moderate	WS	H → Infrequent	N → N	VPA P KD	P KD	Controlled Free for 4y
5	*SCN8A*	10y, M	6m	S	No	Severe → Moderate	WS	H → Infrequent	N → N	ACTH P TPM	ACTH P TPM	Controlled Free for 5y
6	*SCN8A*	3y, M	6m	FS, T, S	No	Severe → Severe	DEE	Frequent → Infrequent	WG → N	LEV CZP TPM OXC	OXC	Controlled Free for 2y
7	*SCN8A*	3y, F	6m	T, FS	No	Severe → Severe	DEE	Frequent → Infrequent	N → N	LEV PB VPA TPM OXC KD	No	Uncontrolled
8	*SCN8A*	5y, F	6m	S, FS, T, AA	No	Severe → Severe	DEE	Frequent → Frequent	N → N	ACTH P OXC TPM LCM	No	Uncontrolled

										KD		
9	*SCN8A*	2y, M	6m	FS	No	Severe → Severe	DEE	Frequent → Infrequent	N → N	PB OXC	OXC	Controlled Free for 1y
10	*KCNQ2*	6m, M	6d	GTCS, SE	No	Severe	DEE	Frequent, Low voltage, BS	N	B6 PB VPA LEV	No	Died of SE at 6m
11	*KCNQ2*	4y, M	1d	T, FS, S	No	Severe → Severe	OS	Frequent, BS → Infrequent	N → N	B6 LEV OXC TPM	No	Uncontrolled
12	*KCNQ2*	4y, F	1d	FS,T	No	Severe → Severe	DEE	BS → Frequent → Frequent	N → N	B6 TPM OXC VPA LCM KD	No	Uncontrolled
13	*KCNQ2*	6y, M	3m	T, GTCS	No	Severe → Severe	DEE	Frequent → Frequent	N → N	TPM OXC LEV VPA LTG KD	No	Uncontrolled

14	*KCNQ2*	7y, M	3d	FS,S, GTCS	No	Severe → Severe	DEE	BS → Frequent → Frequent	N → N	B6 NZP LEV ACTH P OXC TPM KD	TPM KD	Controlled Free for 4y
15	*KCNQ2*	5y, M	1d	FS,T, GTCS	No	Severe → Severe	DEE	BS → Frequent → Frequent	N → N	B6 PB LEV OXC TPM VPA KD	No	Uncontrolled
16	*KCNQ2*	2y, M	10d	FS,T, GTCS	No	Severe → Severe	DEE	Frequent → Frequent	N → N	B6 PB LEV TPM	PB LEV TPM	Controlled Free for 1y
17	*KCNQ2*	2y, M	3d	FS,T, S,GTCS	No	Severe → Severe	DEE	Frequent → Frequent	N → N	B6 PB LEV ACTH TPM	No	Uncontrolled

18	*KCNQ2*	2y, M	4d	FS,T, S,GTCS	No	Severe → Severe	OS → WS	BS → Frequent → H → Frequent	N → N	VPA B6 PB LEV ACTH P TPM VPA OXC KD	No	Uncontrolled
19	*KCNQ3*	2y, M	6m	S,FS,T, GTCS, M	No	Severe → Severe	WS	H → Frequent → Frequent	N → N	VPA LEV TPM ACTH P VGB OXC	No	Uncontrolled
20	*KCTD7*	10y, M	5m	M,FS,T	No	Severe → Severe	DEE	Frequent → Infrequent → N	N → N	LEV VPA TPM	VPA TPM	Controlled Free for 4y
21	*KCNB1*	10y, F	6m	FS,T	No	Severe →	DEE	Frequent →	N →	P VPA	P VPA	Controlled Free for 5y

						Severe		ESES → Infrequent	N			
22	*KCNB1*	4y, M	6m	FS,T	No	Severe → Severe	DEE	Frequent → Infrequent	N → N	LEV VPA	LEV VPA	Controlled Free for 3y
23	*KCNT1*	4y, F	1.5m	FS,T	No	Severe → Severe	MMFSI	Migrating Frequent → Frequent	N → N	PB LEV VPA OXC TPM QD KD	No	Uncontrolled
24	*KCNT1*	2y, M	3d	FS,T	No	Severe → Severe	MMFSI	Migrating Frequent → Frequent	N → N	B6 OXC LEV CZP VPA TPM QD CLB KD	No	Uncontrolled
25	*HCN1*	8y, M	6m	FS,T	No	Severe →	DEE	Frequent →	N →	OXC NZP	VPA	Controlled Free for 4y

						Moderate		Infrequent → N	N	VPA		
26	*CACNB4*	6y, F	1m	FS,T	No	Severe → Moderate	DEE	Frequent → Infrequent → N	N → N	B6 PB OXC	OXC	Controlled Free for 3y
27	*CACNA1H*	6y, M	6m	S	No	Severe → Severe	WS	H → Frequent → Frequent	N → N	MP VPA TPM OXC LTG KD	No	Uncontrolled
28	*CACNA1E*	2y, M	20d	FS,T, GTCS	No	Severe → Severe	DEE	Frequent → Infrequent	N → N	OXC LEV VPA TPM	VPA TPM	Controlled Free for 1y
29	*CDKL5*	8y, F	3m	FS,T, GTCS, S	No	Severe → Severe	WS	H → Frequent → Frequent	N → N	VPA TPM OXC LEV PB LTG	No	Uncontrolled

30	*CDKL5*	8y, F	2m	FS,T,S GTCS	No	Severe → Severe	DEE	Frequent → Frequent	N → N	KD VPA TPM OXC LEV PB LTG	No	Uncontrolled
31	*CDKL5*	8y, F	2m	FS,T,S	No	Severe → Severe	DEE	Frequent → Frequent	N → N	KD VPA TPM OXC LEV PB LTG	No	Uncontrolled
32	*CDKL5*	6y, F	1m	FS,T,S	No	Severe → Severe	DEE	Frequent → Frequent	N → N	KD VPA TPM OXC LEV PB LTG	No	Uncontrolled
33	*CDKL5*	8y, F	2m	FS,T, GTCS	Chorea	Severe →	DEE	Frequent →	N →	KD VPA TPM	No	Uncontrolled

						Severe		Infrequent	N	OXC LEV PB LTG KD			
34	*CDKL5*	5y, F	6m	S, M, FS	Chorea	Severe → Severe	DEE	Frequent → Infrequent	WG → N	VPA TPM OXC LEV PB LTG KD	KD	Controlled Free for 3y	
35	*CDKL5*	4y, F	6m	GTCS,T, S, FS	No	Severe → Severe	DEE	Frequent → Infrequent → N	N → N	ACTH P VPA TPM OXC LEV PB LTG KD	KD	Controlled Free for 3y	
36	*CDKL5*	4y, F	1m	GTCS,T, S, FS	No	Severe → Severe	DEE	Frequent → Infrequent	N → N	ACTH P VPA	KD	Controlled Free for 2y	

										TPM OXC LEV PB LTG KD		
37	*PCDH19*	10y, F	5m	FS,GTCS, T	No	Severe → Severe	DEE	Frequent → Infrequent	N → N	VPA LEV TPM	No	Uncontrolled
38	*PCDH19*	7y, F	5m	FS,T, GTCS, M	No	Severe → Severe	DS	Infrequent → Infrequent	N → N	VPA TPM OXC LEV PB LTG LCM KD	No	Uncontrolled
39	*PCDH19*	8y, F	6m	FS,T, GTCS	No	Severe → Severe	DEE	Frequent → Infrequent	N → N	OXC VPA TPM PB LTG KD	No	Uncontrolled
40	*PCDH19*	4y, F	5m	FS,T,	No	Severe	DEE	Frequent	N	OXC	No	Uncontrolled

				GTCS		→ Severe		→ Infrequent	→ N	VPA TPM PB LTG LCM KD			
41	*SLC2A1*	8y, M	2d	FS,T, GTCS	Dystonia	Severe → Severe	GLUT1DS	Frequent → Infrequent → N	WG → N	VPA LEV KD BAL	KD	Controlled Free for 6y	
42	*STXBP1*	2y, M	1m	FS,T, GTCS	Dystonia	Severe → Severe	DEE	Frequent → Infrequent → Infrequent	N → N	PB LEV BAL	LEV	Controlled Free for 1y	
43	*STXBP1*	2y, M	2m	S, T, FS, GTCS	Dystonia	Severe → Severe	WS	H → Frequent → Infrequent	WG Arachnoid Cysts → Arachnoid Cysts	PB ACTH P VGB LEV BAL	LEV	Controlled Free for 1y	
44	*STXBP1*	7y, M	1m	T, FS, S, GTCS	No	Severe →	OS	BS →	N →	B6 PB	No	Uncontrolled	

						Severe		Frequent	N	TPM LEV VPA OXC KD		
45	*SETBP1*	9y, M	6m	T, FS, GTCS	No	Severe → Severe	DEE	Frequent → Infrequent → N	N → N	PB TPM LEV VPA	VPA	Controlled Free for 5y
46	*ARHGEF9*	8y, M	4m	T, FS, GTCS	No	Severe → Moderate	DEE	Frequent → Infrequent → N	N → N	LEV OXC	OXC	Controlled Free for 4y
47	*GABRG2*	8y, M	5m	T, FS, GTCS	No	Severe → Moderate	DEE	Frequent → Infrequent → N	N → N	PB LEV VPA	LEV VPA	Controlled Free for 5y
48	*GABAA1*	3y, M	4m	S	No	Severe → Moderate	WS	H → Frequent →	N → N	ACTH P KD TPM	TPM VGB	Controlled Free for 1.5y

								Infrequent		VGB		
49	*GABRA2*	2y, M	6m	T, FS	No	Severe → Severe	DEE	Frequent → Infrequent	N → N	LEV VPA	LEV VPA	Controlled Free for 1y
50	*DEPDC5*	5y, M	1m	S, FS	No	Severe → Severe	DEE	Frequent → Infrequent	N → N	LEV TPM P OXC KD VGB	No	Uncontrolled
51	*MECP2*	8y, F	2m	S, FS, T	No	Severe→ Severe	WS	H → Frequent → Infrequent	N → N	P VPA CZP TPM	VPA TPM	Controlled Free for 3y
52	*GRIN3B*	10y, M	6m	FS,T, GTCS	No	Severe → Moderate	DEE	Frequent → Infrequent → N	N → N	PB OXC VPA	OXC VPA	Controlled Free for 6y
53	*GRIA4*	3y, M	6m	FS,T, GTCS	Ataxia	Severe → Moderate	DEE	Frequent → Infrequent	N → N	PB OXC VPA	VPA TPM	Controlled Free for 2y

								→ N		TPM			
54	*DYNC1H1*	5y, M	2m	S, FS,T,	No	Severe → Severe	WS	Frequent → Frequent	N → N	ACTH P VPA CZP TPM	No	Uncontrolled	
55	*ALDH7A1*	5y, F	2d	FS,T, GTCS	No	Severe → Moderate	PDE	Frequent → Infrequent → N	N → N	LEV PB B6	B6	Controlled Free for 4y	
56	*DPYD*	5y, F	2m	FS,T, GTCS	No	Severe → Severe	DEE	Frequent → Frequent	N → N	PB LEV TPM OXC VPA LTG KD	No	Uncontrolled	
57	*ALG11*	2y, F	3m	S,FS,T	No	Severe → Severe	WS	H → Frequent → Frequent	N → N	ACTH P LEV TPM OXC	No	Uncontrolled	

										VPA LTG KD		
58	*CCDC88C*	2y, F	3m	S,FS,T	No	Severe → Severe	WS	H → Frequent → Frequent	N → N	ACTH P LEV TPM OXC VPA LTG KD	No	Uncontrolled
59	*CSNK2B*	2y, F	2m	FS,T, GTCS	No	Severe → Moderate	DEE	Frequent → Infrequent	WG → N	LEV VPA	No	Controlled Free for 1y
60	*CSNK2B*	3y, M	3m	FS,T, GTCS	No	Severe → Moderate	DEE	Frequent → Infrequent	N → N	LEV VPA	No	Controlled Free for 1.5y
61	*IL1RAPL1*	3y, M	6m	S,FS,T	No	Severe → Severe	WS	H → Frequent → Frequent	N → N	ACTH P TPM KD VGB	KD	Controlled Free for 1.5y
62	*IQSEC2*	2y, M	6m	S,FS,T	No	Severe	WS	H	N	ACTH	No	Uncontrolled

						→ Severe		→ Frequent → Frequent	→ N	P TPM KD VGB		
63	*PACS2*	5y, M	10d	FS,T	Special face	Severe → Moderate	DEE	Frequent → Infrequent	N → N	B6 LEV VPA	VPA	Controlled Free for 4y
64	*PACS2*	2y, F	1m	FS,T	Microcephaly Special face	Severe → Severe	DEE	Frequent → Infrequent	N → N	B6 LEV TPM PB KD VPA	VPA	Controlled Free for 1y
65	*PACS2*	2y, F	1m	FS,T	Special face	Severe → Moderate	DEE	Frequent → Infrequent	N → N	B6 LEV VPA	VPA	Controlled Free for 1y
66	*PIGA*	6y, M	6m	FS,T, SE	No	Severe → Severe	DEE	Frequent → Infrequent	N → Atrophy	LEV PB VPA TPM KD	KD	Controlled Free for 3y
67	*QARS*	3y, F	2m	FS,T, S	No	Severe → Severe	WS	H → Frequent	N → N	LEV ACTH P	No	Uncontrolled

										TPM KD		
68	*RNASEH2B*	3y, F	4m	FS,T, S	No	Severe → Severe	WS	H → Frequent	N → N	LEV ACTH P TPM VPA KD	No	Uncontrolled
69	*SMC1A*	4y, F	6m	FS, Cluster Seizures	No	Severe → Severe	DEE	Frequent → Infrequent	N → N	LEV OXC KD	KD	Controlled Free for 3y
70	*SMC1A*	2y, F	2.5m	FS, Cluster seizures	No	Moderate → Moderate	DEE	H → Frequent	N	LEV, TPM, PB OXC	KD	Controlled Free for 1y
71	*SMC1A*	1.5y, F	3m	FS, Cluster seizures	No	Moderate → Moderate	DEE	Frequent → Infrequent	N	LEV OXC KD	KD	Controlled Free for 1y
72	*TBC1D24*	15y, F	6m	M, FS,T, EPC	Dystonia	Severe → Severe	EME	Frequent → Infrequent	N → N	LEV PB VPA LTG CLB KD	No	Uncontrolled

73	*TBC1D24*	8y, F	3m	M, EPC	No	Severe → Severe	EME	Frequent → Infrequent	N → N	LEV PB VPA LTG CLB OXC NZP	No	Uncontrolled
74	*TBC1D24*	5y, M	3m	M, EPC	No	Severe → Severe	EME	Frequent → Infrequent	N → N	LEV PB VPA LTG CLB OXC NZP	No	Uncontrolled
75	*WWOX*	5y, M	6m	FS,T	No	Severe → Moderate	DEE	Frequent → Infrequent	N → N	PB LEV NZP	NZP	Controlled Free for 3y
76	*COL4A2*	2y, M	3m	S	No	Severe → Severe	WS	H → Frequent	N	ACTH P	ACTH P	Controlled Free for 1y
77	*PTEN*	5y, F	6m	S，FS	No	Severe → Severe	WS	H → Frequent	N → N	MP TPM VPA KD	No	Uncontrolled

78	*H3F3A*	2y, M	2m	FS，T	No	Severe → Severe	DEE	Frequent → Frequent	WG	CLB LTG PB LEV	LEV	Controlled Free for 1y
79	*CHD2*	11y, M	6m	FS，T, GTCS, M	No	Severe → Moderate	DEE	Frequent → Infrequent → N	N → N	PB LEV OXC VPA LTG KD	No	Uncontrolled
80	*HNRNPU*	6y, M	6m	FS，T, GTC，AA	No	Moderate → Moderate	DEE	Frequent → Frequent	N → N	PB LEV VPA	VPA	Controlled Free for 2y

P: patient; y: year/years; d: Day; m: month/months; F: female; M: male; ASMs: antiseizure medications; FS: focal seizures; GTCS: generalized tonic-clonic seizures; T: tonic seizures; SE: status epilepticus; S: spasm; AA: atypical absence; M: myoclonic; EPC: epilepsia partialis continua; N: normal; AH: atypical hypsarrhythmia; H: hypsarrhythmia; Frequent: frequent multiple and multifocal sharp waves, spike waves, sharp slow waves or spike slow waves. Infrequent: infrequent sharp waves, spike waves, sharp slow waves or spike slow waves. BS: burst suppression; LEV: levetiracetam; TPM: topiramate; VPA: valproate acid; LTG: lamotrigine; OXC: oxcarbazepine; KD: ketogenic diet; PB: phenobarbital; VNS: vagus nerve stimulation; P: prednisone; LCM: lacosamide; MP: methylprednisolone; CZP: clonazepam; B6: vitamin B6; NZP: nitrazepam; VGB: vigabatrin; QD: quinidine; CLB: clobazam; BAL= baclofen; WG: widen gap in extracerebral space; ESES: electrical status epilepticus during sleep; DEE: developmental and epileptic encephalopathy; EME: early myoclonic encephalopathy; OS: Ohtahara syndrome; WS: west syndrome; MMFSI: malignant migrating focal seizures of infancy; GLUT1DS: GLUT1 deficiency syndrome; PDE: pyridoxine dependent epilepsy.

Supplementary table 2. Summary of the genetic findings in the 80 patients of this study.

P	Coordinates	Gene	Base change	Amino acid change	Predicted effect on protein	Zygosity	Inheritance	NM	Interpretation	Novel/ reported
1	Chr2: 166245951	*SCN2A*	c.5635A>G	p.M1879V	Missense	Heterozygous	De novo	NM_001040143	Likely pathogenic	N
2	Chr2: 166237177	*SCN2A*	c.4384delT	p.F1462Sfs*4	Frameshift	Heterozygous	De novo	NM_001040143	Likely pathogenic	N
3	Chr2: 166170254	*SCN2A*	c.1159G>A	p.E387K	Missense	Heterozygous	De novo	NM_001040143	Likely pathogenic	N

4	Chr2: 166019317	*SCN3A*	c.716C>A	p.A239D	Missense	Heterozygous	De novo	NM_006922	Likely pathogenic	N
5	Chr12: 52082568	*SCN8A*	c.641G>A	p.G214D	Missense	Heterozygous	De novo	NM_014191	Pathogenic	R
6	Chr12: 52162689	*SCN8A*	c.2942G>C	p.S981T	Missense	Heterozygous	De novo	NM_001330260	Likely pathogenic	N
7	Chr12: 52159789	*SCN8A*	c.2879T>A	p.V960D	Missense	Heterozygous	De novo	NM_001330260	Pathogenic	R
8	Chr12: 52082568	*SCN8A*	c.641G>A	p.G214D	Missense	Heterozygous	De novo	NM_014191	Pathogenic	R
9	Chr12: 52200768	*SCN8A*	c.5498A>T	p.D1833V	Missense	Heterozygous	De novo	NM_001330260	Likely pathogenic	N
10	Chr20: 62103803	*KCNQ2*	c.14C>T	p.S5X	Nonsense	Heterozygous	De novo	NM_172107	Likely pathogenic	N
11	Chr20: 62076073	*KCNQ2*	c.629G>A	p.R210H	Missense	Heterozygous	De novo	NM_172107	Likely pathogenic	R
12	Chr20: 62073835	*KCNQ2*	c.740C>T	p.S247L	Missense	Heterozygous	De novo	NM_172107	Pathogenic	N
13	Chr20: 62071057	*KCNQ2*	c.821C>T	p.T274M	Missense	Heterozygous	De novo	NM_172107	Pathogenic	R
14	Chr20: 62044888	*KCNQ2*	c.1678C>T	p.R560W	Missense	Heterozygous	De novo	NM_172107	Pathogenic	R
15	Chr20: 62071057	*KCNQ2*	c.821C>T	p.T274M	Missense	Heterozygous	De novo	NM_172107	Pathogenic	R

16	Chr20: 62076053	*KCNQ2*	c.649A>C	p.T217P	Missense	Heterozygous	De novo	NM_172107	Likely pathogenic	R
17	Chr20: 62059758	*KCNQ2*	c.1179delG	p.L393Lfs*2	Frameshift	Heterozygous	De novo	NM_172107	Likely pathogenic	N
18	Chr20: 62038568	*KCNQ2*	c.2048_2051dup	p.C685Rfs*180	Frameshift	Heterozygous	De novo	NM_172107	Likely pathogenic	N
19	Chr8: 133150241	*KCNQ3*	c.1231A>T	p.K411X	Nonsense	Heterozygous	De novo	NM_001204824	Likely pathogenic	N
20	Chr7: 66103259	*KCTD7*	c.334C>G	p.R112G	Missense	Compound heterozygous	Father	NM_153033	Likely pathogenic	N
	Chr7: 66104035	*KCTD7*	c.686A>T	p.D229V	Missense	Compound heterozygous	Mother	NM_153033	Likely pathogenic	
21	Chr20: 47991181	*KCNB1*	c.916C>T	p.R306C	Missense	Heterozygous	De novo	NM_004975	Pathogenic	R
22	Chr20: 47991462	*KCNB1*	c.635C>A	p.P212H	Missense	Heterozygous	De novo	NM_004975	Likely pathogenic	N
23	Chr9: 138660694	*KCNT1*	c.1421G>A	p.R474H	Missense	Heterozygous	De novo	NM_020822	Likely pathogenic	R
24	Chr9: 138660693	*KCNT1*	c.1420C>T	p.R474C	Missense	Heterozygous	De novo	NM_020822	Likely pathogenic	R
25	Chr5: 45267295	*HCN1*	c.1679G>A	p.R560H	Missense	Heterozygous	De novo	NM_021072	Likely pathogenic	N

26	Chr2: 152698493	*CACNB4*	c.668C>T	p.T223M	Missense	Heterozygous	De novo	NM_001330117	Uncertain significance	N
27	Chr16: 1255153	*CACNA1H*	c.2491G>A	p.V831M	Missense	Heterozygous	De novo	NM_021098	Uncertain significance	R
28	Chr1: 181701989	*CACNA1E*	c.2767C>T	p.H923Y	Missense	Heterozygous	De novo	NM_001205293	Uncertain significance	N
29	ChrX: 18622370-18622371	*CDKL5*	c.1326_1327insA	p.N443Kfs*20	Frameshift	Heterozygous	De novo	NM_001323289	Likely pathogenic	N
30	ChrX: 18622838-18622839	*CDKL5*	c.1794_1795insA	p.T599Nfs*12	Frameshift	Heterozygous	De novo	NM_003159	Likely pathogenic	N
31	ChrX: 18606264	*CDKL5*	c.744+1G>A	Splicing	Splicing	Heterozygous	De novo	NM_001323289	Likely pathogenic	R
32	ChrX: 18664187-18664188	*CDKL5*	c.2774_2775delTG	p.M925Ifs*15	Frameshift	Heterozygous	De novo	NM_003159	Likely pathogenic	N
33	ChrX: 18622289-18622290	*CDKL5*	c.1245_1246delAG	p.T415Tfs*3	Frameshift	Heterozygous	De novo	NM_001323289	Likely pathogenic	N
34	ChrX: 18622744	*CDKL5*	c.1700C>T	p.T567M	Missense	Heterozygous	De novo	NM_003159	Likely pathogenic	N
35	ChrX: 18593566	*CDKL5*	c.238C>T	p.R80C	Missense	Heterozygous	De novo	NM_003159	Likely pathogenic	R

36	ChrX: 18600035	*CDKL5*	c.428T>A	p.I143N	Missense	Heterozygous	De novo	NM_001323289	Likely pathogenic	N
37	ChrX: 99663125	*PCDH19*	c.471C>G	p.D157E	Missense	Heterozygous	De novo	NM_001184880	Likely pathogenic	N
38	ChrX: 99657797	*PCDH19*	c.2341delA	p.I781Sfs*19	Frameshift	Heterozygous	De novo	NM_001184880	Likely pathogenic	R
39	ChrX: 99661483	*PCDH19*	c.2113C>T	p.R705X	Nonsense	Heterozygous	De novo	NM_001184880	Likely pathogenic	R
40	ChrX: 99662798	*PCDH19*	c.798C>G	p.D266E	Missense	Heterozygous	De novo	NM_001184880	Likely pathogenic	R
41	Chr1: 43393245-43393246	*SLC2A1*	c.1278+30_1278+31ins ATTTCTCACC	Splicing	Splicing	Homozygous	AR	NM_006516	Likely pathogenic	N
42	Chr9: 130413914	*STXBP1*	c.69_70insA	p.K24Kfs*40	Frameshift	Heterozygous	De novo	NM_003165	Likely pathogenic	N
43	Chr9: 130423419	*STXBP1*	c.364C>T	p.R122X	Nonsense	Heterozygous	De novo	NM_003165	Likely pathogenic	R
44	Chr9: 130423419	*STXBP1*	c.364C>T	p.R122X	Nonsense	Heterozygous	De novo	NM_003165	Likely pathogenic	R
45	Chr18: 42531644	*SETBP1*	c.2339C>G	p.S780X	Nonsense	Heterozygous	De novo	NM_015559	Likely pathogenic	N
46	ChrX: 62863865	*ARHGEF9*	c.1364A>C	p.G455P	Missense	Heterozygous	De novo	NM_015185	Likely pathogenic	N
47	Chr5:	*GABRG2*	c.929G>T	p.R310I	Missense	Heterozygous	De novo	NM_198903	Likely	N

	161569209								pathogenic	
48	Chr5: 161317979	*GABRA1*	c.779C>T	p.P260L	Missense	Heterozygous	De novo	NM_000806	Pathogenic	N
49	Chr4: 46258291	*GABRA2*	c.995C>T	p.A332V	Missense	Heterozygous	De novo	NM_001286827	Likely pathogenic	N
50	Chr22: 32162570	*DEPDC5*	c.280-1G>A	Splicing	Splicing	Heterozygous	De novo	NM_001242896	Likely pathogenic	R
51	ChrX: 153296842	*MECP2*	c.158G>T	p.G53V	Missense	Heterozygous	De novo	NM_001369391	Likely pathogenic	N
52	Chr19: 1005329	*GRIN3B*	c.1829G>A	p.R610H	Missense	Heterozygous	De novo	NM_138690	Likely pathogenic	N
53	Chr11: 105789546	*GRIA4*	c.1378A>G	p.I460V	Missense	Heterozygous	De novo	NM_001077243	Likely pathogenic	N
54	Chr14: 102452244	*DYNC1H1*	c.1682A>G	p.E561G	Missense	Heterozygous	De novo	NM_001376	Pathogenic	N
55	Chr5: 125894979	*ALDH7A1*	c.961G>A	p.A321T	Missense	Homozygous	AR	NM_001182	Likely pathogenic	R
56	Chr1: 97915746	*DPYD*	c.1774C>T	p.R592W	Missense	Compound heterozygous	Father	NM_000110	Likely pathogenic	N
	Chr1: 97547896	*DPYD*	c.2897C>T	p.S966F	Missense	Compound heterozygous	Mother	NM_000110	Likely pathogenic	
57	Chr13: 52599058	*ALG11*	c.1192G>A	p.E398K	Missense	Compound heterozygous	Mother	NM_001004127	Likely pathogenic	N
	Chr13:	*ALG11*	c.1403G>A	p.R468H	Missense	Compound	Father	NM_001004127	Likely	

	52602650					heterozygous			pathogenic	
58	Chr14: 91792293	*CCDC88C*	c.1158G>C	p.E386D	Missense	Compound heterozygous	Mother	NM_001080414	Likely pathogenic	N
	Chr14: 91739421	*CCDC88C*	c.5635C>T	p.R1879W	Missense	Compound heterozygous	Father	NM_001080414	Likely pathogenic	
59	Chr6: 31637236-31637237	*CSNK2B*	c.508_509delGT	p.V170Afs*75	Frameshift	Heterozygous	De novo	NM_001320	Likely pathogenic	N
60	Chr6: 31635714	*CSNK2B*	c.142C>T	p.Q48X	Nonsense	Heterozygous	De novo	NM_001320	Likely pathogenic	N
61	ChrX: 29973908	*IL1RAPL1*	c.2062G>C	p.E688Q	Missense	Hemizygous	XLR	NM_014271	Likely pathogenic	N
62	ChrX: 53272627	*IQSEC2*	c.2776C>T	p.R926X	Nonsense	Heterozygous	De novo	NM_001111125	Likely pathogenic	R
63	Chr14: 105834449	*PACS2*	c.625G>A	p.E209K	Missense	Heterozygous	De novo	NM_001100913	Likely pathogenic	R
64	Chr14: 105834449	*PACS2*	c.625G>A	p.E209K	Missense	Heterozygous	De novo	NM_001100913	Likely pathogenic	R
65	Chr14: 105834449	*PACS2*	c.625G>A	p.E209K	Missense	Heterozygous	De novo	NM_001100913	Likely pathogenic	R
66	ChrX: 15349812	*PIGA*	c.241C>T	p.R81C	Missense	Hemizygous	XLR	NM_002641	Likely pathogenic	R
67	Chr3: 49136329	*QARS*	c.1852G>A	p.D618N	Missense	Compound heterozygous	Mother	NM_005051	Likely pathogenic	N

	Chr3: 49135802	*QARS*	c.2068C>T	p.R690C	Missense	Compound heterozygous	Father	NM_005051	Likely pathogenic	
68	Chr13: 51522135	*RNASEH2B*	c.629G>A	p.R210H	Missense	Compound heterozygous	Mother	NM_001142279	Likely pathogenic	N
	Chr13: 51530576	*RNASEH2B*	c.905G>A	p.G302E	Missense	Compound heterozygous	Father	NM_024570	Likely pathogenic	
69	ChrX: 53436043	*SMC1A*	c.1495C>T	p.R499X	Nonsense	Heterozygous	De novo	NM_006306	Likely pathogenic	N
70	ChrX: 53432879	*SMC1A*	c.1489C>T	p.L497F	Missense	Heterozygous	De novo	NM_001281463	Likely pathogenic	N
71	ChrX: 53407630	*SMC1A*	c.3463C>T	p.Q1155X	Nonsense	Heterozygous	De novo	NM_001281463	Likely pathogenic	N
72	chr16: 2550850	*TBC1D24*	c.1571G>C	p.R524P	Missense	Compound heterozygous	Mother	NM_001199107	Likely pathogenic	N
	Chr16: 2546829	*TBC1D24*	c.680G>A	p.R227Q	Missense	Compound heterozygous	Father	NM_001199107	Likely pathogenic	
73	Chr16: 2549836	*TBC1D24*	c.1207G>T	p.V403L	Missense	Compound heterozygous	Father	NM_001199107	Likely pathogenic	N
	Chr16: 2550465	*TBC1D24*	c.1499C>T	p.A500V	Missense	Compound heterozygous	Mother	NM_001199107	Likely pathogenic	
74	Chr16: 2549836	*TBC1D24*	c.1207G>T	p.V403L	Missense	Compound heterozygous	Father	NM_001199107	Likely pathogenic	N
	Chr16: 2550465	*TBC1D24*	c.1499C>T	p.A500V	Missense	Compound heterozygous	Mother	NM_001199107	Likely pathogenic	

75	Chr16: 78198138	*WWOX*	c.468G>T	p.R156S	Missense	Homozygous	AR	NM_016373	Likely pathogenic	R
76	Chr13: 111102095	*COL4A2*	c.1148C>T	p.P383L	Missense	Heterozygous	De novo	NM_001846	Likely pathogenic	N
77	Chr10: 89725051	*PTEN*	c.1034T>C	p.L345P	Missense	Heterozygous	De novo	NM_000314	Pathogenic	R
78	Chr1: 226259146	*H3F3A*	c.377A>G	p.Q126R	Missense	Heterozygous	De novo	NM_002107	Likely pathogenic	R
79	Chr15: 93563244	*CHD2*	c.4909C>T	p.R1637X	Nonsense	Heterozygous	De novo	NM_001271	Likely pathogenic	R
80	Chr1: 245021466	*HNRNPU*	c.1341insA	p.V448Sfs*9	Frameshift	Heterozygous	De novo	NM_031844	Likely pathogenic	N

Chr: chromosome; N: novel, i.e. this mutation has not been reported previously; R: reported, i.e. this mutation has been reported previously; R: autosomal recessive inheritance; XLR: X-linked recessive inheritance.