Supplementary Table 1. Detailed phenotypic characteristics of patients with pathogenic or likely pathogenic variants identified by whole exome/genome sequencing

Pt. No.	Sex	Age (yr)	Gene	Development and clinical characteristics	Brain MRI	EEG	Clinical diagnosis
1	F	2.3	*ARID1B*	Global developmental delay, short stature, microcephaly, seizures, facial dysmorphism (thick eyebrows, broad nasal tip, curly hair), horizontal nystagmus, high-arched palate, funnel chest, axial hypotonia	Agenesis of corpus callosum	Normal	Coffin-Siris syndrome
2	M	2.4	*SLC9A6*	Global developmental delay, microcephaly, hypotonia	Normal	Not done	Intellectual developmental disorder, X-linked syndromic, Christianson type
3	F	9.3	*SYNGAP1*	Global developmental delay, microcephaly, seizures, epilepsy, hypotonia, facial dysmorphism (long face, narrow palpebral fissures, broad nasal bridge), intellectual disability	Normal	Focal spikes on both frontal areas and generalized spike	Intellectual developmental disorder, autosomal dominant
4	F	9.4	*SCN2A*	Global developmental delay, seizures, epilepsy, intellectual disability	Normal	Focal spikes on left frontal, central and temporal areas	Developmental and epileptic encephalopathy
5	M	8.2	*NSD1*	Global developmental delay, macrocephaly, hypotonia, seizures, facial dysmorphism (prominent forehead, long face), tall stature, intellectual disability	Ventricular dilatation, thin corpus callosum	Normal	Sotos syndrome
6	F	0.5	*DCX*	Global developmental delay, hypotonia, seizures, epilepsy	Lissencephaly with band heterotopia	Multifocal spikes, hypsarrhythmia	Lissencephaly
7	M	7.8	*SLC6A8*	Global developmental delay, short stature, microcephaly, seizures, epilepsy, hypotonia, facial dysmorphism (triangular face, flat forehead, downslanted palpebral fissures, protruding mouth), long QT syndrome, intellectual disability	Right frontal arachnoid cyst	Focal spikes on both frontal areas	Cerebral creatine deficiency syndrome
8	M	5.6	*CSNK2A1*	Global developmental delay, short stature, microcephaly, hypotonia, facial dysmorphism (round face, broad nasal bridge, synophrys, anteverted nares), atrial septal defect, hydronephrosis, cryptorchidism, intellectual disability	Normal	Normal	Okur-Chung neurodevelopmental syndrome
9	M	1.3	*TAF1*	Global developmental delay, short stature, microcephaly, hypotonia, strabismus, facial dysmorphism (downslanted palpebral fissures, long face, long philtrum, Prominent ears), intellectual disability	Normal	Focal spikes on right and midline central areas	Intellectual developmental disorder, X-linked syndromic
10	F	11.3	*NR2F1*	Global developmental delay, short stature, facial dysmorphism (prominent ears, broad forehead, short philtrum, broad nasal bridge), nystagmus, amblyopia, ADHD, intellectual disability	Partial agenesis of corpus callosum	Normal	Bosch-Boonstra-Schaaf optic atrophy syndrome
11	F	0.5	*NF1*	Global developmental delay, multiple café au lait spot, iris hypopigmentation	Not done	Not done	Neurofibromatosis, type 1
12	F	0.1	*FLNA*	Global developmental delay, hypotonia, atrial septal defect	Subependymal heterotopia, mega cisterna magna, ventricular dilatation	Normal	Heterotopia, periventricular
13	F	4.3	*DYRK1A*	Global developmental delay, microcephaly, seizures, facial dysmorphism (short broad nose, bitemporal narrowing)	Normal	Normal	DYRK1A syndrome
14	F	1.0	*CASK*	Global developmental delay, microcephaly, facial dysmorphism (broad nasal bridge, short chin), spasticity of both legs	Decreased volume of both cerebrum, cerebellum and brain stem	Focal spikes on midline central area	Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia
15	F	0.1	*NEB*	Global developmental delay, short stature, myopathic facies, hypotonia, talipes equinovarus, atrial septal defect, scoliosis, hyporeflexia, high-arch palate	Normal	Not done	Nemaline myopathy
17	M	2.0	*CACNA1A*	Global developmental delay, macrocephaly, hypotonia, nystagmus, ataxia	T2 hyperintense lesions in bilateral subcortical and periventricular white matter	Not done	CACNA1A-related disorders

ADHD, attention-deficit/hyperactivity disorder; EEG, electroencephalography; MRI, magnetic resonance imaging; Pt., patient.

Supplementary Table 1. Detailed phenotypic characteristics of patients with pathogenic or likely pathogenic variants identified by whole exome/genome sequencing (continued)

Pt. No.	Sex	Age (yr)	Gene	Development and clinical characteristics	Brain MRI	EEG	Clinical diagnosis
18	M	0.1	*CREBBP*	Global developmental delay, short stature, microcephaly, hypotonia, facial dysmorphism (synophrys, broad nasal bridge), atrial septal defect, sacral dimple, developmental dysplasia of the hip, broad thumb, radial deviation of thumb, curly toe	Normal	Normal	Rubinstein-Taybi syndrome
19	F	0.9	*CDC42*	Global developmental delay, short stature, microcephaly, hypotonia, seizures, epilepsy, facial dysmorphism (synophrys, hypertelorism, narrow nasal bridge, bulbous nasal tip, thin upper lip), patent ductus arteriosus, ventricular septal defect, atrial septal defect	Decreased volume of cerebral white matter, cerebellar vermis hypoplasia	Focal spikes on midline and left central areas	Takenouchi-Kosaki syndrome
20	F	0.3	*LAMA1*	Global developmental delay, hypotonia, facial dysmorphism (frontal bossing, narrow face), encephalocele	Occipital encephalocele	Not done	Poretti-Boltshauser syndrome
21	M	18.8	*SLC6A8*	Global developmental delay, short stature, microcephaly, seizures, epilepsy, Coarse facial features, severe intellectual disability, aggressive behavior	Normal	Focal spikes on left frontal areas and background slow	Cerebral creatine deficiency syndrome
22	F	0.2	*PTPN11*	Global developmental delay, short stature, microcephaly, hypotonia, facial dysmorphism (downslanted palpebral fissures, prominent forehead, low-set ears), webbed neck	Thin corpus callosum, decreased white matter volume of both cerebrum	Normal	Noonan syndrome
23	F	0.1	*FLNA*	Global developmental delay	Periventricular nodular heterotopia, corpus callosal hypoplasia	Normal	Periventricular nodular heterotopia
24	F	16.3	*SCARB2*	Seizures, tremor, myoclonus, ataxia, nephropathy, proteinuria	Normal	Generalized intermittent slow	Epilepsy, progressive myoclonic, with renal failure
25	F	0.7	*SON*	Global developmental delay, short stature, microcephaly, seizures, epilepsy, facial dysmorphism (frontal bossing, depressed nasal bridge thin vermilion border), strabismus, nystagmus, cleft palate, hypotonia, atrial septal defect, sensorineural hearing impairment	Thin corpus callosum	Focal spikes on right parietal and left central areas	Zhu-Tokita-Takenouchi-Kim [ZTTK] syndrome

ADHD, attention-deficit/hyperactivity disorder; EEG, electroencephalography; MRI, magnetic resonance imaging; Pt., patient.

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