Patient	Gene	OMIM name	OMIM number
1	*POLR1A*	None	NA
4	*KIAA0586*	Joubert syndrome 23	616490
5	*PEX5*	Rhizomelic chondrodysplasia punctata, type 5	616716
8	*TGFB1*	Inflammatory bowel disease, immunodeficiency, and encephalopathy	618213
9	*SLC18A2*	Parkinsonism-dystonia, infantile, 2	618049
11	*TBCK*	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	616900
12	*GRIN1*	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant	614254
		Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive	617820
13	*NGLY1*	Congenital disorder of deglycosylation	615273
14	*ZBTB11*	Intellectual developmental disorder, autosomal recessive 69	618383
15	*SURF1*	Leigh syndrome, due to COX IV deficiency	220110
17	*GRIN2B*	Epileptic encephalopathy, early infantile, 27	616139
		Mental retardation, autosomal dominant 6	613970
18	*SLCO1C1*	None	NA
20	*SZT2*	Epileptic encephalopathy, early infantile, 18	615476
22	KCNQ2	Epileptic encephalopathy, early infantile, 7	613720
		Myokymia. Seizures, benign neonatal, 1	121200
26	KCNQ2	Epileptic encephalopathy, early infantile, 7	613720
		Myokymia. Seizures, benign neonatal, 1	121200
28	*IGHMBP2*	Charcot-Marie-Tooth disease, axonal, type 2S	616155
		Neuronopathy, distal hereditary motor, type VI	604320
29	*PIGT*	Multiple congenital anomalies-hypotonia-seizures syndrome 3	615398
31	*PDE6H*	Achromatopsia 6. Retinal cone dystrophy 3	610024
	LPAR6	Hypotrichosis 8. Woolly hair, autosomal recessive 1, with or without hypotrichosis	278150
32	*YY1*	Gabriele-de Vries syndrome	617557
33	*BBS12*	Bardet-Biedl syndrome 12	615989
34	*CBY1*	None	NA
	NHP2	Dyskeratosis congenita, autosomal recessive 2	613987
37	*UBE3B*	Kaufman oculocerebrofacial syndrome	244450
40	*SETBP1*	Mental retardation, autosomal dominant 29	616078

		Schinzel-Giedion midface retraction syndrome	269150
41	*PYCR2*	Leukodystrophy, hypomyelinating, 10	616420
43	*SLC52A2*	Brown-Vialetto-Van Laere syndrome 2	614707
44	*KMT2B*	Dystonia 28, childhood-onset	617284
45	*UBTF*	Neurodegeneration, childhood-onset, with brain atrophy	617672
46	*FBXL4*	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	615471
47	*GBA2*	Spastic paraplegia 46, autosomal recessive	614409
50	*FADD*	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations	613759
53	*HECW2*	Neurodevelopmental disorder with hypotonia, seizures, and absent language	617268
56	*CLN8*	Ceroid lipofuscinosis, neuronal, 8	600143
57	*POLR3B*	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism	614381
58	*MYH2*	Proximal myopathy and ophthalmoplegia	605637
59	*SCN8A*	Myoclonus, familial, 2	618364
		Cognitive impairment with or without cerebellar ataxia	614306
		Epileptic encephalopathy, early infantile, 13	614558
		Seizures, benign familial infantile, 5	617080
60	*DEAF1*	Dyskinesia, seizures, and intellectual developmental disorder	617171
63 and 64	*SLC39A8*	Congenital disorder of glycosylation, type IIn	616721
67	*TOE1*	Pontocerebellar hypoplasia, type 7	614969
68	*AP4M1*	Spastic paraplegia 50, autosomal recessive	612936
70	*KMT2A*	Wiedemann-Steiner syndrome	605130
71	*CSTB*	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg)	254800
74	*POLR3B*	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism	614381
75	*KREMEN1*	Ectodermal dysplasia 13, hair/tooth type	617392
76	*NDUFA4*	Mitochondrial complex IV deficiency, nuclear type 21	619065
77	*EXOSC5*	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects	606492
78	*CENPF*	Strømme Syndrome	243605

BG = Basal ganglia; BS = Brain stem; CBL = Cerebellum; CCH = Corpus callosum hypoplasia; CIPO = Chronic intestinal pseudo
NA= Not available; PEO = Progressive external ophthalmoplegia; PET = positron emission tomography; PN = Progressive neu

Phenotypic characteristics [diagnosis]	**CT/MRI/PET result**				
Dementia, epilepsy, spasticity [Pelizaeus-Merzbacher]	CBL, WM				
Ataxia, epilepsy; molar tooth sign [Joubert syndrome]	CBL				
Cataract, epilepsy, ID, PN [Conradi-Hünerman-like]	Normal				
Dementia, epilepsy, microcephaly, Autoimmune colitis	CTX				
Dystonia, spasticity [Neurotransmitter disorder]	Normal				
Dementia, epilepsy, PN [GM2 ganglioside storage]	CBL, CTX				
Autism, dystonia, epilepsy, insomnia [Rett-like]	Normal				
Early infantile epilepsy syndrome	CTX				
Cataract, dystonia, spasticity, microcephaly	BG, CBL, WM				
Ataxia, myopathy, PEO, RP [Kearn-Sayre syndrome]					
Ataxia, ID, self-mutilation [Angelman-like]	CTX				
Ataxia, dementia, spasticity; increased NFLP in CSF	CBL, CTX				
Autism, epilepsy, macrocephaly	Normal				
Dystonia (- Dopa), spasticity [Pelizaeus-Merzbacher]	CCH, WM				
Early infantile epilepsy syndrome [Othahara]	CBL, CTX				
Progressive neuropathy, rapid progression	Normal				
Ataxia, early infantile epilepsy [Dravet-like syndrome]	CBL, CTX				
Retinopathy [Achromatopsia] and	Not done				
Hereditary alopecia					
Dystonia (not Dopa responsive), ID; Oesophageal atresia	Normal				
ID, retinopathy; hypogonadism & obesity [Bardet-Biedel]	Not done				
Joubert syndrome	CBL				
Skin pigmentation anomalies, hypogonadism, neuropathy/myopathy, respiratory difficulties					
Oculo-cerebro-facial anomalies [Kaufman syndrome]	Normal				
Epilepsy, ID; Hypertrichosis [Schinzel-Giedion]	CTX				

Ataxia, epilepsy, microcephaly; Hot cross bun sign	BS,CBL,CTX				
Apnea, PN; Aplastic anemia; Hydrops foetalis, Vacuoles	CTX, WM				
Dystonia, spasticity; Non-PKAN neurodegeneration+iron	BG				
Dementia, epilepsy, small feet [Pelizaeus-Merzbacher]	CTX				
Brain cysts, hydrocephalus; Lactacidosis	Ventricles				
[Hereditary spastic paraplegia]; cataracts	Normal				
Epilepsy, learning difficulties [Dravet-like syndrome]	Normal				
Epilepsy, ID, microcephaly [West syndrome like]	CBL, CTX				
Ataxia, dementia, epilepsy, psychosis [Northern epilepsy]	CBL				
Ataxia, epilepsy, spasticity; Leukodystrophy	CBL, WM				
Progressive external ophthalmoplegia, ID [PEO]	Normal				
Epileptic encephalopathy	CTX				
Develop delay, dystonia, hypotonia, pigment change, VSD	CCA, CTX				
Ataxia, hypotonia, scoliosis, seizures (low grade), strabism (converging)	CBL				
Ataxia, general develop delay, spasticity	CBL				
Leukodystrophy, hypoendocrine [4H Leukodystrophy]	CCH, ventr				
Autism, microcephaly, CIPO [MNGIE-like syndrome]	Normal				
Ataxia, dementia, epilepsy [Progressive Myoclonic Epi]	CBL				
Ataxia, psychiatry (anger), spasticity [Leukodystrophy]	WM				
Alopecia, hypodontia [Hair – teeth syndrome]	Not done				
White matter hyperintensities [Leigh syndrome]	WM				
Pontocerebellar hypoplasia, death in infancy	CBL				
Intestinal atresia, microcephaly, ocular anomalies, death in neonatal period.	Ventricles, CC				

ɔ-obstruction; CTX = Cerebral cortex; ID = Intellectual disability; MNGIE = Myo-neural-gastrointestinal encephalopathy; ıropathy; RP = Retinitis pigmentosa; WM = White matter.