Neurology

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Single Gene

Code	Diseases	Gene
sCe2/0001	Gordon Holmes Syndrome	RNF216
sCe2/0002	Spastic paraplegia type 74, autosomal recessive	IBA57
sCe2/0003	SPG10	KIF5A
sCe2/0004	SPG12	RTN2
sCe2/0005	SPG13	HSPD1
sCe2/0006	SPG17	BSCL2
sCe2/0007	SPG31	REEP1
sCe2/0008	SPG33	ZFYVE27
sCe2/0009	SPG3A	ATL1
sCe2/0010	SPG4	SPAST
sCe2/0011	SPG42	SLC33A1
sCe2/0012	SPG6	NIPA1
sCe2/0013	SPG72	REEP2
sCe2/0014	SPG73	CPT1C
sCe2/0015	SPG8	KIAA0196
sCe2/0016	Neuropathy, hereditary sensory, with spastic paraplegia	CCT5
sCe2/0017	Spastic paraplegia type 74, autosomal recessive	IBA57
sCe2/0018	SPG11	SPG11
sCe2/0019	SPG15	ZFYVE26
sCe2/0020	SPG18	ERLIN2
sCe2/0021	SPG20	SPG20
sCe2/0022	SPG21	SPG21
sCe2/0023	SPG26	B4GALNT1
sCe2/0024	SPG28	DDHD1
sCe2/0025	SPG30	KIF1A

sCe2/0026	SPG35	FA2H
sCe2/0027	SPG39	PNPLA6
sCe2/0028	SPG43	C19orf12
sCe2/0029	SPG44	GJC2
sCe2/0030	SPG45	NT5C2
sCe2/0031	SPG47	AP4B1
sCe2/0032	SPG48	AP5Z1
sCe2/0033	SPG49	TECPR2
sCe2/0034	SPG50	AP4M1
sCe2/0035	SPG51	AP4E1
sCe2/0036	SPG52	AP4S1
sCe2/0037	SPG53	VPS37A
sCe2/0038	SPG54	DDHD2
sCe2/0039	SPG55	C12ORF65
sCe2/0040	SPG56	CYP2U1
sCe2/0041	SPG57	TFG
sCe2/0042	SPG59, USP8 related	USP8
sCe2/0043	SPG5A	CYP7B1
sCe2/0044	SPG60, WDR48 related	WDR48
sCe2/0045	SPG61	ARL6IP1
sCe2/0046	SPG62, ERLIN1 related	ERLIN1
sCe2/0047	SPG63	AMPD2
sCe2/0048	SPG64	ENTPD1
sCe2/0049	SPG66, ARSI related	ARSI
sCe2/0050	SPG68, FLRT1 related	FLRT1
sCe2/0051	SPG7	SPG7
sCe2/0052	SPG71, ZFR related	ZFR
sCe2/0053	SPG72	REEP2
sCe2/0054	SPG1	L1CAM

sCe2/0055	SPG2	PLP1
sCe2/0056	Cerebral palsy type 2, spastic quadriplegic	KANK1
sCe2/0057	CMT1A	PMP22
sCe2/0058	CMT1B	MPZ
sCe2/0059	CMT1C	LITAF
sCe2/0060	CMT1D	EGR2
sCe2/0061	CMT1E	PMP22
sCe2/0062	CMT1F	NEFL
sCe2/0063	CMTRIB	KARS
sCe2/0064	Neuropathy with liability to pressure palsies [HNPP]	PMP22
sCe2/0065	Roussy-Levy syndrome	PMP22
sCe2/0066	Cerebral palsy type 1, spastic quadriplegic	GAD1
sCe2/0067	Charcot-Marie-Tooth disease, axonal type 20	DYNC1H1
sCe2/0068	CMT2A1	KIF1B
sCe2/0069	CMT2A2	MFN2
sCe2/0070	CMT2B	RAB7A
sCe2/0071	CMT2B1	LMNA
sCe2/0072	CMT2B2	MED25
sCe2/0073	CMT2C	TRPV4
sCe2/0074	CMT2D	GARS
sCe2/0075	CMT2E	NEFL
sCe2/0076	CMT2F	HSPB1
sCe2/0077	CMT2I	MPZ
sCe2/0078	CMT2J	MPZ
sCe2/0079	CMT2K	GDAP1
sCe2/0080	CMT2L	HSPB8
sCe2/0081	CMT2N	AARS
sCe2/0082	CMT2P	LRSAM1
sCe2/0083	CMTRIB	KARS

sCe2/0084	Dejerine-Sottas disease	EGR2
sCe2/0085	Dejerine-Sottas disease	GJB1
sCe2/0086	Dejerine-Sottas disease	MPZ
sCe2/0087	Dejerine-Sottas disease	PMP22
sCe2/0088	Dejerine-Sottas disease	PRX
sCe2/0089	CMT4, CTDP1 related	CTDP1
sCe2/0090	CMT4A	GDAP1
sCe2/0091	CMT4B1	MTMR2
sCe2/0092	CMT4B2	SBF2
sCe2/0093	CMT4C	SH3TC2
sCe2/0094	CMT4D	NDRG1
sCe2/0095	CMT4E	MPZ
sCe2/0096	CMT4E	EGR2
sCe2/0097	CMT4F	PRX
sCe2/0098	CMT4H	FGD4
sCe2/0099	CMT4J	FIG4
sCe2/0100	CMTRIB	KARS
sCe2/0101	CMTX4	AIFM1
sCe2/0102	46,XY gonadal dysgenesis, partial, with minifascicular neuropathy	DHH
sCe2/0103	Dysautonomia, FRRS1L-related	FRRS1L
sCe2/0104	HSAN1	SPTLC1
sCe2/0105	HSAN2A	WNK1
sCe2/0106	HSAN2B	FAM134B
sCe2/0107	HSAN3	IKBKAP
sCe2/0108	HSAN4	NTRK1
sCe2/0109	HSAN5	NGF
sCe2/0110	HSAN8	PRDM12
sCe2/0111	HSN2C	KIF1A
sCe2/0112	Neuropathy, hereditary sensory and autonomic type 6	DST

sCe2/0113	Dyskinesia, familial, with facial myokymia	ADCY5
sCe2/0114	Hereditary motor and sensory neuropathy, Okinawa type	TFG
sCe2/0115	Neuronopathy distal hereditary motor type 2A	HSPB8
sCe2/0116	Neuronopathy distal hereditary motor type 2B	HSPB1
sCe2/0117	Neuronopathy distal hereditary motor type 5	GARS
sCe2/0118	Neuronopathy distal hereditary motor type 6	IGHMBP2
sCe2/0119	Neuronopathy distal hereditary motor type 7B	DCTN1
sCe2/0120	Agenesis of the corpus callosum with peripheral neuropathy	SLC12A6
sCe2/0121	Amyloidosis	TTR
sCe2/0122	Amyloidosis, finnish type	GSN
sCe2/0123	Amyotrophy hereditary neuralgic	SEPT9
sCe2/0124	Ataxia telangiectasia like disorder	MRE11A
sCe2/0125	Cataracts with facial dysmorphism and neuropathy	CTDP1
sCe2/0126	CMTDIF	GNB4
sCe2/0127	CMTRID	COX6A1
sCe2/0128	CMTX1	GJB1
sCe2/0129	CMTX5	PRPS1
sCe2/0130	DI-CMTB	DNM2
sCe2/0131	DI-CMTC	YARS
sCe2/0132	DI-CMTD	MPZ
sCe2/0133	Episodic pain syndrome type 2, familial	SCN10A
sCe2/0134	Episodic pain syndrome type 3, familial	SCN11A
sCe2/0135	Erythermalgia, primary	SCN9A
sCe2/0136	Facial paresis type 3	HOXB1
sCe2/0137	Giant axonal neuropathy type 1	GAN
sCe2/0138	Neurodegeneration due to cerebral folate transport deficiency	FOLR1
sCe2/0139	Neuronal migration disorder	CTNNA2
sCe2/0140	Neuropathy sensor type 1E	DNMT1
sCe2/0141	Neuropathy with sensory ataxic, dysarthria, and ophthalmoparesis	POLG

sCe2/0142	Neuropathy, distal hereditary motor, type 5A	BSCL2
sCe2/0143	Neuropathy, hereditary sensory and autonomic type 2	SCN9A
sCe2/0144	Neuropathy, hereditary sensory, type 1D	ATL1
sCe2/0145	Neuropathy, hereditary sensory, with spastic paraplegia	CCT5
sCe2/0146	Slowed nerve conduction velocity, autosomanal dominant	ARHGEF10
sCe2/0147	Spinocerebellar ataxia with axonal neuropathy, autosomal recessive	TDP1
sCe2/0148	Convulsions, familial infantile, with paroxysmal choreoathetosis	PRRT2
sCe2/0149	Cervical dystonia	CIZ1
sCe2/0150	Chorea, hereditary benign	NKX2-1
sCe2/0151	Choreoacanthocytosis	VPS13A
sCe2/0152	Dystonia juvenile-onset	ACTB
sCe2/0153	Dystonia, DOPA-responsive, autosomanl recessive	SPR
sCe2/0154	Dystonia-deafness syndrome	TIMM8A
sCe2/0155	DYT1	TOR1A
sCe2/0156	DYT10	PRRT2
sCe2/0157	DYT11	SGCE
sCe2/0158	DYT11, DRD2 related	DRD2
sCe2/0159	DYT12	ATP1A3
sCe2/0160	DYT16	PRKRA
sCe2/0161	DYT18	SLC2A1
sCe2/0162	DYT2	HPCA
sCe2/0163	DYT23	CACNA1B
sCe2/0164	DYT24	ANO3
sCe2/0165	DYT25	GNAL
sCe2/0166	DYT26, myoclonic	KCTD17
sCe2/0167	DYT27	COL6A3
sCe2/0168	DYT3	TAF1
sCe2/0169	DYT4	TUBB4A
sCe2/0170	DYT5A	GCH1

sCe2/0171	DYT6	THAP1
sCe2/0172	DYT8	SLC2A1
sCe2/0173	Hyperphenylalaninemia, BH4-deficient, B	GCH1
sCe2/0174	McLeod syndrome with or without chronic granulomatous disease	XK
sCe2/0175	Myoclonic dystonia, DRD2 related	DRD2
sCe2/0176	Paroxysmal nonkinesigenic dyskinesia	PNKD
sCe2/0177	Segawa syndrome, autosomal recessive	TH
sCe2/0178	Corticobasal Degeneration, CFL1 related	CFL1
sCe2/0179	Mirror movements type 1	DCC
sCe2/0180	Mirror movements type 2	RAD51
sCe2/0181	Mirror movements type 3	DNAL4
sCe2/0182	PARK1 Parkinson	SNCA
sCe2/0183	PARK13 Parkinson	HTRA2
sCe2/0184	PARK14 Parkinson	PLA2G6
sCe2/0185	PARK15 Parkinson	FBXO7
sCe2/0186	PARK17 Parkinson	VPS35
sCe2/0187	PARK19 Parkinson, juvenile-onset	DNAJC6
sCe2/0188	PARK2 Parkinson	PARK2
sCe2/0189	PARK20 Parkinson	SYNJ1
sCe2/0190	PARK21 Parkinson	DNAJC13
sCe2/0191	PARK4 Parkinson	SNCA
sCe2/0192	PARK5 Parkinson	UCHL1
sCe2/0193	PARK6 Parkinson	PINK1
sCe2/0194	PARK7 Parkinson	PARK7
sCe2/0195	PARK8 Parkinson	LRRK2
sCe2/0196	PARK9 Parkinson	ATP13A2
sCe2/0197	Parkinson disease, late-onset, susceptibility to	GBA
sCe2/0198	Parkinson disease, susceptibility to, MT-TT related	MT-TT
sCe2/0199	Parkinsonism with spasticity, X-linked	ATP6AP2

sCe2/0200	Parkinsonism-Dystonia, infantile	SLC6A3
sCe2/0201	Tremor essential type 4	FUS
sCe2/0202	Alternating hemiplegia of childhood type 1	ATP1A2
sCe2/0203	Alternating hemiplegia of childhood type 2	ATP1A3
sCe2/0204	Ataxia and muscle hypotonia	COX20
sCe2/0205	Ataxia, posterior column, with retinitis pigmentosa	FLVCR1
sCe2/0206	Ataxia, progressive seizures, mental deterioration, and hearing loss, MT-TV related	MT-TV
sCe2/0207	Ataxia-oculomotor apraxia type 1	APTX
sCe2/0208	Ataxia-oculomotor apraxia type 2	SETX
sCe2/0209	Ataxia-oculomotor apraxia type 3	PIK3R5
sCe2/0210	Ataxia-oculomotor apraxia type 4	PNKP
sCe2/0211	Ataxia-telangiectasia	ATM
sCe2/0212	CAPOS syndrome	ATP1A3
sCe2/0213	Cerebellar ataxia	CP
sCe2/0214	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion type 3	CA8
sCe2/0215	Cerebellar ataxia with deafness and narcolepsy, autosomal recessive	DNMT1
sCe2/0216	Cerebellar ataxia with mental retardation and dysequilibrium syndrome type 2	WDR81
sCe2/0217	Cerebellar ataxia with spasticity	GBA2
sCe2/0218	Cerebellar ataxia, nonprogressive, with mental retardation	CAMTA1
sCe2/0219	Cerebellar ataxia, SNX14 related	SNX14
sCe2/0220	Coenzyme Q10 deficiency type 1	COQ2
sCe2/0221	Coenzyme Q10 deficiency type 2	PDSS1
sCe2/0222	Coenzyme Q10 deficiency type 3	PDSS2
sCe2/0223	Coenzyme Q10 deficiency type 5	COQ9
sCe2/0224	Dentatorubral-pallidoluysian atrophy	ATN1
sCe2/0225	Episodic ataxia type 1	KCNA1
sCe2/0226	Episodic ataxia type 2	CACNA1A

sCe2/0227	Episodic ataxia type 5	CACNB4
sCe2/0228	Episodic ataxia type 6	SLC1A3
sCe2/0229	Familial hemiplegic migraine type 1	CACNA1A
sCe2/0230	Familial hemiplegic migraine type 2	ATP1A2
sCe2/0231	Familial hemiplegic migraine type 3	SCN1A
sCe2/0232	Fragile X syndrome	FMR1
sCe2/0233	Friedreich ataxia	FXN
sCe2/0234	Huntington disease	HTT
sCe2/0235	Huntington disease, ZDHHC17 related	ZDHHC17
sCe2/0236	Huntington disease-like type 1	PRNP
sCe2/0237	Huntington disease-like type 2	JPH3
sCe2/0238	Infantile neuroaxonal dystrophy type 1	PLA2G6
sCe2/0239	Myotonic dystrophy type 1	DMPK
sCe2/0240	Myotonic dystrophy type 2	CNBP
sCe2/0241	Salih ataxia	RUBCN
sCe2/0242	Spastic ataxia Charlevoix-Saguenay type	SACS
sCe2/0243	Spastic ataxia type 1, autosomal dominant	VAMP1
sCe2/0244	Spastic ataxia type 2, autosomal recessive	KIF1C
sCe2/0245	Spastic ataxia type 3, autosomal recessive	MARS2
sCe2/0246	Spastic ataxia type 5, autosomal recessive	AFG3L2
sCe2/0247	Spinal and bulbar muscular atrophy X-linked	AR
sCe2/0248	Spinocerebellar ataxia type 1, autosomal dominant	ATXN1
sCe2/0249	Spinocerebellar ataxia type 1, X-linked	ATP2B3
sCe2/0250	Spinocerebellar ataxia type 10, autosomal dominant	ATXN10
sCe2/0251	Spinocerebellar ataxia type 10, autosomal recessive	ANO10
sCe2/0252	Spinocerebellar ataxia type 11, autosomal dominant	TTBK2
sCe2/0253	Spinocerebellar ataxia type 12, autosomal dominant	PPP2R2B
sCe2/0254	Spinocerebellar ataxia type 12, autosomal recessive	WWOX
sCe2/0255	Spinocerebellar ataxia type 13, autosomal dominant	KCNC3

sCe2/0256	Spinocerebellar ataxia type 13, autosomal recessive	GRM1
sCe2/0257	Spinocerebellar ataxia type 14, autosomal dominant	PRKCG
sCe2/0258	Spinocerebellar ataxia type 15	ITPR1
sCe2/0259	Spinocerebellar ataxia type 17, autosomal dominant	TBP
sCe2/0260	Spinocerebellar ataxia type 17, autosomal recessive	CWF19L1
sCe2/0261	Spinocerebellar ataxia type 18, autosomal dominant	IFRD1
sCe2/0262	Spinocerebellar ataxia type 18, autosomal recessive	GRID2
sCe2/0263	Spinocerebellar ataxia type 2, autosomal dominant	ATXN2
sCe2/0264	Spinocerebellar ataxia type 21, autosomal dominant	TMEM240
sCe2/0265	Spinocerebellar ataxia type 22, autosomal dominant	KCND3
sCe2/0266	Spinocerebellar ataxia type 23, autosomal dominant	PDYN
sCe2/0267	Spinocerebellar ataxia type 26, autosomal dominant	EEF2
sCe2/0268	Spinocerebellar ataxia type 27, autosomal dominant	FGF14
sCe2/0269	Spinocerebellar ataxia type 28, autosomal dominant	AFG3L2
sCe2/0270	Spinocerebellar ataxia type 29, congenital nonprogressive	ITPR1
sCe2/0271	Spinocerebellar ataxia type 3, autosomal dominant	ATXN3
sCe2/0272	Spinocerebellar ataxia type 31, autosomal dominant	BEAN1
sCe2/0273	Spinocerebellar ataxia type 35, autosomal dominant	TGM6
sCe2/0274	Spinocerebellar ataxia type 36, autosomal dominant	NOP56
sCe2/0275	Spinocerebellar ataxia type 4, autosomal dominant	PLEKHG4
sCe2/0276	Spinocerebellar ataxia type 5, autosomal dominant	SPTBN2
sCe2/0277	Spinocerebellar ataxia type 6, autosomal dominant	CACNA1A
sCe2/0278	Spinocerebellar ataxia type 7, autosomal dominant	ATXN7
sCe2/0279	Spinocerebellar ataxia type 7, autosomal recessive	TPP1
sCe2/0280	Spinocerebellar ataxia type 8, autosomal dominant	ATXN8OS
sCe2/0281	Spinocerebellar ataxia type 8, autosomal recessive	SYNE1
sCe2/0282	Spinocerebellar ataxia type 9, autosomal rececssive	COQ8A
sCe2/0283	Spinocerebellar ataxia with axonal neuropathy, autosomal recessive	TDP1
sCe2/0284	Spinocerebellar ataxia, infantile-onset	TWNK

sCe2/0285	Vitamin E familial deficiency	TTPA
sCe2/0286	Achalasia addisonianism alacrimia syndrome	AAAS
sCe2/0287	Acrocallosal syndrome	KIF7
sCe2/0288	Acyl-CoA peroxisomal oxidase deficiency	ACOX1
sCe2/0289	Adrenoleukodystrophy, x-linked	ABCD1
sCe2/0290	Adrenoleukodystrophy, x-linked	PLXNB3
sCe2/0291	Aicardi-Goutieres syndrome type 1	TREX1
sCe2/0292	Aicardi-Goutieres syndrome type 2	RNASEH2B
sCe2/0293	Aicardi-Goutieres syndrome type 3	RNASEH2C
sCe2/0294	Aicardi-Goutieres syndrome type 4	RNASEH2A
sCe2/0295	Aicardi-Goutieres syndrome type 5	SAMHD1
sCe2/0296	Aicardi-Goutieres syndrome type 6	ADAR
sCe2/0297	Aicardi-Goutieres syndrome type 7	IFIH1
sCe2/0298	Alexander disease	GFAP
sCe2/0299	Allan-Herndon-Dudley syndrome	SLC16A2
sCe2/0300	Asperger syndrome susceptibility X-linked type 2	NLGN3
sCe2/0301	Budd-Chiari syndrome	F5
sCe2/0302	Canavan disease	ASPA
sCe2/0303	Central hypoventilation syndrome with or without Hirschsprung disease	PHOX2B
sCe2/0304	Central hypoventilation syndrome, congenital	ASCL1
sCe2/0305	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion type 1	VLDLR
sCe2/0306	Cerebrotendinous xanthomatosis	CYP27A1
sCe2/0307	COACH syndrome	TMEM67
sCe2/0308	COACH syndrome	RPGRIP1L
sCe2/0309	COACH syndrome	CC2D2A
sCe2/0310	Cohen syndrome	VPS13B
sCe2/0311	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia	IGBP1
sCe2/0312	Encephalomyopathy, mitochondrial, MT-TL2 related	MT-TL2

sCe2/0313	Encephalopathy acute necrotizing type 1	RANBP2
sCe2/0314	Encephalopathy neonatal severe	MECP2
sCe2/0315	Encephalopathy thiamine-responsive	SLC19A3
sCe2/0316	Encephalopathy, progressive, with or without lipodystrophy	BSCL2
sCe2/0317	Epileptic encephalopathy, Lennox-Gastaut type	MAPK10
sCe2/0318	Ethylmalonic encephalopathy	ETHE1
sCe2/0319	Fucosidosis	FUCA1
sCe2/0320	Gaze palsy, horizontal, with progressive scoliosis	ROBO3
sCe2/0321	Glycine encephalopathy	AMT
sCe2/0322	Glycine encephalopathy	GCSH
sCe2/0323	Glycosylation disorde type 2A	MGAT2
sCe2/0324	Glycosylation disorde type 2C	SLC35C1
sCe2/0325	Glycosylation disorder type 1C	ALG6
sCe2/0326	Glycosylation disorder type 1E	DPM1
sCe2/0327	Glycosylation disorder type 1J	DPAGT1
sCe2/0328	Glycosylation disorder type 1M	DOLK
sCe2/0329	Glycosylation disorder type 2D	B4GALT1
sCe2/0330	Glycosylation disorder type 2E	COG7
sCe2/0331	Glycosylation disorder type 2F	SLC35A1
sCe2/0332	Glycosylation disorder type 2G	COG1
sCe2/0333	Glycosylation disorder type 2H	COG8
sCe2/0334	GM1-gangliosidosis	GLB1
sCe2/0335	Griscelli syndrome type 2	RAB27A
sCe2/0336	Hydranencephaly with abnormal genitalia/Lissencephaly X-linked 2	ARX
sCe2/0337	Hydrocephalus with aqueductal stenosis and congenital intestinal pseudoobstraction	L1CAM
sCe2/0338	Hydrocephalus, nonsyndromic, autosomal recessive type 1	CCDC88C
sCe2/0339	Hydrocephalus, nonsyndromic, autosomal recessive type 2	MPDZ
sCe2/0340	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	DARS

sCe2/0341	Insensitivity to pain, channelopathy-associated	SCN9A
sCe2/0342	Joubert syndrome type 1	INPP5E
sCe2/0343	Joubert syndrome type 10	OFD1
sCe2/0344	Joubert syndrome type 13	TCTN1
sCe2/0345	Joubert syndrome type 14	TMEM237
sCe2/0346	Joubert syndrome type 15	CEP41
sCe2/0347	Joubert syndrome type 16	TMEM138
sCe2/0348	Joubert syndrome type 17	C5orf42
sCe2/0349	Joubert syndrome type 18	TCTN3
sCe2/0350	Joubert syndrome type 2	TMEM216
sCe2/0351	Joubert syndrome type 20	TMEM231
sCe2/0352	Joubert syndrome type 21	CSPP1
sCe2/0353	Joubert syndrome type 22	PDE6D
sCe2/0354	Joubert syndrome type 23	KIAA0586
sCe2/0355	Joubert syndrome type 24	TCTN2
sCe2/0356	Joubert syndrome type 3	AHI1
sCe2/0357	Joubert syndrome type 4	NPHP1
sCe2/0358	Joubert syndrome type 5	CEP290
sCe2/0359	Joubert syndrome type 6	TMEM67
sCe2/0360	Joubert syndrome type 7	RPGRIP1L
sCe2/0361	Joubert syndrome type 8	ARL13B
sCe2/0362	Joubert syndrome type 9	CC2D2A
sCe2/0363	Joubert syndrome, EXOC8 related	EXOC8
sCe2/0364	Joubert syndrome, EXOSC8 related	EXOSC8
sCe2/0365	Kenny-Caffey syndrome type 2	FAM111A
sCe2/0366	Krabbe disease	GALC
sCe2/0367	Leigh syndrome	NDUFS7
sCe2/0368	Leigh syndrome	NDUFS4
sCe2/0369	Leigh syndrome	NDUFS8

sCe2/0370	Leigh syndrome	BCS1L
sCe2/0371	Leigh syndrome	NDUFAF6
sCe2/0372	Leigh syndrome	SDHA
sCe2/0373	Leigh syndrome	FOXRED1
sCe2/0374	Leigh syndrome	NUBPL
sCe2/0375	Leigh syndrome	COX15
sCe2/0376	Leigh syndrome	NDUFA2
sCe2/0377	Leigh syndrome	NDUFA10
sCe2/0378	Leigh syndrome	NDUFAF3
sCe2/0379	Leigh syndrome	NDUFAF1
sCe2/0380	Leigh syndrome	NDUFAF2
sCe2/0381	Leigh syndrome	NDUFS3
sCe2/0382	Leigh syndrome	NDUFA9
sCe2/0383	Leigh syndrome and mitochondrial encephalopathy	ACAD9
sCe2/0384	Leigh syndrome due to COX deficiency	SURF1
sCe2/0385	Leigh syndrome due to mitochondrial complex I deficiency	MT-ND3
sCe2/0386	Leigh syndrome due to mitochondrial complex I deficiency	NDUFA12
sCe2/0387	Leigh syndrome due to mitochondrial complex I deficiency	MT-ND6
sCe2/0388	Leigh syndrome due to mitochondrial complex I deficiency	MT-ND5
sCe2/0389	Leigh syndrome due to pyruvate and alpha-ketoglutarate dehydrogenase deficiencies, LIPT1 related	LIPT1
sCe2/0390	Leigh syndrome due to pyruvate carboxylase deficiency	PC
sCe2/0391	Leigh syndrome due to the mitochondrial complex IV deficiency	TACO1
sCe2/0392	Leigh syndrome, French-Canadian type	LRPPRC
sCe2/0393	Leukodystrophy demyelinating adult-onset, autosomal dominant	LMNB1
sCe2/0394	Leukodystrophy hypomyelinating	GJC2
sCe2/0395	Leukodystrophy hypomyelinating type 3	AIMP1
sCe2/0396	Leukodystrophy hypomyelinating type 4	HSPD1
sCe2/0397	Leukodystrophy hypomyelinating type 5	FAM126A
sCe2/0398	Leukodystrophy hypomyelinating type 6	TUBB4A

sCe2/0399	Leukodystrophy hypomyelinating type 7	POLR3A
sCe2/0400	Leukodystrophy hypomyelinating type 8	POLR3B
sCe2/0401	Leukodystrophy hypomyelinating type 9	RARS
sCe2/0402	Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation	DARS2
sCe2/0403	Leukoencephalopathy with dystonia and motor neuropathy	SCP2
sCe2/0404	Leukoencephalopathy with vanishing white matter	EIF2B2
sCe2/0405	Leukoencephalopathy with vanishing white matter	EIF2B1
sCe2/0406	Leukoencephalopathy with vanishing white matter	EIF2B3
sCe2/0407	Leukoencephalopathy with vanishing white matter	EIF2B4
sCe2/0408	Leukoencephalopathy with vanishing white matter	EIF2B5
sCe2/0409	Leukoencephalopathy, cystic without megalencephaly	RNASET2
sCe2/0410	Leukoencephalopathy, diffuse hereditary, with spheroids	CSF1R
sCe2/0411	Leukoencephalopathy, progressive, with ovarian failure	AARS2
sCe2/0412	Lissencephaly type 4 with microcephaly	NDE1
sCe2/0413	Mandibulofacial dysostosis with microcephaly	EFTUD2
sCe2/0414	Megalencephalic leukoencephalopathy with subcortical cysts	MLC1
sCe2/0415	Megalencephalic leukoencephalopathy with subcortical cysts 2A	HEPACAM
sCe2/0416	Mental retardation with language impairment and autistic features	FOXP1
sCe2/0417	Metachromatic leukodystrophy due to Saposin B deficiency	PSAP
sCe2/0418	Mitochondrial complex I deficiency	NDUFAF3
sCe2/0419	Mitochondrial complex I deficiency	NDUFAF5
sCe2/0420	Mitochondrial complex I deficiency	NDUFAF1
sCe2/0421	Mitochondrial complex I deficiency	NDUFS6
sCe2/0422	Mitochondrial complex I deficiency	NDUFS4
sCe2/0423	Mitochondrial complex I deficiency	NDUFV2
sCe2/0424	Mitochondrial complex I deficiency	NDUFV1
sCe2/0425	Mitochondrial complex I deficiency	NDUFA11
sCe2/0426	Mitochondrial complex I deficiency	FOXRED1
sCe2/0427	Mitochondrial complex I deficiency	NDUFS2

sCe2/0428	Mitochondrial complex I deficiency	NDUFS1
sCe2/0429	Mitochondrial complex I deficiency	NDUFAF4
sCe2/0430	Mitochondrial complex I deficiency, MT-TN related	MT-TN
sCe2/0431	Mitochondrial complex II deficiency	SDHAF1
sCe2/0432	Mitochondrial complex III deficiency	UQCRQ
sCe2/0433	Mitochondrial complex III deficiency	UQCRB
sCe2/0434	Mitochondrial complex III deficiency	BCS1L
sCe2/0435	Mitochondrial complex III deficiency	UQCRC2
sCe2/0436	Mitochondrial complex III deficiency, nuclear type 2	TTC19
sCe2/0437	Mitochondrial complex IV deficiency	COX6B1
sCe2/0438	Mitochondrial complex IV deficiency	FASTKD2
sCe2/0439	Mitochondrial complex V (ATP synthase) deficiency	MT-ATP6
sCe2/0440	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1	ATPAF2
sCe2/0441	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	TMEM70
sCe2/0442	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3	ATP5E
sCe2/0443	Mitochondrial DNA depletion syndrome	SUCLA2
sCe2/0444	Mitochondrial DNA depletion syndrome	TK2
sCe2/0445	Mitochondrial DNA depletion syndrome 8B, MNGIE type	RRM2B
sCe2/0446	Mitochondrial DNA depletion syndrome type 11	MGME1
sCe2/0447	Mitochondrial DNA depletion syndrome type 13	FBXL4
sCe2/0448	Mitochondrial DNA depletion syndrome type 4A	POLG
sCe2/0449	Mitochondrial DNA depletion syndrome type 4B	POLG
sCe2/0450	Mitochondrial DNA depletion syndrome type 7	TWNK
sCe2/0451	Mitochondrial DNA depletion syndrome, encephalomyopathic type with methylmalonic aciduria	SUCLG1
sCe2/0452	Mitochondrial encephalomyopathy	MFF
sCe2/0453	Mitochondrial encephalomyopathy	MT-CYB
sCe2/0454	Mitochondrial neurogastrointestinal encephalopathy syndrome without leukoencephalopathy	POLG
sCe2/0455	Multiple congenital anomalies-hypotonia-seizures syndrome type 3	PIGT

sCe2/0456	Multiple mitochondrial dysfunctions syndrome type 1	NFU1
sCe2/0457	Multiple mitochondrial dysfunctions syndrome type 2	BOLA3
sCe2/0458	Multiple mitochondrial dysfunctions syndrome type 3	IBA57
sCe2/0459	Narcolepsy	HCRT
sCe2/0460	Neonatal death due Leigh syndrome, MT-TV related	MT-TV
sCe2/0461	Neurodegeneration with brain iron acculation type 5	WDR45
sCe2/0462	Neurodegeneration with brain iron accumulation type 4	C19orf12
sCe2/0463	Neurodegeneration with brain iron accumulation type 6	COASY
sCe2/0464	Niemann-Pick disease type C1	NPC1
sCe2/0465	Pantothenate kinase-associated neurodegeneration	PANK2
sCe2/0466	Parietal foramina type 2	ALX4
sCe2/0467	Pelizaeus-Merzbacher disease	SLC16A2
sCe2/0468	Pelizaeus-Merzbacher disease	PLP1
sCe2/0469	Periventricular heterotopia with microcephaly	ARFGEF2
sCe2/0470	Peroxisome biogenesis disorder 14B	PEX11B
sCe2/0471	Peroxisome biogenesis disorder type 10A	PEX3
sCe2/0472	Peroxisome biogenesis disorder type 1B	PEX1
sCe2/0473	Peroxisome biogenesis disorder type 2A	PEX5
sCe2/0474	Peroxisome biogenesis disorder type 2B	PEX5
sCe2/0475	Perrault syndrome	HSD17B4
sCe2/0476	Perrault syndrome type 5	TWNK
sCe2/0477	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	TREM2
sCe2/0478	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	TYROBP
sCe2/0479	Polyglucosan body myopathy type 1 with or without immunodeficiency	RBCK1
sCe2/0480	Pontocerebellar hypoplasia type 1A	VRK1
sCe2/0481	Pontocerebellar hypoplasia type 1B	EXOSC3
sCe2/0482	Pontocerebellar hypoplasia type 2A	TSEN54
sCe2/0483	Pontocerebellar hypoplasia type 2B	TSEN2

sCe2/0484	Pontocerebellar hypoplasia type 2C	TSEN34
sCe2/0485	Pontocerebellar hypoplasia type 2D	SEPSECS
sCe2/0486	Pontocerebellar hypoplasia type 2E	VPS53
sCe2/0487	Pontocerebellar hypoplasia type 4	TSEN54
sCe2/0488	Pontocerebellar hypoplasia type 5	TSEN54
sCe2/0489	Pontocerebellar hypoplasia type 6	RARS2
sCe2/0490	Pontocerebellar hypoplasia type 8	CHMP1A
sCe2/0491	Pontocerebellar hypoplasia, type 10	CLP1
sCe2/0492	Pontocerebellar hypoplasia, type 9	AMPD2
sCe2/0493	Porencephaly type 2	COL4A2
sCe2/0494	Progressive external ophthalmoplegia with mitochondrial deletions type 1	POLG
sCe2/0495	Progressive external ophthalmoplegia with mitochondrial deletions type 3	TWNK
sCe2/0496	Progressive external ophthalmoplegia with mitochondrial deletions type 4	POLG2
sCe2/0497	Progressive external ophthalmoplegia with mitochondrial deletions type 5	RRM2B
sCe2/0498	Pyruvate carboxylase deficiency	PC
sCe2/0499	Schizophrenia, CALR related	CALR
sCe2/0500	Schizophrenia, CELSR2 related	CELSR2
sCe2/0501	Schizophrenia, GRID2 related	GRID2
sCe2/0502	Schizophrenia, NOTCH4 related	NOTCH4
sCe2/0503	Sialuria, finish type	SLC17A5
sCe2/0504	Sjogren-Larsson syndrome	ALDH3A2
sCe2/0505	Striatal degeneration	PDE8B
sCe2/0506	Tourette syndrome	SLITRK1
sCe2/0507	Tuberous sclerosis	TSC1
sCe2/0508	Tuberous sclerosis type 2	TSC2
sCe2/0509	Waardenburg syndrome type 2D	SNAI2
sCe2/0510	Waardenburg syndrome/Hirschsprung disease	EDNRB
sCe2/0511	Warburg micro syndrome type 1	RAB3GAP1
sCe2/0512	Zellweger syndrome	PEX1

sCe2/0513	Zellweger syndrome	PEX2
sCe2/0514	Zellweger syndrome	PEX6
sCe2/0515	Zellweger syndrome	PEX10
sCe2/0516	Zellweger syndrome	PEX12
sCe2/0517	Zellweger syndrome	PEX13
sCe2/0518	Zellweger syndrome	PEX14
sCe2/0519	Zellweger syndrome	PEX16
sCe2/0520	Zellweger syndrome	PEX19
sCe2/0521	Zellweger syndrome	PEX26
sCe2/0522	Amyotrophic lateral sclerosis risk factor	CHGB
sCe2/0523	Amyotrophic lateral sclerosis type 1	SOD1
sCe2/0524	Amyotrophic lateral sclerosis type 10	TARDBP
sCe2/0525	Amyotrophic lateral sclerosis type 11	FIG4
sCe2/0526	Amyotrophic lateral sclerosis type 12	OPTN
sCe2/0527	Amyotrophic lateral sclerosis type 14	VCP
sCe2/0528	Amyotrophic lateral sclerosis type 16	SIGMAR1
sCe2/0529	Amyotrophic lateral sclerosis type 17	CHMP2B
sCe2/0530	Amyotrophic lateral sclerosis type 18	PFN1
sCe2/0531	Amyotrophic lateral sclerosis type 2, juvenile	ALS2
sCe2/0532	Amyotrophic lateral sclerosis type 21	MATR3
sCe2/0533	Amyotrophic lateral sclerosis type 4	SETX
sCe2/0534	Amyotrophic lateral sclerosis type 6	FUS
sCe2/0535	Amyotrophic lateral sclerosis type 8	VAPB
sCe2/0536	Amyotrophic lateral sclerosis type 9	ANG
sCe2/0537	Amyotrophic lateral sclerosis with frontotemporal dementia	C9orf72
sCe2/0538	Amyotrophic lateral sclerosis, CREST related	SS18L1
sCe2/0539	Amyotrophic lateral sclerosis, susceptibility to	NEFH
sCe2/0540	Amyotrophic lateral sclerosis, VPS54 related	VPS54
sCe2/0541	Amyotrophic lateral sclerosis, x-linked juvenile and adult-onset ALS	UBQLN2

sCe2/0542	Bethlem myopathy	COL6A2
sCe2/0543	Bethlem myopathy	COL6A1
sCe2/0544	Bethlem myopathy type 1	COL6A3
sCe2/0545	Bethlem myopathy type 2	COL12A1
sCe2/0546	Brody myopathy	ATP2A1
sCe2/0547	Cardiomyopathy, hypertrophic, type 24	LDB3
sCe2/0548	Central core disease	RYR1
sCe2/0549	Centronuclear myopathy type 1	MTMR14
sCe2/0550	Centronuclear myopathy type 3	MYF6
sCe2/0551	Centronuclear myopathy type 4	CCDC78
sCe2/0552	Compton-North congenital myopathy	CNTN1
sCe2/0553	Congenital muscular dystrophy and hypoglycosylation of α-dystroglycan	B3GALNT2
sCe2/0554	Creatine phosphokinase, elevated serum	CAV3
sCe2/0555	Dyssegmental dysplasia, Silverman-Handmaker type	HSPG2
sCe2/0556	Emery-Dreifuss muscular dystrophy type 1	EMD
sCe2/0557	Emery-Dreifuss muscular dystrophy type 2	LMNA
sCe2/0558	Emery-Dreifuss muscular dystrophy type 4	SYNE1
sCe2/0559	Emery-Dreifuss muscular dystrophy type 5	SYNE2
sCe2/0560	Emery-Dreifuss muscular dystrophy type 6	FHL1
sCe2/0561	Encephalopathy lethal, due to defective mitochondrial peroxisomal fission	DNM1L
sCe2/0562	Encephalopathy mitochondrial	VDAC1
sCe2/0563	Encephalopathy mitochondrial with proximal renal tubulopathy due to cytochrome c oxidase deficiency	COX10
sCe2/0564	Encephalopathy, familial, with neuroserpin inclusion bodies	SERPINI1
sCe2/0565	Endplate acetylcholinesterase deficiency	COLQ
sCe2/0566	Epidermolysis bullosa simplex with muscular dystrophy	PLEC
sCe2/0567	Epidermolysis bullosa simplex, autosomal recessive type 2	DST
sCe2/0568	Facioscapulohumeral dystrophy-like phenotype, FAT1 related	FAT1
sCe2/0569	Filaminopathy	FLNC
sCe2/0570	Fukuyama congenital muscular dystrophy	FKTN

sCe2/0571	Hereditary myopathy with early respiratory failure	TTN
sCe2/0572	Hyperkalemic periodic paralysis	SCN4A
sCe2/0573	Hypokalemic periodic paralysis type 1	CACNA1S
sCe2/0574	Inclusion body myopathy	GNE
sCe2/0575	Inclusion body myopathy	MYH2
sCe2/0576	King-Denborough syndrome	RYR1
sCe2/0577	Limb-girdle muscular dystrophy, autosomal dominant type 1A	MYOT
sCe2/0578	Limb-girdle muscular dystrophy, autosomal dominant type 1B	LMNA
sCe2/0579	Limb-girdle muscular dystrophy, autosomal dominant type 1C	CAV3
sCe2/0580	Limb-girdle muscular dystrophy, autosomal dominant type 1E	DNAJB6
sCe2/0581	Limb-girdle muscular dystrophy, autosomal recessice type 2F	SGCD
sCe2/0582	Limb-girdle muscular dystrophy, autosomal recessive type 12C	POMK
sCe2/0583	Limb-girdle muscular dystrophy, autosomal recessive type 2A	CAPN3
sCe2/0584	Limb-girdle muscular dystrophy, autosomal recessive type 2B	DYSF
sCe2/0585	Limb-girdle muscular dystrophy, autosomal recessive type 2C	SGCG
sCe2/0586	Limb-girdle muscular dystrophy, autosomal recessive type 2D	SGCA
sCe2/0587	Limb-girdle muscular dystrophy, autosomal recessive type 2E	SGCB
sCe2/0588	Limb-Girdle Muscular Dystrophy, autosomal recessive type 2G	TCAP
sCe2/0589	Limb-girdle muscular dystrophy, autosomal recessive type 2H	TRIM32
sCe2/0590	Limb-girdle muscular dystrophy, autosomal recessive type 2I	FKRP
sCe2/0591	Limb-girdle muscular dystrophy, autosomal recessive type 2J	TTN
sCe2/0592	Limb-girdle muscular dystrophy, autosomal recessive type 2K	POMT1
sCe2/0593	Limb-girdle muscular dystrophy, autosomal recessive type 2L	ANO5
sCe2/0594	Limb-girdle muscular dystrophy, autosomal recessive type 2M	FKTN
sCe2/0595	Limb-girdle muscular dystrophy, autosomal recessive type 2N	POMT1
sCe2/0596	Limb-girdle muscular dystrophy, autosomal recessive type 2S	TRAPPC11
sCe2/0597	Malignant hyperthermia type 5	CACNA1S
sCe2/0598	Marden-Walker syndrome	PIEZO2
sCe2/0599	MELAS syndrome, MT-TL1 related	MT-TL1

sCe2/0600	MERRF syndrome, MT-TK related	MT-TK
sCe2/0601	MERRF syndrome, MT-TP related	MT-TP
sCe2/0602	Minicore myopathy with external ophthalmoplegia	RYR1
sCe2/0603	Mitochondrial DNA depletion syndrome	DGUOK
sCe2/0604	Mitochondrial DNA depletion syndrome	TK2
sCe2/0605	Mitochondrial myopathy and sideroblastic anemia type 1	PUS1
sCe2/0606	Miyoshi muscular dystrophy type 3	ANO5
sCe2/0607	Miyoshi myopathy	DYSF
sCe2/0608	Multisystemic smooth muscle dysfunction syndrome	ACTA2
sCe2/0609	Muscle hypertrophy	MSTN
sCe2/0610	Muscle-eye-brain disease, POMK related	POMK
sCe2/0611	Muscular dystrophy type 1A	LAMA2
sCe2/0612	Muscular dystrophy type 1C	FKRP
sCe2/0613	Muscular dystrophy type 1D	LARGE
sCe2/0614	Muscular dystrophy, Becker type	DMD
sCe2/0615	Muscular dystrophy, congenital, LMNA related	LMNA
sCe2/0616	Muscular dystrophy, congenital, megaconial type	CHKB
sCe2/0617	Muscular dystrophy, Duchenne type	DMD
sCe2/0618	Muscular dystrophy, limb-girdle type 2A	CAPN3
sCe2/0619	Muscular dystrophy, limb-girdle, type 2Q	PLEC
sCe2/0620	Muscular dystrophy, oculopharyngeal	PABPN1
sCe2/0621	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A10	TMEM5
sCe2/0622	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A13	B4GAT1
sCe2/0623	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A2	POMT2
sCe2/0624	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A3	POMGNT1
sCe2/0625	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A8	POMGNT2

sCe2/0626	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B1	POMT1
sCe2/0627	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B2	POMT2
sCe2/0628	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B3	POMGNT1
sCe2/0629	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C2	POMT2
sCe2/0630	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C3	POMGNT1
sCe2/0631	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C9	DAG1
sCe2/0632	Muscular-skeletal disorder, CAPN1 related	CAPN1
sCe2/0633	Myasthenia congenital with tubular aggregates 1	GFPT1
sCe2/0634	Myasthenic syndrome associated with acetylcholine receptor deficiency	MUSK
sCe2/0635	Myasthenic syndrome due to mutation in SCN4A	SCN4A
sCe2/0636	Myasthenic syndrome fast channel congenital	CHRNA1
sCe2/0637	Myasthenic syndrome slow-channel congenital	CHRNA1
sCe2/0638	Myasthenic syndrome type 10, congenital	DOK7
sCe2/0639	Myasthenic syndrome type 11, congenital, associated with acetylcholine receptor deficiency	RAPSN
sCe2/0640	Myasthenic syndrome, congenital	CHRNB1
sCe2/0641	Myasthenic syndrome, congenital	CHAT
sCe2/0642	Myasthenic syndrome, congenital	CHRNE
sCe2/0643	Myasthenic syndrome, congenital	AGRN
sCe2/0644	Myasthenic syndrome, congenital, type 3C, associated with acetylcholine receptor deficiency	CHRND
sCe2/0645	Myasthenic syndrome, fast-channel, congenital, type 3B	CHRND
sCe2/0646	Myasthenic syndrome, slow channel, congenital, type 3A	CHRND
sCe2/0647	Myoglobinuria acute recurrent	LPIN1
sCe2/0648	Myopathy due to Integrin 7A deficiency	ITGA7
sCe2/0649	Myopathy due to myoadenylate deaminase deficiency	AMPD1
sCe2/0650	Myopathy with fiber-type disproportion	SELENON
sCe2/0651	Myopathy with fiber-type disproportion type 1	ACTA1

sCe2/0652	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset	MEGF10
sCe2/0653	Myopathy, centronuclear	BIN1
sCe2/0654	Myopathy, centronuclear	DNM2
sCe2/0655	Myopathy, COL6A6 related	COL6A6
sCe2/0656	Myopathy, desmin related, associated with mutation in the CRYAB gene	CRYAB
sCe2/0657	Myopathy, distal type 1	MYH7
sCe2/0658	Myopathy, distal type 4	FLNC
sCe2/0659	Myopathy, distal with anterior tibial onset	DYSF
sCe2/0660	Myopathy, distal, Tateyama type	CAV3
sCe2/0661	Myopathy, early-onset with fatal cardiomyopathy	TTN
sCe2/0662	Myopathy, lactic acidosis, and sideroblastic anemia type 2	YARS2
sCe2/0663	Myopathy, limb girdle with bone fragility	MTAP
sCe2/0664	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay	GFER
sCe2/0665	Myopathy, MT-TQ related	MT-TQ
sCe2/0666	Myopathy, myofibrillar type 6	BAG3
sCe2/0667	Myopathy, myofibrillar, Desmin related	DES
sCe2/0668	Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related	CRYAB
sCe2/0669	Myopathy, myofibrillar, ZASP related	LDB3
sCe2/0670	Myopathy, scapulohumeroperoneal	ACTA1
sCe2/0671	Myopathy, tubular aggregate, type 1	STIM1
sCe2/0672	Myosclerosis, autosomal recessive	COL6A2
sCe2/0673	Myosin storage myopathy	MYH7
sCe2/0674	Myotilinopathy	MYOT
sCe2/0675	Myotonia congenital	CLCN1
sCe2/0676	Myotonic dystrophy type 1	DMPK
sCe2/0677	Myotonic dystrophy type 2	CNBP
sCe2/0678	Myotubular myopathy X-linked	MTM1
sCe2/0679	Nemaline myopathy type 1	TPM3
sCe2/0680	Nemaline myopathy type 2, autosomal recessive	NEB

sCe2/0681	Nemaline myopathy type 3	ACTA1
sCe2/0682	Nemaline myopathy type 4	TPM2
sCe2/0683	Nemaline myopathy type 5	TNNT1
sCe2/0684	Nemaline myopathy type 6	KBTBD13
sCe2/0685	Nemaline myopathy type 7	CFL2
sCe2/0686	Neurogenic scapuloperoneal syndrome, Kaeser type	DES
sCe2/0687	Neuromyotonia and axonal neuropathy, autosomal recessive	HINT1
sCe2/0688	Neutral lipid storage disease with myopathy	PNPLA2
sCe2/0689	Nonaka myopathy	GNE
sCe2/0690	Paramyotonia congenita of von Eulenburg	SCN4A
sCe2/0691	Pompe disease	GAA
sCe2/0692	Potassium-aggravated myotonia	SCN4A
sCe2/0693	Primary lateral sclerosis, juvenile	ALS2
sCe2/0694	Progressive external ophthalmoplegia with mitochondrial deletions type 5	RRM2B
sCe2/0695	Ptosis, congenital	ZFHX4
sCe2/0696	Rigid spine muscular dystrophy	SELENON
sCe2/0697	Rippling muscle disease	CAV3
sCe2/0698	Scapuloperoneal myopathy, MYH7 related	MYH7
sCe2/0699	Schwartz-Jampel syndrome type 1	HSPG2
sCe2/0700	Spastic paralysis, infantile onset ascending	ALS2
sCe2/0701	Spheroid body myopathy	MYOT
sCe2/0702	Spinal muscular atrophy (SMA), NAIP related	NAIP
sCe2/0703	Spinal muscular atrophy distal, autosomal recessive type 4	PLEKHG5
sCe2/0704	Spinal muscular atrophy type 1	SMN1
sCe2/0705	Spinal muscular atrophy type 2	SMN1
sCe2/0706	Spinal muscular atrophy type 3	SMN1
sCe2/0707	Spinal muscular atrophy type 3, modifier of	SMN2
sCe2/0708	Spinal muscular atrophy type 4	SMN1
sCe2/0709	Spinal muscular atrophy type 5	DNAJB2

sCe2/0710	Spinal muscular atrophy with progressive myoclonic epilepsy	ASAH1
sCe2/0711	Spinal muscular atrophy, distal, X-linked	ATP7A
sCe2/0712	Spinal muscular atrophy, lower extremity, autosomal dominant, type 2	BICD2
sCe2/0713	Spinal muscular atrophy, lower extremity-predominant type 1, autosomal dominant	DYNC1H1
sCe2/0714	Thyrotoxic periodic paralysis type 1	CACNA1S
sCe2/0715	Thyrotoxic periodic paralysis type 2	KCNJ18
sCe2/0716	Tibial muscular dystrophy, tardive	TTN
sCe2/0717	Ullrich congenital muscular dystrophy	COL6A2
sCe2/0718	Ullrich congenital muscular dystrophy	COL6A1
sCe2/0719	Ullrich congenital muscular dystrophy type 1	COL6A3
sCe2/0720	Ullrich congenital muscular dystrophy type 2	COL12A1
sCe2/0721	Walker-Warburg syndrome	ISPD
sCe2/0722	Walker-Warburg syndrome	FKTN
sCe2/0723	Walker-Warburg syndrome or muscle-eye-brain disease, FKRP related	FKRP
sCe2/0724	Wieacker-Wolff syndrome	ZC4H2
sCe2/0725	Alzheimer disease type 1	APP
sCe2/0726	Alzheimer disease type 2	APOE
sCe2/0727	Alzheimer disease type 3	PSEN1
sCe2/0728	Alzheimer disease type 4	PSEN2
sCe2/0729	Alzheimers disease, early onset, autosomal dominant	SORL1
sCe2/0730	Alzheimers disease, RTN3 related	RTN3
sCe2/0731	Angelman syndrome	UBE3A
sCe2/0732	Angelman syndrome	chr. 15q11
sCe2/0733	Cerebral amyloid angiopathy	CST3
sCe2/0734	Cerebral amyloid angiopathy, APP related	APP
sCe2/0735	Creutzfeldt-Jakob disease	PRNP
sCe2/0736	Dementia, familial, British type	ITM2B
sCe2/0737	Dementia, familial, Danish type	ITM2B
sCe2/0738	Dementia, frontotemporal	GRN

sCe2/0739	Dementia, frontotemporal	MAPT
sCe2/0740	Dementia, frontotemporal	TARDBP
sCe2/0741	Dementia, frontotemporal	PSEN1
sCe2/0742	Dementia, Lewy body	SNCA
sCe2/0743	Fatal familial imsomnia	PRNP
sCe2/0744	Gerstmann-Straussler disease	PRNP
sCe2/0745	Hippocampal longterm potentiation, RFN39 related	RNF39
sCe2/0746	Lewy body dementia, susceptibility to	GBA
sCe2/0747	Peripheral demyelinating neuropathy Waardenburg syndrome and Hirschsprung disease	SOX10
sCe2/0748	Pick disease	PSEN1
sCe2/0749	Prader-Willi syndrome	SNRPN
sCe2/0750	Prader-Willi syndrome	NDN
sCe2/0751	Prader-Willi syndrome	chr. 15q11
sCe2/0752	Amish infantile epilepsy syndrome	ST3GAL5
sCe2/0753	Convulsions, benign familial infantile, 3	SCN2A
sCe2/0754	Cortical dysplasia-focal epilepsy syndrome	CNTNAP2
sCe2/0755	Dravet syndrome	GABRG2
sCe2/0756	Dravet syndrome	SCN2A
sCe2/0757	Dravet syndrome, modifier of	SCN9A
sCe2/0758	Early infantile epileptic encephalopathy type 1	ARX
sCe2/0759	Early infantile epileptic encephalopathy type 10	PNKP
sCe2/0760	Early infantile epileptic encephalopathy type 11	SCN2A
sCe2/0761	Early infantile epileptic encephalopathy type 12	PLCB1
sCe2/0762	Early infantile epileptic encephalopathy type 13	SCN8A
sCe2/0763	Early infantile epileptic encephalopathy type 14	KCNT1
sCe2/0764	Early infantile epileptic encephalopathy type 15	ST3GAL3
sCe2/0765	Early infantile epileptic encephalopathy type 16	TBC1D24
sCe2/0766	Early infantile epileptic encephalopathy type 17	GNAO1
sCe2/0767	Early infantile epileptic encephalopathy type 19	GABRA1

sCe2/0768	Early infantile epileptic encephalopathy type 2	CDKL5
sCe2/0769	Early infantile epileptic encephalopathy type 20	PIGA
sCe2/0770	Early infantile epileptic encephalopathy type 21	NECAP1
sCe2/0771	Early infantile epileptic encephalopathy type 23	DOCK7
sCe2/0772	Early infantile epileptic encephalopathy type 24	HCN1
sCe2/0773	Early infantile epileptic encephalopathy type 25	SLC13A5
sCe2/0774	Early infantile epileptic encephalopathy type 26	KCNB1
sCe2/0775	Early infantile epileptic encephalopathy type 27	GRIN2B
sCe2/0776	Early infantile epileptic encephalopathy type 28	WWOX
sCe2/0777	Early infantile epileptic encephalopathy type 29	AARS
sCe2/0778	Early infantile epileptic encephalopathy type 3	SLC25A22
sCe2/0779	Early infantile epileptic encephalopathy type 30	SIK1
sCe2/0780	Early infantile epileptic encephalopathy type 31	DNM1
sCe2/0781	Early infantile epileptic encephalopathy type 32	KCNA2
sCe2/0782	Early infantile epileptic encephalopathy type 33	EEF1A2
sCe2/0783	Early infantile epileptic encephalopathy type 4	STXBP1
sCe2/0784	Early infantile epileptic encephalopathy type 40	GUF1
sCe2/0785	Early infantile epileptic encephalopathy type 45	GABRB1
sCe2/0786	Early infantile epileptic encephalopathy type 46	GRIN2D
sCe2/0787	Early infantile epileptic encephalopathy type 47	FGF12
sCe2/0788	Early infantile epileptic encephalopathy type 5	SPTAN1
sCe2/0789	Early infantile epileptic encephalopathy type 6	SCN1A
sCe2/0790	Early infantile epileptic encephalopathy type 7	KCNQ2
sCe2/0791	Early infantile epileptic encephalopathy type 8	ARHGEF9
sCe2/0792	Early infantile epileptic encephalopathy type 9	PCDH19
sCe2/0793	Epilepsy with neurodevelopmental defects	GRIN2A
sCe2/0794	Epilepsy, childhood absence type 2	GABRG2
sCe2/0795	Epilepsy, childhood absence type 4, susceptibility to	GABRA1
sCe2/0796	Epilepsy, childhood absence type 5	GABRB3

sCe2/0797	Epilepsy, childhood absence type 6, susceptibility to	CACNA1H
sCe2/0798	Epilepsy, childhood absence, JRK related	JRK
sCe2/0799	Epilepsy, familial focal with variable foci	DEPDC5
sCe2/0800	Epilepsy, familial temporal lobe type 1	LGI1
sCe2/0801	Epilepsy, familial temporal lobe type 5	CPA6
sCe2/0802	Epilepsy, familial temporal lobe type 7	RELN
sCe2/0803	Epilepsy, focal, SCN3A related	SCN3A
sCe2/0804	Epilepsy, HCN2 related	HCN2
sCe2/0805	Epilepsy, hearing loss, and mental retardation syndrome	SPATA5
sCe2/0806	Epilepsy, idiopathic generalized type 10	GABRD
sCe2/0807	Epilepsy, idiopathic generalized type 11	CLCN2
sCe2/0808	Epilepsy, idiopathic generalized type 12	SLC2A1
sCe2/0809	Epilepsy, juvenile absence type 1	EFHC1
sCe2/0810	Epilepsy, nocturnal frontal lobe	KCNT1
sCe2/0811	Epilepsy, nocturnal frontal lobe type 1	CHRNA4
sCe2/0812	Epilepsy, nocturnal frontal lobe type 3	CHRNB2
sCe2/0813	Epilepsy, nocturnal frontal lobe type 4	CHRNA2
sCe2/0814	Epilepsy, progressive myoclonic 4, with or without renal failure	SCARB2
sCe2/0815	Epilepsy, progressive myoclonic type 5	PRICKLE2
sCe2/0816	Epilepsy, X-linked, with learning disabilities and behavior disorders	SYN1
sCe2/0817	Epileptic encephalopathy, childhood-onset	CHD2
sCe2/0818	Familial infantile myoclonic epilepsy	TBC1D24
sCe2/0819	Febrile seizures, familial, type 4	ADGRV1
sCe2/0820	Generalized epilepsy and paroxysmal dyskinesia	KCNMA1
sCe2/0821	Generalized epilepsy with febrile seizures plus type 1	SCN1B
sCe2/0822	Generalized epilepsy with febrile seizures plus type 2	SCN1A
sCe2/0823	Generalized epilepsy with febrile seizures plus type 3	GABRG2
sCe2/0824	Generalized epilepsy with febrile seizures plus type 7	SCN9A
sCe2/0825	GLUT1 deficiency syndrome type 1	SLC2A1

sCe2/0826	Hyperekplexia	SLC6A5
sCe2/0827	Hyperekplexia	GLRB
sCe2/0828	Hyperekplexia	GLRA1
sCe2/0829	Hyperekplexia, EIEE8 related	ARHGEF9
sCe2/0830	Kohlschutter Tonz syndrome	ROGDI
sCe2/0831	MERRF syndrome, MT-TK related	MT-TK
sCe2/0832	MERRF syndrome, MT-TP related	MT-TP
sCe2/0833	Myoclonic epilepsy of Lafora	EPM2A
sCe2/0834	Myoclonic epilepsy of Lafora	NHLRC1
sCe2/0835	Myoclonus, familial cortical	NOL3
sCe2/0836	Paroxysmal exercise-induced dyskinesia with epilepsy and/or hemolytic anemia	SLC2A1
sCe2/0837	Polymicrogyria bilateral occipital	NR2E1
sCe2/0838	Progressive myoclonus epilepsy type 1A	PRICKLE1
sCe2/0839	Progressive myoclonus epilepsy type 3	KCTD7
sCe2/0840	Progressive myoclonus epilepsy type 6	GOSR2
sCe2/0841	Progressive myoclonus epilepsy type 8	CERS1
sCe2/0842	Pyridoxine-dependent epilepsy	ALDH7A1
sCe2/0843	Seizures, benign familial infantile, type 2	PRRT2
sCe2/0844	Seizures, benign neonatal, type 1	KCNQ2
sCe2/0845	Seizures, benign neonatal, type 2	KCNQ3
sCe2/0846	SESAME syndrome	KCNJ10
sCe2/0847	Unverricht-Lundborg disease	CSTB
sCe2/0848	Cytochrome c oxidase 1 deficiency	MT-CO1
sCe2/0849	Cytochrome c oxidase 2 deficiency	MT-CO2
sCe2/0850	Cytochrome c oxidase 3 deficiency	MT-CO3
sCe2/0851	Encephalomyopathy, mitochondrial, MT-TR related	MT-TR
sCe2/0852	Encephalopathy, mitochondrial, MT-TW related	MT-TW
sCe2/0853	MERRF syndrome, MT-TK related	MT-TK
sCe2/0854	MERRF syndrome, MT-TP related	MT-TP

sCe2/0855	MERRF/MELAS overlap syndrome, MT-TS1 related	MT-TS1
sCe2/0856	MERRF/MELAS overlap syndrome, MT-TS2 related	MT-TS2
sCe2/0857	Mitochondrial complex I deficiency	MT-ND4
sCe2/0858	Mitochondrial complex I deficiency	MT-ND3
sCe2/0859	Mitochondrial complex I deficiency	MT-ND2
sCe2/0860	Mitochondrial complex I deficiency	MT-ND6
sCe2/0861	Mitochondrial complex I deficiency	MT-ND1
sCe2/0862	Mitochondrial complex I deficiency	NDUFB3
sCe2/0863	Mitochondrial complex I deficiency	MT-ND5
sCe2/0864	Mitochondrial complex I deficiency	MT-ND4L
sCe2/0865	Mitochondrial complex II deficiency	SDHD
sCe2/0866	Mitochondrial complex III deficiency, nuclear type 7	UQCC2
sCe2/0867	Mitochondrial complex IV deficiency	PET100
sCe2/0868	Mitochondrial complex IV deficiency	APOPT1
sCe2/0869	Mitochondrial complex IV deficiency	MT-CO3
sCe2/0870	Mitochondrial complex V deficiency, nuclear type 4	ATP5A1
sCe2/0871	Mitochondrial Disorders, AKAP1 related	AKAP1
sCe2/0872	Mitochondrial DNA depletion syndrome type 6	MPV17
sCe2/0873	Mitochondrial myopathy and sideroblastic anemia type 1	PUS1
sCe2/0874	Mitochondrial neurogastrointestinal encephalopathy syndrome without leukoencephalopathy	TYMP
sCe2/0875	Mitochondrial respiratory chain complex II deficiency	SDHA
sCe2/0876	Mitochondrial respiratory chain disease, TIMM21 related	TIMM21
sCe2/0877	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	ECHS1
sCe2/0878	Multiple mitochondrial dysfunctions syndrome type 4	ISCA2
sCe2/0879	Progressive external ophthalmoplegia with mitochondrial deletions type 2	SLC25A4
sCe2/0880	Progressive external ophthalmoplegia with mitochondrial deletions type 3	TWNK
sCe2/0881	Progressive external ophthalmoplegia with mitochondrial deletions type 5	RRM2B
sCe2/0882	Thiamine metabolism dysfunction syndrome 4 progressive polyneuropathy type	SLC25A19

sCe2/0883	Adrenoleukodystrophy/Adrenomyeloneuropathy	ABCD1
sCe2/0884	Allan-Herndon-Dudley syndrome	SLC16A2
sCe2/0885	Alpha-thalassemia/mental retardation syndrome	ATRX
sCe2/0886	Al-Raqad syndrome	DCPS
sCe2/0887	Angelman-like syndrome	CDKL5
sCe2/0888	Angelman-like syndrome	MECP2
sCe2/0889	Arts syndrome	PRPS1
sCe2/0890	Attention deficit-hyperactivity disorder	DRD5
sCe2/0891	Attention deficit-hyperactivity disorder	DRD4
sCe2/0892	Autism spectrum disorder	RABGGTA
sCe2/0893	Autism spectrum disorder	ANKS3
sCe2/0894	Autism spectrum disorder	BPIFA3
sCe2/0895	Autism spectrum disorder	AHNAK2
sCe2/0896	Autism spectrum disorder	EN2
sCe2/0897	Autism spectrum disorder, MYO16 related	MYO16
sCe2/0898	Autism spectrum, MXRA5 related	MXRA5
sCe2/0899	Autism spectrum/ hyperactivity/ bipolar disorder, GRM7 related	GRM7
sCe2/0900	Autism susceptibility, X-linked type 1	NLGN3
sCe2/0901	Autism susceptibility, X-linked type 17	SHANK2
sCe2/0902	Autism susceptibility, X-linked type 2	NLGN4X
sCe2/0903	Autism susceptibility, X-linked type 3	MECP2
sCe2/0904	Autism susceptibility, X-linked type 4	PTCHD1
sCe2/0905	Autism susceptibility, X-linked type 5	RPL10
sCe2/0906	Autism, ATP1B4 related	ATP1B4
sCe2/0907	Autism, AVPR1A related	AVPR1A
sCe2/0908	Autism, C7orf43 related	C7orf43
sCe2/0909	Autism, CELF6 related	CELF6
sCe2/0910	Autism, EFCAB13 related	EFCAB13
sCe2/0911	Autism, FAAH2 related	FAAH2

sCe2/0912	Autism, FCRL6 related	FCRL6
sCe2/0913	Autism, GYG2 related	GYG2
sCe2/0914	Autism, IQCE related	IQCE
sCe2/0915	Autism, MBD1 related	MBD1
sCe2/0916	Autism, NTNG1 related	NTNG1
sCe2/0917	Autism, OR13H1 related	OR13H1
sCe2/0918	Autism, OXTR related	OXTR
sCe2/0919	Autism, PKHD1L1 related	PKHD1L1
sCe2/0920	Autism, RNF128 related	RNF128
sCe2/0921	Autism, RRM1 related	RRM1
sCe2/0922	Autism, SETD2 related	SETD2
sCe2/0923	Autism, SLC22A9 related	SLC22A9
sCe2/0924	Autism, UNC13B related	UNC13B
sCe2/0925	Autism, ZNF778 related	ZNF778
sCe2/0926	Autism/Mental retardation/Angelman syndrome, susceptibility to, ATP10A related	ATP10A
sCe2/0927	Borjeson-Forssman-Lehmann syndrome	PHF6
sCe2/0928	Brunner syndrome	MAOA
sCe2/0929	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome type 4	ATP8A2
sCe2/0930	CHILD syndrome	NSDHL
sCe2/0931	CK syndrome	NSDHL
sCe2/0932	Coffin-Lowry syndrome	RPS6KA3
sCe2/0933	Cortical dysplasia, complex, with other brain malformations, type 1	TUBB3
sCe2/0934	Cortical dysplasia, complex, with other brain malformations, type 5	TUBB2A
sCe2/0935	Creatine deficiency syndrome X-linked	SLC6A8
sCe2/0936	Danon disease	LAMP2
sCe2/0937	Dent disease type 2	OCRL
sCe2/0938	Developmental delay and microcephaly, SLC1A4 related	SLC1A4
sCe2/0939	Developmental delay, GNAQ related	GNAQ
sCe2/0940	Developmental delay, KMT2C related	KMT2C

sCe2/0941	Dyslexia	PCDH11X
sCe2/0942	FG syndrome type 1	MED12
sCe2/0943	FG syndrome type 2	FLNA
sCe2/0944	FG syndrome type 4	CASK
sCe2/0945	Fragile X tremor/ataxia syndrome	FMR1
sCe2/0946	Frontometaphyseal dysplasia	FLNA
sCe2/0947	Gillespie syndrome	ITPR1
sCe2/0948	Heterotopia, periventricular, ED variant	FLNA
sCe2/0949	Heterotopia, periventricular, X-linked dominant	FLNA
sCe2/0950	Hoyeraal-Hreidarsson syndrome	DKC1
sCe2/0951	Intellectual disability nonsyndromic, CIC related	CIC
sCe2/0952	Intellectual disability nonsyndromic, CNKSR2 related	CNKSR2
sCe2/0953	Intellectual disability, TBR1 related	TBR1
sCe2/0954	Intestinal pseudoobstraction, neuronal	FLNA
sCe2/0955	Jensen syndrome	TIMM8A
sCe2/0956	Kabuki syndrome type 2	KDM6A
sCe2/0957	Leigh syndrome, X-linked	PDHA1
sCe2/0958	Lesch-Nyham syndrome	HPRT1
sCe2/0959	Lissencephaly/Subcortical laminal heteropia, X-linked	DCX
sCe2/0960	Lowe oculocerebrorenal syndrome	OCRL
sCe2/0961	Major affective disorder	CUX2
sCe2/0962	MASA syndrome	L1CAM
sCe2/0963	Melnick-Needles syndrome	FLNA
sCe2/0964	Mental retardation and microcephaly with pontine and cerebellar hypoplasia	CASK
sCe2/0965	Mental retardation non-syndromic	KLF8
sCe2/0966	Mental retardation non-syndromic	NXF5
sCe2/0967	Mental retardation non-syndromic	ZCCHC12
sCe2/0968	Mental retardation non-syndromic	ELK1
sCe2/0969	Mental retardation with Cerebellar ataxia and dysequilibrium syndrome type 2	WDR81

sCe2/0970	Mental retardation with hypotonic facies syndrome, X-linked	ATRX
sCe2/0971	Mental retardation X-linked, SMARCA1 related	SMARCA1
sCe2/0972	Mental retardation X-linked, syndromic, Claes-Jensen type	KDM5C
sCe2/0973	Mental retardation X-linked, syndromic, Lubs type	MECP2
sCe2/0974	Mental retardation, autosomal dominant type 1	MBD5
sCe2/0975	Mental retardation, autosomal dominant type 12	ARID1B
sCe2/0976	Mental retardation, autosomal dominant type 13	TRAPPC9
sCe2/0977	Mental retardation, autosomal dominant type 13	DYNC1H1
sCe2/0978	Mental retardation, autosomal dominant type 14	ARID1A
sCe2/0979	Mental retardation, autosomal dominant type 15	SMARCB1
sCe2/0980	Mental retardation, autosomal dominant type 16	SMARCA4
sCe2/0981	Mental retardation, autosomal dominant type 17	PACS1
sCe2/0982	Mental retardation, autosomal dominant type 18	GATAD2B
sCe2/0983	Mental retardation, autosomal dominant type 19	CTNNB1
sCe2/0984	Mental retardation, autosomal dominant type 2	DOCK8
sCe2/0985	Mental retardation, autosomal dominant type 20	MEF2C
sCe2/0986	Mental retardation, autosomal dominant type 23	SETD5
sCe2/0987	Mental retardation, autosomal dominant type 24	DEAF1
sCe2/0988	Mental retardation, autosomal dominant type 25	AHDC1
sCe2/0989	Mental retardation, autosomal dominant type 27	SOX11
sCe2/0990	Mental retardation, autosomal dominant type 28	ADNP
sCe2/0991	Mental retardation, autosomal dominant type 31	PURA
sCe2/0992	Mental retardation, autosomal dominant type 32	KAT6A
sCe2/0993	Mental retardation, autosomal dominant type 37	POGZ
sCe2/0994	Mental retardation, autosomal dominant type 38	EEF1A2
sCe2/0995	Mental retardation, autosomal dominant type 5	SYNGAP1
sCe2/0996	Mental retardation, autosomal dominant type 6	GRIN2B
sCe2/0997	Mental retardation, autosomal dominant type 7	DYRK1A
sCe2/0998	Mental retardation, autosomal dominant type 8	GRIN1

sCe2/0999	Mental retardation, autosomal dominant type 9	KIF1A
sCe2/1000	Mental retardation, autosomal recessive type 12	ST3GAL3
sCe2/1001	Mental retardation, autosomal recessive type 14	TECR
sCe2/1002	Mental retardation, autosomal recessive type 15	MAN1B1
sCe2/1003	Mental retardation, autosomal recessive type 18	MED23
sCe2/1004	Mental retardation, autosomal recessive type 2	CRBN
sCe2/1005	Mental retardation, autosomal recessive type 27	LINS1
sCe2/1006	Mental retardation, autosomal recessive type 3	CC2D1A
sCe2/1007	Mental retardation, autosomal recessive type 36	ADAT3
sCe2/1008	Mental retardation, autosomal recessive type 37	ANK3
sCe2/1009	Mental retardation, autosomal recessive type 38	HERC2
sCe2/1010	Mental retardation, autosomal recessive type 39	TTI2
sCe2/1011	Mental retardation, autosomal recessive type 41	KPTN
sCe2/1012	Mental retardation, autosomal recessive type 42	PGAP1
sCe2/1013	Mental retardation, autosomal recessive type 46	NDST1
sCe2/1014	Mental retardation, autosomal recessive type 49	GPT2
sCe2/1015	Mental retardation, autosomal recessive type 5	NSUN2
sCe2/1016	Mental retardation, autosomal recessive type 55	PUS3
sCe2/1017	Mental retardation, autosomal recessive type 7	TUSC3
sCe2/1018	Mental retardation, X-linked	RAB40AL
sCe2/1019	Mental retardation, X-linked syndromic, Christianson type	SLC9A6
sCe2/1020	Mental retardation, X-linked syndromic, Nascimento-type	UBE2A
sCe2/1021	Mental retardation, X-linked syndromic, Raymond type	ZDHHC9
sCe2/1022	Mental retardation, X-linked syndromic, Turner type	HUWE1
sCe2/1023	Mental retardation, X-linked type 1	IQSEC2
sCe2/1024	Mental retardation, X-linked type 101	MID2
sCe2/1025	Mental retardation, X-linked type 102	DDX3X
sCe2/1026	Mental retardation, X-Linked type 13	MECP2
sCe2/1027	Mental retardation, X-linked type 14	UPF3B

sCe2/1028	Mental retardation, X-linked type 15	CUL4B
sCe2/1029	Mental retardation, X-linked type 16	FGD1
sCe2/1030	Mental retardation, X-linked type 17	HSD17B10
sCe2/1031	Mental retardation, X-linked type 19	RPS6KA3
sCe2/1032	Mental retardation, X-linked type 21	IL1RAPL1
sCe2/1033	Mental retardation, X-linked type 29	ARX
sCe2/1034	Mental retardation, X-linked type 3	HCFC1
sCe2/1035	Mental retardation, X-linked type 30	PAK3
sCe2/1036	Mental retardation, X-linked type 32	CLIC2
sCe2/1037	Mental retardation, X-linked type 41	GDI1
sCe2/1038	Mental retardation, X-linked type 44	FTSJ1
sCe2/1039	Mental retardation, X-linked type 45	ZNF81
sCe2/1040	Mental retardation, X-linked type 46	ARHGEF6
sCe2/1041	Mental retardation, X-linked type 58	TSPAN7
sCe2/1042	Mental retardation, X-linked type 59	AP1S2
sCe2/1043	Mental retardation, X-linked type 63	ACSL4
sCe2/1044	Mental retardation, X-linked type 72	RAB39B
sCe2/1045	Mental retardation, X-linked type 88, AGTR2 related	AGTR2
sCe2/1046	Mental retardation, X-linked type 89	ZNF41
sCe2/1047	Mental retardation, X-linked type 90	DLG3
sCe2/1048	Mental retardation, X-linked type 91	ZDHHC15
sCe2/1049	Mental retardation, X-linked type 92	ZNF674
sCe2/1050	Mental retardation, X-linked type 93	BRWD3
sCe2/1051	Mental retardation, X-linked type 94	GRIA3
sCe2/1052	Mental retardation, X-linked type 95	MAGT1
sCe2/1053	Mental retardation, X-linked type 96	SYP
sCe2/1054	Mental retardation, X-linked type 97	ZNF711
sCe2/1055	Mental retardation, X-linked type 99	USP9X
sCe2/1056	Mental retardation, X-linked with epilepsy	ATP6AP2

sCe2/1057	Mental retardation, X-linked, associated with fragile site FRAXE	AFF2
sCe2/1058	Mental retardation, x-linked, EFHC2 related	EFHC2
sCe2/1059	Mental retardation, X-linked, nonsyndromic	NEXMIF
sCe2/1060	Mental retardation, X-linked, Siderius type	PHF8
sCe2/1061	Mental retardation, X-linked, Snyder-Robinson type	SMS
sCe2/1062	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	OPHN1
sCe2/1063	Mental retardation, X-linked, with isolated growth hormone deficiency	SOX3
sCe2/1064	Methionine adenosyltransferase deficiency, autosomal recessive	MAT1A
sCe2/1065	Microcephaly-capillary malformation syndrome	STAMBP
sCe2/1066	Mitochondrial complex I deficiency	NDUFA1
sCe2/1067	Mitochondrial complex I deficiency	NDUFB3
sCe2/1068	Neurodegeneration with brain iron accumulation, GTPBP2 related	GTPBP2
sCe2/1069	Neurodevelopmental disorder, ADAM22 related	ADAM22
sCe2/1070	Neurodevelopmental disorder, APC2-related	APC2
sCe2/1071	Neurodevelopmental disorder, CNTNAP4 related	CNTNAP4
sCe2/1072	Neurodevelopmental disorder, CROCC related	CROCC
sCe2/1073	Neurodevelopmental disorder, FRMPD4 related	FRMPD4
sCe2/1074	Neurodevelopmental disorder, KCTD3 related	KCTD3
sCe2/1075	Neurodevelopmental disorder, MACF1 related	MACF1
sCe2/1076	Neurodevelopmental disorder, MTOR related	MTOR
sCe2/1077	Neurodevelopmental disorder, NCAM1 related	NCAM1
sCe2/1078	Neurodevelopmental disorder, NGEF related	NGEF
sCe2/1079	Neurodevelopmental disorder, PIGQ related	PIGQ
sCe2/1080	Neurodevelopmental disorder, TUBB related	TUBB
sCe2/1081	Neurodevelopmental disorder, ZNF311 related	ZNF311
sCe2/1082	Neurodevelopmental malformation and microcephaly	KIF2A
sCe2/1083	Neurodevelopmental malformation and microcephaly	KIF5C
sCe2/1084	Neurodevelopmental malformation and microcephaly	TUBG1
sCe2/1085	Neuronal migration disorder	EOMES

sCe2/1086	Neuronal migration disorder	SPTBN5
sCe2/1087	Neuronal migration disorder	SRGAP2
sCe2/1088	Norrie disease	NDP
sCe2/1089	Occipital horn syndrome	ATP7A
sCe2/1090	Opitz G syndrome	MID1
sCe2/1091	Oral-facial-digital syndrome type 1	OFD1
sCe2/1092	Partington syndrome	ARX
sCe2/1093	Phosphoglycerate kinase 1 deficiency	PGK1
sCe2/1094	Psychomotor retardation	TANC1
sCe2/1095	Renpenning syndrome	PQBP1
sCe2/1096	Rett syndrome preserved speech variant	MECP2
sCe2/1097	Rolandic epilepsy, mental retardation, and speech dyspraxia	SRPX2
sCe2/1098	Simpson-Golabi-Behmel syndrome type 1	GPC3
sCe2/1099	Smith-Magenis syndrome	RAI1
sCe2/1100	Smith-Magenis syndrome, ULK2 related	ULK2
sCe2/1101	Stocco dos Santos X-linked mental retardation syndrome	SHROOM4
sCe2/1102	Urocanase deficiency	UROC1
sCe2/1103	Ventriculomegaly with cystic kidney disease	CRB2
sCe2/1104	WDR27-related brain disorders	WDR27

Panel

Code	product name	TAT-day
	Clinical Exome Whole exome Solo	
pCe2/0001	Aicardi-Goutieres syndrome panel	25
pCe2/0002	AllNeuro panel	25
pCe2/0003	Alzheimer dementia and dementia panel	25
pCe2/0004	Amyotrophic lateral sclerosis (ALS) panel	25
pCe2/0005	Bethlem myopathy panel	25
pCe2/0006	Cerebellar ataxia panel	25
pCe2/0007	CMT neuropathy panel	25
pCe2/0008	Comprehensive dystonia panel	25
pCe2/0009	Comprehensive epilepsy panel	25
pCe2/0010	Congenital myasthenic syndrome panel	25
pCe2/0011	Congenital myopathy panel	25
pCe2/0012	Dejerine-Sottas syndrome panel	25
pCe2/0013	Dementia panel	25
pCe2/0014	Dopa-responsive dystonia panel	25
pCe2/0015	Dravet syndrome panel	25
pCe2/0016	Early infantile epileptic encephalopathy panel	25
pCe2/0017	Epilepsy (absence) in childhood panel	25
pCe2/0018	Epilepsy (generalized) with febrile seizures panel	25
pCe2/0019	Epilepsy (partial) hereditary panel	25
pCe2/0020	Epileptic encephalopathy panel	25
pCe2/0021	Episodic ataxia panel	25
pCe2/0022	Familial hemiplegic migraine panel	25
pCe2/0023	Frontotemporal dementia panel	25
pCe2/0024	Hyperekplexia panel	25
pCe2/0025	Joubert syndrome panel	25
pCe2/0026	Leukodystrophy and peroxisome biogenesis disorders panel	25

pCe2/0027	Malignant hyperthermia panel	25
pCe2/0028	Mental retardation, X-linked panel	25
pCe2/0029	Metabolic myopathies panel	25
pCe2/0030	Muscular dystrophy panel	25
pCe2/0031	Muscular dystrophy-dystroglycanopathy type A panel	25
pCe2/0032	Myoclonic dystonia panel	25
pCe2/0033	Myoclonic epilepsy panel	25
pCe2/0034	Myofibrillar myopathy panel	25
pCe2/0035	Myopathy-rhabdomyolysis syndrome panel	25
pCe2/0036	Nemaline myopathy panel	25
pCe2/0037	Neuronal migration disorders panel	25
pCe2/0038	Non-dystrophic myotonia congenita panel	25
pCe2/0039	Parkinson disease panel	25
pCe2/0040	Pontocerebellar hypoplasia panel	25
pCe2/0041	SCA comprehensive panel	25
pCe2/0042	SCA repeat expansion panel	25
pCe2/0043	SCA sequencing panel	25
pCe2/0044	SMN negative spinal muscular atrophy panel	25
pCe2/0045	Spastic paraplegia panel complete	25
pCe2/0046	Spastic paraplegia panel, autosomal dominant	25
pCe2/0047	Spastic paraplegia panel, autosomal recessive	25
pCe2/0048	Syndromic autism panel	25
pCe2/0049	Ullrich muscular dystrophy panel	25