Movement Disorders panel

versie V2 (269 genen) Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
AARS1 (AARS)	601065	Epileptic encephalopathy, early infantile, 29, 616339 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant
ACTB	102630	?Dystonia, juvenile-onset, 607371 (3), Autosomal dominant; Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant
ACTL6B	612458	Intellectual developmental disorder with severe speech and ambulation defects, 618470 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 76, 618468 (3), Autosomal recessive
ADAR	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
ADCY5	600293	Dyskinesia, familial, with facial myokymia, 606703 (3), Autosomal dominant
ADH1C	103730	{Parkinson disease, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; {Alcohol dependence, protection against}, 103780 (3), Multifactorial
AFG3L2	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3); Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
ALDH5A1	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
ANO3	610110	Dystonia 24, 615034 (3), Autosomal dominant
AP3D1	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
AP4M1	602296	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
APTX	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
ARSA	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
ARV1	611647	Epileptic encephalopathy, early infantile, 38, 617020 (3), Autosomal recessive
ARX	300382	Epileptic encephalopathy, early infantile, 1, 308350 (3), X-linked recessive; Lissencephaly, X-linked 2, 300215 (3), X-linked; Proud syndrome, 300004 (3), X-linked; Mental retardation, X-linked 29 and others, 300419 (3), X-linked recessive; Partington syndrome, 309510 (3), X-linked recessive; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked

ATM	607585	Lymphoma, mantle cell, somatic (3); Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; T-cell prolymphocytic leukemia, somatic (3)
ATP13A2	610513	Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive; Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive
ATP1A3	182350	CAPOS syndrome, 601338 (3), Autosomal dominant; Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant
ATP6AP2	300556	Congenital disorder of glycosylation, type IIr, 301045 (3), X-linked recessive; Mental retardation, X-linked, syndromic, Hedera type, 300423 (3), X-linked recessive; ?Parkinsonism with spasticity, X-linked, 300911 (3), X-linked recessive
ATP7B	606882	Wilson disease, 277900 (3), Autosomal recessive
ATP8A2	605870	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
ATXN2	601517	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 (3), Autosomal dominant; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; Spinocerebellar ataxia 2, 183090 (3), Autosomal dominant
AUH	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
BCAP31	300398	Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive
BCS1L	603647	Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; GRACILE syndrome, 603358 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive
C19orf12	614297	Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal dominant, Autosomal recessive; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive
CACNA1A	601011	Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 42, 617106 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant
CACNA1B	601012	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497 (3), Autosomal recessive
CACNA1E	601013	Epileptic encephalopathy, early infantile, 69, 618285 (3), Autosomal dominant
CARS2	612800	Combined oxidative phosphorylation deficiency 27, 616672 (3), Autosomal recessive

CHCHD2	616244	Parkinson disease 22, autosomal dominant, 616710 (3), Autosomal dominant
CHMP2B	609512	Amyotrophic lateral sclerosis 17, 614696 (3), Autosomal dominant; Dementia, familial, nonspecific, 600795 (3), Autosomal dominant
CHRNA4	118504	Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant; {Nicotine addiction, susceptibility to}, 188890 (3)
CHRNB2	118507	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)
CIZ1	611420	No OMIM phenotype
CLPB	616254	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271 (3), Autosomal recessive
CNTNAP1	602346	Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive
COASY	609855	Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive
COL6A3	120250	Bethlem myopathy 1, 158810 (3), Autosomal dominant, Autosomal recessive; Dystonia 27, 616411 (3), Autosomal recessive; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal dominant, Autosomal recessive
COQ9	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
COX10	602125	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial; Leigh syndrome due to mitochondrial COX4 deficiency, 256000 (3), Autosomal recessive, Mitochondrial
COX15	603646	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 (3), Autosomal recessive; Leigh syndrome due to cytochrome c oxidase deficiency, 256000 (3), Autosomal recessive, Mitochondrial
CP	117700	[Hypoceruloplasminemia, hereditary], 604290 (3), Autosomal recessive; Hemosiderosis, systemic, due to aceruloplasminemia, 604290 (3), Autosomal recessive; Cerebellar ataxia, 604290 (3), Autosomal recessive
CRAT	600184	?Neurodegeneration with brain iron accumulation 8, 617917 (3), Autosomal recessive
CSF1R	164770	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 (3), Autosomal recessive; Leukoencephalopathy, diffuse hereditary, with spheroids, 221820 (3), Autosomal dominant
CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
CYP27A1	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
DCAF17	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive

DCTN1	601143	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal dominant, Autosomal recessive; Perry syndrome, 168605 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, type VIIB, 607641 (3), Autosomal dominant
DDC	107930	Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive
DEGS1	615843	Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive
DHDDS	608172	Retinitis pigmentosa 59, 613861 (3), Autosomal recessive; Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive
DHX30	616423	Neurodevelopmental disorder with severe motor impairment and absent language, 617804 (3), Autosomal dominant
DLAT	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
DLD	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
DMXL2	612186	?Deafness, autosomal dominant 71, 617605 (3), Autosomal dominant; ?Polyendocrine-polyneuropathy syndrome, 616113 (3), Autosomal recessive; Epileptic encephalopathy, early infantile, 81, 618663 (3), Autosomal recessive
DNAJC13	614334	No OMIM phenotype
DNAJC5	611203	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350 (3), Autosomal dominant
DNAJC6	608375	Parkinson disease 19b, early-onset, 615528 (3), Autosomal recessive; Parkinson disease 19a, juvenile-onset, 615528 (3), Autosomal recessive
DNAL4	610565	?Mirror movements 3, 616059 (3), Autosomal recessive
DRD3	126451	{Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Essential tremor, hereditary, 1}, 190300 (3), Autosomal dominant
EARS2	612799	Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive
ECHS1	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
EEF1A2	602959	Epileptic encephalopathy, early infantile, 33, 616409 (3), Autosomal dominant; Mental retardation, autosomal dominant 38, 616393 (3), Autosomal dominant
EIF4G1	600495	{Parkinson disease 18}, 614251 (3), Autosomal dominant
EPRS1 (EPRS)	138295	Leukodystrophy, hypomyelinating, 15, 617951 (3), Autosomal recessive
ETHE1	608451	Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive
FA2H	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive

FASTKD2	612322	Combined oxidative phosphorylation deficiency 44, 618855 (3), Autosomal recessive
FBXO7	605648	Parkinson disease 15, autosomal recessive, 260300 (3), Autosomal recessive
FITM2	612029	Siddiqi syndrome, 618635 (3), Autosomal recessive
FOXG1	164874	Rett syndrome, congenital variant, 613454 (3), Autosomal dominant
FOXRED1	613622	Mitochondrial complex I deficiency, nuclear type 19, 618241 (3), Autosomal recessive
FTL	134790	Hyperferritinemia-cataract syndrome, 600886 (3), Autosomal dominant; Neurodegeneration with brain iron accumulation 3, 606159 (3), Autosomal dominant; L-ferritin deficiency, dominant and recessive, 615604 (3), Autosomal dominant, Autosomal recessive
FUCA1	612280	Fucosidosis, 230000 (3), Autosomal recessive
FUS	137070	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 (3); Essential tremor, hereditary, 4, 614782 (3), Autosomal dominant
GABRB2	600232	Epileptic encephalopathy, infantile or early childhood, 2, 617829 (3), Autosomal dominant
GAMT	601240	Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive
GBA	606463	Gaucher disease, type III, 231000 (3), Autosomal recessive; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; Gaucher disease, type II, 230900 (3), Autosomal recessive; {Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant
GBE1	607839	Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive; Glycogen storage disease IV, 232500 (3), Autosomal recessive
GCDH	608801	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
GCH1	600225	Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive; Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3), Autosomal dominant, Autosomal recessive
GFAP	137780	Alexander disease, 203450 (3), Autosomal dominant
GFM2	606544	Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive
GIGYF2	612003	{Parkinson disease 11}, 607688 (3)
GJC2	608803	Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Lymphatic malformation 3, 613480 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive

GLB1	611458	GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive
GLUD2	300144	{Parkinson disease, age of onset, modifier}, 168600 (3), Autosomal dominant, Multifactorial
GM2A	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive
GNAL	139312	Dystonia 25, 615073 (3), Autosomal dominant
GNAO1	139311	Epileptic encephalopathy, early infantile, 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant
GNB1	139380	Mental retardation, autosomal dominant 42, 616973 (3), Autosomal dominant; Leukemia, acute lymphoblastic, somatic, 613065 (3)
GPR88	607468	?Chorea, childhood-onset, with psychomotor retardation, 616939 (3), Autosomal recessive
GRIN1	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant
GRIN2B	138252	Epileptic encephalopathy, early infantile, 27, 616139 (3), Autosomal dominant; Mental retardation, autosomal dominant 6, 613970 (3), Autosomal dominant
GRN	138945	Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive; Aphasia, primary progressive, 607485 (3), Autosomal dominant; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant
GSX2	616253	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646 (3), Autosomal recessive
GTPBP2	607434	Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive
HEXA	606869	GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive; [Hex A pseudodeficiency], 272800 (3), Autosomal recessive
HIBCH	610690	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive
HPCA	142622	Dystonia 2, torsion, autosomal recessive, 224500 (3), Autosomal recessive
HPRT1	308000	HPRT-related gout, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive
HSD17B10	300256	HSD10 mitochondrial disease, 300438 (3), X-linked dominant
HTRA2	606441	{Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive

HTT	613004	Huntington disease, 143100 (3), Autosomal dominant; Lopes-Maciel-Rodan syndrome, 617435 (3), Autosomal recessive
IFIH1	606951	Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
IREB2	147582	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451 (3), Autosomal recessive
IRF2BPL	611720	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 (3), Autosomal dominant
JAM2	606870	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824 (3), Autosomal recessive
JPH3	605268	Huntington disease-like 2, 606438 (3), Autosomal dominant
KCNA4	176266	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 (3), Autosomal recessive
KCNMA1	600150	Liang-Wang syndrome, 618729 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 (3), Autosomal dominant; Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant
KCNQ2	602235	Epileptic encephalopathy, early infantile, 7, 613720 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant
KCTD17	616386	Dystonia 26, myoclonic, 616398 (3), Autosomal dominant
KMT2B	606834	Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant
LIPT1	610284	Lipoyltransferase 1 deficiency, 616299 (3), Autosomal recessive
LIPT2	617659	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 (3), Autosomal recessive
LRP10	609921	No OMIM phenotype
LRPPRC	607544	Leigh syndrome, French-Canadian type, 220111 (3), Autosomal recessive
LRRK2	609007	{Parkinson disease 8}, 607060 (3), Autosomal dominant
LYST	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
MAPT	157140	Pick disease, 172700 (3), Autosomal dominant; Dementia, frontotemporal, with or without parkinsonism, 600274 (3), Autosomal dominant; {Parkinson disease, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; Supranuclear palsy, progressive, 601104 (3), Autosomal dominant; Supranuclear palsy, progressive atypical, 260540 (3), Autosomal recessive
MAT1A	610550	Methionine adenosyltransferase deficiency, autosomal recessive, 250850 (3), Autosomal dominant, Autosomal recessive; Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 (3), Autosomal dominant, Autosomal recessive
MCOLN1	605248	Mucolipidosis IV, 252650 (3), Autosomal recessive

MDH2	154100	Epileptic encephalopathy, early infantile, 51, 617339 (3), Autosomal recessive
MECP2	300005	Mental retardation, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Mental retardation, X-linked, syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, atypical, 312750 (3), X-linked dominant; {Autism susceptibility, X-linked 3}, 300496 (3), X-linked; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, preserved speech variant, 312750 (3), X-linked dominant
MECR	608205	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 (3), Autosomal recessive
MED20	612915	No OMIM phenotype
MICU1	605084	Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive
MMADHC	611935	Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive
MPV17	137960	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive
MRE11	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
MRPS34	611994	Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive
MYBPC1	160794	Arthrogryposis, distal, type 1B, 614335 (3), Autosomal dominant; Myopathy, congenital, with tremor, 618524 (3), Autosomal dominant; Lethal congenital contracture syndrome 4, 614915 (3), Autosomal recessive
MYORG	618255	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317 (3), Autosomal recessive
NAXD	615910	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321 (3), Autosomal recessive
NDUFA1	300078	Mitochondrial complex I deficiency, nuclear type 12, 301020 (3), X-linked recessive
NDUFA10	603835	Mitochondrial complex I deficiency, nuclear type 22, 618243 (3), Autosomal recessive
NDUFA12	614530	?Mitochondrial complex I deficiency, nuclear type 23, 618244 (3), Autosomal recessive
NDUFA9	603834	Mitochondrial complex I deficiency, nuclear type 26, 618247 (3), Autosomal recessive
NDUFAF4	611776	Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive

NDUFAF6	612392	Fanconi renotubular syndrome 5, 618913 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 17, 618239 (3), Autosomal recessive
NDUFS1	157655	Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive
NDUFS3	603846	Mitochondrial complex I deficiency, nuclear type 8, 618230 (3), Autosomal recessive
NDUFS7	601825	Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive
NDUFS8	602141	Mitochondrial complex I deficiency, nuclear type 2, 618222 (3), Autosomal recessive
NHLRC2	618277	FINCA syndrome, 618278 (3), Autosomal recessive
NKX2-1	600635	{Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant; Chorea, hereditary benign, 118700 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant
NKX6-2	605955	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560 (3), Autosomal recessive
NPC1	607623	Niemann-Pick disease, type D, 257220 (3), Autosomal recessive; Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive
NPC2	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
NTNG2	618689	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive
NUP62	605815	Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive
PANK2	606157	HARP syndrome, 607236 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
PARK7	602533	Parkinson disease 7, autosomal recessive early-onset, 606324 (3), Autosomal recessive
PCCA	232000	Propionicacidemia, 606054 (3), Autosomal recessive
PCCB	232050	Propionicacidemia, 606054 (3), Autosomal recessive
PCDH12	605622	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive
PCDH19	300460	Epileptic encephalopathy, early infantile, 9, 300088 (3), X-linked
PDE10A	610652	Dyskinesia, limb and orofacial, infantile-onset, 616921 (3), Autosomal recessive; Striatal degeneration, autosomal dominant, 616922 (3), Autosomal dominant
PDE8B	603390	Striatal degeneration, autosomal dominant, 609161 (3), Autosomal dominant; Pigmented nodular adrenocortical disease, primary, 3, 614190 (3)
PDGFB	190040	Dermatofibrosarcoma protuberans, 607907 (3); Basal ganglia calcification, idiopathic, 5, 615483 (3), Autosomal dominant; Meningioma, SIS-related, 607174 (3), Autosomal dominant

PDGFRB	173410	Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant; Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
PDHX	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
PEAR1	610278	No OMIM phenotype
PEX16	603360	Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive; Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive
PINK1	608309	Parkinson disease 6, early onset, 605909 (3), Autosomal recessive
PLA2G6	603604	Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive; Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive
PLEKHG2	611893	Leukodystrophy and acquired microcephaly with or without dystonia, 616763 (3), Autosomal recessive
PLP1	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
PMPCB	603131	Multiple mitochondrial dysfunctions syndrome 6, 617954 (3), Autosomal recessive
PNKD	609023	Paroxysmal nonkinesigenic dyskinesia 1, 118800 (3), Autosomal dominant
PNKP	605610	Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive; Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive
PNPLA8	612123	?Mitochondrial myopathy with lactic acidosis, 251950 (3), Autosomal recessive
PNPT1	610316	Deafness, autosomal recessive 70, 614934 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 13, 614932 (3), Autosomal recessive
PODXL	602632	No OMIM phenotype
POLG	174763	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive

POLR3A	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive; Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive
PON1	168820	{Microvascular complications of diabetes 5}, 612633 (3); {Organophosphate poisoning, sensitivity to} (3); {Coronary artery disease, susceptibility to} (3); {Coronary artery spasm 2, susceptibility to (3)
PRKN	602544	Parkinson disease, juvenile, type 2, 600116 (3), Autosomal recessive; Ovarian cancer, somatic, 167000 (3); Adenocarcinoma of lung, somatic, 211980 (3)
PRKRA	603424	Dystonia 16, 612067 (3), Autosomal recessive
PRNP	176640	Insomnia, fatal familial, 600072 (3), Autosomal dominant; {Kuru, susceptibility to}, 245300 (3); Huntington disease-like 1, 603218 (3), Autosomal dominant; Prion disease with protracted course, 606688 (3), Autosomal dominant; Cerebral amyloid angiopathy, PRNP-related, 137440 (3), Autosomal dominant; Creutzfeldt-Jakob disease, 123400 (3), Autosomal dominant; Gerstmann-Straussler disease, 137440 (3), Autosomal dominant
PRRT2	614386	Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant; Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant
PTRHD1	617342	No OMIM phenotype
PTS	612719	Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive
QDPR	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
RAB11B	604198	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant
RAB39B	300774	Waisman syndrome, 311510 (3), X-linked recessive; Mental retardation, X-linked 72, 300271 (3), X-linked recessive
RAD51	179617	{Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; Mirror movements 2, 614508 (3), Autosomal dominant; Fanconi anemia, complementation group R, 617244 (3), Autosomal dominant
RELN	600514	{Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive
REPS1	614825	?Neurodegeneration with brain iron accumulation 7, 617916 (3), Autosomal recessive
RHOBTB2	607352	Epileptic encephalopathy, early infantile, 64, 618004 (3), Autosomal dominant
RIC3	610509	No OMIM phenotype
RNF216	609948	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive

SCN11A	604385	Neuropathy, hereditary sensory and autonomic, type VII, 615548 (3), Autosomal dominant; Episodic pain syndrome, familial, 3, 615552 (3), Autosomal dominant
SCN4A	603967	Hyperkalemic periodic paralysis, type 2, 170500 (3), Autosomal dominant; Paramyotonia congenita, 168300 (3), Autosomal dominant; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant
SCN8A	600702	Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant; Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; ?Myoclonus, familial, 2, 618364 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 13, 614558 (3), Autosomal dominant
SCP2	184755	?Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3), Autosomal recessive
SDHA	600857	Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Paragangliomas 5, 614165 (3), Autosomal dominant; Mitochondrial respiratory chain complex II deficiency, 252011 (3), Autosomal recessive
SDHAF1	612848	Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive
SDHD	602690	Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive; Paraganglioma and gastric stromal sarcoma, 606864 (3); Pheochromocytoma, 171300 (3), Autosomal dominant
SEPSECS	613009	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive
SERAC1	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
SETX	608465	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant
SGCE	604149	Dystonia-11, myoclonic, 159900 (3), Autosomal dominant
SLC18A2	193001	?Parkinsonism-dystonia, infantile, 2, 618049 (3), Autosomal recessive
SLC19A3	606152	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 (3), Autosomal recessive
SLC20A2	158378	Basal ganglia calcification, idiopathic, 1, 213600 (3), Autosomal dominant
SLC25A42	610823	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 (3), Autosomal recessive

SLC2A1	138140	Dystonia 9, 601042 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal dominant, Autosomal recessive; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant
SLC30A10	611146	Hypermanganesemia with dystonia 1, 613280 (3), Autosomal recessive
SLC30A9	604604	?Birk-Landau-Perez syndrome, 617595 (3), Autosomal recessive
SLC39A14	608736	?Hyperostosis cranalis interna, 144755 (3), Autosomal dominant; Hypermanganesemia with dystonia 2, 617013 (3), Autosomal recessive
SLC44A1	606105	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868 (3), Autosomal recessive
SLC6A3	126455	{Nicotine dependence, protection against}, 188890 (3); Parkinsonism-dystonia, infantile, 1, 613135 (3), Autosomal recessive
SLC6A8	300036	Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive
SMPD1	607608	Niemann-Pick disease, type A, 257200 (3), Autosomal recessive; Niemann-Pick disease, type B, 607616 (3), Autosomal recessive
SNCA	163890	Dementia, Lewy body, 127750 (3), Autosomal dominant; Parkinson disease 1, 168601 (3), Autosomal dominant; Parkinson disease 4, 605543 (3), Autosomal dominant
SNCAIP	603779	No OMIM phenotype
SPG11	610844	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive; Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive
SPR	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), Autosomal recessive, ?Autosomal dominant
SQSTM1	601530	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 (3), Autosomal dominant; Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 (3), Autosomal recessive; Myopathy, distal, with rimmed vacuoles, 617158 (3), Autosomal dominant; Paget disease of bone 3, 167250 (3), Autosomal dominant
STUB1	607207	Spinocerebellar ataxia, autosomal recessive 16, 615768 (3), Autosomal recessive; ?Spinocerebellar ataxia 48, 618093 (3), Autosomal dominant
SUCLA2	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
SUOX	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive

SURF1	185620	Leigh syndrome, due to COX IV deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive
SYNJ1	604297	Epileptic encephalopathy, early infantile, 53, 617389 (3), Autosomal recessive; Parkinson disease 20, early-onset, 615530 (3), Autosomal recessive
SYT1	185605	Baker-Gordon syndrome, 618218 (3), Autosomal dominant
TAF1	313650	Dystonia-Parkinsonism, X-linked, 314250 (3), X-linked recessive; Mental retardation, X-linked, syndromic 33, 300966 (3), X-linked recessive
TANGO2	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
TBC1D24	613577	Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 (3), Autosomal recessive; DOORS syndrome, 220500 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 16, 615338 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive; Deafness , autosomal recessive 86, 614617 (3), Autosomal recessive
TENM4	610084	Essential tremor, hereditary, 5, 616736 (3), Autosomal dominant
TH	191290	Segawa syndrome, recessive, 605407 (3), Autosomal recessive
THAP1	609520	Dystonia 6, torsion, 602629 (3), Autosomal dominant
TIMM8A	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
TOR1A	605204	Dystonia-1, torsion, 128100 (3), Autosomal dominant; Arthrogryposis multiplex congenita 5, 618947 (3); {Dystonia-1, modifier of} (3)
TPI1	190450	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3), Autosomal recessive
TPK1	606370	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 (3), Autosomal recessive
TRAK1	608112	Epileptic encephalopathy, early infantile, 68, 618201 (3), Autosomal recessive
TRAPPC11	614138	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 (3), Autosomal recessive
TRAPPC12	614139	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669 (3), Autosomal recessive
TRIT1	617840	Combined oxidative phosphorylation deficiency 35, 617873 (3), Autosomal recessive
TSEN2	608753	Pontocerebellar hypoplasia type 2B, 612389 (3), Autosomal recessive
TSFM	604723	Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive

TUBB4A	602662	Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant; Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant
TXN2	609063	?Combined oxidative phosphorylation deficiency 29, 616811 (3), Autosomal recessive
UBA5	610552	?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive; Epileptic encephalopathy, early infantile, 44, 617132 (3), Autosomal recessive
UBTF	600673	Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant
UCHL1	191342	Spastic paraplegia 79, autosomal recessive, 615491 (3), Autosomal recessive; {?Parkinson disease 5, susceptibility to}, 613643 (3), Autosomal dominant
UFM1	610553	Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive
UGDH	603370	Epileptic encephalopathy, early infantile, 84, 618792 (3), Autosomal recessive
UQCRQ	612080	Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive
VAC14	604632	Striatonigral degeneration, childhood-onset, 617054 (3), Autosomal recessive
VAMP2	185881	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760 (3), Autosomal dominant
VCP	601023	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2Y, 616687 (3), Autosomal dominant; Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 (3)
VPS13A	605978	Choreoacanthocytosis, 200150 (3), Autosomal recessive
VPS13C	608879	Parkinson disease 23, autosomal recessive, early onset, 616840 (3), Autosomal recessive
VPS13D	608877	Spinocerebellar ataxia, autosomal recessive 4, 607317 (3), Autosomal recessive
VPS16	608550	No OMIM phenotype
VPS35	601501	{Parkinson disease 17}, 614203 (3), Autosomal dominant
WARS2	604733	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 (3), Autosomal recessive
WDR45	300526	Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant
WDR73	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive
XK	314850	McLeod syndrome with or without chronic granulomatous disease, 300842 (3), X-linked
XPR1	605237	Basal ganglia calcification, idiopathic, 6, 616413 (3), Autosomal dominant

YY1	600013	Gabriele-de Vries syndrome, 617557 (3), Autosomal dominant
ZC4H2	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant
ZNF142	604083	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425 (3), Autosomal recessive

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: Aug 20, 2020

Possible phenotype mapping keys
(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
(2) the disorder has been placed on the map by linkage; no mutation has been found
(3) the molecular basis for the disorder is known; a mutation has been found in the gene
(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).
Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).
A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.