Movement Disorders panel

versie	16-Oct-2018 (127 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
ACTB	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; ?Dystonia, juvenile-onset, 607371 (3), Autosomal dominant
ADCY5	600293	Dyskinesia, familial, with facial myokymia, 606703 (3), Autosomal dominant
ANO3	610110	Dystonia 24, 615034 (3), Autosomal dominant
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
ATM	607585	Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant; Lymphoma, B-cell non-Hodgkin, somatic (3); Lymphoma, mantle cell, somatic (3); 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	Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant
ATP7B	606882	Wilson disease, 277900 (3), Autosomal recessive
ATP8A2	605870	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
BCAP31	300398	Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive
C19orf12	614297	Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal recessive; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive
CACNA1A	601011	Epileptic encephalopathy, early infantile, 42, 617106 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant
CACNA1B	601012	?Dystonia 23, 614860 (3), Autosomal dominant
CHCHD2	616244	Parkinson disease 22, autosomal dominant, 616710 (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
COASY	609855	Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive
COL6A3	120250	Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant; Dystonia 27, 616411 (3), Autosomal recessive; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant
CP	117700	Cerebellar ataxia, 604290 (3), Autosomal recessive; Hemosiderosis, systemic, due to aceruloplasminemia, 604290 (3), Autosomal recessive; [Hypoceruloplasminemia, hereditary], 604290 (3), Autosomal recessive
CRAT	600184	?Neurodegeneration with brain iron accumulation 8, 617917 (3), Autosomal recessive
DCAF17	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
DCTN1	601143	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant; Neuropathy, distal hereditary motor, type VIIB, 607641 (3), Autosomal dominant; Perry syndrome, 168605 (3), Autosomal dominant
DLAT	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
DLD	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
DNAJC13	614334	No OMIM phenotype
DNAJC5	611203	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350 (3), Autosomal dominant
DNAJC6	608375	Parkinson disease 19a, juvenile-onset, 615528 (3), Autosomal recessive; Parkinson disease 19b, early-onset, 615528 (3), Autosomal recessive
DNAL4	610565	?Mirror movements 3, 616059 (3), Autosomal recessive
ECHS1	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
EIF4G1	600495	{Parkinson disease 18}, 614251 (3), Autosomal dominant
FA2H	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
FBXO7	605648	Parkinson disease 15, autosomal recessive, 260300 (3), Autosomal recessive
FOXG1	164874	Rett syndrome, congenital variant, 613454 (3), Autosomal dominant
FTL	134790	Hyperferritinemia-cataract syndrome, 600886 (3), Autosomal dominant; L-ferritin deficiency, dominant and recessive, 615604 (3), Autosomal recessive, Autosomal dominant; Neurodegeneration with brain iron accumulation 3, 606159 (3), Autosomal dominant
FUCA1	612280	Fucosidosis, 230000 (3), Autosomal recessive

GBA	606463	Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; {Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Isolated cases, Multifactorial
GCDH	608801	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
GCH1	600225	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3), Autosomal recessive, Autosomal dominant; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
GIGYF2	612003	{Parkinson disease 11}, 607688 (3)
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
GPR88	607468	?Chorea, childhood-onset, with psychomotor retardation, 616939 (3), Autosomal recessive
GRIN1	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive
GRIN2B	138252	Epileptic encephalopathy, early infantile, 27, 616139 (3), Autosomal dominant; Mental retardation, autosomal dominant 6, 613970 (3), Autosomal dominant
GTPBP2	607434	Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive
HEXA	606869	GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; [Hex A pseudodeficiency], 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (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
HTRA2	606441	3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive; {Parkinson disease 13}, 610297 (3)
IRF2BPL	611720	No OMIM phenotype
KCNMA1	600150	?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; Myokymia, 121200 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant
KCTD17	616386	Dystonia 26, myoclonic, 616398 (3), Autosomal dominant
KMT2B	606834	Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant

LRRK2	609007	{Parkinson disease 8}, 607060 (3), Autosomal dominant
MECP2	300005	{Autism susceptibility, X-linked 3}, 300496 (3), Isolated cases, X-linked, Multifactorial; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Mental retardation, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; Mental retardation, X-linked, syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, atypical, 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
MRE11 (MRE11A)	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
MYORG (KIAA1161)	No OMIM gene	No OMIM phenotype
NKX2-1	600635	Chorea, hereditary benign, 118700 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant; {Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant
NPC1	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
NPC2	601015	Niemann-pick disease, type C2, 607625 (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 (DJ1)	602533	Parkinson disease 7, autosomal recessive early-onset, 606324 (3), Autosomal recessive
PDE10A	610652	Dyskinesia, limb and orofacial, infantile-onset, 616921 (3), Autosomal recessive; Striatal degeneration, autosomal dominant, 616922 (3), Autosomal dominant
PDE8B	603390	Pigmented nodular adrenocortical disease, primary, 3, 614190 (3); Striatal degeneration, autosomal dominant, 609161 (3), Autosomal dominant
PDGFB	190040	Basal ganglia calcification, idiopathic, 5, 615483 (3), Autosomal dominant; Dermatofibrosarcoma protuberans, 607907 (3); Meningioma, SIS-related, 607174 (3), Autosomal dominant
PDGFRB	173410	Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant

PINK1	608309	Parkinson disease 6, early onset, 605909 (3), Autosomal recessive
PLA2G6	603604	Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive
PLP1	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
PNKD	609023	Paroxysmal nonkinesigenic dyskinesia 1, 118800 (3), Autosomal dominant
PNKP	605610	Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive
PODXL	602632	No OMIM phenotype
PRKN	602544	Adenocarcinoma of lung, somatic, 211980 (3); Adenocarcinoma, ovarian, somatic, 167000 (3); {Leprosy, susceptibility to}, 607572 (3); Parkinson disease, juvenile, type 2, 600116 (3), Autosomal recessive
PRKRA	603424	Dystonia 16, 612067 (3), Autosomal recessive
PRRT2	614386	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant; Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant
PTRHD1	617342	No OMIM phenotype
RAB39B	300774	Mental retardation, X-linked 72, 300271 (3), X-linked recessive; ?Waisman syndrome, 311510 (3), X-linked recessive
RAD51	179617	{Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant; ?Fanconi anemia, complementation group R, 617244 (3), Autosomal dominant; Mirror movements 2, 614508 (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
RIC3	610509	No OMIM phenotype
RNF216	609948	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive
SCN11A	604385	Episodic pain syndrome, familial, 3, 615552 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type VII, 615548 (3), Autosomal dominant
SCN8A	600702	?Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 13, 614558 (3), Autosomal dominant; Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant
SCP2	184755	?Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3), Autosomal recessive

SERAC1	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
SETX	608465	Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 1, 606002 (3), Autosomal recessive
SGCE	604149	Dystonia-11, myoclonic, 159900 (3), Autosomal dominant
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
SLC2A1	138140	Dystonia 9, 601042 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal recessive, Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant
SLC30A10	611146	Hypermanganesemia with dystonia 1, 613280 (3), Autosomal recessive
SLC39A14	608736	Hypermanganesemia with dystonia 2, 617013 (3), Autosomal recessive; ?Hyperostosis cranalis interna, 144755 (3), Autosomal dominant
SLC6A3	126455	{Nicotine dependence, protection against}, 188890 (3); Parkinsonism-dystonia, infantile, 613135 (3), Autosomal 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
SPG11	610844	Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive
SPR	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), ?Autosomal dominant, Autosomal recessive
SUOX	606887	Sulfite oxidase deficiency, 272300 (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	No OMIM phenotype
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
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, modifier of} (3); Dystonia-1, torsion, 128100 (3), Autosomal dominant
TUBB4A	602662	Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant
UCHL1	191342	{?Parkinson disease 5, susceptibility to}, 613643 (3); Spastic paraplegia 79, autosomal recessive, 615491 (3), Autosomal recessive
VPS13A	605978	Choreoacanthocytosis, 200150 (3), Autosomal recessive
VPS13C	608879	Parkinson disease 23, autosomal recessive, early onset, 616840 (3), Autosomal recessive
VPS13D	608877	No OMIM phenotype
VPS35	601501	{Parkinson disease 17}, 614203 (3), Autosomal dominant
WDR45	300526	Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant
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
ZC4H2	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked 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: July 04, 2018

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.