Movement Disorders

Gene panel

Gene panel information

Gene panel	**Movement Disorders**
Version	4
Total genes	345
Activation date	Friday 21 march 2025
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
AARS1	99.99 %	601065	Developmental and epileptic encephalopathy 29, 616339 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant; ?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661 (3), Autosomal dominant; Trichothiodystrophy 8, nonphotosensitive, 619691 (3), Autosomal recessive
ABCB7	99.58 %	300135	Anemia, sideroblastic, with ataxia, 301310 (3), X-linked
ACBD6	98.86 %	616352	Neurodevelopmental disorder with progressive movement abnormalities, 620785 (3), Autosomal recessive
ACER3	99.76 %	617036	?Leukodystrophy, progressive, early childhood-onset, 617762 (3), Autosomal recessive
ACOX1	99.98 %	609751	Mitchell syndrome, 618960 (3), Autosomal dominant; Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
ACSF3	99.99 %	614245	Combined malonic and methylmalonic aciduria, 614265 (3), Autosomal recessive
ACTB	100 %	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; Becker nevus, syndromic or isolated, somatic mosaic, 604919 (3); Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475 (3), Autosomal dominant; Dystonia-deafness syndrome 1, 607371 (3), Autosomal dominant; Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620479 (3)
ACTL6B	99.9 %	612458	Developmental and epileptic encephalopathy 76, 618468 (3), Autosomal recessive; Intellectual developmental disorder with severe speech and ambulation defects, 618470 (3), Autosomal dominant
ADAR	99.84 %	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
ADCY5	99.98 %	600293	Dyskinesia with orofacial involvement, autosomal dominant, 606703 (3), Autosomal dominant; Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651 (3), Autosomal recessive; Dyskinesia with orofacial involvement, autosomal recessive, 619647 (3), Autosomal recessive
AFG3L2	99.97 %	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3), Autosomal dominant; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
ALDH18A1	99.96 %	138250	Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive; Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant
ALDH5A1	96.19 %	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
ANO3	99.98 %	610110	Dystonia 24, 615034 (3), Autosomal dominant
AOPEP	99.99 %	619600	Dystonia 31, 619565 (3), Autosomal recessive
AP1S2	99.56 %	300629	Pettigrew syndrome, 304340 (3), X-linked recessive

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AP3D1	100 %	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
AP4M1	99.98 %	602296	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
APTX	99.92 %	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
ARFGEF3	99.93 %	617411	*No OMIM phenotypes*
ARSA	99.99 %	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
ARV1	99.85 %	611647	Developmental and epileptic encephalopathy 38, 617020 (3), Autosomal recessive
ARX	95.36 %	300382	Proud syndrome, 300004 (3), X-linked; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Partington syndrome, 309510 (3), X-linked recessive; Developmental and epileptic encephalopathy 1, 308350 (3), X-linked recessive; Lissencephaly, X-linked 2, 300215 (3), X-linked; Intellectual developmental disorder, X-linked 29, 300419 (3), X-linked recessive
ASL	99.98 %	608310	Argininosuccinic aciduria, 207900 (3), Autosomal recessive
ATCAY	100 %	608179	Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive
ATM	99.83 %	607585	Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; T-cell prolymphocytic leukemia, somatic (3); Lymphoma, mantle cell, somatic (3)
ATP13A2	99.96 %	610513	Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive; Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive
ATP1A2	99.85 %	182340	Developmental and epileptic encephalopathy 98, 619605 (3), Autosomal dominant; Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 (3), Autosomal recessive; Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant
ATP1A3	99.98 %	182350	Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant; Developmental and epileptic encephalopathy 99, 619606 (3), Autosomal dominant
ATP5MC3	99.97 %	602736	Dystonia, early-onset, and/or spastic paraplegia, 619681 (3), Autosomal dominant
ATP6AP2	99.55 %	300556	Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423 (3), X-linked recessive; ?Parkinsonism with spasticity, X-linked, 300911 (3), X-linked recessive; Congenital disorder of glycosylation, type IIr, 301045 (3), X-linked recessive
ATP7B	100 %	606882	Wilson disease, 277900 (3), Autosomal recessive
ATP8A2	100 %	605870	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
AUH	99.95 %	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
BCAP31	99.95 %	300398	Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive
BCS1L	99.99 %	603647	GRACILE syndrome, 603358 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive
C12orf57	100 %	615140	Temtamy syndrome, 218340 (3), Autosomal recessive
C19orf12	99.99 %	614297	Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal dominant, Autosomal recessive; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive
CA8	99.71 %	114815	Spinocerebellar ataxia, autosomal recessive 34, 613227 (3), Autosomal recessive

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Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CACNA1A	98.16 %	601011	Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Developmental and epileptic encephalopathy 42, 617106 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant
CACNA1B	100 %	601012	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497 (3), Autosomal recessive
CACNA1E	99.82 %	601013	Developmental and epileptic encephalopathy 69, 618285 (3), Autosomal dominant
CACNA1G	99.95 %	604065	Spinocerebellar ataxia 42, 616795 (3), Autosomal dominant; Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 (3), Autosomal dominant
CAMK4	99.78 %	114080	*No OMIM phenotypes*
CARS2	99.99 %	612800	Combined oxidative phosphorylation deficiency 27, 616672 (3), Autosomal recessive
CHCHD2	99.89 %	616244	Parkinson disease 22, autosomal dominant, 616710 (3), Autosomal dominant
CHMP2B	99.8 %	609512	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795 (3), Autosomal dominant
CHRNA4	100 %	118504	{Nicotine addiction, susceptibility to}, 188890 (3); Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant
CHRNB2	99.99 %	118507	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)
CIZ1	99.94 %	611420	*No OMIM phenotypes*
CLN3	99.92 %	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
CLN5	100 %	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
CLN8	100 %	607837	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive
CLPB	99.97 %	616254	Neutropenia, severe congenital, 9, autosomal dominant, 619813 (3), Autosomal dominant; 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 (3), Autosomal recessive; 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 (3), Autosomal dominant
CNTNAP1	99.98 %	602346	Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive
COASY	99.98 %	609855	Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive
COL6A3	99.99 %	120250	Bethlem myopathy 1C, 620726 (3), Autosomal dominant, Autosomal recessive; Ullrich congenital muscular dystrophy 1C, 620728 (3), Autosomal dominant, Autosomal recessive; Dystonia 27, 616411 (3), Autosomal recessive
COQ8A	100 %	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
COQ9	99.62 %	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
COX10	99.99 %	602125	Mitochondrial complex IV deficiency, nuclear type 3, 619046 (3), Autosomal recessive
COX15	100 %	603646	Mitochondrial complex IV deficiency, nuclear type 6, 615119 (3), Autosomal recessive
COX20	99.67 %	614698	Mitochondrial complex IV deficiency, nuclear type 11, 619054 (3), Autosomal recessive
CP	99.95 %	117700	Aceruloplasminemia, 604290 (3), Autosomal recessive
CRAT	99.99 %	600184	?Neurodegeneration with brain iron accumulation 8, 617917 (3), Autosomal recessive
CSF1R	99.92 %	164770	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 (3), Autosomal recessive; Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820 (3), Autosomal dominant
CTC1	100 %	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CTSD	100 %	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
CWF19L1	99.91 %	616120	Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive
CYP27A1	100 %	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
DCAF17	99.84 %	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
DCC	99.96 %	120470	Mirror movements 1 and/or agenesis of the corpus callosum, 157600 (3), Autosomal dominant; Esophageal carcinoma, somatic, 133239 (3); Colorectal cancer, somatic, 114500 (3); Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 (3), Autosomal recessive
DCTN1	99.98 %	601143	Perry syndrome, 168605 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal dominant, Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal dominant 14, 607641 (3), Autosomal dominant
DDC	99.67 %	107930	Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive
DEGS1	99.99 %	615843	Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive
DHDDS	98.65 %	608172	Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive; Retinitis pigmentosa 59, 613861 (3), Autosomal recessive
DHX30	99.96 %	616423	Neurodevelopmental disorder with variable motor and speech impairment, 617804 (3), Autosomal dominant
DLAT	99.65 %	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
DLD	99.89 %	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
DMXL2	99.86 %	612186	Developmental and epileptic encephalopathy 81, 618663 (3), Autosomal recessive; ?Deafness, autosomal dominant 71, 617605 (3), Autosomal dominant; ?Polyendocrine-polyneuropathy syndrome, 616113 (3), Autosomal recessive
DNAJC12	99.72 %	606060	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 (3), Autosomal recessive
DNAJC5	99.99 %	611203	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350 (3), Autosomal dominant
DNAJC6	99.48 %	608375	Parkinson disease 19a, juvenile-onset, 615528 (3), Autosomal recessive; Parkinson disease 19b, early-onset, 615528 (3), Autosomal recessive
DNAL4	99.89 %	610565	?Mirror movements 3, 616059 (3), Autosomal recessive
DRD2	100 %	126450	*No OMIM phenotypes*
EARS2	99.96 %	612799	Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive
ECHS1	100 %	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
EEF1A2	100 %	602959	Developmental and epileptic encephalopathy 33, 616409 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 38, 616393 (3), Autosomal dominant
EIF2AK2	99.7 %	176871	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877 (3), Autosomal dominant; Dystonia 33, 619687 (3), Autosomal dominant, Autosomal recessive
EIF4G1	100 %	600495	{Parkinson disease 18}, 614251 (3), Autosomal dominant
EPRS1	99.53 %	138295	Leukodystrophy, hypomyelinating, 15, 617951 (3), Autosomal recessive
ETHE1	84.97 %	608451	Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive
FA2H	99.98 %	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
FASTKD2	99.93 %	612322	Combined oxidative phosphorylation deficiency 44, 618855 (3), Autosomal recessive
FBXO7	99.98 %	605648	Parkinson disease 15, autosomal recessive, 260300 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FGF14	99.99 %	601515	Spinocerebellar ataxia 27A, 193003 (3), Autosomal dominant; Spinocerebellar ataxia 27B, late-onset, 620174 (3), Autosomal dominant
FITM2	99.99 %	612029	Siddiqi syndrome, 618635 (3), Autosomal recessive
FOXG1	99.91 %	164874	Rett syndrome, congenital variant, 613454 (3), Autosomal dominant
FOXRED1	100 %	613622	Mitochondrial complex I deficiency, nuclear type 19, 618241 (3), Autosomal recessive
FTL	99.99 %	134790	Hyperferritinemia-cataract syndrome, 600886 (3), Autosomal dominant; L-ferritin deficiency, dominant and recessive, 615604 (3), Autosomal dominant, Autosomal recessive; Neurodegeneration with brain iron accumulation 3, 606159 (3), Autosomal dominant
FUCA1	98.72 %	612280	Fucosidosis, 230000 (3), Autosomal recessive
FUS	99.93 %	137070	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 (3); Essential tremor, hereditary, 4, 614782 (3), Autosomal dominant
GABRB2	99.92 %	600232	Developmental and epileptic encephalopathy 92, 617829 (3), Autosomal dominant
GAMT	100 %	601240	Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive
GBA	96.92 %	606463	{Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial
GBE1	99.73 %	607839	Glycogen storage disease IV, 232500 (3), Autosomal recessive; Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive
GCDH	100 %	608801	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
GCH1	99.94 %	600225	Dystonia, DOPA-responsive, 128230 (3), Autosomal dominant, Autosomal recessive; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
GFAP	99.99 %	137780	Alexander disease, 203450 (3), Autosomal dominant
GFM2	99.87 %	606544	Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive
GJC2	100 %	608803	Lymphatic malformation 3, 613480 (3), Autosomal dominant; ?Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive
GLB1	100 %	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive
GLRA1	100 %	138491	Hyperekplexia 1, 149400 (3), Autosomal dominant, Autosomal recessive
GLRB	99.79 %	138492	Hyperekplexia 2, 614619 (3), Autosomal recessive
GLUD2	100 %	300144	*No OMIM phenotypes*
GM2A	100 %	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive
GNAL	100 %	139312	Dystonia 25, 615073 (3), Autosomal dominant
GNAO1	99.86 %	139311	Developmental and epileptic encephalopathy 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant
GNB1	100 %	139380	Myelodysplastic syndrome, somatic, 614286 (3); Leukemia, acute lymphoblastic, somatic, 613065 (3); Intellectual developmental disorder, autosomal dominant 42, 616973 (3), Autosomal dominant
GPR88	100 %	607468	?Chorea, childhood-onset, with psychomotor retardation, 616939 (3), Autosomal recessive

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GRIN1	100 %	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive; Developmental and epileptic encephalopathy 101, 619814 (3), Autosomal recessive; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant
GRIN2B	99.99 %	138252	Developmental and epileptic encephalopathy 27, 616139 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 (3), Autosomal dominant
GRN	100 %	138945	Frontotemporal dementia 2, 607485 (3), Autosomal dominant, Autosomal recessive; Aphasia, primary progressive, 607485 (3), Autosomal dominant, Autosomal recessive; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive
GSX2	100 %	616253	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646 (3), Autosomal recessive
GTPBP2	99.98 %	607434	Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive
HCFC1	99.99 %	300019	Methylmalonic aciduria and homocysteinemia, cblX type, 309541 (3), X-linked recessive
HEXA	99.99 %	606869	[Hex A pseudodeficiency], 272800 (3), Autosomal recessive; GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive
HEXB	99.91 %	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
HIBCH	99.7 %	610690	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive
HNRNPH1	99.98 %	601035	Neurodevelopmental disorder with craniofacial dysmorphism and skeletal defects, 620083 (3), Autosomal dominant
HPCA	99.98 %	142622	Dystonia 2, torsion, autosomal recessive, 224500 (3), Autosomal recessive
HPRT1	97.8 %	308000	Hyperuricemia, HRPT-related, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive
HSD17B10	99.98 %	300256	HSD10 mitochondrial disease, 300438 (3), X-linked dominant
HSPD1	83.42 %	118190	Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive
HTRA2	99.99 %	606441	{Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive
HTT	99.98 %	613004	Lopes-Maciel-Rodan syndrome, 617435 (3), Autosomal recessive; Huntington disease, 143100 (3), Autosomal dominant
IFIH1	99.84 %	606951	Immunodeficiency 95, 619773 (3), Autosomal recessive; Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
IREB2	99.9 %	147582	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451 (3), Autosomal recessive
IRF2BPL	99.21 %	611720	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 (3), Autosomal dominant
JAM2	91.82 %	606870	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824 (3), Autosomal recessive
KCNA1	100 %	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
KCNA4	100 %	176266	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 (3), Autosomal recessive
KCND3	99.98 %	605411	Spinocerebellar ataxia 19, 607346 (3), Autosomal dominant; Brugada syndrome 9, 616399 (3), Autosomal dominant

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KCNMA1	99.89 %	600150	{Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 (3), Autosomal dominant; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant; Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Liang-Wang syndrome, 618729 (3), Autosomal dominant
KCNN2	91.25 %	605879	?Dystonia 34, myoclonic, 619724 (3), Autosomal dominant; Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725 (3), Autosomal dominant
KCNQ2	100 %	602235	Developmental and epileptic encephalopathy 7, 613720 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant
KCTD17	100 %	616386	Dystonia 26, myoclonic, 616398 (3), Autosomal dominant
KIF1C	99.99 %	603060	Spastic ataxia 2, autosomal recessive, 611302 (3), Autosomal recessive
KMT2B	99.99 %	606834	Intellectual developmental disorder, autosomal dominant 68, 619934 (3), Autosomal dominant; Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant
L2HGDH	99.92 %	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
LIPT1	99.89 %	610284	Lipoyltransferase 1 deficiency, 616299 (3), Autosomal recessive
LIPT2	99.99 %	617659	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 (3), Autosomal recessive
LRP10	99.99 %	609921	*No OMIM phenotypes*
LRPPRC	99.8 %	607544	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111 (3), Autosomal recessive
LRRK2	99.44 %	609007	{Parkinson disease 8}, 607060 (3), Autosomal dominant
LYST	99.87 %	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
MAPT	99.8 %	157140	Supranuclear palsy, progressive, 601104 (3), Autosomal dominant; Frontotemporal dementia 1, with or without parkinsonism, 600274 (3), Autosomal dominant; Supranuclear palsy, progressive atypical, 260540 (3), Autosomal recessive; {Parkinson disease, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; Pick disease, 172700 (3), Autosomal dominant
MARS2	100 %	609728	?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive; Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive
MAT1A	99.7 %	610550	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 (3), Autosomal dominant, Autosomal recessive; Methionine adenosyltransferase deficiency, autosomal recessive, 250850 (3), Autosomal dominant, Autosomal recessive
MCOLN1	100 %	605248	Lisch epithelial corneal dystrophy, 620763 (3), Autosomal dominant; Mucolipidosis IV, 252650 (3), Autosomal recessive
MDH2	99.54 %	154100	Developmental and epileptic encephalopathy 51, 617339 (3), Autosomal recessive
MECP2	99.95 %	300005	Rett syndrome, atypical, 312750 (3), X-linked dominant; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; {Autism susceptibility, X-linked 3}, 300496 (3), X-linked; Intellectual developmental disorder, X-linked syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, preserved speech variant, 312750 (3), X-linked dominant
MECR	99.63 %	608205	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 (3), Autosomal recessive; Optic atrophy 16, 620629 (3), Autosomal recessive
MED20	99.99 %	612915	*No OMIM phenotypes*
MED27	99.99 %	605044	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MICU1	99.56 %	605084	Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive
MMADHC	99.76 %	611935	Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive
MPV17	99.98 %	137960	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive
MRE11	99.93 %	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
MRPS34	100 %	611994	Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive
MTFMT	99.98 %	611766	Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive
MYBPC1	99.77 %	160794	Congenital myopathy 16, 618524 (3), Autosomal dominant; Lethal congenital contracture syndrome 4, 614915 (3), Autosomal recessive; Arthrogryposis, distal, type 1B, 614335 (3), Autosomal dominant
MYORG	99.99 %	618255	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317 (3), Autosomal recessive
NAA60	99.99 %	614246	Basal ganglia calcification, idiopathic, 9, autosomal recessive, 620786 (3), Autosomal recessive
NAXD	99.99 %	615910	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321 (3), Autosomal recessive
NDUFA1	99.93 %	300078	Mitochondrial complex I deficiency, nuclear type 12, 301020 (3), X-linked recessive
NDUFA10	99.98 %	603835	Mitochondrial complex I deficiency, nuclear type 22, 618243 (3), Autosomal recessive
NDUFA12	99.21 %	614530	Mitochondrial complex I deficiency, nuclear type 23, 618244 (3), Autosomal recessive
NDUFA2	99.95 %	602137	Mitochondrial complex I deficiency, nuclear type 13, 618235 (3), Autosomal recessive
NDUFA9	100 %	603834	Mitochondrial complex I deficiency, nuclear type 26, 618247 (3), Autosomal recessive
NDUFAF4	99.95 %	611776	Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive
NDUFAF5	99.89 %	612360	Mitochondrial complex I deficiency, nuclear type 16, 618238 (3), Autosomal recessive
NDUFAF6	99.86 %	612392	Mitochondrial complex I deficiency, nuclear type 17, 618239 (3), Autosomal recessive; Fanconi renotubular syndrome 5, 618913 (3), Autosomal recessive
NDUFS1	99.79 %	157655	Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive
NDUFS3	100 %	603846	Mitochondrial complex I deficiency, nuclear type 8, 618230 (3), Autosomal recessive
NDUFS4	99.99 %	602694	Mitochondrial complex I deficiency, nuclear type 1, 252010 (3), Autosomal recessive
NDUFS7	99.99 %	601825	Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive
NDUFS8	100 %	602141	Mitochondrial complex I deficiency, nuclear type 2, 618222 (3), Autosomal recessive
NDUFV1	99.99 %	161015	Mitochondrial complex I deficiency, nuclear type 4, 618225 (3), Autosomal recessive
NGLY1	99.93 %	610661	Congenital disorder of deglycosylation 1, 615273 (3), Autosomal recessive
NHLRC2	99.83 %	618277	FINCA syndrome, 618278 (3), Autosomal recessive
NKX2-1	100 %	600635	Chorea, hereditary benign, 118700 (3), Autosomal dominant; {Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant
NKX6-2	100 %	605955	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560 (3), Autosomal recessive
NPC1	99.99 %	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
NPC2	100 %	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
NR4A2	99.97 %	601828	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NTNG2	99.98 %	618689	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive
NUP54	99.95 %	607607	Dystonia 37, early-onset, with striatal lesions, 620427 (3), Autosomal recessive
NUP62	100 %	605815	Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive
NUS1	99.9 %	610463	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1aa, 617082 (3), Autosomal recessive
OPA3	100 %	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
PANK2	99.99 %	606157	Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
PARK7	99.93 %	602533	Parkinson disease 7, autosomal recessive early-onset, 606324 (3), Autosomal recessive
PCCA	99.9 %	232000	Propionicacidemia, 606054 (3), Autosomal recessive
PCCB	99.97 %	232050	Propionicacidemia, 606054 (3), Autosomal recessive
PCDH12	100 %	605622	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive
PCDH19	99.98 %	300460	Developmental and epileptic encephalopathy 9, 300088 (3), X-linked
PDE10A	87.37 %	610652	Striatal degeneration, autosomal dominant, 616922 (3), Autosomal dominant; Dyskinesia, limb and orofacial, infantile-onset, 616921 (3), Autosomal recessive
PDE2A	99.95 %	602658	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150 (3), Autosomal recessive
PDE8B	99.98 %	603390	Pigmented nodular adrenocortical disease, primary, 3, 614190 (3); Striatal degeneration, autosomal dominant, 609161 (3), Autosomal dominant
PDGFB	99.99 %	190040	Meningioma, SIS-related, 607174 (3), Autosomal dominant; Basal ganglia calcification, idiopathic, 5, 615483 (3), Autosomal dominant; Dermatofibrosarcoma protuberans, 607907 (3)
PDGFRB	99.99 %	173410	Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant
PDHA1	99.04 %	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
PDHX	99.64 %	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
PET100	99.98 %	614770	Mitochondrial complex IV deficiency, nuclear type 12, 619055 (3), Autosomal recessive
PEX16	99.94 %	603360	Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive; Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive
PINK1	99.69 %	608309	Parkinson disease 6, early onset, 605909 (3), Autosomal recessive
PLA2G6	99.98 %	603604	Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive
PLEKHG2	99.99 %	611893	Leukodystrophy and acquired microcephaly with or without dystonia, 616763 (3), Autosomal recessive
PLP1	99.98 %	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
PMPCB	99.94 %	603131	Multiple mitochondrial dysfunctions syndrome 6, 617954 (3), Autosomal recessive
PNKD	100 %	609023	Paroxysmal nonkinesigenic dyskinesia 1, 118800 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PNKP	100 %	605610	?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive; Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive
PNPLA8	99.92 %	612123	?Mitochondrial myopathy with lactic acidosis, 251950 (3), Autosomal recessive
PNPT1	99.56 %	610316	Spinocerebellar ataxia 25, 608703 (3), Autosomal dominant; Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 13, 614932 (3), Autosomal recessive
PODXL	93.29 %	602632	*No OMIM phenotypes*
POLG	100 %	174763	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
POLR3A	99.97 %	614258	Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
PON1	99.7 %	168820	{Coronary artery spasm 2, susceptibility to} (3); {Organophosphate poisoning, sensitivity to} (3); {Coronary artery disease, susceptibility to} (3); {Microvascular complications of diabetes 5}, 612633 (3)
PRKN	99.99 %	602544	Adenocarcinoma of lung, somatic, 211980 (3); Parkinson disease, juvenile, type 2, 600116 (3), Autosomal recessive; Ovarian cancer, somatic, 167000 (3)
PRKRA	99.94 %	603424	Dystonia 16, 612067 (3), Autosomal recessive
PRNP	100 %	176640	Spongiform encephalopathy with neuropsychiatric features, 606688 (3), Autosomal dominant; Gerstmann-Straussler disease, 137440 (3), Autosomal dominant; Huntington disease-like 1, 603218 (3), Autosomal dominant; Insomnia, fatal familial, 600072 (3), Autosomal dominant; {Kuru, susceptibility to}, 245300 (3); Cerebral amyloid angiopathy, PRNP-related, 137440 (3), Autosomal dominant; Creutzfeldt-Jakob disease, 123400 (3), Autosomal dominant
PRRT2	99.97 %	614386	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant; Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant
PTRHD1	100 %	617342	Neurodevelopmental disorder with early-onset parkinsonism and behavioral abnormalities, 620747 (3), Autosomal recessive
PTS	99.93 %	612719	Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive
QDPR	99.92 %	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
RAB11B	100 %	604198	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant
RAB39B	99.99 %	300774	Intellectual developmental disorder, X-linked 72, 300271 (3), X-linked recessive; Waisman syndrome, 311510 (3), X-linked recessive
RAD51	90.17 %	179617	Mirror movements 2, 614508 (3), Autosomal dominant; {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; Fanconi anemia, complementation group R, 617244 (3), Autosomal dominant
RELN	99.98 %	600514	{Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive
REPS1	99.93 %	614825	?Neurodegeneration with brain iron accumulation 7, 617916 (3), Autosomal recessive
RHOBTB2	100 %	607352	Developmental and epileptic encephalopathy 64, 618004 (3), Autosomal dominant
RNASEH2A	99.95 %	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RNASEH2B	99.94 %	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
RNASEH2C	99.99 %	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
RNASET2	99.99 %	612944	Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive
RNF216	99.99 %	609948	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive
RNU7-1	33.9 %	617876	Aicardi-Goutieres syndrome 9, 619487 (3), Autosomal recessive
SAMHD1	99.98 %	606754	?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive
SCN11A	99.94 %	604385	Episodic pain syndrome, familial, 3, 615552 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type VII, 615548 (3), Autosomal dominant
SCN1A	99.94 %	182389	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant; Dravet syndrome, 607208 (3), Autosomal dominant; Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 2, 604403 (3), Autosomal dominant
SCN4A	99.98 %	603967	Paramyotonia congenita, 168300 (3), Autosomal dominant; Hyperkalemic periodic paralysis, 170500 (3), Autosomal dominant; Congenital myopathy 22B, severe fetal, 620369 (3), Autosomal recessive; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Congenital myopathy 22A, classic, 620351 (3), Autosomal recessive
SCN8A	99.77 %	600702	?Myoclonus, familial, 2, 618364 (3), Autosomal dominant; Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant; Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; Developmental and epileptic encephalopathy 13, 614558 (3), Autosomal dominant
SCP2	94.94 %	184755	?Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3), Autosomal recessive
SDHA	99.98 %	600857	Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Mitochondrial complex II deficiency, nuclear type 1, 252011 (3), Autosomal recessive; Neurodegeneration with ataxia and late-onset optic atrophy, 619259 (3), Autosomal dominant; Pheochromocytoma/paraganglioma syndrome 5, 614165 (3), Autosomal dominant
SDHAF1	99.99 %	612848	Mitochondrial complex II deficiency, nuclear type 2, 619166 (3), Autosomal recessive
SDHD	82.93 %	602690	Pheochromocytoma/paraganglioma syndrome 1, 168000 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Mitochondrial complex II deficiency, nuclear type 3, 619167 (3), Autosomal recessive
SEPSECS	99.78 %	613009	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive
SERAC1	99.9 %	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
SETX	99.97 %	608465	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant
SGCE	93.12 %	604149	Dystonia-11, myoclonic, 159900 (3), Autosomal dominant
SHQ1	99.81 %	613663	Neurodevelopmental disorder with dystonia and seizures, 619922 (3), Autosomal recessive; ?Dystonia 35, childhood-onset, 619921 (3), Autosomal recessive
SLC16A2	99.97 %	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
SLC18A2	100 %	193001	Parkinsonism-dystonia, infantile, 2, 618049 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SLC19A3	99.95 %	606152	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483 (3), Autosomal recessive
SLC20A2	99.95 %	158378	Basal ganglia calcification, idiopathic, 1, 213600 (3), Autosomal dominant
SLC25A42	99.99 %	610823	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 (3), Autosomal recessive
SLC2A1	99.93 %	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; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant
SLC30A10	99.99 %	611146	Hypermanganesemia with dystonia 1, 613280 (3), Autosomal recessive
SLC30A9	99.75 %	604604	Birk-Landau-Perez syndrome, 617595 (3), Autosomal recessive
SLC39A14	92.87 %	608736	?Hyperostosis cranalis interna, 144755 (3), Autosomal dominant; Hypermanganesemia with dystonia 2, 617013 (3), Autosomal recessive
SLC44A1	99.9 %	606105	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868 (3), Autosomal recessive
SLC6A3	99.96 %	126455	Parkinsonism-dystonia, infantile, 1, 613135 (3), Autosomal recessive; {Nicotine dependence, protection against}, 188890 (3)
SLC6A5	99.97 %	604159	Hyperekplexia 3, 614618 (3), Autosomal dominant, Autosomal recessive
SLC6A8	99.99 %	300036	Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive
SMPD1	100 %	607608	Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive
SNCA	99.97 %	163890	Dementia, Lewy body, 127750 (3), Autosomal dominant; Parkinson disease 1, 168601 (3), Autosomal dominant; Parkinson disease 4, 605543 (3), Autosomal dominant
SNCAIP	99.99 %	603779	*No OMIM phenotypes*
SNORD118	100 %	616663	Leukoencephalopathy, brain calcifications, and cysts, 614561 (3), Autosomal recessive
SPATA5L1	99.91 %	619578	Deafness, autosomal recessive 119, 619615 (3), Autosomal recessive; Neurodevelopmental disorder with hearing loss and spasticity, 619616 (3), Autosomal recessive
SPG11	99.89 %	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	99.99 %	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), ?Autosomal dominant, Autosomal recessive
SQSTM1	100 %	601530	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 (3), Autosomal recessive; Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 (3), Autosomal dominant; Myopathy, distal, with rimmed vacuoles, 617158 (3), Autosomal dominant; Paget disease of bone 3, 167250 (3), Autosomal dominant
STUB1	99.99 %	607207	Spinocerebellar ataxia 48, 618093 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 16, 615768 (3), Autosomal recessive
SUCLA2	99.96 %	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
SUCLG1	99.64 %	611224	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive
SUOX	100 %	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SURF1	100 %	185620	Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Mitochondrial complex IV deficiency, nuclear type 1, 220110 (3), Autosomal recessive
SYNJ1	99.91 %	604297	Parkinson disease 20, early-onset, 615530 (3), Autosomal recessive; Developmental and epileptic encephalopathy 53, 617389 (3), Autosomal recessive
SYT1	99.7 %	185605	Baker-Gordon syndrome, 618218 (3), Autosomal dominant
TAF1	99.84 %	313650	Intellectual developmental disorder, X-linked syndromic 33, 300966 (3), X-linked recessive; Dystonia-Parkinsonism, X-linked, 314250 (3), X-linked recessive
TANGO2	99.85 %	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
TARS2	99.77 %	612805	Combined oxidative phosphorylation deficiency 21, 615918 (3), Autosomal recessive
TBC1D24	100 %	613577	Deafness, autosomal recessive 86, 614617 (3), Autosomal recessive; Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Developmental and epileptic encephalopathy 16, 615338 (3), Autosomal recessive; DOORS syndrome, 220500 (3), Autosomal recessive
TBCD	100 %	604649	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 (3), Autosomal recessive
TGM6	99.99 %	613900	Spinocerebellar ataxia 35, 613908 (3), Autosomal dominant
TH	99.99 %	191290	Segawa syndrome, recessive, 605407 (3), Autosomal recessive
THAP1	99.96 %	609520	Dystonia 6, torsion, 602629 (3), Autosomal dominant
TIMM8A	100 %	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
TMEM107	100 %	616183	Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; ?Joubert syndrome 29, 617562 (3), Autosomal recessive
TMEM151A	100 %	620108	Episodic kinesigenic dyskinesia 3, 620245 (3), Autosomal dominant
TMEM240	99.99 %	616101	Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant
TOR1A	100 %	605204	{Dystonia-1, modifier of} (3); Arthrogryposis multiplex congenita 5, 618947 (3), Autosomal recessive; Dystonia-1, torsion, 128100 (3), Autosomal dominant
TPI1	99.95 %	190450	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3), Autosomal recessive
TPK1	99.96 %	606370	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 (3), Autosomal recessive
TPP1	99.99 %	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
TRAK1	99.98 %	608112	Developmental and epileptic encephalopathy 68, 618201 (3), Autosomal recessive
TRAPPC11	99.93 %	614138	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 (3), Autosomal recessive
TRAPPC12	99.96 %	614139	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669 (3), Autosomal recessive
TREX1	100 %	606609	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal dominant, Autosomal recessive; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant
TRIT1	99.25 %	617840	Combined oxidative phosphorylation deficiency 35, 617873 (3), Autosomal recessive
TSEN2	99.98 %	608753	Pontocerebellar hypoplasia type 2B, 612389 (3), Autosomal recessive
TSFM	100 %	604723	Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TSPOAP1	99.95 %	610764	Dystonia 22, juvenile-onset, 620453 (3), Autosomal recessive; ?Dystonia 22, adult-onset, 620456 (3), Autosomal recessive
TUBB4A	100 %	602662	Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant
TXN2	99.88 %	609063	?Combined oxidative phosphorylation deficiency 29, 616811 (3), Autosomal recessive
UBA5	99.95 %	610552	?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive; Developmental and epileptic encephalopathy 44, 617132 (3), Autosomal recessive
UBR5	99.87 %	608413	*No OMIM phenotypes*
UBTF	99.99 %	600673	Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant
UCHL1	99.99 %	191342	{?Parkinson disease 5, susceptibility to}, 613643 (3), Autosomal dominant; Spastic paraplegia 79A, autosomal dominant, 620221 (3), Autosomal dominant; Spastic paraplegia 79B, autosomal recessive, 615491 (3), Autosomal recessive
UFM1	99.25 %	610553	Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive
UGDH	99.77 %	603370	Developmental and epileptic encephalopathy 84, 618792 (3), Autosomal recessive
UQCRC1	99.99 %	191328	Parkinsonism with polyneuropathy, 619279 (3), Autosomal dominant
UQCRQ	99.96 %	612080	Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive
VAC14	99.91 %	604632	Striatonigral degeneration, childhood-onset, 617054 (3), Autosomal recessive
VAMP1	100 %	185880	Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive; Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant
VAMP2	100 %	185881	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760 (3), Autosomal dominant
VCP	99.99 %	601023	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2Y, 616687 (3), Autosomal dominant; Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 (3), Autosomal dominant
VPS13A	99.68 %	605978	Choreoacanthocytosis, 200150 (3), Autosomal recessive
VPS13C	99.65 %	608879	Parkinson disease 23, autosomal recessive, early onset, 616840 (3), Autosomal recessive
VPS13D	99.96 %	608877	Spinocerebellar ataxia, autosomal recessive 4, 607317 (3), Autosomal recessive
VPS16	100 %	608550	Dystonia 30, 619291 (3), Autosomal dominant
VPS35	99.83 %	601501	{Parkinson disease 17}, 614203 (3), Autosomal dominant
VPS41	99.92 %	605485	Spinocerebellar ataxia, autosomal recessive 29, 619389 (3), Autosomal recessive
VPS4A	99.98 %	609982	CIMDAG syndrome, 619273 (3), Autosomal dominant
WARS2	97.72 %	604733	Parkinsonism-dystonia 3, childhood-onset, 619738 (3), Autosomal recessive; Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 (3), Autosomal recessive
WDR45	99.99 %	300526	Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant
WDR73	99.92 %	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive
WFS1	99.99 %	606201	Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; ?Cataract 41, 116400 (3), Autosomal dominant; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive
XK	99.98 %	314850	McLeod syndrome, 300842 (3), X-linked
XPR1	99.13 %	605237	Basal ganglia calcification, idiopathic, 6, 616413 (3), Autosomal dominant
YIF1B	99.8 %	619109	Kaya-Barakat-Masson syndrome, 619125 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
YY1	100 %	600013	Gabriele-de Vries syndrome, 617557 (3), Autosomal dominant
ZC4H2	99.98 %	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant
ZNF142	100 %	604083	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425 (3), Autosomal recessive
ZSWIM6	98.94 %	615951	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 (3), Autosomal dominant; Acromelic frontonasal dysostosis, 603671 (3), Autosomal dominant

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2024-09-05**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

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.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.