Neuropathy panel

versie	v1 (211 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
ABCA1	600046	HDL deficiency, familial, 1, 604091 (3); Tangier disease, 205400 (3), Autosomal recessive
ABHD12	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
AGTPBP1	606830	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 (3), Autosomal recessive
AGXT	604285	Hyperoxaluria, primary, type 1, 259900 (3), Autosomal recessive
AHNAK2	No OMIM gene	No OMIM phenotype
AIFM1	300169	Cowchock syndrome, 310490 (3), X-linked recessive; Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 (3), X-linked recessive; Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive
AMACR	604489	Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive; Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive
AP1S1	603531	MEDNIK syndrome, 609313 (3), Autosomal recessive
APOA1	107680	Hypoalphalipoproteinemia, primary, 2, with or without corneal clouding, 618463 (3); Amyloidosis, 3 or more types, 105200 (3), Autosomal dominant; ApoA-I and apoC-III deficiency, combined, 618463 (3)
APTX	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
AR	313700	Hypospadias 1, X-linked, 300633 (3), X-linked recessive; Androgen insensitivity, 300068 (3), X-linked recessive; {Prostate cancer, susceptibility to}, 176807 (3), Autosomal dominant, Somatic mutation; Androgen insensitivity, partial, with or without breast cancer, 312300 (3), X-linked recessive; Spinal and bulbar muscular atrophy of Kennedy, 313200 (3), X-linked recessive
ARHGEF10	608136	?Slowed nerve conduction velocity, AD, 608236 (3), Autosomal dominant
ARSA	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
ASAH1	613468	Farber lipogranulomatosis, 228000 (3), Autosomal recessive; Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive

ASCC1	614215	Spinal muscular atrophy with congenital bone fractures 2, 616867 (3), Autosomal recessive; Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
ATAD3A	612316	Harel-Yoon syndrome, 617183 (3), Autosomal recessive, Autosomal dominant; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive
ATL1	606439	Spastic paraplegia 3A, autosomal dominant, 182600 (3), Autosomal dominant; Neuropathy, hereditary sensory, type ID, 613708 (3), Autosomal dominant
ATL3	609369	Neuropathy, hereditary sensory, type IF, 615632 (3), Autosomal dominant
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)
ATP1A1	182310	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 (3), Autosomal dominant; Hypomagnesemia, seizures, and mental retardation 2, 618314 (3), Autosomal dominant
ATP7A	300011	Occipital horn syndrome, 304150 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive
B4GALNT1	601873	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive
BAG3	603883	Cardiomyopathy, dilated, 1HH, 613881 (3), Autosomal dominant; Myopathy, myofibrillar, 6, 612954 (3), Autosomal dominant
BCKDHB	248611	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
BICD2	609797	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290 (3), Autosomal dominant
BSCL2	606158	Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive
C12orf65	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive
C1orf194	618682	No OMIM phenotype
CCT5	610150	Neuropathy, hereditary sensory, with spastic paraplegia, 256840 (3), Autosomal recessive
CD59	107271	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 (3), Autosomal recessive

CHCHD10	615903	Spinal muscular atrophy, Jokela type, 615048 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 (3), Autosomal dominant; ?Myopathy, isolated mitochondrial, autosomal dominant, 616209 (3), Autosomal dominant
CNTNAP1	602346	Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive
COA7	615623	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387 (3), Autosomal recessive
COX6A1	602072	Charcot-Marie-Tooth disease, recessive intermediate D, 616039 (3), Autosomal recessive
CPOX	612732	Harderoporphyria, 618892 (3), Autosomal recessive; Coproporphyria, 121300 (3), Autosomal recessive, Autosomal dominant
CTDP1	604927	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive
CYP27A1	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
DARS2	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
DCAF8	615820	?Giant axonal neuropathy 2, autosomal dominant, 610100 (3), Autosomal dominant
DCTN1	601143	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant; Perry syndrome, 168605 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, type VIIB, 607641 (3), Autosomal dominant
DEGS1	615843	Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive
DGAT2	606983	No OMIM phenotype
DGUOK	601465	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 (3), Autosomal recessive; Portal hypertension, noncirrhotic, 617068 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3), Autosomal recessive
DHTKD1	614984	2-aminoadipic 2-oxoadipic aciduria, 204750 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 (3), Autosomal dominant
DNAJB2	604139	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881 (3), Autosomal recessive
DNAJC3	601184	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192 (3), Autosomal recessive
DNM2	602378	Lethal congenital contracture syndrome 5, 615368 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal type 2M, 606482 (3), Autosomal dominant; Centronuclear myopathy 1, 160150 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate B, 606482 (3), Autosomal dominant

DNMT1	126375	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IE, 614116 (3), Autosomal dominant
DRP2	300052	No OMIM phenotype
DST	113810	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 (3), Autosomal recessive; Epidermolysis bullosa simplex, autosomal recessive 2, 615425 (3), Autosomal recessive
DYNC1H1	600112	Mental retardation, autosomal dominant 13, 614563 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 20, 614228 (3), Autosomal dominant
EGR2	129010	Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; Hypomyelinating neuropathy, congenital, 1, 605253 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, type 1D, 607678 (3), Autosomal dominant
ELP1	603722	Dysautonomia, familial, 223900 (3), Autosomal recessive
ERCC6	609413	{Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); {Lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; Premature ovarian failure 11, 616946 (3), Autosomal dominant; UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive
ERCC8	609412	Cockayne syndrome, type A, 216400 (3), Autosomal recessive; UV-sensitive syndrome 2, 614621 (3), Autosomal recessive
ETFDH	231675	Glutaric acidemia IIC, 231680 (3), Autosomal recessive
EXOSC3	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
EXOSC8	606019	Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive
EXOSC9	606180	Pontocerebellar hypoplasia, type 1D, 618065 (3), Autosomal recessive
FAH	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive
FAM126A	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
FBLN5	604580	Macular degeneration, age-related, 3, 608895 (3), Autosomal dominant; ?Cutis laxa, autosomal dominant 2, 614434 (3), Autosomal dominant; Neuropathy, hereditary, with or without age-related macular degeneration, 608895 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IA, 219100 (3), Autosomal recessive
FBXO38	608533	Neuronopathy, distal hereditary motor, type IID, 615575 (3), Autosomal dominant
FGD4	611104	Charcot-Marie-Tooth disease, type 4H, 609311 (3), Autosomal recessive
FIG4	609390	Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Charcot-Marie-Tooth disease, type 4J, 611228 (3),

		Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant
FLVCR1	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
FXN	606829	Friedreich ataxia with retained reflexes, 229300 (3), Autosomal recessive; Friedreich ataxia, 229300 (3), Autosomal recessive
GALC	606890	Krabbe disease, 245200 (3), Autosomal recessive
GAN	605379	Giant axonal neuropathy-1, 256850 (3), Autosomal recessive
GARS1 (GARS)	600287	Charcot-Marie-Tooth disease, type 2D, 601472 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant
GBA2	609471	Spastic paraplegia 46, autosomal recessive, 614409 (3), Autosomal recessive
GBF1	603698	No OMIM phenotype
GDAP1	606598	Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 (3), Autosomal recessive; Charcot-Marie-Tooth disease, type 4A, 214400 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2K, 607831 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 (3), Autosomal recessive
GJB1	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant
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
GLA	300644	Fabry disease, 301500 (3), X-linked; Fabry disease, cardiac variant, 301500 (3), X-linked
GNB4	610863	Charcot-Marie-Tooth disease, dominant intermediate F, 615185 (3), Autosomal dominant
GNE	603824	Sialuria, 269921 (3), Autosomal dominant; Nonaka myopathy, 605820 (3), Autosomal recessive
GSN	137350	Amyloidosis, Finnish type, 105120 (3), Autosomal dominant
HADHA	600890	LCHAD deficiency, 609016 (3), Autosomal recessive; HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; Mitochondrial trifunctional protein deficiency, 609015 (3), Autosomal recessive; Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive
HADHB	143450	Trifunctional protein deficiency, 609015 (3), Autosomal recessive
HARS1 (HARS)	142810	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 (3), Autosomal dominant; Usher syndrome type 3B, 614504 (3), Autosomal recessive
HINT1	601314	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200 (3), Autosomal recessive

HK1	142600	Hemolytic anemia due to hexokinase deficiency, 235700 (3), Autosomal recessive; Neuropathy, hereditary motor and sensory, Russe type, 605285 (3), Autosomal recessive; Neurodevelopmental disorder with visual defects and brain anomalies, 618547 (3), Autosomal dominant; Retinitis pigmentosa 79, 617460 (3), Autosomal dominant
HMBS	609806	Porphyria, acute intermittent, 176000 (3), Autosomal dominant; Porphyria, acute intermittent, nonerythroid variant, 176000 (3), Autosomal dominant
HSPB1	602195	Neuronopathy, distal hereditary motor, type IIB, 608634 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2F, 606595 (3), Autosomal dominant
HSPB3	604624	?Neuronopathy, distal hereditary motor, type IIC, 613376 (3), Autosomal dominant
HSPB8	608014	Neuronopathy, distal hereditary motor, type IIA, 158590 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2L, 608673 (3), Autosomal dominant
IARS2	612801	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 (3), Autosomal recessive
IGHMBP2	600502	Neuronopathy, distal hereditary motor, type VI, 604320 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2S, 616155 (3), Autosomal recessive
INF2	610982	Glomerulosclerosis, focal segmental, 5, 613237 (3); Charcot-Marie-Tooth disease, dominant intermediate E, 614455 (3), Autosomal dominant
ITPR3	147267	{Diabetes, type 1, susceptibility to}, 222100 (2), Autosomal recessive
JAG1	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3); Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
KARS1 (KARS)	601421	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive
KCNA2	176262	Epileptic encephalopathy, early infantile, 32, 616366 (3), Autosomal dominant
KIF1A	601255	NESCAV syndrome, 614255 (3), Autosomal dominant; Spastic paraplegia 30, autosomal dominant, 610357 (3), Autosomal recessive, Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal recessive, 610357 (3), Autosomal recessive, Autosomal dominant
KIF1B	605995	Pheochromocytoma, 171300 (3), Autosomal dominant; ?Charcot-Marie-Tooth disease, type 2A1, 118210 (3), Autosomal dominant; {Neuroblastoma, susceptibility to, 1}, 256700 (3), Autosomal dominant, Somatic mutation
KIF26B	614026	No OMIM phenotype

KIF5A	602821	Myoclonus, intractable, neonatal, 617235 (3), Autosomal dominant; Spastic paraplegia 10, autosomal dominant, 604187 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 (3), Autosomal dominant
LITAF	603795	Charcot-Marie-Tooth disease, type 1C, 601098 (3), Autosomal dominant
LMNA	150330	Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal recessive, Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive; Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Malouf syndrome, 212112 (3), Autosomal dominant
LRSAM1	610933	Charcot-Marie-Tooth disease, axonal, type 2P, 614436 (3), Autosomal recessive, Autosomal dominant
LYST	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
MARS1 (MARS)	156560	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant; Interstitial lung and liver disease, 615486 (3), Autosomal recessive
MCM3AP	603294	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124 (3), Autosomal recessive
MFN2	608507	Hereditary motor and sensory neuropathy VIA, 601152 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 (3), Autosomal dominant
MICAL1	607129	No OMIM phenotype
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
MME	120520	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 (3), Autosomal recessive, Autosomal dominant; ?Spinocerebellar ataxia 43, 617018 (3), Autosomal dominant
MORC2	616661	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 (3), Autosomal dominant
MPV17	137960	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive

MPZ	159440	Charcot-Marie-Tooth disease, type 2J, 607736 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1B, 118200 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; Hypomyelinating neuropathy, congenital, 2, 618184 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate D, 607791 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2I, 607677 (3), Autosomal dominant
MTMR2	603557	Charcot-Marie-Tooth disease, type 4B1, 601382 (3), Autosomal recessive
MTTP	157147	Abetalipoproteinemia, 200100 (3), Autosomal recessive; {Metabolic syndrome, protection against}, 605552 (3), Autosomal dominant
MYH14	608568	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 (3), Autosomal dominant; Deafness, autosomal dominant 4A, 600652 (3), Autosomal dominant
NAGA	104170	Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type I, 609241 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive
NAGLU	609701	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant
NDRG1	605262	Charcot-Marie-Tooth disease, type 4D, 601455 (3), Autosomal recessive
NEFH	162230	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924 (3), Autosomal dominant; ?{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant
NEFL	162280	Charcot-Marie-Tooth disease, type 1F, 607734 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate G, 617882 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2E, 607684 (3), Autosomal dominant
NGF	162030	Neuropathy, hereditary sensory and autonomic, type V, 608654 (3), Autosomal recessive
NMNAT2	608701	No OMIM phenotype
NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive
OPA1	605290	{Glaucoma, normal tension, susceptibility to}, 606657 (3); Behr syndrome, 210000 (3), Autosomal recessive; Optic atrophy 1, 165500 (3), Autosomal dominant; Optic atrophy plus syndrome, 125250 (3), Autosomal dominant; ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 (3), Autosomal recessive
OPA3	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant

PDK3	300906	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905 (3), X-linked dominant
PDYN	131340	Spinocerebellar ataxia 23, 610245 (3), Autosomal dominant
PEX10	602859	Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive; Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive
PEX7	601757	Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
PHYH	602026	Refsum disease, 266500 (3), Autosomal recessive
PLEKHG5	611101	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate C, 615376 (3), Autosomal recessive
PLP1	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
PMM2	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
PMP2	170715	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279 (3), Autosomal dominant
PMP22	601097	Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; ?Neuropathy, inflammatory demyelinating, 139393 (3), ?Autosomal dominant; Charcot-Marie-Tooth disease, type 1E, 118300 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant; Neuropathy, recurrent, with pressure palsies, 162500 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1A, 118220 (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
PNPLA6	603197	Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive
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
POLG2	604983	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 (3), Autosomal dominant;

		Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 (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
PPOX	600923	Porphyria variegata, 176200 (3), Autosomal dominant
PRDM12	616458	Neuropathy, hereditary sensory and autonomic, type VIII, 616488 (3), Autosomal recessive
PRKCG	176980	Spinocerebellar ataxia 14, 605361 (3), Autosomal dominant
PRPS1	311850	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Arts syndrome, 301835 (3), X-linked recessive; Gout, PRPS-related, 300661 (3), X-linked recessive
PRUNE1	617413	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481 (3), Autosomal recessive
PRX	605725	Charcot-Marie-Tooth disease, type 4F, 614895 (3), Autosomal recessive; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant
PTEN	601728	Prostate cancer, somatic, 176807 (3); {Glioma susceptibility 2}, 613028 (3); Cowden syndrome 1, 158350 (3), Autosomal dominant; Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant
PTPN11	176876	LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Noonan syndrome 1, 163950 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
PTRH2	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
RAB7A	602298	Charcot-Marie-Tooth disease, type 2B, 600882 (3), Autosomal dominant
RBM7	612413	No OMIM phenotype
REEP1	609139	Spastic paraplegia 31, autosomal dominant, 610250 (3), Autosomal dominant; ?Neuronopathy, distal hereditary motor, type VB, 614751 (3), Autosomal dominant
RETREG1	613114	Neuropathy, hereditary sensory and autonomic, type IIB, 613115 (3), Autosomal recessive
SACS	604490	Spastic ataxia, Charlevoix-Saguenay type, 270550 (3), Autosomal recessive
SBF1	603560	Charcot-Marie-Tooth disease, type 4B3, 615284 (3), Autosomal recessive

SBF2	607697	Charcot-Marie-Tooth disease, type 4B2, 604563 (3), Autosomal recessive
SCARB2	602257	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive
SCN10A	604427	Episodic pain syndrome, familial, 2, 615551 (3), Autosomal dominant
SCN11A	604385	Neuropathy, hereditary sensory and autonomic, type VII, 615548 (3), Autosomal dominant; Episodic pain syndrome, familial, 3, 615552 (3), Autosomal dominant
SCN9A	603415	Neuropathy, hereditary sensory and autonomic, type IID, 243000 (3), Autosomal recessive; Generalized epilepsy with febrile seizures plus, type 7, 613863 (3), Autosomal dominant; Small fiber neuropathy, 133020 (3), Autosomal dominant; Paroxysmal extreme pain disorder, 167400 (3), Autosomal dominant; Insensitivity to pain, congenital, 243000 (3), Autosomal recessive; Erythermalgia, primary, 133020 (3), Autosomal dominant; Febrile seizures, familial, 3B, 613863 (3), Autosomal dominant
SCO2	604272	Myopia 6, 608908 (3), Autosomal dominant; Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 (3), Autosomal recessive
SCYL1	607982	Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive
SEPTIN9 (SEPT9)	604061	Amyotrophy, hereditary neuralgic, 162100 (3), Autosomal dominant
SETX	608465	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant
SGPL1	603729	Nephrotic syndrome, type 14, 617575 (3), Autosomal recessive
SH3TC2	608206	Charcot-Marie-Tooth disease, type 4C, 601596 (3), Autosomal recessive; Mononeuropathy of the median nerve, mild, 613353 (3), Autosomal dominant
SIGMAR1	601978	?Amyotrophic lateral sclerosis 16, juvenile, 614373 (3), Autosomal recessive; ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 (3), Autosomal recessive
SLC12A6	604878	Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3), Autosomal recessive
SLC25A19	606521	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 (3), Autosomal recessive; Microcephaly, Amish type, 607196 (3), Autosomal recessive
SLC25A21	607571	?Mitochondrial DNA depletion syndrome 18, 618811 (3), Autosomal recessive
SLC25A46	610826	Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive
SLC52A2	607882	Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive

SLC52A3	613350	Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive; ?Fazio-Londe disease, 211500 (3), Autosomal recessive
SLC5A7	608761	Neuronopathy, distal hereditary motor, type VIIA, 158580 (3), Autosomal dominant; Myasthenic syndrome, congenital, 20, presynaptic, 617143 (3), Autosomal recessive
SMN1	600354	Spinal muscular atrophy-2, 253550 (3), Autosomal recessive; Spinal muscular atrophy-3, 253400 (3), Autosomal recessive; Spinal muscular atrophy-1, 253300 (3), Autosomal recessive; Spinal muscular atrophy-4, 271150 (3), Autosomal recessive
SMN2	601627	{Spinal muscular atrophy, type III, modifier of}, 253400 (3), Autosomal recessive
SORD	182500	Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912 (3), Autosomal recessive
SOX10	602229	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant; PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant
SPAST	604277	Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant
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
SPG7	602783	Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal recessive, Autosomal dominant
SPTAN1	182810	Epileptic encephalopathy, early infantile, 5, 613477 (3), Autosomal dominant
SPTBN4	606214	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 (3), Autosomal recessive
SPTLC1	605712	Neuropathy, hereditary sensory and autonomic, type IA, 162400 (3), Autosomal dominant
SPTLC2	605713	Neuropathy, hereditary sensory and autonomic, type IC, 613640 (3), Autosomal dominant
SUCLA2	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (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
TFG	602498	?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive; Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant
TRIM2	614141	Charcot-Marie-Tooth disease, type 2R, 615490 (3), Autosomal recessive

TRIP4	604501	Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive; ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive
TRPA1	604775	?Episodic pain syndrome, familial, 1, 615040 (3), Autosomal dominant
TRPV4	605427	Spondylometaphyseal dysplasia, Kozlowski type, 184252 (3), Autosomal dominant; Parastremmatic dwarfism, 168400 (3), Autosomal dominant; SED, Maroteaux type, 184095 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, type VIII, 600175 (3), Autosomal dominant; [Sodium serum level QTL 1], 613508 (3); Scapuloperoneal spinal muscular atrophy, 181405 (3), Autosomal dominant; Metatropic dysplasia, 156530 (3), Autosomal dominant; Digital arthropathy-brachydactyly, familial, 606835 (3), Autosomal dominant; Hereditary motor and sensory neuropathy, type IIc, 606071 (3), Autosomal dominant; Brachyolmia type 3, 113500 (3), Autosomal dominant; ?Avascular necrosis of femoral head, primary, 2, 617383 (3), Autosomal dominant
TTPA	600415	Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive
TTR	176300	Amyloidosis, hereditary, transthyretin-related, 105210 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant
TUBB3	602661	Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant
TWNK	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive
TYMP	131222	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive
UBA1	314370	Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive
VAPB	605704	Spinal muscular atrophy, late-onset, Finkel type, 182980 (3), Autosomal dominant; Amyotrophic lateral sclerosis 8, 608627 (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
VRK1	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive
WARS1 (WARS)	191050	Neuronopathy, distal hereditary motor, type IX, 617721 (3), Autosomal dominant

WNK1	605232	Pseudohypoaldosteronism, type IIC, 614492 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type II, 201300 (3), Autosomal recessive
XK	314850	McLeod syndrome with or without chronic granulomatous disease, 300842 (3), X-linked
XPA	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
XRCC1	194360	?Spinocerebellar ataxia, autosomal recessive 26, 617633 (3), Autosomal recessive
YARS1 (YARS)	603623	Charcot-Marie-Tooth disease, dominant intermediate C, 608323 (3), Autosomal dominant
ZFYVE26	612012	Spastic paraplegia 15, autosomal recessive, 270700 (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: Sep 01, 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.