

<b>Neuropathy panel</b>		
<b>versie</b>	v1 (211 genen)	Centrum voor Medische Genetica Gent
<b>Gene</b>	<b>OMIM gene ID</b>	<b>Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern</b>
<i>AARS1</i> ( <i>AARS</i> )	601065	Epileptic encephalopathy, early infantile, 29, 616339 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant
<i>ABCA1</i>	600046	HDL deficiency, familial, 1, 604091 (3); Tangier disease, 205400 (3), Autosomal recessive
<i>ABHD12</i>	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
<i>AGTPBP1</i>	606830	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 (3), Autosomal recessive
<i>AGXT</i>	604285	Hyperoxaluria, primary, type 1, 259900 (3), Autosomal recessive
<i>AHNAK2</i>	No OMIM gene	No OMIM phenotype
<i>AIFM1</i>	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
<i>AMACR</i>	604489	Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive; Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive
<i>AP1S1</i>	603531	MEDNIK syndrome, 609313 (3), Autosomal recessive
<i>APOA1</i>	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)
<i>APTX</i>	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
<i>AR</i>	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
<i>ARHGEF10</i>	608136	?Slowed nerve conduction velocity, AD, 608236 (3), Autosomal dominant
<i>ARSA</i>	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
<i>ASAH1</i>	613468	Farber lipogranulomatosis, 228000 (3), Autosomal recessive; Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive

<i>ASCC1</i>	614215	Spinal muscular atrophy with congenital bone fractures 2, 616867 (3), Autosomal recessive; Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
<i>ATAD3A</i>	612316	Harel-Yoon syndrome, 617183 (3), Autosomal recessive, Autosomal dominant; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive
<i>ATL1</i>	606439	Spastic paraplegia 3A, autosomal dominant, 182600 (3), Autosomal dominant; Neuropathy, hereditary sensory, type ID, 613708 (3), Autosomal dominant
<i>ATL3</i>	609369	Neuropathy, hereditary sensory, type IF, 615632 (3), Autosomal dominant
<i>ATM</i>	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)
<i>ATP1A1</i>	182310	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 (3), Autosomal dominant; Hypomagnesemia, seizures, and mental retardation 2, 618314 (3), Autosomal dominant
<i>ATP7A</i>	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
<i>B4GALNT1</i>	601873	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive
<i>BAG3</i>	603883	Cardiomyopathy, dilated, 1HH, 613881 (3), Autosomal dominant; Myopathy, myofibrillar, 6, 612954 (3), Autosomal dominant
<i>BCKDHB</i>	248611	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
<i>BICD2</i>	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
<i>BSCL2</i>	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
<i>C12orf65</i>	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive
<i>C1orf194</i>	618682	No OMIM phenotype
<i>CCT5</i>	610150	Neuropathy, hereditary sensory, with spastic paraplegia, 256840 (3), Autosomal recessive
<i>CD59</i>	107271	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 (3), Autosomal recessive

<i>CHCHD10</i>	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
<i>CNTNAP1</i>	602346	Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive
<i>COA7</i>	615623	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387 (3), Autosomal recessive
<i>COX6A1</i>	602072	Charcot-Marie-Tooth disease, recessive intermediate D, 616039 (3), Autosomal recessive
<i>CPOX</i>	612732	Harderoporphyria, 618892 (3), Autosomal recessive; Coproporphyria, 121300 (3), Autosomal recessive, Autosomal dominant
<i>CTDP1</i>	604927	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive
<i>CYP27A1</i>	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
<i>DARS2</i>	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
<i>DCAF8</i>	615820	?Giant axonal neuropathy 2, autosomal dominant, 610100 (3), Autosomal dominant
<i>DCTN1</i>	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
<i>DEGS1</i>	615843	Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive
<i>DGAT2</i>	606983	No OMIM phenotype
<i>DGUOK</i>	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
<i>DHTKD1</i>	614984	2-aminoadipic 2-oxoadipic aciduria, 204750 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 (3), Autosomal dominant
<i>DNAJB2</i>	604139	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881 (3), Autosomal recessive
<i>DNAJC3</i>	601184	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192 (3), Autosomal recessive
<i>DNM2</i>	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

<i>DNMT1</i>	126375	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IE, 614116 (3), Autosomal dominant
<i>DRP2</i>	300052	No OMIM phenotype
<i>DST</i>	113810	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 (3), Autosomal recessive; Epidermolysis bullosa simplex, autosomal recessive 2, 615425 (3), Autosomal recessive
<i>DYNC1H1</i>	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
<i>EGR2</i>	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
<i>ELP1</i>	603722	Dysautonomia, familial, 223900 (3), Autosomal recessive
<i>ERCC6</i>	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
<i>ERCC8</i>	609412	Cockayne syndrome, type A, 216400 (3), Autosomal recessive; UV-sensitive syndrome 2, 614621 (3), Autosomal recessive
<i>ETFDH</i>	231675	Glutaric acidemia IIC, 231680 (3), Autosomal recessive
<i>EXOSC3</i>	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
<i>EXOSC8</i>	606019	Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive
<i>EXOSC9</i>	606180	Pontocerebellar hypoplasia, type 1D, 618065 (3), Autosomal recessive
<i>FAH</i>	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive
<i>FAM126A</i>	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
<i>FBLN5</i>	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
<i>FBXO38</i>	608533	Neuronopathy, distal hereditary motor, type IID, 615575 (3), Autosomal dominant
<i>FGD4</i>	611104	Charcot-Marie-Tooth disease, type 4H, 609311 (3), Autosomal recessive
<i>FIG4</i>	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

<i>FLVCR1</i>	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
<i>FXN</i>	606829	Friedreich ataxia with retained reflexes, 229300 (3), Autosomal recessive; Friedreich ataxia, 229300 (3), Autosomal recessive
<i>GALC</i>	606890	Krabbe disease, 245200 (3), Autosomal recessive
<i>GAN</i>	605379	Giant axonal neuropathy-1, 256850 (3), Autosomal recessive
<i>GARS1</i> ( <i>GARS</i> )	600287	Charcot-Marie-Tooth disease, type 2D, 601472 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant
<i>GBA2</i>	609471	Spastic paraplegia 46, autosomal recessive, 614409 (3), Autosomal recessive
<i>GBF1</i>	603698	No OMIM phenotype
<i>GDAP1</i>	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
<i>GJB1</i>	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X- linked dominant
<i>GJC2</i>	608803	Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Lymphatic malformation 3, 613480 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive
<i>GLA</i>	300644	Fabry disease, 301500 (3), X-linked; Fabry disease, cardiac variant, 301500 (3), X-linked
<i>GNB4</i>	610863	Charcot-Marie-Tooth disease, dominant intermediate F, 615185 (3), Autosomal dominant
<i>GNE</i>	603824	Sialuria, 269921 (3), Autosomal dominant; Nonaka myopathy, 605820 (3), Autosomal recessive
<i>GSN</i>	137350	Amyloidosis, Finnish type, 105120 (3), Autosomal dominant
<i>HADHA</i>	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
<i>HADHB</i>	143450	Trifunctional protein deficiency, 609015 (3), Autosomal recessive
<i>HARS1</i> ( <i>HARS</i> )	142810	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 (3), Autosomal dominant; Usher syndrome type 3B, 614504 (3), Autosomal recessive
<i>HINT1</i>	601314	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200 (3), Autosomal recessive

<i>HK1</i>	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
<i>HMBS</i>	609806	Porphyria, acute intermittent, 176000 (3), Autosomal dominant; Porphyria, acute intermittent, nonerythroid variant, 176000 (3), Autosomal dominant
<i>HSPB1</i>	602195	Neuronopathy, distal hereditary motor, type IIB, 608634 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2F, 606595 (3), Autosomal dominant
<i>HSPB3</i>	604624	?Neuronopathy, distal hereditary motor, type IIC, 613376 (3), Autosomal dominant
<i>HSPB8</i>	608014	Neuronopathy, distal hereditary motor, type IIA, 158590 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2L, 608673 (3), Autosomal dominant
<i>IARS2</i>	612801	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 (3), Autosomal recessive
<i>IGHMBP2</i>	600502	Neuronopathy, distal hereditary motor, type VI, 604320 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2S, 616155 (3), Autosomal recessive
<i>INF2</i>	610982	Glomerulosclerosis, focal segmental, 5, 613237 (3); Charcot-Marie-Tooth disease, dominant intermediate E, 614455 (3), Autosomal dominant
<i>ITPR3</i>	147267	{Diabetes, type 1, susceptibility to}, 222100 (2), Autosomal recessive
<i>JAG1</i>	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3); Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
<i>KARS1 (KARS)</i>	601421	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive
<i>KCNA2</i>	176262	Epileptic encephalopathy, early infantile, 32, 616366 (3), Autosomal dominant
<i>KIF1A</i>	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
<i>KIF1B</i>	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
<i>KIF26B</i>	614026	No OMIM phenotype

<i>KIF5A</i>	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
<i>LITAF</i>	603795	Charcot-Marie-Tooth disease, type 1C, 601098 (3), Autosomal dominant
<i>LMNA</i>	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
<i>LRSAM1</i>	610933	Charcot-Marie-Tooth disease, axonal, type 2P, 614436 (3), Autosomal recessive, Autosomal dominant
<i>LYST</i>	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
<i>MARS1 (MARS)</i>	156560	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant; Interstitial lung and liver disease, 615486 (3), Autosomal recessive
<i>MCM3AP</i>	603294	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124 (3), Autosomal recessive
<i>MFN2</i>	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
<i>MICAL1</i>	607129	No OMIM phenotype
<i>MMACHC</i>	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
<i>MME</i>	120520	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 (3), Autosomal recessive, Autosomal dominant; ?Spinocerebellar ataxia 43, 617018 (3), Autosomal dominant
<i>MORC2</i>	616661	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 (3), Autosomal dominant
<i>MPV17</i>	137960	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive

<i>MPZ</i>	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
<i>MTMR2</i>	603557	Charcot-Marie-Tooth disease, type 4B1, 601382 (3), Autosomal recessive
<i>MTTP</i>	157147	Abetalipoproteinemia, 200100 (3), Autosomal recessive; {Metabolic syndrome, protection against}, 605552 (3), Autosomal dominant
<i>MYH14</i>	608568	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 (3), Autosomal dominant; Deafness, autosomal dominant 4A, 600652 (3), Autosomal dominant
<i>NAGA</i>	104170	Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type I, 609241 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive
<i>NAGLU</i>	609701	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant
<i>NDRG1</i>	605262	Charcot-Marie-Tooth disease, type 4D, 601455 (3), Autosomal recessive
<i>NEFH</i>	162230	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924 (3), Autosomal dominant; ?{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant
<i>NEFL</i>	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
<i>NGF</i>	162030	Neuropathy, hereditary sensory and autonomic, type V, 608654 (3), Autosomal recessive
<i>NMNAT2</i>	608701	No OMIM phenotype
<i>NTRK1</i>	191315	Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive
<i>OPA1</i>	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
<i>OPA3</i>	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
<i>PDHA1</i>	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant



<i>PDK3</i>	300906	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905 (3), X-linked dominant
<i>PDYN</i>	131340	Spinocerebellar ataxia 23, 610245 (3), Autosomal dominant
<i>PEX10</i>	602859	Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive; Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive
<i>PEX7</i>	601757	Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
<i>PHYH</i>	602026	Refsum disease, 266500 (3), Autosomal recessive
<i>PLEKHG5</i>	611101	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate C, 615376 (3), Autosomal recessive
<i>PLP1</i>	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
<i>PMM2</i>	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
<i>PMP2</i>	170715	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279 (3), Autosomal dominant
<i>PMP22</i>	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
<i>PNKP</i>	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
<i>PNPLA6</i>	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
<i>POLG</i>	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
<i>POLG2</i>	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
<i>POLR3A</i>	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive; Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive
<i>PPOX</i>	600923	Porphyria variegata, 176200 (3), Autosomal dominant
<i>PRDM12</i>	616458	Neuropathy, hereditary sensory and autonomic, type VIII, 616488 (3), Autosomal recessive
<i>PRKCG</i>	176980	Spinocerebellar ataxia 14, 605361 (3), Autosomal dominant
<i>PRPS1</i>	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
<i>PRUNE1</i>	617413	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481 (3), Autosomal recessive
<i>PRX</i>	605725	Charcot-Marie-Tooth disease, type 4F, 614895 (3), Autosomal recessive; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant
<i>PTEN</i>	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
<i>PTPN11</i>	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)
<i>PTRH2</i>	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
<i>RAB7A</i>	602298	Charcot-Marie-Tooth disease, type 2B, 600882 (3), Autosomal dominant
<i>RBM7</i>	612413	No OMIM phenotype
<i>REEP1</i>	609139	Spastic paraplegia 31, autosomal dominant, 610250 (3), Autosomal dominant; ?Neuronopathy, distal hereditary motor, type VB, 614751 (3), Autosomal dominant
<i>RETREG1</i>	613114	Neuropathy, hereditary sensory and autonomic, type IIB, 613115 (3), Autosomal recessive
<i>SACS</i>	604490	Spastic ataxia, Charlevoix-Saguenay type, 270550 (3), Autosomal recessive
<i>SBF1</i>	603560	Charcot-Marie-Tooth disease, type 4B3, 615284 (3), Autosomal recessive

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

<i>SLC52A3</i>	613350	Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive; ?Fazio-Londe disease, 211500 (3), Autosomal recessive
<i>SLC5A7</i>	608761	Neuronopathy, distal hereditary motor, type VIIA, 158580 (3), Autosomal dominant; Myasthenic syndrome, congenital, 20, presynaptic, 617143 (3), Autosomal recessive
<i>SMN1</i>	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
<i>SMN2</i>	601627	{Spinal muscular atrophy, type III, modifier of}, 253400 (3), Autosomal recessive
<i>SORD</i>	182500	Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912 (3), Autosomal recessive
<i>SOX10</i>	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
<i>SPAST</i>	604277	Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant
<i>SPG11</i>	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
<i>SPG7</i>	602783	Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal recessive, Autosomal dominant
<i>SPTAN1</i>	182810	Epileptic encephalopathy, early infantile, 5, 613477 (3), Autosomal dominant
<i>SPTBN4</i>	606214	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 (3), Autosomal recessive
<i>SPTLC1</i>	605712	Neuropathy, hereditary sensory and autonomic, type IA, 162400 (3), Autosomal dominant
<i>SPTLC2</i>	605713	Neuropathy, hereditary sensory and autonomic, type IC, 613640 (3), Autosomal dominant
<i>SUCLA2</i>	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
<i>SURF1</i>	185620	Leigh syndrome, due to COX IV deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive
<i>TFG</i>	602498	?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive; Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant
<i>TRIM2</i>	614141	Charcot-Marie-Tooth disease, type 2R, 615490 (3), Autosomal recessive

<i>TRIP4</i>	604501	Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive; ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive
<i>TRPA1</i>	604775	?Episodic pain syndrome, familial, 1, 615040 (3), Autosomal dominant
<i>TRPV4</i>	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
<i>TTPA</i>	600415	Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive
<i>TTR</i>	176300	Amyloidosis, hereditary, transthyretin-related, 105210 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant
<i>TUBB3</i>	602661	Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant
<i>TWINK</i>	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
<i>TYMP</i>	131222	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive
<i>UBA1</i>	314370	Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive
<i>VAPB</i>	605704	Spinal muscular atrophy, late-onset, Finkel type, 182980 (3), Autosomal dominant; Amyotrophic lateral sclerosis 8, 608627 (3), Autosomal dominant
<i>VCP</i>	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)
<i>VPS13A</i>	605978	Choreoacanthocytosis, 200150 (3), Autosomal recessive
<i>VRK1</i>	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive
<i>WARS1 (WARS)</i>	191050	Neuronopathy, distal hereditary motor, type IX, 617721 (3), Autosomal dominant

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