NEUROPATHIES GENE PANEL DG 2.16 (137 genes)

Releasedate: 07-06-2019

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AARS	103,7	100.0%	99.5%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
ABCD1	87,4	77.2%	75.0%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABHD12	93,1	100.0%	98.9%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
AIFM1	90	99.8%	96.7%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
ARHGEF10	119,5	99.8%	98.1%	?Slowed nerve conduction velocity, AD, 608236
ARSA	138,5	100.0%	100.0%	Metachromatic leukodystrophy, 250100
ATAD3A	90,3	93.6%	87.5%	Harel-Yoon syndrome, 617183
ATL1	134,7	99.9%	99.0%	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, autosomal dominant, 182600
ATL3	115,4	99.8%	97.7%	Neuropathy, hereditary sensory, type IF, 615632
ATP1A1	111,1	100.0%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 Hypomagnesemia, seizures, and mental retardation 2, 618314
ATP7A	111,2	99.5%	96.7%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
BAG3	171,4	100.0%	99.9%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BICD2	150,5	100.0%	99.6%	Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290 Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291
BSCL2	105,2	100.0%	100.0%	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685
C12orf65	110,4	100.0%	99.6%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035

CCT5	117,9	99.9%	98.9%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CHCHD10	26,1	63.1%	38.4%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048
CNTNAP1	157,2	99.9%	99.1%	Hypomyelinating neuropathy, congenital, 3, 618186 Lethal congenital contracture syndrome 7, 616286
COX6A1	148,3	100.0%	99.9%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
CTDP1	128,1	95.1%	88.0%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CYP27A1	173	100.0%	99.7%	Cerebrotendinous xanthomatosis, 213700
DCAF8	110,7	100.0%	99.7%	?Giant axonal neuropathy 2, autosomal dominant, 610100
DCTN1	112,6	99.9%	99.2%	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605 {Amyotrophic lateral sclerosis, susceptibility to}, 105400
DCTN2	89,7	100.0%	99.1%	No OMIM phenotype
DHTKD1	122,4	99.9%	98.8%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DNAH10	126,8	99.9%	99.1%	No OMIM phenotype
DNAJB2	118,4	100.0%	100.0%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB5	129,1	99.0%	95.2%	No OMIM phenotype
DNM2	123,9	99.7%	96.7%	Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368
DNMT1	114,3	99.2%	98.7%	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DRP2	82,4	98.6%	94.6%	No OMIM phenotype
DST	144,8	99.9%	99.2%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DYNC1H1	140,6	100.0%	99.6%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
EGR2	130	100.0%	100.0%	Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253
ELP1	118,7	99.7%	98.4%	Dysautonomia, familial, 223900

EXOSC8	89	98.2%	90.3%	Pontocerebellar hypoplasia, type 1C, 616081
FBLN5	96,6	91.8%	91.5%	Cutis laxa, autosomal dominant 2, 614434 Cutis laxa, autosomal recessive, type IA, 219100 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, with or without age-related macular degeneration, 608895
FBXO38	159,8	99.7%	98.3%	Neuronopathy, distal hereditary motor, type IID, 615575
FGD4	104,3	99.7%	97.8%	Charcot-Marie-Tooth disease, type 4H, 609311
FIG4	157,5	100.0%	99.6%	?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340
FLVCR1	146,1	99.9%	99.2%	Ataxia, posterior column, with retinitis pigmentosa, 609033
GALC	102,9	99.8%	98.8%	Krabbe disease, 245200
GAN	142,2	99.9%	99.4%	Giant axonal neuropathy-1, 256850
GARS	128,9	100.0%	99.7%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GBE1	157,4	99.9%	99.7%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GDAP1	145,6	99.8%	99.0%	Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, type 4A, 214400
GJB1	150,9	100.0%	99.9%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB3	228,5	100.0%	100.0%	Deafness, autosomal dominant 2B, 612644 Deafness, autosomal dominant, with peripheral neuropathy, 0 Deafness, autosomal recessive, 0 Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva 1, 133200
GLA	73,6	99.5%	95.8%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GNB4	150,3	100.0%	100.0%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
HADHB	77,6	96.7%	83.8%	Trifunctional protein deficiency, 609015
HARS	134,8	100.0%	100.0%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HINT1	63,3	91.2%	79.1%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200

HK1	116,5	100.0%	99.7%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Retinitis pigmentosa 79, 617460
HMBS	97,3	100.0%	98.4%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HOXD10	157,3	100.0%	100.0%	Charcot-Marie-Tooth disease, foot deformity of, 192950 Vertical talus, congenital, 192950
HSPB1	67,2	98.0%	91.9%	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 Neuropathy, distal hereditary motor, type IIB, 608634
HSPB3	226,1	100.0%	100.0%	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	213,9	100.0%	100.0%	Charcot-Marie-Tooth disease, axonal, type 2L, 608673 Neuropathy, distal hereditary motor, type IIA, 158590
IFRD1	138,3	99.8%	99.1%	No OMIM phenotype
IGHMBP2	108,3	99.6%	97.4%	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320
INF2	99,5	85.6%	83.5%	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 Glomerulosclerosis, focal segmental, 5, 613237
KARS	104,1	100.0%	98.8%	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KIF1A	115	99.7%	97.6%	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	139,6	100.0%	99.6%	?Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700
KIF5A	116	100.0%	99.9%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187 {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921
LITAF	102,2	95.2%	91.1%	Charcot-Marie-Tooth disease, type 1C, 601098
LMNA	104,7	97.7%	91.9%	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Malouf syndrome, 212112

				Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Restrictive dermopathy, lethal, 275210
LRIG3	154,4	99.9%	99.4%	No OMIM phenotype
LRSAM1	135,4	100.0%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
MARS	100,4	99.9%	98.0%	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 Interstitial lung and liver disease, 615486
MCM3AP	130,4	99.9%	99.2%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MED25	132,7	100.0%	99.7%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome, 616449
MFN2	122,8	100.0%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152
MME	112,7	99.7%	98.1%	?Spinocerebellar ataxia 43, 617018 Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MORC2	123,8	100.0%	99.5%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688
MPZ	125	100.0%	98.9%	Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Charcot-Marie-Tooth disease, type 1B, 118200 Charcot-Marie-Tooth disease, type 2I, 607677 Charcot-Marie-Tooth disease, type 2J, 607736 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 2, 618184 Roussy-Levy syndrome, 180800
MTMR2	99,1	99.9%	98.5%	Charcot-Marie-Tooth disease, type 4B1, 601382
MYH14	109,8	99.0%	95.1%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYO1A	101,9	100.0%	99.9%	?deafness,autosomal dominant 48,607841
NAGLU	117,7	97.1%	94.1%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NDRG1	114	100.0%	100.0%	Charcot-Marie-Tooth disease, type 4D, 601455
NEFH	110,6	99.5%	97.6%	?{Amyotrophic lateral sclerosis, susceptibility to}, 105400 Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
NEFL	178,7	99.8%	97.8%	Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, type 2E, 607684
NGF	199	100.0%	100.0%	Neuropathy, hereditary sensory and autonomic, type V, 608654

NIPA1	156,7	100.0%	99.9%	Spastic paraplegia 6, autosomal dominant, 600363
NTRK1	133	100.0%	99.3%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
PDK3	108,4	97.4%	94.5%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PEX1	127,9	99.9%	99.3%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX7	111	91.2%	89.3%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHYH	74	99.9%	96.9%	Refsum disease, 266500
PLA2G6	111,9	99.8%	98.2%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLEKHG5	101,6	99.8%	97.7%	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 Spinal muscular atrophy, distal, autosomal recessive, 4, 611067
PMM2	127,7	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PMP22	96,3	98.9%	94.3%	?Neuropathy, inflammatory demyelinating, 139393 Charcot-Marie-Tooth disease, type 1A, 118220 Charcot-Marie-Tooth disease, type 1E, 118300 Dejerine-Sottas disease, 145900 Neuropathy, recurrent, with pressure palsies, 162500 Roussy-Levy syndrome, 180800
PRDM12	138,9	92.6%	90.4%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRPS1	111,6	100.0%	99.9%	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661
PRX	156,5	100.0%	99.9%	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	98,1	100.0%	99.3%	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
RAB7A	115,3	100.0%	99.9%	Charcot-Marie-Tooth disease, type 2B, 600882
REEP1	71,5	78.6%	76.2%	?Neuronopathy, distal hereditary motor, type VB, 614751

				Spastic paraplegia 31, autosomal dominant, 610250
RETREG1	126,9	99.7%	98.8%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
SBF1	122	99.4%	97.9%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	107,7	99.9%	99.0%	Charcot-Marie-Tooth disease, type 4B2, 604563
SCN10A	133,3	100.0%	99.4%	Episodic pain syndrome, familial, 2, 615551
SCN11A	122,1	99.3%	97.1%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	128,4	99.1%	97.7%	Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863 HSAN2D, autosomal recessive, 243000 Insensitivity to pain, congenital, 243000 Paroxysmal extreme pain disorder, 167400 Small fiber neuropathy, 133020 {Dravet syndrome, modifier of}, 607208
SCO2	115,7	100.0%	99.9%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SEPT9	NC	NC	NC	Amyotrophy, hereditary neuralgic, 162100 Leukemia, acute myeloid, therapy-related, 0 Ovarian carcinoma, 0
SETX	151,6	100.0%	99.6%	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002
SH3TC2	102,1	100.0%	99.4%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SIGMAR1	146,6	100.0%	100.0%	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
SLC12A6	118,9	100.0%	99.9%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC25A46	173	99.8%	98.3%	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC52A2	185,4	100.0%	100.0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	118,8	100.0%	99.8%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A7	100,2	100.0%	99.9%	Myasthenic syndrome, congenital, 20, presynaptic, 617143 Neuronopathy, distal hereditary motor, type VIIA, 158580
SOX10	88,2	100.0%	99.1%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266

SPG11	116,1	99.7%	98.4%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPTBN4	103,8	99.8%	98.1%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SPTLC1	108,6	98.5%	93.4%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	142,8	100.0%	99.9%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SPTLC3	132,9	100.0%	99.7%	No OMIM phenotype
SURF1	84,8	91.3%	88.4%	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000
SYT2	96	99.8%	98.4%	Myasthenic syndrome, congenital, 7, presynaptic, 616040
TDP1	103,9	99.9%	99.5%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TFG	106,2	96.8%	95.3%	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TRIM2	136,6	93.9%	93.6%	Charcot-Marie-Tooth disease, type 2R, 615490
TRPV4	138,4	100.0%	99.8%	?Avascular necrosis of femoral head, primary, 2, 617383 Brachyolmia type 3, 113500 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Metatropic dysplasia, 156530 Parastremmatic dwarfism, 168400 Scapuloperoneal spinal muscular atrophy, 181405 SED, Maroteaux type, 184095 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 [Sodium serum level QTL 1], 613508
TTR	122,6	94.6%	94.6%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680
TUBB3	121,3	99.8%	98.4%	Cortical dysplasia, complex, with other brain malformations 1, 614039 Fibrosis of extraocular muscles, congenital, 3A, 600638
TWNK	159,6	100.0%	100.0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
VCP	100,3	100.0%	99.2%	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320

VRK1	129,6	99.8%	98.7%	Pontocerebellar hypoplasia type 1A, 607596
WARS	97,6	99.5%	97.0%	Neuronopathy, distal hereditary motor, type IX, 617721
WNK1	134,4	100.0%	99.5%	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
YARS	105,5	100.0%	99.2%	Charcot-Marie-Tooth disease, dominant intermediate C, 608323

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors