

NEUROPATHIES HMSN GENE PANEL DG 2.11 (78 genes)

<i>Gene</i>	<i>Median</i>	<i>% covered > 10x</i>	<i>% covered > 20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
AARS	124.4	100	99	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
ABHD12	107	97	87	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
AIFM1	107	100	99	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614
ATL1	160.3	99	97	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, autosomal dominant, 182600
ATP7A	134.1	99	97	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
BSCL2	113.7	100	100	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
C10orf2	179	100	100	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
COX6A1	181.3	100	99	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
CTDP1	105.3	86	83	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
DCTN1	131.9	99	98	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605 {Amyotrophic lateral sclerosis, susceptibility to}, 105400
DHH	117.7	100	100	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DHTKD1	141.1	99	98	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DNAJB2	102.1	100	100	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881

DNM2	127.9	97	94	Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Lethal congenital contracture syndrome 5, 615368 Myopathy, centronuclear, 160150
DNMT1	113.5	99	98	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DYNC1H1	180	100	99	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
EGR2	124.6	100	100	Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900 Neuropathy, congenital hypomyelinating, 1, 605253
EXOSC8	79.9	91	76	Pontocerebellar hypoplasia, type 1C, 616081
FAM134B	126.1	95	89	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
FGD4	110.2	98	96	Charcot-Marie-Tooth disease, type 4H, 609311
FIG4	154.9	99	98	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691
GAN	190.2	100	99	Giant axonal neuropathy-1, 256850
GARS	125.1	99	98	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy, distal hereditary motor, type VA, 600794
GDAP1	163	99	96	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	231.2	100	99	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GNB4	152.5	100	99	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
HINT1	60	98	88	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HOXD10	137.8	100	99	Charcot-Marie-Tooth disease, foot deformity of, 192950 Vertical talus, congenital, 192950
HSPB1	39.9	93	81	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 Neuropathy, distal hereditary motor, type IIB, 608634
HSPB8	156.4	100	100	Charcot-Marie-Tooth disease, axonal, type 2L, 608673

				Neuropathy, distal hereditary motor, type IIA, 158590
IGHMBP2	107.9	99	96	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320
IKBKAP	142.7	99	98	Dysautonomia, familial, 223900
INF2	88.2	93	90	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 Glomerulosclerosis, focal segmental, 5, 613237
KARS	122.7	100	99	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KIF1A	114.2	99	96	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	154.8	100	99	Pheochromocytoma, 171300 ?Charcot-Marie-Tooth disease, type 2A1, 118210 {Neuroblastoma, susceptibility to, 1}, 256700
KIF5A	136.3	100	99	Spastic paraplegia 10, autosomal dominant, 604187
LITAF	126.6	94	91	Charcot-Marie-Tooth disease, type 1C, 601098
LMNA	89.4	98	91	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, AD, 181350 Emery-Dreifuss muscular dystrophy 3, AR, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Muscular dystrophy, limb-girdle, type 1B, 159001 Restrictive dermopathy, lethal, 275210
LRSAM1	130.5	100	99	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
MED25	104.1	99	95	Basel-Vanagait-Smirin-Yosef syndrome, 616449 ?Charcot-Marie-Tooth disease, type 2B2, 605589
MFN2	150.9	100	99	Charcot-Marie-Tooth disease, type 2A2A, 609260 Charcot-Marie-Tooth disease, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152

MME	100.6	98	93	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 ?Spinocerebellar ataxia 43, 617018
MPZ	118.7	99	97	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 Neuropathy, congenital hypomyelinating, 605253 Roussy-Levy syndrome, 180800
MTMR2	106.5	100	99	Charcot-Marie-Tooth disease, type 4B1, 601382
NDRG1	128.7	99	98	Charcot-Marie-Tooth disease, type 4D, 601455
NEFL	165	99	98	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, type 2E, 607684
NGF	258.2	100	100	Neuropathy, hereditary sensory and autonomic, type V, 608654
NTRK1	130.9	99	97	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
PHYH	74.7	97	90	Refsum disease, 266500
PLEKHG5	87	96	88	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 Spinal muscular atrophy, distal, autosomal recessive, 4, 611067
PMP22	111.4	96	91	Charcot-Marie-Tooth disease, type 1A, 118220 Charcot-Marie-Tooth disease, type 1E, 118300 Dejerine-Sottas disease, 145900 Neuropathy, inflammatory demyelinating, 139393 Neuropathy, recurrent, with pressure palsies, 162500 Roussy-Levy syndrome, 180800
PRPS1	150.5	100	100	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	115.8	99	98	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900

PSAP	114.3	99	99	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
RAB7A	158.2	100	100	Charcot-Marie-Tooth disease,type 2B, 600882
SBF2	116.9	99	96	Charcot-Marie-Tooth disease, type 4B2, 604563
SCN10A	165.5	100	99	Episodic pain syndrome, familial, 2, 615551
SCN11A	138	99	97	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	148.3	98	97	Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Erythralgia, 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
SEPT9	118.8	99	96	Amyotrophy, hereditary neuralgic, 162100 Leukemia, acute myeloid, therapy-related Ovarian carcinoma
SH3TC2	122.8	100	99	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SLC12A6	141.8	100	99	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC52A2	177.8	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	119.7	100	100	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SLC5A7	117.2	100	99	Neuronopathy, distal hereditary motor, type VIIA, 158580
SMN1	116.3	99	96	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150
SOX10	65.8	98	91	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SPTLC1	115.4	98	94	Neuropathy, hereditary sensory and autonomic, type IA, 162400

SPTLC2	160.3	100	100	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SYT2	101.3	100	99	Myasthenic syndrome, congenital, 7, presynaptic, 616040
TDP1	123	98	95	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TFG	125.3	96	93	Hereditary motor and sensory neuropathy, Okinawa type, 604484 ?Spastic paraplegia 57, autosomal recessive, 615658
TRPV4	172.7	99	98	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	159.7	100	100	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680
VCP	144.9	99	99	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
WNK1	167.2	99	99	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypaldosteronism, type IIC, 614492
YARS	122.6	100	100	Charcot-Marie-Tooth disease, dominant intermediate C, 608323

Gene symbols used follow HGNC 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 : April 14th, 2017.

This list is accurate for panel version DG 2.11

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors