NEUROPATHIES HMSN GENE PANEL DG 2.5/2.6

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AARS	116.4	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy,early infantile,29,616339
AIFM1	76.8	100%	100%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome,310490
ATL1	165.2	97%	91%	Neuropathy,hereditary sensory,type 1D,613708 Spastic paraplegia 3A, autosomal dominant, 182600
BSCL2	107.4	100%	98%	Encephalopathy,progressive,with/without lipodystrophy,615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy,distal hereditary motor,type VA,600794 Silver spastic paraplegia syndrome,270685
C10orf2	154	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type),271245 Perrault syndrome 5,616138 Progressive external ophthalmoplegia with mitochondrial DNA depletions, autosomal dominant,609286
COX6A1	160.3	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
DCTN1	119.2	100%	98%	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome,168605 {Amyotrophic lateral sclerosis,susceptibility to},105400
DHTKD1	139.3	98%	97%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DNAJB2	104.9	100%	100%	?Charcot-Marie-Tooth disease,axonal, type 2T,616233 Spinal muscular atrophy, distal, autosomal recessive,5, 614881
DNM2	116	96%	93%	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	104.1	99%	98%	Cerebellar ataxia,deafness,and narcolepsy,autosomal dominant,604121 Neuropathy, hereditary sensory, type IE, 614116

DYNC1H1	166.1	99%	98%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation,autosomal dominant 13,614563 Spinal muscular atrophy,lower extremity-predominant 1,AD,158600
EGR2	110	100%	100%	Charcot-Marie-Tooth disease,type 1D,607678 Dejerine-Sottas disease,145900 Neuropathy, congenital hypomyelinating, 1, 605253
EXOSC8	75	85%	72%	Pontocerebellar hypoplasia,type 1C,616081
FAM134B	104.6	93%	81%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
FGD4	108.6	96%	92%	Charcot-Marie-Tooth disease, type 4H, 609311
FIG4	155.7	100%	99%	?Polymicrogyria,bilateral temporooccipital,612691 Amyotrophic lateral sclerosis 11,612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome,216340
GAN	179.2	99%	98%	Giant axonal neuropathy-1, 256850
GARS	127.7	100%	99%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy,distal hereditary motor,type VA,600794
GDAP1	170.7	100%	99%	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	114.6	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GNB4	140.3	100%	98%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
HINT1	49.4	96%	81%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HSPB1	28.6	95%	63%	Charcot-Marie-Tooth disease,axonal,type 2F,606595 Neuropathy, distal hereditary motor, type IIB, 608634
HSPB3	305.4	100%	100%	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	126.7	100%	100%	Charcot-Marie-Tooth disease,axonal,type 2L,608673 Neuropathy, distal hereditary motor, type IIA, 158590
IGHMBP2	94.6	100%	87%	Charcot-Marie-Tooth disease,axonal,type 2S,616155 Neuronopathy, distal hereditary motor, type VI, 604320
IKBKAP	140.6	100%	99%	Dysautonomia, familial, 223900
INF2	76.5	92%	86%	Charcot-Marie-Tooth disease,dominant intermediate E,614455 Glomerulosclerosis, focal segmental, 5, 613237
KARS	116.5	100%	98%	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641

				Deafness,autosomal recessive 89,613916
KIF1A	107.9	98%	94%	Mental retardation,autosomal dominant 9,614255 Neuropathy,hereditary sensory,type IIC,614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	149.8	99%	98%	?Charcot-Marie-Tooth disease,type 2A1,118210 Pheochromocytoma,171300 {Neuroblastoma,susceptibility to,1},256700
KIF5A	126.5	100%	99%	Spastic paraplegia 10, autosomal dominant, 604187
LITAF	115.7	99%	93%	Charcot-Marie-Tooth disease, type 1C, 601098
LMNA	72	95%	87%	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
LRSAM1	122.7	100%	99%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
MED25	82.6	97%	85%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome,616449
MFN2	138	100%	100%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VIA,601152
MPZ	88.5	100%	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
MTMR2	115	100%	98%	Charcot-Marie-Tooth disease, type 4B1, 601382
NDRG1	119.4	100%	97%	Charcot-Marie-Tooth disease, type 4D, 601455
NEFL	131.5	99%	97%	Charcot-Marie-Tooth disease, type 1F,607734 Charcot-Marie-Tooth disease, type 2E, 607684
NGF	227.1	100%	100%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NTRK1	115.4	97%	93%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma,familial,155240
PLEKHG5	79.2	92%	85%	Charcot-Marie-Tooth disease,recessive intermediate C,615376 Spinal muscular atrophy, distal, autosomal recessive, 4, 611067

PMP22	93.2	87%	84%	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	113.7	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	92	100%	99%	Charcot-Marie-Tooth disease,type 4F,614895 Dejerine-Sottas disease, autosomal recessive, 145900
RAB7A	151.6	100%	100%	Charcot-Marie-Tooth disease,type 2B, 600882
SBF2	117	99%	95%	Charcot-Marie-Tooth disease, type 4B2, 604563
SCN10A	164	99%	98%	Episodic pain syndrome,familial 2,615551
SCN11A	131.8	98%	97%	Episodic pain syndrome, familial, 3, 615552 Neuropathy,hereditary sensory and autonomic,type VIII,615548
SCN9A	146.4	96%	94%	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
SH3TC2	103.3	100%	99%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve,mild,613353
SLC12A6	144.7	100%	100%	Agenesis of the corpus callosum with peripheral neuropathy,218000
SLC52A3	104.7	100%	98%	Brown-Vialetto-Van Laere syndrome,211530 Fazio-Londe disease,211500
SLC5A7	121	100%	100%	Neuronopathy, distal hereditary motor, type VIIA, 158580
SMN1	73.3	99%	91%	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150

SPTLC1	107.7	100%	92%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	159.1	100%	100%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SYT2	92.6	100%	98%	Myasthenic syndrome, presynaptic, congenital, with or without motor neuropathy, 616040
TFG	118.8	95%	90%	?Spastic paraplegia 57,autosomal recessive,615658 Hereditary motor and sensory neuropathy,Okinawa type,604484
TRPV4	173.3	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
TTR	138.6	100%	100%	Amyloidosis,hereditary,transthyretin-related,105210 Carpal tunnel syndrome,familial,115430 [Dystransthyretinemic hyperthyroxinemia],145680
VCP	147	100%	99%	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
WNK1	159.4	98%	97%	Neuropathy,hereditary sensory and autonomic, type II,201300 Pseudohypoaldosteronism, type IIC, 614492
YARS	119.6	100%	100%	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 : April 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.

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