NEUROPATHIES HMSN GENE PANEL DG 2.4.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AARS	91.7	98%	95%	*Charcot-Marie-Tooth disease, axonal, type 2N, 613287* *Epileptic encephalopathy,early infantile,29,616339*
AIFM1	58.2	99%	87%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome,310490
ATL1	106.5	100%	99%	Neuropathy,hereditary sensory,type 1D,613708 Spastic paraplegia 3A, autosomal dominant, 182600
BSCL2	110.2	100%	100%	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	138.3	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	99.9	74%	74%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
DCTN1	113.6	99%	96%	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome,168605 {Amyotrophic lateral sclerosis,susceptibility to},105400
DHTKD1	103.3	100%	98%	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DNAJB2	107.5	100%	96%	?Charcot-Marie-Tooth disease,axonal, type 2T,616233 Spinal muscular atrophy, distal, autosomal recessive,5, 614881
DNM2	80.9	100%	96%	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	101.7	99%	96%	Cerebellar ataxia,deafness,and narcolepsy,autosomal dominant,604121 Neuropathy, hereditary sensory, type IE, 614116

DYNC1H1	117.4	99%	96%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation,autosomal dominant 13,614563 Spinal muscular atrophy,lower extremity-predominant 1,AD,158600
EGR2	73.2	100%	99%	Charcot-Marie-Tooth disease,type 1D,607678 Dejerine-Sottas disease,145900 Neuropathy, congenital hypomyelinating, 1, 605253
EXOSC8	69.2	98%	92%	Pontocerebellar hypoplasia,type 1C,616081
FAM134B	89.2	100%	99%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
FGD4	114.5	93%	93%	Charcot-Marie-Tooth disease, type 4H, 609311
FIG4	132.9	100%	99%	?Polymicrogyria,bilateral temporooccipital,612691 Amyotrophic lateral sclerosis 11,612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome,216340
GAN	147.9	100%	99%	Giant axonal neuropathy-1, 256850
GARS	108	98%	95%	Charcot-Marie-Tooth disease, type 2D, 601472 Neuropathy,distal hereditary motor,type VA,600794
GDAP1	106.2	100%	100%	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	85.4	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GNB4	144.4	100%	100%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
HINT1	79	97%	84%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HSPB1	50.1	98%	80%	Charcot-Marie-Tooth disease,axonal,type 2F,606595 Neuropathy, distal hereditary motor, type IIB, 608634
HSPB3	208.7	100%	100%	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	98.5	100%	100%	Charcot-Marie-Tooth disease,axonal,type 2L,608673 Neuropathy, distal hereditary motor, type IIA, 158590
IGHMBP2	70.4	97%	88%	Charcot-Marie-Tooth disease,axonal,type 2S,616155 Neuronopathy, distal hereditary motor, type VI, 604320
IKBKAP	106.1	100%	99%	Dysautonomia, familial, 223900

INF2	67.9	93%	85%	Charcot-Marie-Tooth disease,dominant intermediate E,614455 Glomerulosclerosis, focal segmental, 5, 613237
KARS	116.2	100%	100%	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness,autosomal recessive 89,613916
KIF1A	71	98%	92%	Mental retardation,autosomal dominant 9,614255 Neuropathy,hereditary sensory,type IIC,614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	130.6	100%	100%	?Charcot-Marie-Tooth disease,type 2A1,118210 Pheochromocytoma,171300 {Neuroblastoma,susceptibility to,1},256700
KIF5A	94.4	99%	98%	Spastic paraplegia 10, autosomal dominant, 604187
LITAF	75.6	94%	89%	Charcot-Marie-Tooth disease, type 1C, 601098
LMNA	72.5	97%	90%	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	81.4	100%	97%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
MED25	98.4	99%	87%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Basel-Vanagait-Smirin-Yosef syndrome,616449
MFN2	103.5	100%	97%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VIA,601152
MPZ	100.9	100%	95%	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	106.9	100%	100%	Charcot-Marie-Tooth disease, type 4B1, 601382
NDRG1	82.9	97%	92%	Charcot-Marie-Tooth disease, type 4D, 601455
NEFL	133.8	100%	100%	Charcot-Marie-Tooth disease, type 1F,607734 Charcot-Marie-Tooth disease, type 2E, 607684
NGF	180	100%	100%	Neuropathy, hereditary sensory and autonomic, type V, 608654

NTRK1	67.9	99%	94%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma,familial,155240
PLEKHG5	80.3	96%	93%	Charcot-Marie-Tooth disease,recessive intermediate C,615376 Spinal muscular atrophy, distal, autosomal recessive, 4, 611067
PMP22	97.9	100%	99%	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	62.3	99%	97%	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	113.4	99%	98%	Charcot-Marie-Tooth disease,type 4F,614895 Dejerine-Sottas disease, autosomal recessive, 145900
RAB7A	78.9	100%	100%	Charcot-Marie-Tooth disease,type 2B, 600882
SBF2	104.6	99%	97%	Charcot-Marie-Tooth disease, type 4B2, 604563
SCN10A	125.2	99%	98%	Episodic pain syndrome,familial 2,615551
SCN11A	121.5	99%	98%	Episodic pain syndrome, familial, 3, 615552 Neuropathy,hereditary sensory and autonomic,type VIII,615548
SCN9A	113.4	100%	99%	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	98.3	97%	96%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve,mild,613353
SLC12A6	96.5	100%	99%	Agenesis of the corpus callosum with peripheral neuropathy,218000
SLC52A3	69.1	100%	95%	Brown-Vialetto-Van Laere syndrome,211530 Fazio-Londe disease,211500

SLC5A7	109.8	100%	100%	Neuronopathy, distal hereditary motor, type VIIA, 158580
SMN1	2.3	10%	0%	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150
SPTLC1	81	95%	90%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	110	100%	100%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SYT2	91.5	88%	85%	Myasthenic syndrome, presynaptic, congenital, with or without motor neuropathy, 616040
TFG	108.2	100%	97%	?Spastic paraplegia 57,autosomal recessive,615658 Hereditary motor and sensory neuropathy,Okinawa type,604484
TRPV4	94.3	100%	99%	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	87.4	100%	99%	Amyloidosis,hereditary,transthyretin-related,105210 Carpal tunnel syndrome,familial,115430 [Dystransthyretinemic hyperthyroxinemia],145680
VCP	111.2	99%	96%	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
WNK1	143.2	100%	99%	Neuropathy,hereditary sensory and autonomic, type II,201300 Pseudohypoaldosteronism, type IIC, 614492
YARS	107.6	100%	97%	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
OMIM release used for OMIM disease identifiers and descriptions : November 15th, 2015
This list is accurate for all panel versions starting with DG 2.4. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

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