NEUROPATHIES (HMSN) PANEL DGD20062014

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
AARS	91,7	98%	95%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287
AIFM1	60,8	99%	87%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490
ATL1	106,6	100%	99%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
BSCL2	114,3	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type V, 600794
DCTN1	113,7	99%	96%	Neuropathy, distal hereditary motor, type VIIB, 607641 {Amyotrophic lateral sclerosis, susceptibility to}, 105400 Perry syndrome, 168605
DHTKD1	103,3	100%	98%	2-aminoadipic 2-oxoadipic aciduria, 204750 Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DNM2	81,5	100%	96%	Charcot-Marie-Tooth disease, dominant intermediate B, 606482 Myopathy, centronuclear, 160150 Charcot-Marie-Tooth disease, axonal, type 2M, 606482 Lethal congenital contracture syndrome 5, 615368
DNMT1	102,8	99%	97%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
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, AD, 158600
EGR2	73,2	100%	99%	Neuropathy, congenital hypomyelinating, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900
FAM134B	88,7	100%	99%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115

FGD4	124,3	100%	100%	Charcot-Marie-Tooth disease, type 4H, 609311
FIG4	132,9	100%	99%	Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577 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 V, 600794
GDAP1	106,2	100%	100%	Charcot-Marie-Tooth disease, type 4A, 214400 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340
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	80,6	96%	85%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HSPB1	50,1	98%	80%	Neuropathy, distal hereditary motor, type IIB, 608634 Charcot-Marie-Tooth disease, axonal, type 2F, 606595
HSPB3	208,7	100%	100%	Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	98,5	100%	100%	Neuropathy, distal hereditary motor, type IIA, 158590 Charcot-Marie-Tooth disease, axonal, type 2L, 608673
IGHMBP2	70,4	97%	88%	Neuronopathy, distal hereditary motor, type VI, 604320
IKBKAP	106,1	100%	99%	Dysautonomia, familial, 223900
INF2	67,4	93%	85%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
KARS	117,9	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916
KIF1A	71	98%	92%	Spastic paraplegia 30, autosomal recessive, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255

KIF1B	118,8	100%	100%	Charcot-Marie-Tooth disease, type 2A1, 118210 Pheochromocytoma, 171300 {Neuroblastoma, susceptibility to, 1}, 256700
LITAF	91,6	100%	100%	Charcot-Marie-Tooth disease, type 1C, 601098
LMNA	72,2	97%	89%	Emery-Dreifuss muscular dystrophy 2, AD, 181350 Cardiomyopathy, dilated, 1A, 115200 Lipodystrophy, familial partial, 2, 151660 Emery-Dreifuss muscular dystrophy 3, AR, 181350 Charcot-Marie-Tooth disease, type 2B1, 605588 Muscular dystrophy, congenital, 613205 Muscular dystrophy, limb-girdle, type 1B, 159001 Mandibuloacral dysplasia, 248370 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210 Heart-hand syndrome, Slovenian type, 610140 Malouf syndrome, 212112
LRSAM1	81,4	100%	97%	Charcot-Marie-Toothe disease, axonal, type 2P, 614436
MED25	98,4	99%	87%	Charcot-Marie-Tooth disease, type 2B2, 605589
MFN2	103,5	100%	97%	Charcot-Marie-Tooth disease, type 2A2, 609260 Hereditary motor and sensory neuropathy VI, 601152
MPZ	103,2	100%	95%	Charcot-Marie-Tooth disease, type 1B, 118200 Dejerine-Sottas disease, 145900 Neuropathy, congenital hypomyelinating, 605253 Charcot-Marie-Tooth disease, type 2J, 607736 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, type 2I, 607677 Charcot-Marie-Tooth disease, dominant intermediate D, 607791
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 2E, 607684 Charcot-Marie-Tooth disease, type 1F, 607734

NGF	180	100%	100%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NTRK1	67,6	98%	93%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
PLEKHG5	86,1	99%	97%	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PMP22	104,1	100%	100%	Charcot-Marie-Tooth disease, type 1A, 118220 Dejerine-Sottas disease, 145900 Neuropathy, recurrent, with pressure palsies, 162500 Charcot-Marie-Tooth disease, type 1E, 118300 Roussy-Levy syndrome, 180800 Neuropathy, inflammatory demyelinating, 139393
PRPS1	62,8	99%	97%	Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Arts syndrome, 301835 Deafness, X-linked 1, 304500
PRX	114	99%	98%	Dejerine-Sottas disease, autosomal recessive, 145900 Charcot-Marie-Tooth disease, type 4F, 614895
SBF2	104,6	99%	97%	Charcot-Marie-Tooth disease, type 4B2, 604563
SCN11A	121,6	99%	98%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	113,8	100%	99%	Erythermalgia, primary, 133020 Insensitivity to pain, channelopathy-associated, 243000 Paroxysmal extreme pain disorder, 167400 Febrile seizures, familial, 3B, 613863 Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Small fiber neuropathy, 133020 {Dravet syndrome, modifier of}, 607208
SH3TC2	97,8	97%	96%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
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	80,9	95%	90%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	110	100%	100%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
TRPV4	94,3	100%	99%	Brachyolmia type 3, 113500 Spondylometaphyseal dysplasia, Kozlowski type, 184252 Metatropic dysplasia, 156530 Hereditary motor and sensory neuropathy, type IIc, 606071 Scapuloperoneal spinal muscular atrophy, 181405 [Sodium serum level QTL 1], 613508 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Digital arthropathy-brachydactyly, familial, 606835
WNK1	148,5	100%	99%	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300
YARS	107,6	100%	97%	Charcot-Marie-Tooth disease, dominant intermediate C, 608323

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
Median Coverage describes the average number of reads seen across 50 exomes
% Covered 10x describes the percentage of a gene's coding region that is covered at least 10x
% Covered 20x describes the percentage of a gene's coding region that is covered at least 20x
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors