NEUROPATHIES (HMSN) GENE PANEL DGD141114

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
AARS	107.0	98%	96%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287
AIFM1	71.7	96%	92%	Combined oxidative phosphorylation deficiency 6, 300816
ATL1	121.3	100%	99%	Spastic paraplegia 3A, autosomal dominant, 182600
BSCL2	140.0	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700
COX6A1	125.8	74%	74%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
DCTN1	134.3	100%	98%	Neuropathy, distal hereditary motor, type VIIB, 607641
DHTKD1	130.2	100%	99%	-2-aminoadipic 2-oxoadipic aciduria, 204750 Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DNAJB2	127.9	100%	99%	Spinal muscular atrophy, distal, autosomal recessive,5, 614881
DNM2	96.9	100%	98%	Charcot-Marie-Tooth disease, dominant intermediate B, 606482
DNMT1	125.0	100%	97%	Neuropathy, hereditary sensory, type IE, 614116
DYNC1H1	137.9	99%	98%	Charcot-Marie-Tooth disease, axonal, type 20, 614228
EGR2	94.1	100%	99%	Neuropathy, congenital hypomyelinating, 1, 605253
FAM134B	102.7	100%	98%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
FGD4	153.7	100%	100%	Charcot-Marie-Tooth disease, type 4H, 609311
FIG4	165.4	100%	98%	Charcot-Marie-Tooth disease, type 4J, 611228
GAN	160.9	100%	99%	Giant axonal neuropathy-1, 256850
GARS	129.5	99%	96%	Charcot-Marie-Tooth disease, type 2D, 601472
GDAP1	124.0	100%	100%	Charcot-Marie-Tooth disease, type 4A, 214400
GJB1	100.0	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GNB4	171.0	100%	100%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
HINT1	98.6	99%	91%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HSPB1	58.0	98%	81%	Neuropathy, distal hereditary motor, type IIB, 608634
HSPB3	234.3	100%	100%	Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB8	133.3	100%	100%	Neuropathy, distal hereditary motor, type IIA, 158590
IGHMBP2	84.6	100%	96%	Neuronopathy, distal hereditary motor, type VI, 604320
IKBKAP	123.6	100%	99%	Dysautonomia, familial, 223900
INF2	90.4	95%	91%	Glomerulosclerosis, focal segmental, 5, 613237
KARS	140.0	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate, B, 613641
KIF1A	85.9	99%	95%	Spastic paraplegia 30, autosomal recessive, 610357

KIF5A	110.2	100%	97%	Spastic paraplegia 10, autosomal dominant, 604187
LITAF	107.3	100%	100%	Charcot-Marie-Tooth disease, type 1C, 601098
LMNA	87.9	98%	92%	Emery-Dreifuss muscular dystrophy 2, AD, 181350
LRSAM1	100.1	100%	97%	Charcot-Marie-Toothe disease, axonal, type 2P, 614436
MED25	113.0	98%	92%	Charcot-Marie-Tooth disease, type 2B2, 605589
MFN2	119.1	100%	99%	Charcot-Marie-Tooth disease, type 2A2, 609260
MPZ	129.5	100%	100%	Charcot-Marie-Tooth disease, type 1B, 118200
MTMR2	129.5	100%	100%	Charcot-Marie-Tooth disease, type 4B1, 601382
NDRG1	96.1	96%	90%	Charcot-Marie-Tooth disease, type 4D, 601455
NEFL	147.0	100%	100%	Charcot-Marie-Tooth disease, type 2E, 607684
NGF	182.9	100%	100%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NTRK1	88.2	99%	92%	Insensitivity to pain, congenital, with anhidrosis, 256800
PLEKHG5	116.2	100%	100%	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067
PMP22	126.7	100%	100%	Charcot-Marie-Tooth disease, type 1A, 118220
PRPS1	75.5	100%	96%	Gout, PRPS-related, 300661
PRX	140.7	99%	98%	Dejerine-Sottas disease, autosomal recessive, 145900
RAB7A	100.5	100%	94%	Charcot-Marie-Tooth disease,type 2B, 600882
SBF2	126.8	100%	99%	Charcot-Marie-Tooth disease, type 4B2, 604563
SCN11A	138.5	100%	99%	Episodic pain syndrome, familial, 3, 615552
SCN9A	136.4	100%	99%	Erythermalgia, primary, 133020
SH3TC2	121.9	98%	96%	Charcot-Marie-Tooth disease, type 4C, 601596
SLC5A7	134.4	100%	100%	Neuronopathy, distal hereditary motor, type VIIA, 158580
SMN1	3.0	10%	10%	Spinal muscular atrophy-1, 253300
SPTLC1	100.1	97%	92%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	128.4	100%	99%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SYT2	93.2	90%	85%	Myasthenic syndrome, presynaptic, congenital, with or without motor neuropathy, 616040
TRPV4	116.0	100%	99%	Brachyolmia type 3, 113500
VCP	138.0	100%	95%	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Inclusion body myopathy,Paget disease and frontotemporal dementia 1, 167320
WNK1	166.8	100%	99%	Pseudohypoaldosteronism, type IIC, 614492
YARS	124.6	100%	98%	Charcot-Marie-Tooth disease, dominant intermediate C, 608323

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
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 : 31 october 2014

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