Supplementary Table S1. Updated list of genes known to cause Charcot–Marie–Tooth disease and related disorders. ALS, amyotrophic lateral sclerosis; CFEOM3, congenital fibrosis of the extraocular muscles type 3; CHN, congenital hypomyelinating neuropathy; CMT, Charcot–Marie–Tooth disease; DSD, Dejerine-Sottas disease; DSMA, distal spinal muscular atrophy; EDS, Ehlers-Danlos syndrome; HGNC, HUGO Gene Nomenclature Committee; HMN, hereditary motor neuropathy; HNPP, hereditary neuropathy with liability to pressure palsies; HSN, hereditary sensory neuropathy; MCV, motor nerve conduction velocity; NCV, nerve conduction velocity; NEFL, neurofilament light chain polypeptide; OMIM, Online Mendelian Inheritance in Man database; PCH, pontocerebellar hypoplasia; SMA, spinal muscular atrophy; SMAJ, spinal muscular atrophy Jokela type; SPG17, spastic paraplegia 17; SPSMA, scapuloperoneal spinal muscular atrophy; UMN, upper motor neuron.

CMT Type (OMIM number)	**HGNC Approved Gene Symbol**	**HGNC Approved Gene Name**	**Phenotype**
Autosomal dominant CMT1			
CMT1A (118220) CMT1E (118300)	17p dup. (*PMP22*) *PMP22* point mutation	Peripheral Myelin Protein 22kD	Classic CMT1 Classic CMT1; DSD; CHN (rarely recessive)
CMT1B (118200)	*MPZ*	Myelin Protein Zero	CMT1; DSD; CHN2; CMT2 (rarely recessive)
CMT1C (601098)	*LITAF*	Lipopolysaccharide-Induced Tumor necrosis factor-Alpha Factor	Classic CMT1
CMT1D (607678)	*EGR2*	Early Growth Response 2	Classic CMT1; DSD; CHN
CMT1F (607734)	*NEFL*	Neurofilament Light polypeptide	CMT2 but can have slow MCV in the CMT1 range (rarely recessive)
CMT1 plus (608895)	*FBLN5*	Fibulin 5	Macular degeneration; cutis laxa; HMN; slow NCV
CMT1G (618279)	*PMP2*	Peripheral Myelin Protein 2	Classic CMT1
SNCV / CMT1 (608236)	*ARHGEF10*	Rho Guanine Nucleotide Exchange Factor 10	Asymptomatic slow conduction velocities
Hereditary neuropathy with liability to pressure palsies			
HNPP (162500)	17p del. (*PMP22*) *PMP22* point mutation	Peripheral Myelin Protein 22kD	Typical HNPP Typical HNPP

Autosomal recessive CMT1			
CMT4A (214400)	*GDAP1*	Ganglioside-induced Differentiation-Associated Protein 1	CMT1 or CMT2, usually severe early onset (dominant and recessive); vocal cord and diaphragmatic paralysis described; CMTRIA; CMT2K
CMT4B1 (601382)	*MTMR2*	Myotubularin-Related protein 2	Severe CMT1; facial; bulbar; focally folded myelin
CMT4B2 (604563)	*SBF2 (MTMR13)*	Set-Binding Factor 2	Severe CMT1; glaucoma; focally folded myelin
CMT4B3 (615284)	*SBF1 (MTMR5)*	Set-Binding Factor 1	CMT1; focally folded myelin; in some cases associated with microcephaly and developmental delay, pyramidal signs, ophthalmoplegia. Also causes early onset CMT2 (recessive)
CMT4C (601596)	*SH3TC2*	SH3 domain and Tetratricopeptide repeats 2	Severe CMT1; scoliosis; cytoplasmic inclusions
CMT4D or HMSN-Lom (601455)	*NDRG1*	N-myc Downstream-Regulated Gene 1	Severe CMT1; gypsy; deafness; tongue atrophy
CHN1 or CMT4E (605253)	*EGR2*	Early Growth Response 2	Congenital hypomyelinating phenotype; CMT1; DSD
CMT4F (614895)	*PRX*	Periaxin	CMT1; predominantly sensory; focally folded myelin
CMT4G or HMSN-Russe (605285)	*HK1*	Hexokinase 1	Severe early-onset CMT1; gypsy
CMT4H (609311)	*FGD4*	FYVE, RhoGEF and PH domain-containing 4	Classic CMT1
CMT4J (611228)	*FIG4*	FIG4 phosphoinositide 5-phosphatase	CMT1; predominantly motor; progressive, complex syndromes including the Yunis-Varon syndrome
CMT4K (616684)	*SURF1*	SURF1 cytochrome c oxidase assembly factor	CMT1; encephalopathy; ataxia; reduced life span; Leigh syndrome
CCFDN (604168)	*CTDP1*	Carboxy-Terminal Domain Phosphatase subunit 1	CMT1; gypsy; cataracts; dysmorphic features

Lethal congenital contracture syndrome 7 (616286) or CHN3 (618186)	*CNTNAP1*	Contactin-Associated Protein 1	Congenital severe arthrogryposis multiplex congenital, demyelinating neuropathy; congenital hypomyelinating phenotype
Lethal congenital contracture syndrome 8 (616287)	*ADCY6*	Adenylate Cyclase 6	Distal joint contractures, reduced fetal movements, death in neonatal period. Hypomyelination on EM.
Autosomal dominant CMT2			
CMT2A (609260)	*MFN2*	Mitofusin 2	CMT2; progressive; optic atrophy (rarely recessive / semi-dominant)
CMT2B (600882)	*RAB7A*	Ras-Associated protein RAB7	CMT2 with sensory complications (ulcero mutilating); HSN
CMT2C (606071)	*TRPV4*	Transient Receptor Potential cation channel subfamily V, member 4	CMT2; vocal cord paralysis; SPSMA
CMT2D (601472)	*GARS*	Glycyl-tRNA Synthetase	CMT2 with predominant hand wasting; HMN5A
CMT2	*BSCL2*	BSCL2 lipid droplet biogenesis associated, seipin	CMT2 with predominant hand wasting; HMN5A; SPG17
CMT2E (607684)	*NEFL*	Neurofilament Light polypeptide	CMT2 but can have slow NCV in the CMT1 range (rarely recessive) or intermediate NCV
CMT2F (606595)	*HSPB1*	Heat-Shock 27-kD Protein family B, member 1	Motor-predominant CMT2; HMN2B
CMT2I (607677)	*MPZ*	Myelin Protein Zero	Late-onset CMT2
CMT2J (607736)	*MPZ*	Myelin Protein Zero	CMT2 with hearing loss and pupillary abnormalities
CMT2K (607831)	*GDAP1*	Ganglioside-induced Differentiation-Associated Protein 1	Late-onset CMT2 (dominant); severe CMT2 (recessive); CMT4A; CMTRIA
CMT2L (608673)	*HSPB8*	Heat-Shock 22-kD Protein family B, member 8	Motor-predominant CMT2; HMN2A
CMT2M or CMTDIB (606482)	*DNM2*	Dynamin 2	Intermediate CMT or CMT2; cataracts; ophthalmoplegia; ptosis

CMT2N (613287)	*AARS*	Alanyl-tRNA Synthetase	Classic CMT2
CMT2P (614436)	*LRSAM1*	Leucine-Rich repeat and Sterile Alpha Motif-containing 1	Mild sensory-predominant CMT2 (dominant and recessive)
CMT2Q (615025)	*DHTKD1*	Dehydrogenase E1 and Transketolase Domain-containing protein 1	CMT2
CMT2V (616491)	*NAGLU*	N-Acetyl-alpha-Glucosaminidase	Late onset, painful, sensory predominant CMT2
CMT2W (616625)	*HARS*	Histidyl-tRNA Synthetase	CMT2; HMN
CMT2Y (616687)	*VCP*	Valosin-Containing Protein	CMT2
CMT2Z (616688)	*MORC2*	MORC Family CW-type Zing Finger Protein 2	CMT2 with pyramidal signs; learning difficulties; cerebellar; retinal pigmentation
CMT2CC (616924)	*NEFH*	Neurofilament Heavy polypeptide	CMT2, proximal weakness, early plantarflexion weakness. Frameshift variants in terminal exon.
CMT2DD (618036)	*ATP1A1*	ATPase Na^+/K^+ transporting subunit, Alpha-1 polypeptide	CMT2, slowly progressive
HMSNO (604484)	*TFG*	Trafficking from ER to Golgi regulator	CMT2 with proximal involvement
SPG10 (604187)	*KIF5A*	Kinesin Family member 5A	CMT2; hereditary spastic paraplegia
CMT2	*MT-ATP6*	Mitochondrial encoded ATP Synthase membrane subunit 6	CMT2; pyramidal signs; relapsing
CMT2 with giant axons (GAN2 / 610100)	*DCAF8*	DDB1 and CUL4-associated factor 8	CMT2 with childhood onset, NEFL accumulations on nerve biopsy
CMT2 (600638)	*TUBB3*	Tubulin Beta-3	CMT2; CFEOM3
CMT2	*DGAT2*	Diacylglycerol O-Acyltransferase 2	Early onset axonal neuropathy with sensory ataxia
Autosomal recessive CMT2			
CMT2B1 (605588)	*LMNA*	Lamin A/C	CMT2 rapid progression

CMT2B2	*PNKP*	Polynucleotide Kinase 3'-Phosphatase	CMT2 and cerebellar ataxia (previously attributed to MED25)
CMT2R (615490)	*TRIM2*	Tripartite Motif-Containing Protein 2	Infantile-onset CMT2
CMT2S (616155)	*IGHMBP2*	Immunoglobulin Mu DNA-Binding Protein 2	CMT2; autonomic dysfunction GI tract
CMT2T (617017)	*MME*	Membrane Metalloendopeptidase	Late onset CMT2
CMT2X (616668)	*SPG11/KIAA8140*	SPG11 vesicle trafficking associated, spatacsin	Onset in the second decade, CMT2 with UMN signs
CMT6B or HMSN6B (616505)	*SLC25A46*	Solute Carrier family 25, member 46	CMT2, optic atrophy, cerebellar syndrome
NMAN (137200)	*HINT1*	Histidine Triad Nucleotide-binding protein 1	Neuromyotonia and axonal neuropathy; motor predominant CMT2; also HMN with neuromyotonia
Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development (618124)	*MCM3AP*	Minichromosome Maintenance complex component 3-Associated Protein	Early onset CMT2, loss of ambulation by second decade. Mild to moderate intellectual disability
AR-CMT2	*DNAJB2* (*HSJ1*)	DNAJ Heat Shock Protein family (Hsp40) member B2	CMT2, motor predominant; DSMA5
AR-CMT2 (607706)	*GDAP1*	Ganglioside-induced Differentiation-Associated Protein 1	Severe CMT2 with vocal cord involvement; CMT4A; CMT2K; CMTRIA
AR-CMT2	*SCO2*	SCO Cytochrome c Oxidase assembly protein 2	Early onset, motor predominant CMT2; also causative of fatal infantile cardioencephalomyopathy
AR-CMT2	*MPV17*	Mitochondrial inner membrane protein MPV17	Early onset severe CMT2; variant specific (p.Arg41Gln)
AR-CMT6	*C12orf65*	Chromosome 12 Open Reading Frame 65	CMT2 and optic atrophy

X-linked CMT			
CMTX1 (302800)	*GJB1*	Gap-Junction protein Beta-1	Males CMT1 (patchy NCV); females CMT2
CMTX3	*78kb insertion from Chr8 into Xq27.1*	-	Early-onset, male specific, slightly milder phenotype compared to CMTX1
CMTX4 or Cowchock syndrome (310490)	*AIFM1*	Apoptosis-Inducing Factor Mitochondria-associated 1	CMT2; infantile onset; developmental delay; deafness; learning difficulties; X-linked recessive
CMTX5 (311070)	*PRPS1*	Phosphoribosyl Pyrophosphate Synthetase 1	CMT2; deafness; optic atrophy; X-linked recessive
CMTX6 (300905)	*PDK3*	Pyruvate Dehydrogenase Kinase 3	CMT2; X-linked dominant
CMTX	*DRP2*	Dystrophin Related Protein 2	Intermediate MCV
Dominant intermediate CMT			
CMTDIB or CMT2M (606482)	*DNM2*	Dynamin 2	Intermediate CMT or CMT2; cataracts; ophthalmoplegia; ptosis
CMTDIC (608323)	*YARS*	Tyrosyl-tRNA Synthetase	Intermediate CMT
CMTDID (607791)	*MPZ*	Myelin Protein Zero	Intermediate CMT
CMTDIE (614455)	*INF2*	Inverted Formin FH2 and WH2 domain containing	Intermediate CMT; focal segmental glomerulosclerosis; end-stage renal failure
CMTD1F (615185)	*GNB4*	Guanine Nucleotide-binding subunit Beta-4	Intermediate CMT
Recessive intermediate CMT			
CMTRIA (608340)	*GDAP1*	Ganglioside-induced Differentiation-Associated Protein 1	Intermediate CMT; severe early onset; CMT4A, CMT2K; recessive CMT2
CMTRIC (615376)	*PLEKHG5*	Pleckstrin Homology domain and RhoGEF domain-containing protein G5	Intermediate CMT; DSMA4
CMTRID (616039)	*COX6A1*	Cytochrome c Oxidase subunit 6A1	Intermediate CMT; onset 1st decade
Hereditary motor neuropathy			

HMN1 or DHMN1 (182960)	*1.35Mb insertion from Chr7q36.3 into Chr7q34*	-	Median onset 10 years; upper motor neuron signs, plantar flexion weakness; dominant
HMN2A (158590)	*HSPB8*	Heat-Shock 22-kD Protein family B, member 8	Classical HMN; dominant; CMT2L
HMN2B (608634)	*HSPB1*	Heat-Shock 27-kD Protein family B, member 1	Classical HMN; dominant; CMT2F
HMN2C (613376)	*HSPB3*	Heat-Shock 27-kD Protein family B, member 3	Classical HMN; dominant
HMN2D (615575)	*FBXO38*	F-Box Only protein 38	Classical HMN; dominant
HMN5A (600794) or SPG17 (270685)	*BSCL2*	BSCL2 lipid droplet biogenesis associated, seipin	Predominant hand wasting; silver syndrome but can have sensory involvement as in CMT2D; dominant
HMN5A (600794)	*GARS*	Glycyl-tRNA Synthetase	Predominant hand wasting; dominant; CMT2D
HMN5B (614751) or SPG31 (610250)	*REEP1*	Receptor Accessory Protein 1	Predominant hand wasting; pyramidal signs; dominant
HMN6 or SMARD1 (604320)	*IGHMBP2*	Immunoglobulin Mu DNA-Binding Protein 2	Infantile onset; respiratory distress; autonomic dysfunction; recessive
HMN7A (158580)	*SLC5A7*	Solute Carrier family 5 member 7	Classical HMN; vocal cord palsy; dominant
HMN7B (607641)	*DCTN1*	Dynactin subunit 1	HMN; bulbar and facial weakness; vocal cord palsy; dominant
DHMN9 or HMNIX (617721)	*WARS*	Tryptophanyl-tRNA Synthetase	Typical HMN; dominant
DSMA2 or HMNJ (605726)	*SIGMAR1*	Sigma non-opioid intracellular Receptor 1	HMN with pyramidal signs; recessive; Juvenile ALS
DSMA4 (611067)	*PLEKHG5*	Pleckstrin Homology domain and RhoGEF domain-containing protein G5	SMA; CMTRIC
DSMA5 (614881)	*DNAJB2* (*HSJ1*)	DNAJ Heat Shock Protein family (Hsp40) member B2	Classical HMN; recessive; also motor predominant CMT2

SMALED 1 (158600)	*DYNC1H1*	Dynein Cytoplasmic 1 Heavy chain 1	Congenital; contractures; lower-limb predominant; pyramidal signs; cortical migration defects; learning difficulties; dominant
SMALED2 (615290)	*BICD2*	Bicaudal D cargo adaptor 2	Congenital; contractures; lower-limb predominant; pyramidal signs; dominant
SMARD2 or SMAX	*LAS1L*	LAS1-like Ribosome Biogenesis factor	Infantile onset; respiratory distress; X-linked recessive
SMAX2 (301830)	*UBA1*	Ubiquitin-like modifier-activating enzyme 1	Neonatal onset, arthrogryposis, loss of anterior horn cells, infantile death; X-linked recessive
SMAX3 (300489)	*ATP7A*	ATPase Cu^{2+}-Transporting alpha polypeptide	Classical HMN; X-linked recessive
SPSMA (181405)	*TRPV4*	Transient Receptor Potential cation channel subfamily V, member 4	HMN; scapular winging; vocal cord palsy; dominant; CMT2C
SMA	*MORC2*	MORC Family CW-type Zing Finger Protein 2	Infantile onset motor neuropathy, cerebellar atrophy and diaphragmatic palsy
SMAJ (615048)	*CHCHD10*	Coiled-coil-Helix-Coiled-coil-Helix Domain-containing protein 10	SMAJ; CMT2; Late onset; Finnish; dominant
HMN	*VRK1*	Vaccinia-Related serine/threonine Kinase 1	HMN with UMN signs; recessive; SMA with PCH
HMN	*AARS*	Alanyl-tRNA Synthetase	Typical HMN; dominant; CMT2N
HMN	*HARS*	Histidyl-tRNA Synthetase	Typical HMN; dominant; CMT2W
HMN	*HINT1*	Histidine Triad Nucleotide-binding protein 1	HMN with neuromyotonia; recessive; also CMT2 with neuromyotonia
PNMHH (614369)	*MYH14*	Myosin Heavy chain 14	Typical HMN; distal myopathy; hoarseness; hearing loss; dominant
HMN with pyramidal features or ALS4 (602433)	*SETX*	Senataxin	HMN with pyramidal signs; dominant
LCCS1 (253310) and LAAHD (611890)	*GLE1*	GLE RNA export mediator	Fetal akinesia, multiple contractures and facial anomalies. Motor neuron loss. Fatal during the fetal or neonatal period.

Hereditary sensory neuropathy (also called Hereditary sensory and autonomic neuropathy (HSAN))			
HSAN1A (162400)	*SPTLC1*	Serine Palmitoyltransferase Long-Chain subunit 1	HSN with sensory complications (ulcero mutilating); dominant
HSAN1C (613640)	*SPTLC2*	Serine Palmitoyltransferase Long-Chain subunit 2	HSN with sensory complications (ulcero mutilating); dominant
CMT2B (600882)	*RAB7A*	Ras-Associated protein RAB7	HSN with sensory complications (ulcero mutilating); dominant
HSN1D (613708) or SPG3A (182600)	*ATL1*	Atlastin GTPase 1	HSN with sensory complications (ulcero mutilating); spasticity; dominant
HSN1E (614116)	*DNMT1*	DNA Methyltransferase 1	HSN; hearing loss; dementia; dominant
HSN1F (615632)	*ATL3*	Atlastin GTPase 3	HSN; bone destruction; dominant
HSAN2A (201300)	*WNK1*	WNK lysine deficient protein Kinase 1	HSN with sensory complications (ulcero mutilating); recessive
HSAN2B (613115)	*RETREG1 (FAM134B)*	Reticulophagy regulator 1	HSN with sensory complications (ulcero mutilating); recessive
HSN2C (614213) or SPG30 (610357)	*KIF1A*	Kinesin Family member 1A	HSN with sensory complications (ulcero mutilating); recessive
HSAN2D or insensitivity to pain (243000), paroxysmal extreme pain disorder (167400), primary erythermalgia (133020), small-fibre neuropathy	*SCN9A*	Sodium voltage-gated Channel alpha subunit 9	Recessive: insensitivity to pain Dominant: paroxysmal extreme pain disorder; primary erythermalgia; small fibre neuropathy
HSAN3, familial dysautonomia or Riley–Day (223900)	*ELP1 (IKBKAP)*	Elongation complex Protein 1	Ashkenazi Jewish; autonomic dysfunction; HSN; absent fungiform papillae; recessive
HSAN4 or CIPA (256800)	*NTRK1*	Neurotrophic Receptor Tyrosine Kinase 1	Congenital insensitivity to pain with anhydrosis; recessive
HSAN5 (608654)	*NGF*	Nerve Growth Factor	Insensitivity to pain; recessive

HSAN6 (614653)	*DST*	Dystonin	Ashkenazi Jewish; autonomic dysfunction; HSN; absent fungiform papillae; death by age 2; recessive
HSAN7 (615548)	*SCN11A*	Sodium voltage-gated Channel alpha subunit 11	Congenital insensitivity to pain with hyperhidrosis and gastrointestinal dysfunction; painful peripheral neuropathy; dominant
HSAN8 (616488)	*PRDM12*	PR/SET domain 12	Congenital insensitivity to pain; recessive
SNAX1 (608984)	*RNF170*	Ring Finger Protein 170	Preganglionic sensory neuroaxonal loss; normal sensory action potentials; dominant
CIP	*CLTCL1*	Clathrin heavy chain-like 1	Congenital insensitivity to pain and severe global developmental delay; dysmorphic, delayed myelination on brain MRI; Iranian; recessive
HSAN and dementia	*PRNP*	Prion Protein	Autonomic dysfunction; sensory loss; dementia; dominant
Hereditary sensory neuropathy with spastic paraplegia (256840)	*CCT5*	Chaperonin Containing T-complex polypeptide 1 subunit 5	HSN with sensory complications (ulcero mutilating) and spastic paraplegia; recessive
Posterior column ataxia & Retinitis pigmentosa (PCARP / 609033)	*FLVCR1*	Feline Leukemia Virus subgroup C cellular Receptor 1	Retinitis pigmentosa, sensory ganglionopathy and abnormal posterior columns on MRI; recessive.
Familial Neuropathic Chronic Itch	*COL6A5*	Collagen type 6 alpha 5 chain	Dominant, Chronic itch, EDS in 1 of 3 families
Arthrogryposis, distal, with impaired proprioception and touch (617146)	*PIEZO2*	PIEZO-type mechanosensitive ion channel component 2	Loss of mechanosensation, arthrogryposis, scoliosis; recessive