Supplementary table 1. CMT genes: inheritance, phenotype and cell pathophysiology.

Gene	OMIM (*)	MoI	Phenotype Neuropathy type				OMIM (#)	Localization		Protein function / Pathway	Disrupted process
			A	D	I			Schwann cell	Body / axon		
Other genes											
AARS1	601065	AD	✓			CMT2N	613287		✓	Aminoacyl tRNA synthetase	Nuclear envelope, mRNA processing
ABHD12	613599	AR		✓		Polyneuropathy	612674	NA	NA	Lipid hydrolysis	ER and Golgi
AIFM1	300169	XL	✓			CMTX4	310490		✓	Oxidative phosphorylation - apoptosis	Mitochondria
ARHGEF10	608136	AD		✓		Slowed nerve conduction velocities	608236	✓		Guanine nucleotide exchange factor	Endosomal sorting and cell signaling
ATP1A1	182310	AD	✓			CMT2DD	618036		✓	Cation transport ATPase	Channel
ATP7A	300011	XL	✓			Spinal muscular atrophy/distal motor neuropathy Menkes disease	300489 309400		✓	Copper transport ATPase	Channel
BAG3	603883	AD	✓			Myopathy, myofibrillar, 6	612954	NA	NA	Co-chaperone	Apoptosis
BSCL2	606158	AD	✓			dHMN type VC	619112		✓	Formation of lipid droplets	ER and Golgi
CADM3	609743	AD	✓			CMT2FF	619519	NA	NA	Calcium-independent cell-cell adhesion	Cell-cell interaction
CNTNAP1	602346	AR	✓	✓		Hypomyelinating neuropathy, congenital, 3	618186	NA	NA	Signaling between axons and myelinating glial cells	Axon guidance
COA7	615623	AR	✓			Spinocerebellar ataxia, AR, with axonal neuropathy 3	618387	NA	NA	Assembly of mitochondrial oxidative phosphorylation complexes	NA
DCTN1	601143	AD				dHMN type VIIB	607641		✓	Dynein-mediated retrograde transport of vesicles and organelles along microtubules	Cytoskeleton, axonal transport
DCTN2	607376	AD	✓			NA	NA	NA	NA	Modulation of dynein-organelles binding	Cytoskeleton
DGAT2	606983	AD	✓			NA	NA		✓	Synthesis of triglycerides	ER and Golgi
DHTKD1	614984	AD	✓			CMT2Q	615025		✓	Degradation of amino acids	Mitochondria
DNAJB2	604139	AR	✓			Spinal muscular atrophy, distal, AR, 5	614881		✓	Co-chaperone	Proteasome and protein aggregation

DNMT1	126375	AD	✓			dHSN type IE	614116		✓	Maintenance of methylation patterns of genomic DNA	Nuclear envelope, mRNA processing
DNM2	602378	AD			✓	CMT2B CMT2M Centronuclear myopathy 1	606482 606482 160150	✓		GTPase, fussion-fision apparatus	Endosomal sorting and cell signaling
DRP2	300052	XL			✓	NA	NA	✓		Maintenance of membrane-associated complexes	Cytoskeleton
DYNC1H1	600112	AD	✓			CMT20	614228		✓	Retrograde axonal transport	Cytoskeleton, axonal transport
EGR2	129010	AD AR		✓		CMT1D	607678	✓		Transcription regulator	Transcription, mRNA processing
FGD4	611104	AR		✓		CMT4H	609311	✓		Regulates cell signaling involved in myelin production and actin cytoskeleton	Cytoskeleton
FIG4	609390	AR		✓		CMT4J	611228	✓		Abnormal transport of intracellular organelles	Endosomal sorting and cell signaling
GARS1	600287	AD	✓			CMT2D dHMN type VA	601472 600794		✓	Aminoacyl tRNA synthetase	Nuclear envelope, mRNA processing
GNB4	610863	AD			✓	CMTDIF	615185		✓	Signal transduction	Endosomal sorting and cell signaling
HARS1	142810	AD	✓	✓		CMT2W	616625		✓	Aminoacyl tRNA synthetase	Nuclear envelope, mRNA processing
HSPB1	602195	AD	✓			CMT2F dHMN type IIB	606595 608634		✓	Microtubule regulator	Proteasome and protein aggregation
HSPB3	604624	AD	✓			dHMN type IIC	613376		✓	Microtubule regulator	Proteasome and protein aggregation
HSPB8	608014	AD	✓			CMT2L dHMN type IIA	608673 158590		✓	Microtubule regulator	Proteasome and protein aggregation
IGHMBP2	600502	AR	✓			CMT2S dHMN type VI	616155 604320		✓	Helicase	Nuclear envelope, mRNA processing
INF2	610982	AD			✓	CMTDIE	614455	✓		Actin polymerization and filament severing	Cytoskeleton
KARS	609574	NA				NA	NA		✓	Aminoacyl tRNA synthetase	Nuclear envelope, mRNA processing
KIF1B	605995	AD	✓			CMT2A1	118210		✓	Intracellular organelle transport	Cytoskeleton, axonal transport

KIF5A	602821	AD	✓			Spastic paraplegia 10	604187		✓	Intracellular organelle transport	Cytoskeleton, axonal transport
LITAF	603795	AD			✓	CMT1C	603795	✓		Regulation of endosome to lysosome trafficking and cell signaling	Endosomal sorting and cell signaling
LMNA	150330	AR	✓			CMT2B1	605588		✓	Intermediate filament protein of nuclear envelope	Nuclear envelope, mRNA processing
LRSAM1	610933	AD AR	✓			CMT2P	614436		✓	E3 ubiquitin ligase, regulates cell adhesion molecules	Proteasome and protein aggregation
MARS1	156560	AD	✓			CMT2U	616280		✓	Aminoacyl tRNA synthetase	Nuclear envelope, mRNA processing
MCM3AP	603294	AR	✓	✓		Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development	618124		✓	DNA replication	
MME	120520	AR AD	✓			CMT2T	120520		✓	Destruction of opioid peptides	Endosomal sorting and cell signaling
MORC2	616661	AD	✓			CMT2Z	616688		✓	Regulation of heterochromatin condensation	Nuclear envelope, mRNA processing
MPV17	137960	AR	✓			CMT2EE	137960		✓	Metabolism of reactive oxygen species	Mitochondria
MTMR2	603557	AR			✓	CMT4B1	603557	✓		Modification of chemical messengers involved in signal transduction	Endosomal sorting and cell signaling
NAGLU	609701	AD	✓			CMT2V	616491		✓	Degradation of heparan sulfate	Endosomal sorting and cell signaling
NDRG1	605262	AR			✓	CMT4D	605262	✓		Signaling protein shuttling between cytoplasm and nucleus	Endosomal sorting and cell signaling
NEFH	162230	AD	✓			CMT2CC	616924		✓	Growth arrest and cell differentiation	Cytoskeleton
NEFL	162280	AD AR	✓	✓		CMTDIG CMT1F CMT2E	617882 607734 607684		✓	Intermediate filament in neurons	Cytoskeleton, axonal transport
PDK3	300906	XL	✓			CMTX6	300905		✓	Regulates pyruvate dehydrogenase complex	Mitochondria
PLEKHG5	611101	AR			✓	CMTRIC	615376		✓	Nuclear factor kB-activator	Nuclear envelope, mRNA processing
PMP2	170715	AD			✓	CMT1G	618279	✓		Myelin assembly	Myelin assembly

PNKP	605610	AR	✓			CMT2B2	605589		✓	DNA repair	Nuclear envelope, mRNA processing
PRPS1	311850	XL				CMTX5	311070		✓	Purine and pyrimidine biosynthesis	Nuclear envelope, mRNA processing
PRX	605725	AR	✓			CMT4F	614895	✓		Membrane-protein interaction stabilizing myelin sheath	Cytoskeleton
PTRH2	608625	AR				Infantile-onset multisystem neurologic, endocrine, and pancreatic disease	616263	NA	NA	Peptidyl-tRNA hydrolase	Proteasome and protein metabolism
RAB7A	602298	AD	✓			CMT2B	600882		✓	Vesicular transport and membrane trafficking	Endosomal sorting and cell signaling
SBF1	603560	AR		✓		CMT4B3	615284	✓		Endosomal trafficking	Endosomal sorting and cell signaling
SBF2	607697	AR		✓		CMT4B2	604563	✓		Development of Schwann cells	Endosomal sorting and cell signaling
SCO2	604272	AR	✓			Motor neuropathy [7]	NA	NA	NA	Cytochrome c oxidase synthesis	Gene expression
SETX	608465	AD	✓			Amyotrophic lateral sclerosis 4, juvenile Spinocerebellar ataxia, AR, with axonal neuropathy 2	602433 606002		✓	Probable RNA/DNA helicase	Nuclear envelope, mRNA processing
SIGMAR1	601978	AR	✓			Amyotrophic lateral sclerosis 16, juvenile Spinal muscular atrophy, distal, AR, 2	614373 605726		✓	Chaperone	ER and Golgi
SGPL1	603729	AR	✓			Motoneuropathy	NA		✓	Sphingolipid catabolism	ER and Golgi
SPG11	610844	AR	✓			CMT2X	616668		✓	Cytoskeleton stability and synaptic vesicle transport	Cytoskeleton, axonal transport
SPTLC1	605712	AD	✓			Neuropathy, hereditary sensory and autonomic, type IA	162400		✓	Serine palmitoyltransferase	Toxin
TRIM2	614141	AR	✓			CMT2R	614141		✓	E3 ubiquitin ligase	Proteasome and protein aggregation
TRPV4	605427	AD	✓			CMT2C dHMN type VIII	606071 600175		✓	Calcium homeostasis, cytoskeleton remodeling	Channel
VCP	611745	AD	✓			NA	NA		✓	AAA ATPase family	ER and Golgi
VWA1	611901	AR	✓			dHMN	619216	NA	NA	Matrix assembly	

WARS	191050	AD	✓			dHMN type IX	617721		✓	Aminoacyl tRNA synthetase	Nuclear envelope, mRNA processing
YARS1	603623	AD			✓	CMTDIC	608323		✓	Aminoacyl tRNA synthetase	Nuclear envelope, mRNA processing

MoI: Mode of Inheritance; A: axonal; D: demyelinating; I: intermediate; AR: autosomal recessive; AD: autosomal dominant; XL: X-linked; CMT: Charcot-Marie-Tooth; dHMN: distal hereditary motor neuropathy; dHSN: distal hereditary sensory neuropathy DI: dominant intermediate; NA: data not available. Information about genes and protein functions was extracted from databases such as OMIM (https://www.omim.org/), GeneCards (https://www.genecards.org/), Reactome (https://reactome.org/) and UniProt (https://www.uniprot.org/).