

GENE SYMBOL	GENE NAME	RefSeq NM_DIAG	DISEASE*	INHERITANCE	ClinGen evaluation	Classification	Genetic evidence PMIDs/Notes 2 Publications	Experimental evidence PMIDs/ Note 1 publication
<i>AARS1</i>	Alanyl-tRNA-synthetase	NM_001605	CMT 2N	AD		Definitive	20045102[1] 22206013[2]	22009580 [3]
<i>AIFM1</i>	Apoptosis inducing factor mitochondria associated 1	NM_145812	CMT X4	X	Definitive	Definitive	20362274[4] 23217327[5]	20362274[4]
<i>ARHGEF10</i>	Rho guanine nucleotide exchange factor (GEF) 10	NM_014629	HMSN /Slow nerve conduction velocity	AD	Limited	Limited	14508709 [6]	21719701 [7]
<i>ATL1</i>	Atlastin GTPase 1	NM_001127713	HSAN 1D / SPG 3A	AD		Limited	21194679[8] 22340599[9]	33287888 [10]
<i>ATL3</i>	Atlastin GTPase 3	NM_015459	HSAN 1F	AD	Moderate	Definitive	24459106[11] 24736309[12]	30339187 [13]
<i>ATP1A1</i>	ATPase, Na+/K+ transporting, alpha 1 polypeptide	NM_000701	CMT2 DD	AD		Definitive	29499166 [14] 31373411 [15]	31707753 [16]
<i>ATP7A</i>	ATPase, Cu++ transporting, alpha polypeptide	NM_000052	dHMN / SMA	XLD		Definitive	20170900[17] 31558336[18]	31969342 [19]
<i>B4GALNT1</i>	Beta-1,4-N-acetyl-galactosaminyltransferase 1	NM_001478	HSP 26 / CMT2	AR		Limited	28536081[20] 34595861[21]	
<i>BICD2</i>	Bicaudal D homolog 2 (Drosophila)	NM_001003800	SMALED 2A / 2B dHMN	AD		Definitive	23664116[22] 23664119[23] 30536747[24]	27784775 [25]
<i>BSCL2</i>	Berardinelli-Seip congenital lipodystrophy 2 (seipin)	NM_001122955	dHMN 5A	AD	Definitive	Definitive	14981520[26] 26815532[27]	17387721 [28]

<i>CADM3</i>	Cell adhesion molecule 3	NM_001127173	CMT 2FF	AD		Limited	33889941[29]	
<i>CCT5</i>	Chaperonin containing TCP1, subunit 5 (epsilon)	NM_012073	HSAN SP	AR		Limited	16399879[30]	25345891 [31]
<i>CHCHD10</i>	Coiled-coil-helix-coiled-coil-helix domain containing 10	NM_213720	SMA Jokela/ CMT 2	AD		Limited	25428574[32] 27066538[33]	
<i>CNTNAP1</i>	contactin associated protein 1	NM_003632	CMT 2/ congenital hypomyelinating neuropathy type 3 (CHN3)	AR		Limited	28374019[34] 31397905[35]	
<i>COX6A1</i>	Cytochrome c oxidase subunit VIa polypeptide 1	NM_004373	CMT ID	AR		Definitive	25152455 [36] 26302975 [37]	25152455 [36]
<i>CTDP1</i>	CTD (carboxy-terminal domain, RNA polymerase II, polypeptide A) phosphatase, subunit 1	NM_004715	CCFDN	AR		Definitive	14517542 [38] 24690360 [39]	14517542 [38]
<i>DCTN1</i>	Dynactin 1	NM_004082	dHMN VIIB	AD		Definitive	12627231 [40] 27573046 [41]	16505168 [42]
<i>DCTN2</i>	Dynactin 2	NM_001261413	CMT I	AD		Limited	26517670[43]	
<i>DHTKD1</i>	Dehydrogenase E1 and transketolase domain containing 1	NM_018706	CMT 2Q	AD		Limited	23141294 [44]	29661920 [45]
<i>DGAT2</i>	diacylglycerol O-acyltransferase 2	NM_032564	CMT 2	AD		Limited	31372974[46]	26786738[47]
<i>DNAJB2</i>	DnaJ (Hsp40) homolog, subfamily B, member 2	NM_006736	SMA 5	AR		Definitive	22522442 [48] 25274842 [49]	22522442 [48]

<i>DNM2</i>	Dynamin 2	NM_001005361	CMT 2N / CMT 1B / Lethal congenital contracture syndrome	AD AR	Definitive	Definitive	15731758 [50] 17636067 [51]	26842864 [52]
<i>DNMT1</i>	DNA (cytosine-5-)-methyltransferase 1	NM_001130823	HSAN 1E	AD		Definitive	21532572 [53] 23365052 [54]	21532572 [53]
<i>DRP2</i>	dystrophin related protein 2	NM_001939	CMT I	X		Definitive	26227883[55] 31217940[56]	29623298[57]
<i>DST</i>	Dystonin	NM_001144769	HSAN 6	AR	Definitive	Definitive	22522446 [58] 32802955[59]	29982604 [60]
<i>DST</i>	Dystonin	NM_001144769	HMSN	AR		Limited	32802955[59]	
<i>DYNC1H1</i>	Dynein, cytoplasmic 1, heavy chain 1	NM_001376	CMT 2O	AD		Limited	21820100 [61]	
<i>DYNC1H1</i>	Dynein, cytoplasmic 1, heavy chain 1	NM_001376	HMSN	AD		Definitive	22459677 [62] 22847149 [63]	23742762 [64]
<i>EGR2</i>	Early growth response 2	NM_000399	CMT 1D / Dejerine-Sottas Disease / Hypomyelinating congenital neuropathy 1	AD / AD;AR / AD;AR	Definitive	Definitive	9537424 [65] 10371530 [66]	7935840 [67]
<i>ELP1/ IKBKAP</i>	Inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein	NM_003640	FD	AR		Definitive	11179008 [68] 12687659 [69]	17644305[70]
<i>FBLN5</i>	Fibulin 5	NM_006329	HNARMD / CMT1	AD		Limited	21576112 [71] 23328402 [72] 28332470 [73]	33107705 [74]
<i>FBXO38</i>	F-box protein 38	NM_030793	SMALED 1 / dHMN2D	AD/AR		Limited	24207122 [75] 31420593 [76]	
<i>FGD4</i>	FYVE, RhoGEF and PH domain containing 4	NM_139241	CMT 4H	AR	Definitive	Definitive	15744041 [77] 17564972 [78]	17564959 [79]

<i>FIG4</i>	Phosphoinositide 5-phosphatase	NM_014845	CMT 4J	AR	Definitive	Definitive	17572665 [80] 21705420 [81]	18556664 [82]
<i>GARS1</i>	Glycyl-tRNA synthetase	NM_002047	CMT 2D / dSMA V	AD / AD		Definitive	8872480 [83] 12690580 [84]	25168514 [85] 26503042 [86]
<i>GBF1</i>	Golgi brefeldin A resistant guanine nucleotide exchange factor 1	NM_001377137	CMT2GG	AD		Limited	32937143[87]	32937143[87]
<i>GDAP1</i>	Ganglioside induced differentiation associated protein 1	NM_018972	CMT 2K / CMT 4A	AD/ AR	Definitive	Definitive	11743579 [88] 15805163 [89]	21753178 [90]
<i>GJB1</i>	Gap junction protein, beta 1, 32kDa	NM_001097642	CMT X	X	Definitive	Definitive	8266101 [91] 10732813 [92]	15852376 [93]
<i>GLA</i>	Galactosidase alpha	NM_000169	Fabry disease		Definitive	Definitive	27164712[94] 12359124[95]	12359124[95]
<i>GNB4</i>	Guanine nucleotide binding protein (G protein), beta polypeptide 4	NM_021629	CMT IF	AD	Moderate	Definitive	23434117 [96] 27908631 [97]	23434117 [96]
<i>HARS1</i>	Histidyl-tRNA synthetase	NM_002109	CMT 2W	AD		Definitive	22930593 [98] 26072516 [99]	22930593 [98]
<i>HINT1</i>	Histidine triad nucleotide binding protein 1	NM_005340	ARAN-NM	AR	Definitive	Definitive	22961002 [100] 25342199 [101]	22961002 [100]
<i>HK1</i>	Hexokinase 1	NM_033497	HMSN type Russe	AR		Limited	19536174 [102] 22978647 [103]	
<i>HSPB1</i>	Heat shock 27kDa protein 1	NM_001540	CMT 2F/dHMN IIB	AD		Definitive	15122254 [104] 18832141 [105]	21785432 [106]
<i>HSPB3</i>	Heat shock 27kDa protein 3	NM_006308	HMN 2C	AD		Limited	20142617 [107] 29341343 [108]	27567740 [109]

<i>HSPB8</i>	Heat shock 22kDa protein 8	NM_014365	CMT 2L	AD		Definitive	15122253 [110] 15565283 [111]	20538880 [112]
<i>HSPB8</i>	Heat shock 22kDa protein 8	NM_014365	dHMN IIA	AD	Definitive	Definitive	15122253 [110] 15565283 [111]	20538880 [112]
<i>IGHMBP2</i>	Immunoglobulin mu binding protein 2	NM_002180	CMT 2S	AR		Definitive	25439726 [113] 25568292 [114]	25439726 [113]
<i>IGHMBP2</i>	Immunoglobulin mu binding protein 2	NM_002180	dHMN VI	AR		Definitive	25439726 [113] 25568292 [114]	25439726 [113]
<i>INF2</i>	Inverted formin, FH2 and WH2 domain containing	NM_022489	CMT IE	AD	Definitive	Definitive	22187985 [115] 30680856 [116]	22187985 [115]
<i>ITPR3</i>	inositol 1,4,5-trisphosphate receptor type 3	NM_002224	CMT I	AD		Limited	32949214[117]	
<i>JAG1</i>	canonical Notch ligand Jagged1	NM_000214	CMT 2H	AD		Limited	32065591[118]	
<i>JPH1</i>	Junctophilin 1	NM_020647	CMT 2K	AD		Definitive		
<i>KARS1</i>	Lysyl-tRNA synthetase	NM_001130089	CMT IB	AR		Limited	20920668 [119]	
<i>KIF1A</i>	Kinesin family member 1A	NM_001244008	HSAN 2C / HSP 30	AR		Definitive	21820098 [120] 22258533 [121]	27263974 [122]
<i>KIF1B</i>	Kinesin family member 1B	NM_015074	CMT 2A1	AD	No Known Disease Relationship	Limited	11389829 [123] 25802885 [124]	19503091 [125] 30126838 [126]
<i>KIF5A</i>	Kinesin family member 5A	NM_004984	CMT 2/ HSP 10	AD		Limited	25008398[127] 29892902[128]	
<i>LITAF</i>	Lipopolysaccharide-induced TNF factor	NM_001136472	CMT 1C	AD	Moderate	Definitive	12525712 [129] 15776429 [130]	23359569 [131]
<i>LMNA</i>	Lamin A/C	NM_170707	CMT 2B1	AR		Definitive	11799477 [132] 17536044 [133]	11799477 [132]
<i>LRSAM1</i>	Leucine rich repeat and sterile alpha motif containing 1	NM_001005374	CMT 2P	AD / AR		Definitive	20865121 [134] 27686364 [135]	22012984 [136]

<i>MARS1</i>	Methionyl-tRNA synthetase	NM_004990	CMT 2U	AD	Limited	Limited	23729695 [137] 24354524 [138]	31356216 [139]
<i>MED25</i>	Mediator complex subunit 25	NM_030973	CMT2B2	AR	Disputed	Disputed	19290556[140] 30039206[141]	
<i>MFN2</i>	Mitofusin 2	NM_001127660	CMT 2A2A / CMT 2A2B	AD / AR		Definitive	15064763 [142] 16835246 [143]	17959936 [144]
<i>MFN2</i>	Mitofusin 2	NM_001127660	dHMN VIA	AD / AR		Definitive	15064763 [142] 16835246 [143]	17959936 [144]
<i>MME</i>	Membrane metallo-endopeptidase	NM_007288	CMT 2T	AD AR		Definitive in AR inheritance / Limited in AD inheritance	26991897 [145] 27588448 [146]	27588448 [146]
<i>MORC2</i>	MORC family CW-type zinc finger 2	NM_014941	CMT 2Z	AD		Definitive	26497905 [147] 26659848 [148]	30624633 [149]
<i>MPV17</i>	Mitochondrial inner membrane protein 17	NM_002437	CMT 2	AR		Definitive	26437932[150] 30298599[151]	26437932[150]
<i>MPZ</i>	Myelin protein zero	NM_000530	CMT 1B CMT 2I	AD	Definitive	Definitive	7693129 [152] 19293842 [153]	10704451 [154]
<i>MTMR2</i>	Myotubularin related protein 2	NM_016156	CMT 4B1	AR	Definitive	Definitive	10802647 [155] 28509084 [156]	15557122 [157]
<i>MYH14</i>	Myosin, heavy chain 14, non-muscle	NM_001145809	PNMHH	AD		Definitive	21480433 [158] 27875632 [159]	31231018 [160]
<i>NAGLU</i>	N-acetylglucosaminidase, alpha	NM_000263	CMT 2V	AD		Limited	25818867 [161]	25818867 [161]
<i>NDRG1</i>	N-myc downstream regulated 1	NM_006096	CMT 4D	AR		Definitive	10831399 [90] 12872253 [163]	15082788 [164]
<i>NEFH</i>	Neurofilament, heavy polypeptide	NM_021076	CMT 2CC	AD	Definitive	Definitive	27040688 [165] 30992180 [166]	27040688 [165]
<i>NEFL</i>	Neurofilament, light polypeptide	NM_006158	CMT 2E / CMT 1F / CMT IG	AD / AD-AR		Definitive	10841809 [167] 12566280 [168]	12393795 [169]
<i>NGF</i>	Nerve growth factor (beta polypeptide)	NM_002506	HSAN 5	AR		Definitive	14976160 [170] 20978020 [171]	29483280 [172]

<i>NRG2</i>	Neuregulin 2	NM_004883		AR		No correlation with CMT disease	10369162[173]	
<i>NTRK1</i>	Neurotrophic tyrosine kinase, receptor, type 1	NM_002529	CIPA / HSAN 4	AR	Definitive	Definitive	10330344 [174] 28177573 [175]	27676246 [176]
<i>PDK3</i>	Pyruvate dehydrogenase kinase, isozyme 3	NM_001142386	CMT X6	AD		Definitive	23297365 [177] 26801680 [178]	23297365[177]
<i>PHYH</i>	<u>Phytanoyl-coa hydroxylase</u>	<u>NM_006214</u>	<u>Refsum disease</u>	<u>AR</u>		<u>Definitive</u>	<u>11555634[179]</u> <u>32773395[180]</u>	
<i>PLEKHG5</i>	Pleckstrin homology domain containing, family G (with RhoGef domain) member 5	NM_001265593	CMT IC	AR		Definitive	23777631 [181] 23844677 [182]	
<i>PMP2</i>	Peripheral myelin protein 2	NM_002677	CMT 1G	AD	Limited	Limited	26257172 [183] 26828946 [184]	26828946 [184]
<i>PMP22</i>	Peripheral myelin protein 22	NM_153322	CMT 1A / CMT 1E / DSS / HNPP	AD / AR in some DSS		Definitive	(Chicago: Mosby- Yearbook) [185] 1303230 [186] 32538861 [187]	1999826 [187] 32538861[188]
<i>PNKP</i>	Polynucleotide kinase 3'-phosphatase	NM_007254	CMT 2B2	AR		Limited	23224214[189] 30039206[141]	
<i>POLR3B</i>	RNA polymerase 3 subunit B	NM_018082	CMT 1	AD		Limited	33417887[190] 34666706[191]	
<i>PRDM12</i>	PR domain containing 12	NM_021619	HSAN 8	AR		Definitive	26005867 [192] 26975306 [193]	25891934 [194]
<i>PRPS1</i>	Phosphoribosyl pyrophosphate synthetase 1	NM_002764	CMT X5	X		Definitive	17701900 [195] 20380929 [196]	31434166 [197]
<i>PRX</i>	Periaxin	NM_181882	CMT 4F / DSS	AR / AD-AR		Definitive	10848494 [198] 15197604 [199]	10839370 [200]
<i>RAB7A</i>	RAB7A, member RAS oncogene family	NM_004637	CMT 2B	AD		Definitive	12545426 [201] 15455439 [202]	24521780 [203]

<i>REEP1</i>	Receptor accessory protein 1	NM_022912	HMN 5B / SPG31	AD / AD		Definitive	22703882 [204] 30637453 [205]	22703882 [204]
<i>RETREG1/ FAM134B</i>	Family with sequence similarity 134, member B	NM_001034850	HSAN 2B	AR		Definitive	19838196 [206] 24327336 [207]	26040720 [208]
<i>SBF1</i>	SET binding factor 1	NM_002972	CMT 4B3	AR	Moderate	Limited	23749797 [209] 24799518 [210]	
<i>SBF2</i>	SET binding factor 2	NM_030962	CMT 4B2	AR	Definitive	Definitive	12554688 [211] 12687498 [212]	17855448 [213]
<i>SCN10A</i>	Sodium channel, voltage-gated, type X, alpha subunit	NM_006514	FEPS 2	AD		Definitive	23115331 [214] 29448912 [215]	23115331 [214]
<i>SCN11A</i>	Sodium channel, voltage-gated, type XI, alpha subunit	NM_014139	HSAN 7	AD		Definitive	24036948 [216] 25118027 [217]	15964986 [218]
<i>SCN9A</i>	Sodium channel, voltage-gated, type IX, alpha subunit	NM_002977	HSAN 2D / CIPA Small fiber neuropathy (SFN)	AR AD in SFN		Limited	20692858 [219] 23596073 [220]	
<i>SETX</i>	Senataxin	NM_015046	SCAN2	AR		Definitive	15732101 [221] 14770181 [222]	19593598 [223]
<i>SH3TC2</i>	SH3 domain and tetratricopeptide repeats 2	NM_024577	CMT 4C	AR		Definitive	14574644 [224] 16326826 [225]	19805030 [226]
<i>SIGMAR1</i>	Sigma non-opioid intracellular receptor 1	NM_005866	SMA 2	AR		Definitive	26078401 [227] 28503617 [228]	27402882 [229]
<i>SLC25A46</i>	Solute carrier family 25 member 46	NM_138773.4	HSMN6B	AR	Definitive	Definitive	26168012[230] 27430653[231]	26168012[230]
<i>SLC5A7</i>	Solute carrier family 5 (sodium/choline cotransporter), member 7	NM_021815	HMN 7A	AD	Moderate	Definitive	23141292 [232] 29782645 [233]	23141292 [232]
<i>SLC9A3R1</i>	SLC9A3 regulator 1	NM_004252	CMT1			Limited	32077526[234]	
<i>SOD1</i>	Superoxide dismutase 1	NM_000454.5	HMSN			Limited	9462467[235]	

<i>SORD</i>	Sorbitol dehydrogenase	NM_003104	dHMN/ CMT 2	AR		Definitive	32367058 [236] 33397963 [237]	32367058 [236]
<i>SPG11</i>	Spastic paraplegia 11	NM_025137	CMT 2X	AR		Definitive	26556829 [238] 27544499 [239]	28237315 [240]
<i>SPG11</i>	Spastic paraplegia 11	NM_025137	SPG	AR		Definitive	14732628[241]	28237315 [240]
<i>SPTAN1</i>	Alpha II spectrin	NM_001130438	HMN	AD		Limited	31332438[242] 33206935[243]	
<i>SPTLC1</i>	Serine palmitoyltransferase, long chain base subunit 1	NM_006415	HSAN 1A	AD		Definitive	11242114 [244] 11242106 [245]	16210380 [246]
<i>SPTLC2</i>	Serine palmitoyltransferase, long chain base subunit 2	NM_004863	HSAN 1C	AD		Definitive	20920666 [247] 23658386 [248]	20920666 [247]
<i>SURF1</i>	Surfeit 1	NM_003172	CMT 4K	AR		Limited	24027061 [249]	
<i>SYT2</i>	Synaptotagmin II	NM_177402	CMS 7	AD		Definitive	26519543 [250] 30533528 [251]	30533528 [251]
<i>TECPR2</i>	Tectonin β -propeller repeat-containing protein	NM_014844	HSAN 3	AR		Definitive	26542466[252] 33847017[253]	33218264[254]
<i>TFG</i>	TRK-fused gene	NM_001195478	HMSN Okinawa	AD		Definitive	22883144 [255] 23553329 [256]	25098539 [257]
<i>TRIM2</i>	Tripartite motif containing 2	NM_015271	CMT 2R	AR		Definitive	23562820 [258] 25893792 [259]	18687884 [260]

<i>TRPV4</i>	Transient receptor potential cation channel, subfamily V, member 4	NM_021625	CMT 2C	AD	Definitive	Definitive	20037588 [261] 20037587 [262]	21288981 [263]
<i>TTR</i>	Transthyretin	NM-000371	Amyloidosis hereditary	AD	Definitive	Definitive	11385707[264] 1848299[265]	15820680[266]
<i>VAPB</i>	VAMP (vesicle-associated membrane protein)-associated protein B and C	NM_004738	ALS 8 / SMA Finkel Type	AD		Definitive	15372378 [267] 20940299 [268]	26362257 [269]
<i>VCP</i>	Valosin containing protein	NM_007126	CMT 2Y	AD		Definitive	25125609 [270] 25878907 [271]	25125609 [270]
<i>VRK1</i>	Vaccinia related kinase 1	NM_003384	SMA	AR		Definitive	24126608 [272] 30847374 [273]	31090908[274]
<i>VRK1</i>	Vaccinia related kinase 1	NM_003384	dHMN	AR		Definitive	31090908[274] 34990802[275]	31090908 [274]
<i>WARS1</i>	Tryptophanyl-tRNA synthetase	NM_173701	dHMN IX	AD		Definitive	28369220 [276] 31069783 [277]	28369220 [276]
<i>WNK1</i>	WNK lysine deficient protein kinase 1	NM_001184985	HSAN 2	AR		Definitive	15060842 [278] 15911806 [279]	23300475 [280]
<i>YARS1</i>	Tyrosyl-tRNA synthetase	NM_003680	CMT IC	AD	Definitive	Definitive	16429158 [281] 24354524 [138]	19561293 [282]

Supplementary Table S1: National French consensual gene list for the genetic diagnosis of NSIPN

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