Table 1: Non-syndromic inherited neuropathies.
These are grouped by their pattern of inheritance and phenotypes, and largely follows OMIM (http://www.ncbi.nlm.nih.gov/Omim/), which uses inconsistent nomenclature, and includes some rare forms that have not been confirmed. Dominant diseases, including X-linked dominant diseases, are **bolded**. The genes are named by the HUGO gene nomenclature (http://www.genenames.org/). Table 2 list the inherited syndromes that typically overshadowed by the other aspects of the syndrome; Table 3 lists the neuropathies in Table 2 that can be the presenting clinical issue.

disease (OMIM)	gene (OMIM)	clinical
CMT1 (autosomal dominant demyelinating neuropathy)		
HNPP (162500)	*PMP22* (601097)	focal neuropathies, sometimes severe, transient palsies
CMT1A (118220)	*PMP22* (601097)	CMT1 phenotype
CMT1B (118200)	*MPZ* (159440)	phenotypes range from CHN and DSS to typical CMT1
CMT1C (601098)	*LITAF* (603795)	CMT1 phenotype
CMT1D (607678)	*EGR2* (129010)	phenotypes range from CHN and DSS to typical CMT1
CMT1E (118300)	*PMP22* (601097)	phenotypes range from CHN and DSS to typical CMT1, hearing loss
CMT1F (607734)	*NEFL* (162280)	severe neuropathy, slow nerve conduction velocity owing to small, myelinated axons, hearing loss
CMT1G (618279**)**	*PMP2* (170715)	CMT1 phenotype
Slowed nerve conduction velocity (608236)	*ARHGEF10* (608136)	slowed conduction velocities but no clinical neuropathy
autosomal recessive demyelinating neuropathy (“CMT4”)		
CMT4A (214400)	*GDAP1* (606598)	SEOAN phenotype, vocal cord and diaphragmatic weakness
CMT4B1 (601382)	*MTMR2* (603557)	severe CMT1 or DSS; focally folded myelin sheaths
CMT4B2 (604563)	*SBF2* (607697)	severe CMT1 or DSS, glaucoma; focally folded myelin sheaths
CMT4B3 (615284)	*SBF1*(603560)	demyelinating CMT phenotype or syndromic axonal neuropathy with strabismus, microcephaly, syndactally; focally folded myelin sheaths
CMT4C (601596)	*SH3TC2* (608206)	CMT phenotype, scoliosis, hearing loss
CMT4D (601455)1	*NDRG1* (605262)	severe CMT, hearing loss; founder mutation in Roma
CMT4E (605253)	*EGR2* (129010)	DSS/CHN phenotype
CMT4F (614895)	*PRX* (605725)	DSS/CHN or CMT1 phenotypes
CMT4G (605285)	*HK1* (142600)	CMT phenotype; founder mutation in Roma; allelic with hemolytic anemia due to hexokinase deficiency (235700) and retinitis pigmentosa 79 (617460)
CMT4H (609311)	*FGD4* (11104)	CMT1 phenotype, often early onset; focally folded myelin sheaths
CMT4J (611228)	*FIG4* (609390)	variable onset, variable severity, with non-length dependent weakness, can rapidly progress;

		allelic with ALS11 (612577)	
CMT4K (616684)1	*SURF1* (185620)	The designation of CMT is misleading; they have Leigh syndrome with neuropathy as the presenting issue (see Table 3).	
no OMIM (145900)	*PMP22* (601097)	DSS phenotype	
no OMIM (145900)	*MPZ* (159440)	DSS phenotype	
severe, demyelinating neuropathies; dominant or recessive			
Déjérine-Sottas neuropathy (DSN) (145900)	*PMP22* (601097) *MPZ* (159440) *EGR2* (129010) *PRX* (605725) *FGD4* (11104)	*FIG4* (609390) *MTMR2* (603557) *SBF2* (607697) *SH3TC2* (608206)	dominant or recessive mutations that cause a severe, demyelinating neuropathy with an onset in early childhood
congenital hypo-myelinating neuropathy (CHN) (605253)	*PMP22* (601097) *MPZ* (159440) *EGR2* (129010)	dominant or recessive mutations that cause a severe, demyelinating neuropathy with an infantile onset	
CMT, dominant intermediate (CMTDI)			
CMTDIA (606483)		CMT phenotype	
CMTDIB (606482)	*DNM2* (602378)	CMT phenotype; also myopathy, neutropenia, cataracts; allelic to centronuclear myopathy (160150)	
CMTDIC (608323)	*YARS* (603623)	CMT phenotype	
CMTDID (607791)	*MPZ* (159440)	"axonal phenotype" associated with some *MPZ* mutations; allelic to CMT1B (118200)	
CMTDIE (614455)1	*INF2* (610982)	CMT phenotype, most have focal, segmental glomerulosclerosis; allelic to focal segmental glomerulosclerosis type 5 (613237)	
CMTDIF (615185)	*GNB4* (610863)	CMT phenotype	
CMT2 (autosomal dominant axonal)			
CMT2A (609260)	*MFN2* (608507)	severity ranges from SEOAN to mild, adult-onset axonal neuropathy, optic neuropathy, myelopathy, encephalopathy; allelic to ARCMT2A2B (617087)	
CMT2B (600882)	*RAB7A* (602298)	CMT phenotype, ulcers and infections lead to amputations	
CMT2C (606071)	*TRPV4* (605427)	motor predominant neuropathy of variable severity, diaphragm and vocal cord involvement; allelic to SMA (600175) and scapuloperoneal neuropathy (181405), and many kinds of skeletal defects	
CMT2D (601472)	*GARS* (600287)	motor predominant neuropathy, atrophy intrinsic hand muscles; allelic to dHMN5A (600794)	
CMT2E (607684)	*NEFL* (162280)	severity ranges from severe, childhood-onset to mild, adult-onset axonal neuropathy, hearing loss	
CMT2F (606595)	*HSPB1* (602195)	motor predominant neuropathy, allelic to dHMN2B (608634)	

CMT2I (607677)	*MPZ* (159440)	late-onset axonal CMT2
CMT2J (607736)	*MPZ* (159440)	late-onset axonal CMT2 with hearing loss and unreactive pupils
CMT2K (607831)	*GDAP1* (606598)	CMT2 phenotype to asymptomatic; allelic to CMT4A (214400) and CMTRIA (608340)
CMT2L (608673)	*HSPB8* (608014)	motor predominant neuropathy, allelic to dHMN2A (158590)
CMT2M (606482**)**	*DNM2* (602378)	See CMTDIB (606482)
CMT2N (613287)	*AARS* (601065)	CMT2 phenotype, hearing loss, myelopathy
CMT2O (614228)2	*DYNC1H1* (600112)	motor > > sensory; allelic to SMALED1 (158600) and MRD13 (614563)
CMT2P (614436)	*LRSAM1* (610933)	late-onset CMT2
CMT2Q (615025)	*DHTKD1* (614984)	CMT2 phenotype; this is problematic because recessive mutations cause 2-aminoadipic 2-oxoadipic aciduria (204750), which does not cause neuropathy
CMT2T (617017**)**	*MME* (120520)	late-onset CMT2; allelic to SCA43 (617018)
CMT2U (616280**)**	*MARS* (156560)	late-onset CMT2
CMT2V (616491**)**	*NAGLU* (609701)	late-onset CMT2, neuropathic pain; allelic to mucopolysaccharidosis type IIIB (252920)
CMT2W (616625**)**	*HARS* (142810)	onset early childhood to late adulthood
CMT2Y (616687)	*VCP* (601023)	CMT2; allelic to ALS14 (613954) and IBMPFD (167320)
CMT2Z (616688)	*MORC2* (616661)	CMT2, onset childhood to young adulthood, asymmetric, cramps
CMTCC (616924)	*NEFH* (162230)	CMT2, childhood onset
CMT2DD (618036**)**	*ATP1A1* (182310)	CMT2 phenotype; allelic to hypomagnesemia, seizures, and mental retardation 2 (618314)
HMSNO (604484)3	*TFG* (602498)	adult-onset, proximal and distal weakness, cramping, fasciculation; motor neuropathy and sensory neuropathy; allelic to SPG57 (615658)
no OMIM	*DGAT2* (606983)	CMT2 phenotype
giant axonal neuropathy 2 (610100)	*DCAF8* (615820)	CMT2, childhood onset, slowly progressive, giant axons
no OMIM	*HSPB3* (604624)	motor predominant neuropathy
hereditary neuralgic amyotrophy (162100)	*SEPT9* (604061)	acute, recurrent episode of painful neuropathies related to the brachial plexus
no OMIM	*CHCHD10* (615903)	p.Gly66Val causes CMT2, other mutations cause SMA, Jokela type (615048) and frontotemporal dementia and/or amyotrophic lateral sclerosis 2 (615911)
HNARMD (608895)	*FBLN5* (604580)	childhood onset, hyperelastic skin, macular degeneration; allelic to dominant (614434) and recessive (219100) cutix laxis
autosomal recessive axonal neuropathy (ARCMT2); some are severe early-onset axonal neuropathy (SEOAN) phenotype		
ARCMT2B1 (605588)	*LMNA* (150330)	onset first to third decade, variable severity, p.R298C founder effect NW Africa; allelic to many disorders

ARCMT2B2 (605589)	*PNKP* (605610)	adult onset, cerebellar findings and atrophy; allelic to AOA4 (605610)
ARCMT2A2B (617087)	*MFN2* (608507)	SEOAN phenotype, vocal cord and diaphragmatic involvement
CMT1F (607734)	*NEFL* (162280)	SEOAN phenotype, slow nerve conduction velocities owing to small, myelinated axons
CMT2P (614436)	*LRSAM1* (610933)	young adult onset
CMT2R (615490)	*TRIM2* (614141)	infantile onset
CMT2S (616155)	*IGHMBP2* (600502)	infantile to childhood onset, allelic to DSMA1/SMARD1 (604320)
CMT2T (617017)	*MME* (120520)	late-onset; allelic to SCA43 (617018)
CMT2X (616668)	*SPG11* (610844)	allelic to ALS5 (602099) and SGP11 (604360), identical mutations can cause different phenotypes
neuromyotonia and axonal neuropathy (137200)	*HINT1* (601314)	motor>sensory, onset first or second decade, neuromyotonia
DSMA5 (614881)	*DNAJB2* (604139)	motor > sensory, onset teens to twenties, progresses to proximal muscles
no OMIM	*REEP1* (609139)	congenital neuropathy; allelic with SPG19 (610250)
PNRIID (618124)	*MCM3AP* (603294)	childhood onset, mild intellectual disability
no OMIM	*SGPL1* (603729)	teenage onset, episodic worsening
CMT, recessive intermediate (CMTRI)		
CMTRIA (608340)	*GDAP1* (606598)	axonal CMT, infantile to childhood onset, often progressing to wheel chair, vocal cord and diaphragmatic involvement; allelic to CMT4A (214400) and CMT2K (607831)
CMTRIB (613641)	*KARS* (601421)	axonal CMT
CMTRIC (615376)	*PLEKHG5* (611101)	axonal CMT, onset first to fifth decade; allelic to DSMA4 (611067)
CMTRID (616039)	*COX6A1* (602072)	axonal CMT, childhood onset
hereditary sensory (and autonomic) neuropathy (HSN or HSAN)		
HSN1A (162400)	*SPTLC1* (605712)	neuropathy can have slow conduction, neuropathic pain, loss of distal pain sensation, ulcers, infections, women less affected
HSN1B (608088)	3p24-p22	gastroesophageal reflux and cough
HSN1C (613640)	*SPTLC2* (605713)	neuropathy can have slow conduction, loss of distal pain sensation, ulcers, infections, but no neuropathic pain, variable penetrance, especially in women
HSN1D (613708)	*ATL1* (606439)	severe distal sensory loss and mild atrophy in early adulthood, allelic to SPG3A (182600)
HSN1E (614116)	*DNMT1* (126375)	adult-onset, hearing loss, frontotemporal dementia; allelic to cerebellar ataxia, deafness, and narcolepsy, autosomal dominant (604121)
HSN1F (615632**)**	*ATL3* (609369)	loss of pain sensation, ulcers, infections, no neuropathic pain, childhood to adult onset
HSAN2A (201300)	*WNK1* (605232)	early-onset, loss of pain sensation, ulcers, infections, amputations
HSN2B (613115)	*FAM134B* (613114)	early-onset, ulcero-mutilative neuropathy

HSN2C (614213)	*KIF1A* (601255)	early-onset, ulcero-mutilative neuropathy, allelic to SPG30 (610357) and mental retardation, autosomal dominant 9 (614255)
HSAN2D (243000)	*SCN9A* (603415)	progressive sensory loss, including pain, ulcers, infections, no neuropathic pain, childhood onset, hyposmia, progressive loss of sensory axons
HSAN3 (223900)	*IKBKAP* (603722)	progressive sensory loss, autonomic dysfunction, lack of fungiform papillae, progressive loss of sensory and autonomic neurons, reduced unmyelinated axons
HSAN4 (256800)1, 2	*NTRK1* (191315)	congenital insensitivity to pain and anhidrosis, mild static encephalopathy, no unmyelinated axons
HSAN5 (608654)1, 2	*NGF* (162030)	congenital insensitivity to pain and anhidrosis, mild static encephalopathy, loss of unmyelinated axons, immunodeficiency
HSAN6 (614653)1	*DST* (113810)	neonatal hypotonia, poor feeding and respiratory effort, dysautonomia and early death; also painless, progressive ulcero-mutilative neuropathy; allelic to epidermolysis bullosa simplex-2 (615425)
HSAN7 (615548)	*SCN11A* (604385)	congenital inability to experience pain, ulcers, infections, amputations, no loss of unmyelinated or small myelinated axons
HSAN8 (616488)	*PRDM12* (616458)	congenital insensitivity to pain, loss of small myelinated axons
Marsili syndrome (147430**)**	*ZFHX2* (147430)	hyposensitive to heat, capsaicin, and bone fractures
congenital indifference to pain (243000)4	*SCN9A* (603415)	L1302F mutation; congenital lack of pain, anosmia, no demonstrable axonal loss
no OMIM	*SCN9A* (603415)	congenital lack of pain, no demonstrable axonal loss
primary erythermalgia (133020)4	*SCN9A* (603415)	burning neuropathic pain, usually in the feet, without neuropathy
small fiber neuropathy (133020)	*SCN9A* (603415)	burning neuropathic pain, usually in the feet, with neuropathy
paroxysmal extreme pain disorder (167400)4	*SCN9A* (603415)	painful attacks to the rectal, ocular, or jaw area, even entire body, often triggered, carbamazepine often effective
FEPD 1 (615040)4	*TRPA1* (604775)	infantile onset, debilitating attacks of upper body pain, usually triggered
FEPD 2 (615551)4	*SCN10A* (604427)	adult-onset of paroxysmal pain mainly affecting the distal lower extremities
FEPD 3 (615552)4	*SCN11A* (604385)	early childhood, episodic, intense pain localized primarily to the distal lower extremities
no OMIM	*COL6A5* (611916)	chronic itch, decreased intra-epidermal nerve fibers
DAIPT (617146)4	*PIEZO2* (613629)	profoundly reduced proprioception, selective loss of discriminative touch perception, scoliosis, distal arthrogryposis; allelic to distal arthrogryposis type 3 (114300) and 5 (108145)

CISS1 (272430)4	*CRLF1* (604237)	impaired thermoregulation, hyperhidrosis in response to cold temperatures, dysmorphic features, progressive kyphoscoliosis
CISS2 (610313)4	*CLCF1* (607672)	impaired thermoregulation, hyperhidrosis in response to cold temperatures, dysmorphic features, progressive kyphoscoliosis
hereditary motor neuropathies (HMN or distal spinal muscular atrophy; DSMA)		
HMN1 (182960)	1.35 Mb insertion	onset first-fourth decade, distal weakness, myelopathic features
HMN2A (158590)	*HSPB8* (608014)	onset first or second decade, allelic to CMT2L (608673)
HMN2B (608634)	*HSPB1* (602195)	onset second decade, allelic to CMT2F; 606595)
HMN2C (613376)	*HSPB3* (604624)	onset early 20s
HMN2D (615575)	*FBXO38* (608533)	onset second to fourth decade, progressing to proximal weakness
no OMIM	*HARS* (142810)	onset in first decade; allelic to CMT2W **(**616625**)**
no OMIM	*AARS* (601065)	onset in first decade; allelic to CMT2N (613287)
no OMIM	*FBLN5* (604580)	childhood onset, hyperelastic skin, macular degeneration; allelic to HNARMD (608895), dominant (614434) and recessive (219100) cutix laxis
no OMIM	*SLC12A6* (604878)	T991A mutation; infantile onset; allelic to agenesis of the corpus callosum with peripheral neuropathy (218000)
HMN5A (600794)	*GARS* (600287)	onset first to third decade, starting in the hands, allelic to CMT2D (601472)
HMN5A (600794)5	*BSCL2 (*606158)	onset second to fifth decade, some patients are asymptomatic, starting in the hands, myelopathy; allelic to SPG17 (270685)
HMN5B (614751)5	*REEP1* (609139)	onset second or second decade, starting in the hands, myelopathy; allelic to SPG31 (610250)
HMN/ALS4 (602433)5	*SETX* (608465)	typical onset second decade, spasticity; allelic to SCA1 (606002)
DSMA1/SMARD1 (604320)	*IGHMBP2* (600502)	intrauterine growth retardation, diaphragmatic paralysis
DSMA2 (605726)	*SIGMAR1* (9601978)	onset in first decade, myelopathic features fade as neuropathy predominates, allelic to ALS16 (614373)
DSMA3 (607088)		
DSMA4 (611067)	*PLEKHG5* (611101)	infancy to childhood onset, progressive to respiratory insufficiency; allelic to CMTRIC (615376)
DSMA5 (614881)	*DNAJB2* (604139)	onset teen to twenties, progresses to proximal muscles
no OMIM	*HSPB1* (602195)	motor >> sensory neuropathy, childhood onset; allelic to CMT2F (606595) and HMN2B (608634), both of which are dominant
HMN7A (158580)	*SLC5A7* (608761)	with vocal cord involvement; allelic to CMS20 (617143)
HMN7B (607641)	*DCTN1* (601143)	with vocal cord involvement; allelic to Perry syndrome (168605)
HMN9 (617721)	*WARS* (191050)	onset second decade

scapuloperoneal SMA (181405)	*TRPV4* (605427)	allelic to CMT2C (606071) and congenital distal SMA (181405), and a host of skeletal defects
congenital distal SMA (600175)	*TRPV4* (605427)	allelic to CMT2C (606071) and scapuloperoneal SMA (181405), and a host of skeletal defects
SMALED1 (158600)2	*DYNC1H1* (600112)	childhood onset, followed by minimal progression; allelic to CMT2O (614228) and MRD13 (614563)
SMALED2 (615290)2	*BICD2* (609797)	childhood onset, followed by minimal progression
X-linked (**dominant** or recessive, demyelinating or axonal) CMT or HMN/DSMA		
CMT1X (302800)	*GJB1* (304040)	males more affected than females, intermediate slowing of conduction
CMTX5 (311070)	*PRPS1* (311850)	hearing loss, optic atrophy; allelic to Arts syndrome (304150), X-linked deafness 1 (304500), PRPS-related gout (300661)
CMTX6 (300905)	*PDK3 (300906)*	onset first decade, males more affected than females; the only mutation found to date (R158H) causes a toxic gain of function
SMAX3 (300489)	*ATP7A* (300011)	onset first to third decades; allelic to occipital horn syndrome (304150) and Menkes disease (309400)
no OMIM	*DRP2* (300052)	one case, onset at age 50

CISS: cold-induced sweating syndrome; CMS: congenital myasthenic syndrome; DAIPT: distal arthrogryposis with impaired proprioception and touch; FEPD: familial episodic pain disorder; HNARD: Neuropathy, hereditary, with or without age-related macular degeneration; HNPP: hereditary neuropathy with liability to pressure palsies; IBMPFD: inclusion body myopathy with early-onset Paget disease and frontotemporal dementia; MRD: mental retardation, autosomal dominant; PNRIID: peripheral neuropathy, autosomal recessive, with or without impaired intellectual development; SMALED: spinal muscular atrophy, lower extremity predominant; SMARD: spinal muscular atrophy with respiratory distress; SCA: spinocerebellar ataxia; SPG: spastic paraplegia;

1: Syndromic neuropathy that is classified as CMT or HSAN in OMIM.
2: Static clinical conditions, more likely the result of neuronal loss during development than an ongoing peripheral neuropathy.
3: CMT2O has been reconsidered to be a progressive sensory neuropathy superimposed on a static congenital motor neuronopathy.
4: Altered peripheral sensory and/or autonomic function without known evidence of axonal loss.
5: These patients typically have a myelopathy and a length-dependent motor axonopathy.