Supplementary Digital Content Table 1 Hereditary Neuropathies by Gene

Gene	Primary relevant phenotype		New nomenclature	Inheritance pattern	Distinguishing neuropathy features and other associated phenotypes
PMP22	CMT1A	Duplication	CMT-De-AD-PMP22	AD	Classic demyelinating CMT phenotype Roussy-Levy syndrome (tremor, ataxia)
	CMT1E	Point mutation	CMT-De-AD-PMP22	AD	Classic demyelinating CMT phenotype Sensorineural hearing loss
	HNPP	Deletion	HNPP-De-AD-PMP22	AD	Early-adult–onset demyelinating neuropathy Episodic peripheral nerve palsies at common compression sites, often precipitated by minor trauma or compression Prolonged distal motor latencies Slowing of focal conduction velocity (median and peroneal nerves most affected), sometimes with conduction block
MPZ	CMT1B		CMT-De-AD-MPZ	AD	Early-onset classic demyelinating CMT Roussy-Levy syndrome (tremor, ataxia) Very slow conduction velocities (often <15 meters per second)
	CMT2I		CMT-Ax-AD-MPZ	AD	Late-onset sensory-predominant axonal CMT
	CMT2J		CMT-Ax-AD-MPZ	AD	Late-onset sensory-predominant axonal CMT Pupillary abnormalities and deafness
	CMTDID		CMT-In-AD-MPZ	AD	Late-onset intermediate neuropathy
LITAF (*SIMPLE*)	CMT1C		CMT-De-AD-LITAF	AD	Mild classic demyelinating CMT phenotype Occasional conduction block Sensory neuropathy
EGR2	CMT1D		CMT-De-AD-EGR2	AD	Dejerine-Sottas disease Congenital hypomyelinating neuropathy Adult-onset demyelinating neuropathy

				Respiratory and cranial nerve involvement
	CMT2	CMT-Ax-AD-EGR2	AD	Adult-onset axonal or intermediate neuropathy
	CMT4E	CMT-De-AR-EGR2	AR	Congenital hypomyelinating neuropathy Infantile hypotonia, areflexia
NEFL	CMT1F	CMT-De-AD/AR-NEFL	AD, AR	Early-onset severe-demyelinating neuropathy
	CMT2E	CMT-Ax-AD-NEFL	AD	Adolescent-onset and early-adult–onset axonal neuropathy Cerebellar dysfunction (ataxia, dysarthria)
	CMTDIG	CMT-In-AD-NEFL	AD	Childhood-onset through adult-onset intermediate neuropathy Spastic gait, cerebellar ataxia, dysarthria
PMP2	CMT1G	CMT-De-AD-PMP2	AD	Classic demyelinating CMT phenotype
FBLN5	CMT1H	CMT-De-AD-FBLN5	AD	Early-adult–onset sensorimotor neuropathy
				Neuropathy with macular degeneration
POLR3B	CMT1I	CMT-De-AD-POLR3B	AR	Early-childhood–onset sensorimotor neuropathy Ataxia, spasticity, pyramidal signs, seizures
			AR	Hypomyelinating leukodystrophy-8
ITPR3	CMT1J	CMT-De-AD-ITPR3	AD	Variable-onset demyelinating or intermediate sensorimotor neuropathy
CFAP276 (*C1orf194*)	CMT1/CMTDI	CMT-De/In-AD-CFAP276	AD	Variable-onset demyelinating or intermediate sensorimotor neuropathy
MFN2	CMT2A	CMT-Ax-AD/AR-MFN2	AD, AR	Childhood-onset through adult-onset Positive sensory symptoms More progressive course Optic nerve atrophy
KIF1B	CMT2A1	CMT-Ax-AD-KIF1B	AD	Childhood-onset through adult-onset axonal neuropathy Cranial nerve involvement

RAB7A (*RAB7*)	CMT2B	CMT-Ax-AD-RAB7	AD	Early-childhood–onset or adolescence-onset axonal neuropathy Classic motor-predominant axonal neuropathy
	HSAN	HSAN-AD-RAB7	AD	Adolescent or adult-onset distal axonal sensory neuropathy Skin ulcerations and amputations Some distal weakness and atrophy
LMNA	CMT2B1	CMT-Ax-AR-LMNA	AR	Adolescent-onset axonal neuropathy Can have rapid and severe progression or be mild Can have both proximal and distal weakness Upper and lower extremity involvement Founder mutation in Northern African descent
			AD	Emery-Dreifuss muscular dystrophy
PNKP (previously MED25)	CMT2B2	CMT-Ax-AR-PNKP	AR	Early-onset or late-onset axonal neuropathy Slowly progressive CMT2 Cerebellar dysfunction (slurred speech, ataxia, oculomotor involvement)
TRPV4	CMT2C	CMT-Ax-AD-TRPV4	AD	Infantile through adult-onset axonal neuropathy Diaphragmatic and vocal cord paresis or stridor Sensorineural hearing loss Urinary urgency and incontinence Skeletal dysplasia and short stature
	dHMN8	dHMN-AD-TRPV4		
				Congenital SMA with arthrogryposis
				Scapuloperoneal muscular atrophy: scapular winging, distal lower extremity weakness and atrophy, vocal cord paralysis
NEFH	CMT2CC	CMT-Ax-AD-NEFH	AD	Adult-onset nonlength-dependent axonal neuropathy Proximal and distal weakness Early ankle plantar flexion weakness

GARS1	CMT2D	CMT-Ax-AD-GARS1	AD	Infantile-onset to early-adult–onset axonal neuropathy Can disproportionately affect the upper extremities
	dHMN5A/ James type of infantile SMA	dHMN-AD-GARS1		Infantile to early-adult–onset distal hereditary motor neuropathies, upper limb onset Respiratory insufficiency and poor feeding (SMARD-like)
			AR	Mitochondrial disorder with intellectual disability, structural brain abnormalities, cardiac involvement
ATP1A1	CMT2DD	CMT-Ax-AD-ATP1A1	AD	Early-onset intellectual disability, spasticity, peripheral motor-predominant axonal neuropathy Hereditary spastic paraplegia
MPV17	CMT2EE	CMT-Ax-AR-MPV17	AR	Adolescent-onset pure sensorimotor neuropathy Neuropathy and leukoencephalopathy Restrictive lung disease Navajo neurohepatopathy
HSPB1 (*HSP27*)	CMT2F	CMT-Ax-AD/AR-HSPB1	AD, AR	Variable age of onset axonal neuropathy Occasional hyperreflexia and pyramidal signs
	dHMN2B	dHMN-AD-HSPB1	AD	Juvenile-onset or early-adult–onset distal hereditary motor neuropathies Disproportionate plantar flexion weakness Occasional spastic paraplegia with hyperreflexia and mild cerebellar signs
				Distal vacuolar myopathy
CADM3	CMT2FF	CMT-Ax-AD-CADM3	AD	Early-onset through adult-onset axonal motor-predominant neuropathy Marked upper extremity involvement Preserved or increased reflexes

GBF1	CMT2GG	CMT-Ax-AD-GBF1	AD	Adult-onset motor-predominant axonal neuropathy Preserved or increased reflexes
	dHMN	dHMN-Ax-AD-GBF1		
GDAP1	CMT2H	CMT-Ax-AR-GDAP1	AR	Early-onset severe axonal or intermediate neuropathy Upper extremity involvement (claw hands) Vocal cord paralysis
	CMT4A		AR	Infantile or early childhood-onset progressive mixed axonal and demyelinating neuropathy Vocal cord paresis
	CMT2K		AD, AR	Childhood through late adult-onset axonal neuropathy Finnish founding mutation Heterozygous patients can have mild symptoms
	CMTRIA	CMT-In-AR-GDAP1	AR	No distinguishing features
JAG1	CMT2HH	CMT-Ax-AD-JAG1	AD	Mild distal motor and sensory axonal neuropathy Vocal cord paralysis with stridor in infancy or early childhood, sometimes requiring tracheostomy
SLC12A6 (*KCC3*)	CMTII	CMT-Ax-AD/AR-SLC12A6	AD, AR	Variable age of onset axonal or intermediate neuropathy Intellectual disability in some
	dHMN	dHMN-Ax-AD-SLC12A6	AD	Infantile-onset progressive motor neuropathy Mild demyelinating features
			AR	Agenesis of the corpus callosum with peripheral neuropathy
HSPB8	CMT2L	CMT-Ax-AD-HSPB8	AD	Adolescent-onset to adult-onset axonal neuropathy

				Some with proximal weakness in addition to distal weakness Scoliosis
	dHMN2A	dHMN-AD-HSPB8	AD	Juvenile-onset or early-adult–onset distal hereditary motor neuropathies Concurrent distal myofibrillar myopathy with aggregates and vacuoles
DNM2	CMT2M/CMTDIB	CMT-Ax/In-AD-DNM2	AD	Adolescent-onset axonal or intermediate neuropathy Neutropenia Cataracts
				Centronuclear myopathy
AARS1	CMT2N	CMT-Ax-AD-AARS1	AD	Childhood-onset through adult-onset axonal neuropathy Some patients with upper extremity involvement Case report of stroke-like episodes with reversible white matter lesions
DYNC1H1	CMT2O	CMT-Ax-AD-DYNC1H1	AD	Early-childhood–onset axonal neuropathy Delayed motor milestones or abnormal gait Variable sensory loss
	dSMA1/SMALED1	dSMA-AD-SYNC1H1	AD	Childhood-onset DSMA
LRSAM1	CMT2P	CMT-Ax-AD/AR-LRSAM1	AD, AR	Classic adult-onset axonal CMT
DHTKD1	CMT2Q	CMT-Ax-AD-DHTKD1	AD	Adolescent-onset through early-adult–onset axonal neuropathy
				Amyotrophic lateral sclerosis
			AR	Infantile SMA with cognitive delay
				2-aminoadipic and 2-oxoadipic aciduria, hypotonia, developmental delay, epilepsy

TRIM2	CMT2R	CMT-Ax-AR-TRIM	AR	Infantile-onset axonal neuropathy with severe weakness and areflexia Hypotonia and club foot, delayed motor milestones Vocal cord paralysis and other cranial nerve involvement
IGHMBP2	CMT2S	CMT-Ax-AR-IGHMBP2	AR	Childhood-onset axonal neuropathy Delayed motor development Scoliosis
	dHMN6/SMARD1	dHMN-AR-IGHMBP2	AR	Infantile-onset hypotonia with distal weakness and respiratory failure
MME	CMT2T	CMT-Ax-AD/AR-MME	AD, AR	Late-onset axonal CMT
			AD	Spinocerebellar ataxia 43
MARS1	CMT2U	CMT-Ax-AD-MARS1	AD	Late-adult–onset axonal neuropathy
			AR	Hereditary spastic paraplegia 70
NAGLU	CMT2V	CMT-Ax-AD-NAGLU	AD	Late-onset painful axonal sensory neuropathy
			AR	Mucopolysaccharidosis type IIIB (Sanfilippo B)
HARS1	CMT2W	CMT-Ax-AD-HARS1	AD	Variable-onset classic axonal CMT
	CMT1	CMT-De-AD-HARS1	AD	Late-onset demyelinating neuropathy Cognitive impairment and cerebellar atrophy
SPG11	CMT2X	CMT-Ax-AR-SPG11	AR	Childhood-onset through early-adult–onset axonal neuropathy Tremor, fasciculations, bladder dysfunction, sexual dysfunction
				Hereditary spastic paraplegia 11
				Juvenile amyotrophic lateral sclerosis-5

VCP	CMT2Y	CMT-Ax-AD-VCP	AD	Variable onset classic axonal CMT
				Amyotrophic lateral sclerosis type 6 and/or frontotemporal dementia
				Inclusion body myositis
MORC2	CMT2Z	CMT-Ax-AD-MORC2	AD	Axonal sensorimotor neuropathy that can be sensory predominant or motor neuropathy/neuronopathy Proximal weakness Scoliosis, hearing loss, pigmentary retinopathy
				Infantile SMA phenotype with variable CNS involvement (cerebellar hypoplasia, white matter abnormalities, spasticity, intellectual disability) +/-diaphragmatic paralysis
				Developmental delay, impaired growth, dysmorphic features, and axonal peripheral neuropathy
B4GALNT1	CMT2	CMT-Ax-AR-B4GALNT1	AR	Adult-onset axonal neuropathy Intellectual disability
				Hereditary spastic paraplegia 26
NF-L	CMT2	CMT-Ax-AD-NF-L	AD	Late-onset axonal sensorimotor neuropathy
DGAT1	CMT2	CMT-Ax-AD-DGAT1	AD	Early-onset axonal sensorimotor neuropathy Sensory ataxia
AGRN	CMT2	CMT-Ax-AR-AGRN	AR	Adolescent-onset axonal sensorimotor neuropathy
				Congenital myasthenic syndrome 8
ARHGEF28	CMT2	CMT-Ax-AR-ARHGEF28	AR	Childhood-onset through adult-onset sensorimotor axonal neuropathy Severe scoliosis
			AD	Amyotrophic lateral sclerosis

HINT1	CMT2	CMT-Ax-AR-HINT1	AR	Childhood-onset axonal neuropathy, pure motor or mixed sensorimotor Neuromyotonia
	dHMN	dHMN-Ax-AR-HINT1		
AHMAK2	CMT2	CMT-Ax-AR-AHMAK2	AR	Unknown
MCM3AP (*GANP*)	CMT2	CMT-Ax-AR-MCM3AP	AR	Childhood-onset progressive sensorimotor axonal neuropathy Intellectual disability
MYO9A	CMT2	CMT-Ax-AR-MYO9	AR	Childhood-onset or adolescent-onset sensorimotor axonal neuropathy Optic nerve atrophy
NRG1	CMT2	CMT-Ax/In-AR-NRG1	AR	Early-adult–onset sensorimotor axonal or intermediate neuropathy
MTMR2	CMT4B1	CMT-De-AR-MTMR2	AR	Early-onset severe-demyelinating neuropathy Tremor and dysmetria Cranial nerve involvement Vocal cord and respiratory dysfunction
SBF2 (previously *MTMR13*)	CMT4B2	CMT-De-AR-SBF2	AR	Early-childhood–onset demyelinating neuropathy Congenital glaucoma Vocal cord and respiratory dysfunction
SBF1	CMT4B3	CMT-De-AR-SBF1	AR	Childhood-onset demyelinating neuropathy Microcephaly and cognitive impairment Cranial nerve involvement
SH3TC2 (previously *KIAA1985*)	CMT4C	CMT-De-AR-SH3T2	AR	Early-onset demyelinating neuropathy Severe spine and foot deformities Cranial nerve involvement Respiratory insufficiency
NDRG1	CMT4D	CMT-De-AR-NDRG1	AR	Early-onset demyelinating neuropathy Sensorineural hearing loss Founder mutation in the Romani population in Europe

PRX	CMT4F	CMT-De-AR-PRX	AR	Variable-onset demyelinating neuropathy Dejerine-Sottas disease phenotype Sensory ataxia Very slow conduction velocities with temporal dispersion
HK1	CMT4G	CMT-De-AR-HK1	AR	Childhood-onset demyelination neuropathy Russe type due to founder mutation in the Romani population in Europe
			AD	Neurodevelopmental disorder, structural brain abnormalities, visual impairment
FGD4 (previously *FDG1*)	CMT4H	CMT-De-AR-FGD4	AR	Variable-onset demyelinating neuropathy Severe scoliosis
FIG4	CMT4J	CMT-De-AR-FIG4	AR	Early-childhood–onset through adult-onset demyelinating neuropathy Features of acquired demyelinating (temporal dispersion, conduction block) More severe childhood-onset with proximal>distal Asymmetry Scoliosis
	dHMN	dHMN-Ax-AR-FIG4		Childhood-onset axonal motor neuropathy
			AD	Amyotrophic lateral sclerosis type 11
SURF1	CMT4K	CMT-De-AR-SURF1	AR	Childhood-onset demyelinating neuropathy Nystagmus and late-onset cerebellar ataxia Lactic acidosis and white matter lesions
				Mitochondrial complex IV deficiency nuclear type 1
PCK2	CMT4	CMT-De-AR-PCK2	AR	Childhood distal weakness and ataxia Temporal dispersion and conduction block
SORD	CMT4	CMT-Ax-AR-SORD	AR	Adolescent-onset or early-adult–onset axonal neuropathy

				Disproportionately weaker plantar flexion
	dHMN	dHMN-Ax-AR-SORD		Adolescent-onset or early-adult–onset distal hereditary motor neuropathies
YARS1	CMTDIC	CMT-In-AD-YARS1	AD	Adolescent-onset or adult-onset intermediate neuropathy or motor neuronopathy
			AR	Multisystem disorders with global developmental delay, microcephaly, hypotonia, ataxia, and brain anomalies; hematologic, endocrine, and liver dysfunction
INF2	CMTDIE	CMT-In-AD-INF2	AD	Childhood-onset through early-adult–onset intermediate neuropathy Focal segmental glomerulonephritis
GNB4	CMTDIG	CMT-In/De-AD-GNB4	AD	Adolescent-onset intermediate or demyelinating neuropathy
KARS1	CMTRIB	CMT-In/Ax-AR-KARS	AR	Developmental delay and self-injurious behavior Vestibular schwannoma Dysmorphic features
				Deafness and/or progressive leukoencephalopathy, congenital or adult onset
COX6A1	CMTRID	CMT-In/Ax-AR-COX6A1	AR	Adult-onset intermediate or axonal neuropathy
GJB1	CMTX1	CMT-De/In-XLD-GJB1	XLD	Adolescent-onset demyelinating or intermediate CMT Disproportionately weaker thumb abduction/APB atrophy Transient neurologic episodes with white matter abnormalities Males more severely affected than females
AIFM1	CMTX4	CMT-Ax-XLR-AIFM1	XLR	Cowchock syndrome

				Infancy-onset through adult-onset axonal sensory neuropathy greater than motor neuropathy Delayed motor development Hearing loss Cerebellar ataxia Intellectual disability White matter T2 hyperintensities Mitochondrial disorder
PRPS1	CMTX5	CMT-Ax-XLR-PRPS1	XLR	Infantile-onset through adult-onset mixed demyelinating and axonal neuropathy Optic atrophy and deafness Arts syndrome (Intellectual disability, hypotonia, ataxia, motor impairment, hearing loss, optic atrophy) and X-linked deafness-1 Females with mild neuropathy and hearing loss
PDK3	CMTX6	CMT-Ax-XLD-PDK3	XLD	Adolescent-onset axonal sensorimotor polyneuropathy Occasional hearing loss Females mildly affected
DRP2	CMTX	CMT-In-XLR-DRP2	XLR	Early-adult–onset intermediate neuropathy
HSPB3	dHMN2C	dHMN-Ax-AD-HSPB3	AD	Early-adult–onset distal hereditary motor neuropathies Can have sensory involvement
	CMT2	CMT-Ax-AD-HSPB3		
FBXO38	dHMN2D	dHMN-Ax-AD/AR-FBXO38	AD, AR	Juvenile-onset or adult-onset distal hereditary motor neuropathies with prominent plantar flexion weakness Early-onset autosomal recessive form
BSCL2	dHMN5C	dHMN-Ax-AD-BSCL2	AD	Upper extremity predominant distal hereditary motor neuropathies

				Can have pyramidal signs
	CMT2	CMT-Ax-AD-BSCL2		
				Amyotrophic lateral sclerosis
				Hereditary spastic paraplegia; pure or complex (Silver syndrome)
REEP1	dHMN5B	dHMN-Ax-AD-REEP1	AD	Childhood-onset or adolescent-onset distal hereditary motor neuropathies, upper limb onset
				Hereditary spastic paraplegia 31
	dSMA6/SMARD	dSMA-Ax-AR-REEP1	AR	Infantile-onset distal weakness and contractures Diaphragmatic weakness
SLC5A7	dHMN7A	dHMN-Ax-AD-SLC5A7	AD	Vocal cord paralysis
DCTN1	dHMN7B	dHMN-Ax-AD-DCTN1	AD	Early-adult–onset distal hereditary motor neuropathies with facial and arm weakness Vocal cord paralysis
				Juvenile amyotrophic lateral sclerosis
				Perry syndrome: parkinsonism, weight loss, depression, central hypoventilation
SETX	dHMN	dHMN-Ax-AD-SETX	AD	Distal hereditary motor neuropathies with pyramidal signs
				Juvenile amyotrophic lateral sclerosis type 4
			AR	Spinocerebellar ataxia with axonal neuropathy-2
WARS1	dHMN9	dHMN-Ax-AD-WARS1	AD	Juvenile-onset distal hereditary motor neuropathies
			AR	Neurodevelopmental disorder with microcephaly and epilepsy
EMILIN1	dHMN10	dHMN-Ax-AD-EMILIN1	AD	Early childhood–onset motor neuropathy Pyramidal signs Connective tissues disease

AAAS	dHMN	dHMN-Ax-AR-AAAS	AR	Early-childhood–onset motor neuropathy Triple A or Allgrove syndrome: achalasia, alacrimia, adrenal insufficiency Intellectual disability, spasticity, dysautonomia, cranial neuropathies
VRK1	dHMN	dHMN-Ax-AD-VRK1	AR	Childhood-onset to early-adult–onset motor neuropathy
				Juvenile amyotrophic lateral sclerosis
				SMA with pontocerebellar hypoplasia
KBTBD13	dHMN	dHMN-Ax-AD-KBTBD13	AR	Early-adult–onset motor neuropathy Upper motor neuron signs
			AD	Nemaline myopathy type 6
MYH14	dHMN	dHMN-Ax-AD-MYH14	AD	Childhood-onset distal hereditary motor neuropathies, peripheral neuropathy-myopathy-hoarseness-hearing loss (PNMHH)
	CMT2	CMT-Ax-AD-MYH14		Mild sensory involvement
SPTAN1	dHMN	dHMN-Ax-AD-SPTAN1	AD	Adolescent-onset motor or sensorimotor neuropathy
	CMT2	CMT2-Ax-AD-SPTAN1		
				Developmental and epileptic encephalopathy 5
				Hereditary spastic paraplegia with cerebellar ataxia
			AR	Pontocerebellar atrophy and intellectual disability
COQ7	dHMN	dHMN-Ax-AR-COQ7	AR	Childhood-onset motor neuropathy
				Encephalopathy and multisystem mitochondrial disorder
TBCK			AR	

BICD2	dSMA2/ SMALED2	dSMA-Ax-AD-BICD2	AD	Congenital contractures and infantile-onset distal lower extremity weakness Pyramidal or hereditary spastic paraplegias–like features
SIGMAR1	dSMA2	dSMA-Ax-AR-SIGMAR	AR	Childhood-onset distal hereditary motor neuropathies with pyramidal features Jerash type
				Juvenile amyotrophic lateral sclerosis type 16
ATP7A	dSMA3	dSMA-Ax-XLR-ATP7A	XLR	Childhood-onset and early-adult–onset distal hereditary motor neuropathies Mild sensory involvement Skin features and dysautonomia (Occipital horn syndrome)
PLEKHG5	dSMA4	dSMA-Ax-AR-PLEKHG5	AR	Early childhood–onset distal hereditary motor neuropathies or SMA with proximal weakness
	CMTRIC	CMT-In-AR-PLEKHG5	AR	Childhood-onset through adult-onset intermediate neuropathy
DNAJB2 (previously *HSJ1*)	dSMA5	dSMA-Ax-AR-DNAJB2	AR	Early-adult–onset distal hereditary motor neuropathies
	CMT2	CMT-Ax-AR-DNAJB2		Axonal sensorimotor neuropathy Hearing loss, parkinsonism, pyramidal signs
SPTLC1	HSAN1A	HSAN-Ax-AD-SPTLC1	AD	Adult-onset HSAN with mild motor involvement Early-onset sensorimotor neuropathy (“S331 syndrome”) Juvenile amyotrophic lateral sclerosis
SPTLC2	HSAN1C	HSAN-Ax-AD-SPTLC2	AD	Adult-onset HSAN with mild motor involvement
ATL1	HSAN1D	HSAN-Ax-AD-ATL1	AD	Sensory neuropathy Sometimes with subtle upper motor neuron findings Hereditary spastic paraplegia 3A

DNMT1	HSAN1E	HSAN-Ax-AD-DNMT1	AD	Sensory axonal neuropathy with dementia and hearing loss Cerebellar ataxia, deafness, and narcolepsy
ATL3	HSAN1F	HSAN-Ax-AD-ATL3	AD	Adolescent-onset or adult-onset HSAN Spasticity, fractures, poor wound healing
WNK1 (previously *HSN2*)	HSAN2A	HSAN-Ax-AR-WNK1	AR	Childhood-onset HSAN Severe acral mutilation
RETREG1 (previously *FAM134B*)	HSAN2B	HSAN-Ax-AR-RETREG1	AR	Childhood-onset or adult-onset HSAN Spasticity, can have weakness Risk of osteomyelitis Hyperhidrosis
KIF1A	HSAN2C	HSAN-Ax-AR-KIF1A	AR	Childhood-onset HSAN Muscle weakness, intellectual disability
			AD, AR	Hereditary spastic paraplegia 30
			AD	Congenital ataxia syndrome
			AD	Amyotrophic lateral sclerosis
SCN9A	HSAN2D	HSAN-Ax-AR-SCN9A	AR	Congenital-onset HSAN/congenital insensitivity to pain Anosmia and fractures
			AD	Primary erythromelalgia, small fiber neuropathy
ELP1 (previously *IKBKAP*)	HSAN3/ Riley-Day syndrome	HSAN-Ax-AR-ELP1	AR	Infantile-onset predominantly autonomic neuropathy with proprioceptive loss Absence of fungiform papillae of the tongue Spinal deformities High carrier frequency in the Ashkenazi Jewish population
NTRK1	HSAN4	HSAN-Ax-AR-NTRK1	AR	Congenital-onset HSAN Intellectual disability Fractures, osteomyelitis Anhidrosis leading to hyperthermia

NGF	HSAN5	HSAN-Ax-AR-NGF	AR	Congenital-onset HSAN Intellectual disability Fractures, osteomyelitis Anhidrosis leading to hyperthermia
DST	HSAN6	HSAN-Ax-AR-DST	AR	Severe psychomotor retardation, joint contractures, alacrimia, feeding difficulties, cardiovascular instability, hypomimia and muscle weakness
SCN11A	HSAN7	HSAN-Ax-AD-SCN11A	AD	Congenital-onset sensory neuropathy Delayed motor development Pruritis and skin ulcers Gastrointestinal dysmotility Familial episodic pain syndrome-3
PRDM12	HSAN8	HSAN-AR-PRDM12	AR	Congenital-onset HSAN/congenital insensitivity to pain Facial injuries, corneal scarring Hypohidrosis
TECPR2	HSAN9	HSAN-AR-TECPR2	AR	Early-childhood–onset sensory and autonomic neuropathy Global developmental delay and intellectual disability Dysarthria and ataxic gait Dysmorphic features Central hypoventilation
CLTCL1	CIP	CIP-AR-CLTCL1	AR	Congenital insensitivity to pain with normal motor function Developmental delay and intellectual disability Dysmorphic features, visual impairment, seizures
FAAHP1 (*FAAH-OUT*)	CIP	CIP-Ax-FAAHP1	AD	Congenital insensitivity to pain Lack of anxiety

ZFHX2	CIP	CIP-AD-ZFHX2	AD	Marsili syndrome Anhidrosis
FLVCR1	HSAN	HSAN-De-AR-FLVCR-1	AR	Childhood-onset hypomyelinating HSAN Retinitis pigmentosa, sensory ataxia
GMPPA	HSAN	HSAN-Ax-AR-GMPPA	AR	Childhood-onset autonomic neuropathy Alacrimia and achalasia Intellectual disability
MADD	HSAN	HSAN-Ax-AR-MADD	AR	Autonomic neuropathy with reduced pain sensation Developmental delay with endocrine, exocrine, autonomic, and hematologic abnormalities
CCT5	HSAN	HSAN-Ax-AR-CCT5	AR	Infantile-onset or childhood-onset mutilating sensory neuropathy Spastic paraplegia
PIEZO2	HSAN	HSAN-AR-PIEZO22	AR	Childhood-onset proprioceptive loss, sensory ataxia, scoliosis
			AD, AR	Arthrogryposis
COX20	HSN/MC4DN11		AR	Childhood-onset sensory neuropathy or neuronopathy Tremor, dystonia, choreoathetosis, dysarthria Intellectual disability Mitochondrial complex IV deficiency nuclear type 11

AD = autosomal dominant; APB = abductor pollicis brevis; AR = autosomal recessive; CIP = congenital insensitivity to pain; CMT = Charcot-Marie-Tooth; CNS = central nervous system; dSMA = distal spinal muscular atrophy; HSAN = hereditary sensory and autonomic neuropathies; HSP = hereditary spastic paraplegias; SMA = spinal muscular atrophy; SMARD = spinal muscular atrophy with respiratory distress.