Table S1. List of 103 CMT and related neuropathies genes.
AARS
ABHD12
AIFM1
ARHGEF10
ATL1
ATL3
ATP7A
BICD2
BSCL2
TWNK
C12orf65
CCT5
COX6A1
CTDP1
DCAF8
DCTN1
DHTKD1
DNAJB2
DNM2
DNMT1
DST
DYNC1H1

EGR2
EXOSC8
RETREG1
FBLN5
FBXO38
FGD4
FIG4
GAN
GARS1
GDAP1
GJB1
GJB3
GNB4
HADHA
HADHB
HARS1
HINT1
KCNA4
HOXD10
HSPB1
HSPB3
HSPB8
IFRD1
IGHMBP2
ELP1
INF2
KARS1
KIF1A

KIF1B
KIF5A
LITAF
LMNA
LRSAM1
MARS1
MED25
MFN2
MME
MPZ
MTMR2
MYH14
NDRG1
NEFL
NGF
NGFB
NTRK1
OPA1
PDK3
PLEKHG5
PMP22
POLG
PRPS1
PRX
RAB7A
REEP1
SBF1
SBF2

SCN10A
SCN11A
SCN9A
SEPTIN9
SETX
SH3TC2
SLC12A6
SLC52A3
SLC5A7
SMN1
SOX10
SPTLC1
SPTLC2
SURF1
SYT2
TFG
TRIM2
TRPV4
TTR
TUBB3
TYMP
VAPB
VCP
WNK1
YARS1

Table S2A. Information of identified variants in CMT and related IPN genes in this study.

Family Number	Gene Symbol (MIM number)	Gene Description	Gender	Age (age of onset) years	inheritance	Variant Description	Gene Related Disorders	Reference for relation of variant with CMT or other Neuropathies	Observed phenotype and EMG/NCV result	Published and expected phenotype
Pathogenic										
1	GJB1 (304040)	Gap junction beta-1 protein	M	31 (18)	XLD	c.370A>G (p.Lys124Glu)	**Charcot-Marie-Tooth neuropathy, X-linked dominant, 1**	PMID:27234031	-Muscular weakness and distal muscle atrophy,pes cavus **-Axonal sensorimotor polyneuropathy**	Pes cavus, Paraparesis, Distal limb muscle weakness and atrophy due to peripheral neuropathy, Reduced motor nerve conduction velocity (NCV) (range less than 38 m/s to normal)and Axonal degeneration
2	GJB1 (304040)	Gap junction beta-1 protein	M	28 (20)	XLD	c.491G>A (p.Arg164Gln)	**Charcot-Marie-Tooth neuropathy, X-linked dominant, 1**	PMID: 16301507, 15006706, 12499506, 8733054, 7580242	-Muscular weakness and distal muscle atrophy, pes cavus,sensory loss,hearing impairment **-Demyelinating sensorimotor polyneuropathy with Secondary axonal degeneration**	- Pes cavus, Paraparesis, Distal limb muscle weakness and atrophy due to peripheral neuropathy - Sensorineural hearing loss (uncommon), Reduced motor nerve conduction velocity (NCV) (range less than 38 m/s to normal)and Axonal degeneration
3	GJB1 (304040)	Gap junction beta-1 protein	M	23 (15)	XLD	c.491G>A (p.Arg164Gln)	**Charcot-Marie-Tooth neuropathy, X-linked dominant, 1**	PMID: 16301507, 15006706, 12499506, 8733054, 7580242	-Muscular weakness and distal muscle atrophy, sensory loss **-Axonal sensorimotor polyneuropathy**	- Pes cavus, Paraparesis, Distal limb muscle weakness and atrophy due to peripheral neuropathy - Sensorineural hearing loss (uncommon), Reduced motor nerve conduction velocity (NCV) (range less than 38 m/s to normal)and Axonal degeneration
4	GJB1 (304040)	Gap junction beta-1 protein	M	25 (15)	XLR	c.472C>T (p.Pro158Ser)	**Charcot-Marie-Tooth neuropathy, X-linked dominant, 1**	PMID:9888385	-Muscular weakness and distal muscle atrophy,pes cavus **-no EMG**	- Pes cavus, Paraparesis, Distal limb muscle weakness and atrophy due to peripheral neuropathy - Sensorineural hearing loss (uncommon), Reduced motor nerve conduction velocity (NCV) (range less than 38 m/s to normal)and Axonal degeneration
6	SH3TC2	SH3	M	32	AR	c.233_239delT	**-Charcot-Marie-**	NA	-Muscular weakness and distal	Abnormal pupillary light reflexes ,

	(608206)	domain and tetratrico peptide repeats-containin g protein 2		(15)		CTGGGC (p.Leu78Hisfs* 49)	**Tooth disease, type 4C, AR**/ -Mononeuropathy of the median nerve, mild , AD		muscle atrophy,pes cavus, arefelxia,sensory loss, scoliosis **-Demyelinating sensorimotor polyneuropathy with Secondary axonal degeneration**	Scoliosis, early-onset, severe, Onset usually in first or second decades, Distal lower limb muscle weakness and atrophy due to peripheral neuropathy, Distal upper limb involvement may occur later, Distal sensory impairment of touch, vibration, proprioception Secondary axonal degeneration and regeneration, Deafness and Decreased motor nerve conduction velocity (NCV) (less than 38 m/s)
7	SH3TC2 (608206)	SH3 domain and tetratrico peptide repeats-containin g protein 2	M	48 (16)	AR	c.1972C>T (p.Arg658Cys)	**-Charcot-Marie-Tooth disease, type 4C, AR**/ -Mononeuropathy of the median nerve, mild, AD	PMID: 21291453, 19744956, 16924012, 14574644, 20301514 , 14574644	-Muscular weakness and distal muscle atrophy, pes cavus, sensory loss hearing loss **-Demyelinating sensorimotor polyneuropathy**	Abnormal pupillary light reflexes , Scoliosis, early-onset, severe, Onset usually in first or second decades, Distal lower limb muscle weakness and atrophy due to peripheral neuropathy, Distal upper limb involvement may occur later, Distal sensory impairment of touch, vibration, proprioception Secondary axonal degeneration and regeneration, Deafness and Decreased motor nerve conduction velocity (NCV) (less than 38 m/s)
8	SH3TC2 (608206)	SH3 domain and tetratrico peptide repeats-containin g protein 2	F	40 (31)	AR	c.1366delC (p.Leu456*)	Charcot-Marie-Tooth disease, type 4C, AR/ - Mononeuropathy of the median nerve, mild, AD	NA	-Muscular weakness and distal muscle atrophy,pes cavus, sensory loss **-Demyelinating sensorimotor polyneuropathy**	Abnormal pupillary light reflexes , Scoliosis, early-onset, severe , Onset usually in first or second decades, Distal lower limb muscle weakness and atrophy due to peripheral neuropathy, Distal upper limb involvement may occur later, Distal sensory impairment of touch, vibration, proprioception Secondary axonal degeneration and regeneration, Deafness and Decreased motor nerve conduction velocity (NCV) (less than 38 m/s)
10	GDAP1 (606598)	Ganglios ide-induced differenti ation-associate	F	20 (6)	spora dic	c.458C>T (p.Pro153Leu)	**-Charcot-Marie-Tooth disease, axonal, type 2K , AD, AR**/ -Charcot-Marie-Tooth disease,	PMID: 18421898, 18504680, 28751717	-Muscular weakness and distal muscle atrophy,pes cavus,lordosis, tremor **-Axonal sensorimotor polyneuropathy**	Early onset, Kyphoscoliosis, Claw hand deformities, Talipes equinovarus, Distal limb muscle weakness and atrophy due to peripheral neuropathy , Normal or mildly reduced motor nerve

		d protein 1					axonal, with vocal cord paresis, AR/ -Charcot-Marie-Tooth disease, recessive intermediate, A, AR/ - Charcot-Marie-Tooth disease, type 4A, AR			conduction velocities (NCV) (greater than 38 m/s) and Axonal regeneration on nerve biopsy
13	NEFL (162280)	Neurofilament Light	M	15 (13)	AD	c.803T>C (p.Leu268Pro)	-Charcot-Marie-Tooth disease, dominant intermediate G, AD/ **-Charcot-Marie-Tooth disease, type 1F, AD, AR/** -Charcot-Marie-Tooth disease, type 2E, AD	NA	-Muscular weakness and distal muscle atrophy,pes cavus **-Demyelinating sensorimotor polyneuropathy**	Pes cavus, Distal lower limb muscle weakness and atrophy due to peripheral neuropathy , Upper limb involvement usually occurs later , Onset in infancy or childhood Decreased motor nerve conduction velocity (NCV) (less than 38 m/s)
14	MFN2 (608507)	Mitofusin-2	F	19 (7)	AR	c.839G>A (p.Arg280His)	**-Charcot-Marie-Tooth disease, axonal, type 2A2A, AD** -Charcot-Marie-Tooth disease, axonal, type 2A2B , AR -Hereditary motor and sensory neuropathy VIA , AD	PMID:15064763, 16714318,16835246	-Muscular weakness and distal muscle atrophy,pes cavus,sensory loss **-Axonal sensorimotor polyneuropathy**	Distal muscle weakness and atrophy due to peripheral neuropathy, Distal sensory impairment, Normal or mildly decreased motor nerve conduction velocity (NCV) (greater than 38 m/s), Axonal degeneration/ regeneration on nerve biopsy, Pes cavus, Hammer toes and Foot deformities are **CMT2A2A** manifestations**.**
16	PRX (605725)	Periaxin	M	40 (15)	AR	c.1090C>T (p.Arg364*)	**-Charcot-Marie-Tooth disease, type 4F**, AR -Dejerine-Sottas disease, AD, AR	PMID:21741241, 27391121	-Muscular weakness and distal muscle atrophy,pes cavus -**Demyelinating sensorimotor polyneuropathy**	Scoliosis , - Pes cavus, Distal limb muscle weakness and atrophy due to peripheral neuropathy and Decreased motor nerve conduction velocity (NCV) (less than 38 m/s)
17	PRX (605725)	Periaxin	M	35 (20)	AR	c.1090C>T (p.Arg364*)	**-Charcot-Marie-Tooth disease, type 4F, AR/** -Dejerine-Sottas disease, AR, AD	PMID:21741241, 27391121	-Muscular weakness and distal muscle atrophy,pes cavus,scoliosis, hearing loss,areflexia, sensory loss -**Demyelinating sensorimotor polyneuropathy**	Scoliosis, - Pes cavus, Distal limb muscle weakness and atrophy due to peripheral neuropathy, Distal upper limb involvement, Distal sensory impairment, Areflexia , and Decreased motor nerve conduction velocity (NCV) (less than 38 m/s)

20	MME (120520)	Membrane metallo endopeptidase	M	27 (NA)	sporadic	c.1342C>T (p.Arg448*)	-Spinocerebellar ataxia 43, AD / -Charcot-Marie-Tooth disease, axonal, type 2T, AD, AR	PMID: 25565308, 15464186, 30415211	-Muscular weakness and distal muscle atrophy **-Axonal sensorimotor polyneuropathy**	Distal limb muscle weakness and atrophy due to peripheral neuropathy, Axonal sensorimotor neuropathy, Distal sensory impairment, Foot drop, Gait instability, Adult onset
21	MME (120520)	Membrane metallo endopeptidase	F	47 (NA)	AR	c.1861T>C (p.Cys621Arg)	-Spinocerebellar ataxia 43, AD/ **-Charcot-Marie-Tooth disease, axonal, type 2T, AD, AR**	PMID: 26991897	-Muscular weakness and distal muscle atrophy,pes cavus, tremor, scoliosis **-Axonal sensorimotor polyneuropathy**	Distal limb muscle weakness and atrophy due to peripheral neuropathy, Axonal sensorimotor neuropathy, Distal sensory impairment, Foot drop, Gait instability, Adult onset
28	IGHMBP2 (600502)	DNA-binding protein SMUBP-2, or immunoglobulin helicase μ-binding protein 2	F	8 (4m)	sporadic	c.449+1G>T	**-Charcot-Marie-Tooth disease, axonal, type 2S, AR**/ -Neuronopathy, distal hereditary motor, type VI ,AR	PMID:23566544, 25568292 , 27450922	-Muscular weakness and distal muscle atrophy,pes cavus, lordosis, hypotonia, hyporeflexia **-Demyelinating sensorimotor polyneuropathy**	- Onset in first decade, Distal muscle weakness and atrophy due to peripheral neuropathy, Distal motor impairment, Impaired gait, Steppage gait, Reduction in large myelinated fibers seen on sural nerve biopsy and axonal sensorimotor neuropathy affecting upper and lower limbs are **CMT2S** manifestations
29	IGHMBP2 (600502)	DNA-binding protein SMUBP-2, or immunoglobulin helicase μ-binding protein 2	F	47 (NA)	AR	c.449+1G>T	**-Charcot-Marie-Tooth disease, axonal, type 2S, AR**/ -Neuronopathy, distal hereditary motor, type VI ,AR	PMID:23566544	-Muscular weakness and distal muscle atrophy,pes cavus **- Axonal sensorimotor polyneuropathy**	- Onset in first decade, Distal muscle weakness and atrophy due to peripheral neuropathy, Distal motor impairment, Impaired gait, Steppage gait, Reduction in large myelinated fibers seen on sural nerve biopsy and axonal sensorimotor neuropathy affecting upper and lower limbs are **CMT2S** manifestations
35	BAG3 (603883)	BAG family molecular chaperone regulator 3	F	48 (NA)	AD	c.625C>T (p.Pro209Ser)	-Myopathy, myofibrillar, 6, AD/ Cardiomyopathy, dilated, 1HH, AD **-Charcot-Marie-Tooth disease, axonal, related to BAG3**	PMID:28754666	-Muscular weakness and distal muscle atrophy,pes cavus **-Axonal sensorimotor polyneuropathy**	- Muscle atrophy and weakness, Pes cavus and Axonal and demyelinating peripheral neuropathy is one of **myofibrillar myopathy 6** neurologic manifestations -Axonal sensorimotor polyneuropathy in main characteristic of **CMT2**
Likely pathogenic										
5	SH3TC2	SH3	M	34 (4)	AR	c.1712T>C	**Charcot-Marie-**	NA	-Muscular weakness and distal	Abnormal pupillary light reflexes ,

	(608206)	domain and tetratricopeptide repeats-containing protein 2				(p.Leu571Pro)	**Tooth disease, type 4C, AR/** - Mononeuropathy of the median nerve, mild , AD		muscle atrophy,pes cavus,facial weakness, hearing loss, scoliosis **-Demyelinating sensorimotor polyneuropathy**	Scoliosis, Facial weakness, early-onset, severe , Onset usually in first or second decades, Distal lower limb muscle weakness and atrophy due to peripheral neuropathy, Distal upper limb involvement may occur later, Distal sensory impairment of touch, vibration, proprioception Secondary axonal degeneration and regeneration, Deafness and Decreased motor nerve conduction velocity (NCV) (less than 38 m/s)
12	NEFL (162280)	Neurofilament Light	F	35 (6)	sporadic	c.837G>A (p.Trp279*)	-Charcot-Marie-Tooth disease, dominant intermediate G, AD/ **-Charcot-Marie-Tooth disease, type 1F , AD, AR/** -Charcot-Marie-Tooth disease, type 2E , AD	NA	-Muscular weakness and distal muscle atrophy,pes cavus, scoliosis **-Demyelinating sensorimotor polyneuropathy**	Pes cavus, Distal lower limb muscle weakness and atrophy due to peripheral neuropathy , Upper limb involvement usually occurs later , Onset in infancy or childhood Decreased motor nerve conduction velocity (NCV) (less than 38 m/s)
15	MFN2 (608507)	Mitofusin-2	F	25(NA)	AR	c.334G>A (p.Val112Met)	**-Charcot-Marie-Tooth disease, axonal, type 2A2A, AD** -Charcot-Marie-Tooth disease, axonal, type 2A2B , AR -Hereditary motor and sensory neuropathy VIA , AD	NA	-Muscular weakness and distal muscle atrophy,pes cavus - **Axonal sensorimotor polyneuropathy**	Distal muscle weakness and atrophy due to peripheral neuropathy, Distal sensory impairment, Normal or mildly decreased motor nerve conduction velocity (NCV) (greater than 38 m/s), Axonal degeneration/regeneration on nerve biopsy, Pes cavus, Hammer toes and Foot deformities are **CMT2A2A** manifestations**.**
18	MTMR2 (603557)	Myotubularin-related protein 2	M	29(7)	AR	c.1810C>T (p.Arg604*)	Charcot-Marie-Tooth disease, type 4B1, AR	NA	-Muscular weakness and distal muscle atrophy **-Demyelinating sensorimotor polyneuropathy**	Scoliosis may be present , Talipes equinovarus, Foot deformities , Delayed motor development , Distal limb muscle weakness and atrophy due to peripheral neuropathy , Proximal limb muscle weakness , Facial weakness , Distal sensory impairment , Severely decreased motor nerve conduction velocity (NCV) (15 m/s) and Abnormal auditory evoked potentials

19	MTMR2 (603557)	Myotubularin-related protein 2	F	7 (1.5)	AR	c.1098_1099insC (p.Lys367Glnfs*9)	**Charcot-Marie-Tooth disease, type 4B1, AR**	NA	-Muscular weakness and distal muscle atrophy,pes cavus **-Demyelinating sensorimotor polyneuropathy**	Scoliosis may be present , Talipes equinovarus, Foot deformities , Delayed motor development , Distal limb muscle weakness and atrophy due to peripheral neuropathy , Proximal limb muscle weakness , Facial weakness , Distal sensory impairment , Severely decreased motor nerve conduction velocity (NCV) (15 m/s) and Abnormal auditory evoked potentials
22	ARHGEF 10 (608136)	Rho guanine nucleotide exchange factor 10	F	67 (63)	AR	c.415G>T (p.Glu139*)	-Slowed nerve conduction velocity, AD, **-CMT****	PMID:25091364	-Muscular weakness and distal muscle atrophy,pes cavus, hearing loss **-Axonal sensorimotor polyneuropathy**	- Slowed nerve conduction velocities (NCV), Nerve biopsy may show demyelination and remyelination, Adult onset and Usually clinically asymptomatic in Slowed nerve conduction velocity, AD **-ARHGEF10-associated CMT patient reported with axonal polyneuropathy**
25	DNM2 (602378)	Dynamin-2	F	22 (2)	sporadic	c.1022A>G (p.Glu341Gly)	Centronuclear myopathy 1 ,AD/ -Charcot-Marie-Tooth disease, axonal type 2M ,AD/ **-Charcot-Marie-Tooth disease, dominant intermediate B , AD/** - Lethal congenital contracture syndrome 5, AR	NA	-Muscular weakness and distal muscle atrophy,sensory loss **-Demyelinating sensorimotor polyneuropathy with Secondary axonal degeneration**	Onset in first or second decade , Distal limb muscle weakness due to peripheral neuropathy , Distal limb muscle atrophy due to peripheral neuropathy , Hyporeflexia , Areflexia , Distal sensory impairment, Low to normal range of motor nerve conduction velocity (NCV) (25-54 m/s) ('intermediate' CMT, CMTDIB), Individuals with normal NCV values have axonal CMT (CMT2M) Axonal degeneration and loss of myelinated fibers on nerve biopsy
30	PMP22 (601097)	Peripheral Myelin Protein 22	M	7 (2)	AD	c.53T>G (p.Lue18Arg)	**Charcot-Marie-Tooth disease, type 1E, AD /** ?Neuropathy, inflammatory demyelinating, ?AD/ Charcot-Marie-Tooth disease, type 1A, AD / **Dejerine-Sottas disease, AD, AR/**	PMID:23313019	-Muscular weakness and distal muscle atrophy,pes cavus,scoliosis **-no EMG**	Kyphoscoliosis may be present , Claw hand deformities , Pes calcaneovarus , Pes cavus , Hammertoes , Foot deformities, Distal limb muscle weakness due to peripheral neuropathy, Decreased motor nerve conduction velocity (NCV), Childhood onset, Usually begins in feet and legs (peroneal distribution) and Upper limb

							Neuropathy, recurrent, with pressure palsies, AD / Roussy-Levy syndrome, AD **(overlapping phenotypes)**			involvement usually occurs later
31	ATP1A1 (182310)	Sodium/ Potassium-Transporting ATPase Subunit Alpha-1	M	40 (NA)	AR	c.1645G>A (p.Gly549Arg)	**-Charcot-Marie-Tooth disease, axonal, type 2DD, AD**/ -Hypomagnesemia, seizures, and mental retardation 2, AD	NA	-Muscular weakness and distal muscle atrophy,pes cavus, sensory loss **-Axonal sensorimotor polyneuropathy**	Pes cavus, Distal limb muscle weakness and atrophy due to peripheral neuropathy, Muscle cramps, Sensorimotor peripheral neuropathy, Steppage gait, Foot drop, Decreased vibratory sensation, distal and Normal nerve conduction velocities (NCV)
VUS (Variant of Uncertain Significance)										
9	GDAP1 (606598)	Ganglioside-induced differentiation-associated protein 1	F	6 (2)	sporadic	c.802_803delTG (p.Trp268Glyfs*22)	-Charcot-Marie-Tooth disease, axonal, type 2K , AD, AR/ -Charcot-Marie-Tooth disease, axonal, with vocal cord paresis , AR/ - Charcot-Marie-Tooth disease, recessive intermediate, A , AR/ -Charcot-Marie-Tooth disease, type 4A, AR	NA	-Muscular weakness and distal muscle atrophy,pes cavus,areflexia **-Demyelinating sensorimotor polyneuropathy**	- Axonal regeneration on nerve biopsy / - Decreased motor nerve conduction velocity (NCV) (less than 38 m/s) - Axonal features may coexist (normal NCV, axonal degeneration on biopsy)
11	GDAP1 (606598)	Ganglioside-induced differentiation-associated protein 1	F	21 (19)	sporadic	c.602A>G (p.Asn201Ser)	-Charcot-Marie-Tooth disease, axonal, type 2K , AD, AR/ -Charcot-Marie-Tooth disease, axonal, with vocal cord paresis , AR/ **- Charcot-Marie-Tooth disease, recessive intermediate, A,**	NA	-Muscular weakness and distal muscle atrophy,pes cavus,kyphoscoliosis ,sensory impairment **-Demyelinating sensorimotor polyneuropathy**	Early onset, Kyphoscoliosis, Claw hand deformities , Talipes equinovarus, Distal limb muscle weakness and atrophy due to peripheral neuropathy , Normal or mildly reduced motor nerve conduction velocities (NCV) (greater than 38 m/s) and Axonal regeneration on nerve biopsy

							AR/ -Charcot-Marie-Tooth disease, type 4A, AR			
23	ARHGEF10 (608136)	Rho guanine nucleotide exchange factor 10	F	27 (14)	AR	c.1315C>G (p.Leu439Val)	?Slowed nerve conduction velocity, AD, **-CMT****	PMID:26392352 , 25091364	-Muscular weakness and distal muscle atrophy,pes cavus , blurred vision,dizziness **-Axonal sensorimotor polyneuropathy**	- Slowed nerve conduction velocities (NCV), Nerve biopsy may show demyelination and remyelination, Adult onset and Usually clinically asymptomatic in Slowed nerve conduction velocity, AD **-ARHGEF10-associated CMT patient reported with axonal polyneuropathy**
24	DNM2 (602378)	Dynamin-2	M	28(21)	sporadic	c.869G>A (p.Arg290Gln)	Centronuclear myopathy 1 ,AD/ **-Charcot-Marie-Tooth disease, axonal type 2M, AD/** -Charcot-Marie-Tooth disease, dominant intermediate B, AD/ - Lethal congenital contracture syndrome 5 , AR	NA	-Muscular weakness and distal muscle atrophy,pes cavus **-Axonal sensorimotor polyneuropathy**	Onset in first or second decade , Distal limb muscle weakness due to peripheral neuropathy , Distal limb muscle atrophy due to peripheral neuropathy , Hyporeflexia , Areflexia , Distal sensory impairment, Low to normal range of motor nerve conduction velocity (NCV) (25-54 m/s) ('intermediate' CMT, CMTDIB), Individuals with normal NCV values have axonal CMT (CMT2M) Axonal degeneration and loss of myelinated fibers on nerve biopsy
26	NDRG1 (605262)	N-Myc Downstream Regulated	F	29 (3)	AR	c.771G>C (p.Leu257Phe)	**Charcot-Marie-Tooth disease, type 4D, AR**	NA	-Muscular weakness and distal muscle atrophy,areflexia ,dysphagia,dysarthria,sensory loss **-Demyelinating sensorimotor polyneuropathy**	- Deafness (often in third decade), Hand deformities , Foot deformities, Talipes cavus equinovarus , Distal limb muscle weakness due to peripheral neuropathy , Distal limb muscle atrophy due to peripheral neuropathy , Gait disorder , Hyporeflexia , Areflexia , Distal sensory loss , Severely reduced nerve conduction velocities (NCV) (may become unattainable) , Segmental demyelination/remyelination on nerve biopsy and Axonal loss
27	NDRG1 (605262)	N-Myc Downstream	M	16 (5)	AR	c.287C>T (p.Ala96Val)	**Charcot-Marie-Tooth disease, type 4D, AR**	NA	-Muscular weakness and distal muscle atrophy,pes cavus,tremor **-no EMG**	- Deafness (often in third decade), Hand deformities, Foot deformities, Talipes cavus equinovarus , Distal

		Regulated								limb muscle weakness due to peripheral neuropathy , Distal limb muscle atrophy due to peripheral neuropathy , Gait disorder , Hyporeflexia , Areflexia , Distal sensory loss , Severely reduced nerve conduction velocities (NCV) (may become unattainable) , Segmental demyelination/remyelination on nerve biopsy and Axonal loss
32	FGD4 (611104)	FYVE, RhoGEF and PH domain-containing protein 4	M	4 (2)	AR	c.1525_1527delAAA (p.Lys509del)	**Charcot-Marie-Tooth disease, type 4H, AR**	NA	-Muscular weakness and distal muscle atrophy, pes cavus **-Demyelinating sensorimotor polyneuropathy**	Scoliosis, Pes cavus, Pes equinus, Distal lower limb muscle weakness and atrophy due to peripheral neuropathy, Upper limb involvement may occur later, 'Waddling' gait Decreased motor nerve conduction velocity (NCV) (less than 38 m/s) and Nerve biopsy shows demyelination/remyelination
34	LMNA (613205)	Lamin A/C	M	43 (NA)	sporadic	c.1696C>G (p.His566Asp)	-Emery-Dreifuss muscular dystrophy 2, autosomal dominant -Malouf syndrome, -Muscular dystrophy, congenital, -Lipodystrophy, familial partial, type 2, **-Charcot-Marie-Tooth disease, type 2B1 (AR)**	NA	-Muscular weakness and distal muscle atrophy,pes cavus, sensory loss, mild cerebellar atroph **-Demyelinating sensorimotor polyneuropathy with Secondary axonal degeneration**	Kyphoscoliosis may be present, Pes cavus , Foot deformities, Distal limb muscle weakness and atrophy due to peripheral neuropathy , Proximal muscle involvement may occur , 'Steppage' gait , Foot drop , Distal sensory impairment , Hyporeflexia , Areflexia , Normal or mildly decreased motor nerve conduction velocity (NCV) (greater than 38 m/s**), Axonal degeneration/regeneration and atrophy on nerve biopsy**
38	KIF5A (602821)	Kinesin heavy chain isoform 5A	M	30 (2)	sporadic	c.2005G>A (p.Ala669Thr)	Myoclonus, intractable, neonatal , AD/ -**Spastic paraplegia 10, autosomal dominant, AD/** -{Amyotrophic lateral sclerosis, susceptibility to, 25} , AD, **-CMT2****	PMID:25008398 , 29892902	-Muscular weakness and distal muscle atrophy, spastic gait, seizure, pes cavus,facial weakness,sensory loss **-Demyelinating sensorimotor polyneuropathy**	-Scoliosis, Pes cavus , Lower limb spasticity , Lower limb weakness , Spastic gait, Upper limb weakness may occur later, Cerebellar ataxia (rare), Axonal sensorimotor peripheral neuropathy, Decreased vibratory sense in the lower limbs, Distal sensory impairment, Upper limb sensory loss may occur later in **SPG10**. **-CMT2AD** reported recently.

39	FBLN5 (604580)	Fibulin-5	M	20 (NA)	AR	c.889C>G (p.His297Asp)	-Cutis laxa, autosomal dominant 2 / -Cutis laxa, autosomal recessive, type IA , AR/ **- Macular degeneration, age-related, 3 ,AD/ -Neuropathy, hereditary, with or without age-related macular degeneration ,AD, - CMT1****	PMID:28332470 , 23328402	-Muscular weakness and distal muscle atrophy,sensory loss- **Axonal sensorimotor polyneuropathy**	Distal muscle weakness and atrophy due to peripheral neuropathy, lower limbs more affected than upper limbs , Peripheral neuropathy, demyelinating , Distal sensory impairment (in most patients) , Decreased nerve conduction velocities (in most patients) , Axonal peripheral neuropathy (in some patients)
**Genes that recently were reported in CMT or dMND. EMG/NCV reports and presumable disease for each variant specified with bold words. MIM, Mendelian Inheritance in Man; M, Male; F, Female; XLD, X-linked dominant; XLR, X-linked recessive; AR, Autosomal recessive ; AD, Autosomal dominant; PMID, PubMed IDentifier; EMG, electromyography; NA, Not Available; CMT, Charcot-Marie-Tooth disease.										

Table S2B. Information of identified variants in non-CMT and related IPN genes in this study.

Family Number	Gene Symbol	Gene	Gender	age	inheritance	Variant Description	Gene Related Disorders	Reference for relation of variant with CMT or other Neuropathies	Observed phenotype and EMG/NCV	Published or expected phenotype
Likely pathogenic										
42	HADHA (600890)	Trifunctional enzyme subunit alpha,	F	27 (NA)	AR	c.539C>T (p.Pro180Leu)	**Trifunctional protein deficiency , AR, Fatty liver, acute, of pregnancy, AR/** -HELLP syndrome,	NA	-Distal Muscular weakness -**Axonal sensorimotor polyneuropathy**	Hypotonia , Generalized weakness, Limb-girdle myopathy, slowly progressive , Muscle pain , Rhabdomyolysis, episodic, Poor spontaneous movements , Delayed psychomotor

		mitochondrial					maternal, of pregnancy , AR/ LCHAD deficiency ,AR/			development , Sensorimotor axonopathy
VUS (Variant of Uncertain Significance)										
33	KCNJ10 (602208)	ATP-sensitive inward rectifier potassium channel 10	M	10 (NA)	AR	c.967T>C (p.Tyr323His)	Enlarged vestibular aqueduct, digenic , AR/ **-SESAME syndrome, AR**	NA	-Muscular eakness ,seizure, dysarthria, dysdiadochokinesis, epilepsy,physical disability **-Sensory polyneuropathy and meunoneuritis multiplex**	- Hearing loss, sensorineural, Seizures , Psychomotor delay, Mental retardation , Ataxia, Poor speech development , Hypotonia, Intention tremor, Dysdiadochokinesis , Cerebellar atrophy, Axonal neuropathy (rare) and Hypomyelination of sural nerve (rare) in SESAME syndrome
36	REEP1 (609139)	Receptor expression-enhancing protein	M	55 (45)	AR	c.601G>A (p.Ala201Thr)	Neuronopathy, distal hereditary motor, type VB , AD/ **-Spastic paraplegia 31, autosomal dominant, AD**	NA	-Muscular weakness and distal muscle atrophy,sensory loss **-Axonal sensorimotor polyneuropathy**	-Lower limb weakness and spasticity, Upper limbs may be affected , Spastic gait , Hyperreflexia, Proximal weakness of the lower extremities , Dysarthria , Extensor plantar responses , Ankle clonus, Distal sensory loss, Bimodal age of onset: Most have onset in first or second decade and a minority of patients have onset after age 30 years in **SPG39**. -Decreased motor nerve conduction velocities, Areflexia and Hyporeflexia in **NHMV**
37	MYH14 (608568)	Myosin heavy chain-14	F	9(2)	AR	c.3667G>C (p.Gly1223Arg)	**Peripheral neuropathy, myopathy, hoarseness, and hearing loss, AD**/ - Deafness, autosomal dominant 4A, AD	NA	-Muscular weakness and distal muscle atrophy,pes cavus **-Axonal sensorimotor polyneuropathy**	Early onset, Hearing loss and hoarseness occur later , Foot deformities, Distal muscle weakness (first affects anterior leg muscles, then posterior leg muscles), Distal muscle atrophy (lower limbs more affected than upper limbs), Proximal weakness of the lower limbs with longer disease duration ,Areflexia , Hyporeflexia and Nerve conduction studies show mildly

										reduced or normal sensory values
40	ATL3 (609369)	Atlastin GTPase 3	M	33 (13)	AD	c.1217G>A (p.Gly406Glu)	**Neuropathy, hereditary sensory, type IF, AD**	NA	-Muscular weakness and distal muscle atrophy,pes cavus, lordosis **-no EMG**	Onset in first or second decade, Hallux valgus , Foot arthropathy, Chronic ulceration due to sensory neuropathy, Distal sensory impairment to pain, temperature, and touch, lower limbs, Sensory axonal neuropathy and Hyporeflexia of the lower limbs
41	SYT2 (600104)	Synaptotagmin-2	M	63 (NA)	AR	c.322A>G (p.Met108Val)	**-Myasthenic syndrome, congenital, 7, presynaptic , AD, -dMND***	PMID:30533528	-Muscular weakness and distal muscle atrophy,pes cavus, hearing loss, sensory loss,seizures **-Axonal sensorimotor polyneuropathy**	- Hearing loss, - Distal muscle weakness (lower limbs more severely affected than upper limbs) - Proximal muscle weakness (in some patients) , Easy fatigability with exercise , Gait abnormalities, Impaired toe-walking, Impaired heel-walking , Muscle atrophy (in some patients) , Motor neuropathy (1 family) , Onset in early childhood in Myasthenic syndrome, congenital, 7, presynaptic , AD **Unusual dMN reported recently**
43	SLC12A6 (604878)	Solute carrier family 12 member 6	F	12 (9M)	sporadic	c.2075T>G (p.Leu692Arg)	**Agenesis of the corpus callosum with peripheral neuropathy, AR**	NA	-Developmental delay, scoliosis, generalized hypotonia,areflexia **-Demyelinating sensorimotor polyneuropathy**	Onset within the first year of life, Delayed motor milestones, Developmental delay, Hypotonia, generalized , Mental retardation, mild to severe, Seizures, Agenesis of the corpus callosum, Axonal swelling of spinal nerve roots and cranial nerves, Peripheral motor and sensory neuropathy, Areflexia , Limb tremor, Sural nerve biopsy shows absence of large myelinated fibers , Axonal neuropathy , Axonal degeneration/regeneration, Decreased motor and sensory nerve conduction velocities and Demyelinating neuropathy
44	TECPR2 (615000)	Tectonin beta-	F	14 (4	AR	c.818G>A (p.Arg273His)	**Spastic paraplegia 49, autosomal**	NA	-Spastic gait , Foot deformity, Distal limb muscle weakness,claw	Delayed psychomotor development, Hypotonia, Ataxic

		propeller repeat-containing protein 2		)			**recessive**		hand, hammer toes -**Demyelinating sensorimotor polyneuropathy**	gait, Spastic gait , Rigid gait , Dysarthria , Dysmetria , Seizures (in some patients), Thin corpus callosum , Cerebral atrophy and Areflexia
45	SPTBN2 (604985)	Spectrin beta chain, brain 2	F	7 (2)	AR	c.5314G>A (p.Val1772Met)	-Spinocerebellar ataxia 5, AD / -**Spinocerebellar ataxia, autosomal recessive 14**	NA	-Distal lower limb muscle weakness,distal lower limb muscle atrophy,ataxic gait, Scoliosis,Distal sensory impairment -**Axonal sensorimotor polyneuropathy**	- Delayed psychomotor development, Gait ataxia, Cognitive impairment, mild to moderate, Speech delay, Dysmetria , Dysdiadochokinesis, Intention tremor , Spasticity (in some patients) , Hyperreflexia (in some patients) , Cerebellar atrophy in **SCA14**. -Decreased vibration sense in **SCA5**
46	SYNE1 (608441)	Spectrin Repeat Containing Nuclear Envelope Protein 1	F	21 (NA)	AR	c.25157C>A (p.Ser8386Tyr)	**-Emery-Dreifuss muscular dystrophy 4, autosomal dominant** / - Spinocerebellar ataxia, autosomal recessive 8	NA	-Muscular weakness and distal muscle atrophy,pes cavus, sensory loss,cleft palate **-no EMG**	-Muscle weakness and atrophy, proximal, Muscle biopsy shows dystrophic pattern and No neuropathic event in **Emery-Dreifuss.** - Cerebellar ataxia, Gait ataxia, Limb ataxia and Cerebellar atrophy in **SCA8**
47	MYH8 (160741)	Myosin heavy chains 8	F	8 (7)	AR	c.4688G>A (p.Arg1563His)	-Carney complex variant / -**Trismus-pseudocamptodactyly syndrome, AD**	NA	-Distal lower limb muscle weakness, Frequent fallings, Exercise-induced fatigue, Pes cavus, distal upper limb weakness, sensory impairment -**Demyelinating sensorimotor polyneuropathy**	Short stature (3rd-25th percentile) and head and neck anomalies, Shortening of flexor profundus muscle-tendon unit , Shortening of various muscle-tendon groups in legs , Shortening of various muscle-tendon groups in feet and No neuropathic event

**Genes that recently were reported in CMT or dMND. EMG/NCV reports and presumable disease for each variant specified with bold words.
M, Male; F, Female; AR, Autosomal recessive; AD, Autosomal dominant; PMID, PubMed IDentifier; EMG, electromyography; NA, Not Available; CMT, Charcot-Marie-Tooth disease; NCV, nerve conduction velocity; SESAME, Seizures, Sensorineural deafness, Ataxia, Mental retardation, and Electrolyte imbalance.

Table S2C. Information of identified variants in novel candidate genes in this study.

Family Number	Gene Symbol	Gene	Gender	age	inheritance	Variant Description	Gene Related Disorders	Reference for relation of variant with CMT or other Neuropathies	Observed phenotype and EMG/NCV	Published or expected phenotype
VUS (Variant of Uncertain Significance)										
49	TES (606085)	Testin LIM Domain Protein	M	17 (13)	AR	c.1161G>C (p.Glu387Asp)	NA	NA	-Distal lower limb muscle weakness,Pes cavus, Claw hand , Foot drop, slow eye pursuit ,toe walking,distal sensory impairment,exercise fatigue induced **-Demyelinating sensorimotor polyneuropathy**	No neuropathic event
50	YLPM1 (NA)	YLP Motif Containing 1	M	31 (20)	sporadic	c.5194C>T (p.Arg1732*)	NA	NA	-Muscular weakness and distal muscle atrophy,pes cavus,hoarseness **-no EMG**	No omim entry
51	ARHGAP6 (300118)	Rho GTPase Activating Protein 6	M	31 (16)	AR	c.2236_2237delTC (p.Ser746Glnfs*14)	NA	NA	-Muscular weakness and distal muscle atrophy,pes cavus **-Demyelinating sensorimotor polyneuropathy**	no dissease in OMIM
52	SYT8 (607719)	Synaptotagmin 8	F	53 (48)	AR	c.1099C>T (p.Arg367Trp)	NA	NA	**-Axonal sensorimotor polyneuropathy**	no dissease in OMIM
53	MCAM (155735)	Melanoma Cell Adhesion Molecule	M	54 (NA)	AR	c.739+2_739+3delTG	NA	NA	-Muscular weakness and distal muscle atrophy,pes cavus, sensory loss, ataxia **-Demyelinating sensorimotor polyneuropathy**	no dissease in OMIM

*Observed variants in patients with more than one causative gene. **Genes that recently were reported in CMT or dMND. EMG/NCV reports and presumable disease for each variant specified with bold words.
M, Male; F, Female; AR, Autosomal recessive; EMG, electromyography; NA, Not Available; OMIM, Online Mendelian Inheritance in Man; NCV, nerve conduction velocity.

Table S2D. Information of identified variants in unsolved cases in this study.

Family Number	Gene Symbol	Gene	Gender	age	inheritance	Variant Description	Gene Related Disorders	Reference for relation of variant with CMT or other Neuropathies	Observed phenotype and EMG/NCV	Published or expected phenotype
VUS (Variant of Uncertain Significance)										
48	HSPG2* (142461)	Heparan Sulfate Proteoglycan 2	M	46 (8)	AR	c.12329G>A (p.Arg4110His)	-Dyssegmental dysplasia, Silverman-Handmaker type, AR / -**Schwartz-Jampel syndrome, type 1, AR**	NA	-Muscle weakness, Impaired distal sensation -**Demyelinating sensorimotor polyneuropathy with Secondary axonal degeneration**	Myotonia, Muscular hypertrophy, Muscle weakness, Muscle wasting, EMG - repetitive muscle discharges, Hyporeflexia and Mental retardation (25%)
	Dock6* (614194)	Dedicator of cytokinesis 6	M	46 (8)	AR	c.4732C>T (p.Lue1578Phe)	Adams-Oliver syndrome 2, AR	PMID:25824905	-Muscle weakness, Impaired distal sensation -**Demyelinating sensorimotor polyneuropathy with Secondary axonal degeneration**	Psychomotor retardation , Seizures – Hypotonia, Calcifications of cerebral ventricles and Dilation of cerebral ventricles
54	NBPF10* (614000)	Neuroblastoma Breakpoint Family Member 10	F	55 (5)	sporadic	c.1367C>A (p.Ser456*)	NA	NA	-Muscular weakness and distal muscle atrophy, pes cavus, tremor **-no EMG**	no disease in OMIM
	MYH15* (609929)	Myosin heavy chains 15	F	55 (5)	sporadic	c.4066C>T (p.Arg1356*)	NA	NA	-Muscular weakness and distal muscle atrophy,pes cavus, tremor **-no EMG**	no disease in OMIM

*Observed variants in patients with more than one causative gene. **Genes that recently were reported in CMT or dMND. EMG/NCV reports and presumable disease for each variant specified with bold words.
M, Male; F, Female; AR, Autosomal recessive; EMG, electromyography; NA, Not Available; OMIM, Online Mendelian Inheritance in Man.

Table S3. Different classifications of whole detected variants in 58 investigated families underwent whole exome sequencing.

Solved cases (89.7%) 52/58											**Unsolved cases (10.3%) 6/58**		
CMT and related IPN						Non-CMT and related IPN neuropathies				Novel candidate	With 2 candidate		Negative result
37						10				5	4 variants (2 patients)		4 patients
Known variants			Novel variants			Known variants	Novel variants			Novel variants	Known variants	Novel variants	0
17			20			1	9			5	1	3	0
pathogenic	Likely pathogenic	VUS	pathogenic	Likely pathogenic	VUS	VUS	pathogenic	Likely pathogenic	VUS	VUS	VUS	VUS	0
15	1	1	2	8	10	1	0	1	8	5	1	3	0
CMT, Charcot-Marie-Tooth disease; IPN, Inherited Peripheral Neuropathy; VUS, Variants of Uncertain Significance.													