E-TABLE 121-4 CHARCOT-MARIE-TOOTH DISEASE

DISORDER	LOCUS/PROTEIN	INHERITANCE	USUAL ONSET	SPECIFIC CLINICAL FEATURES
	CMT1: DEMYELINATING			
CMT1A	17p11.2 Peripheral myelin protein 22 (PMP22)	AD	1st decade	Distal weakness
CMT1B	1q22 Myelin protein zero (P0)	AD	1st decade	Distal weakness More severe
CMT1C	16p13.1-p12.3 LITAF	AD	2nd decade	Distal weakness
CMT1D	10q21.1-q22.1 Early growth response protein 2 (EGR2)	AD	2nd decade	Distal weakness Ptosis
CMT1F	8p21 NF-68	AD	1 to 40 years	Distal weakness Ataxia
CMTX	Xq13.1 Connexin 32	X-linked	2nd decade	Distal weakness Hearing loss Encephalopathy
HNPP	17p11.2 Peripheral myelin protein 22 (PMP22)	AD	3rd decade	Focal episodic weakness
Dejerine-Sottas (HSMN3)	8q23,17p11,10q21 Early growth response protein 2 (EGR2) Peripheral myelin protein 22 (PMP22)	AD	2 yrs	Severe weakness
	CMT2: DOMINANT; AXONAL			
CMT2A	1p36.2 Kinesin-like protein (KIF1B)	AD	10 years	Distal weakness
CMT2A2	1p36 Mitofusin 2 (MFN2)	AD	10 years	Distal weakness Hearing loss
CMT2B	3q21 RAB7	AD	2nd decade	Distal weakness Sensory loss Acromutilation
CMT2C	12q23-24 TRPV4	AD	1st decade	Vocal cord and distal weakness
CMT2D	7p15 Glycyl-tRNA (GARS)	AD	16 to 30 years	Distal weakness arms > legs
CMT2E	8p21 Synthetase Neurofilament triplet L protein (NEFL Sequencing)	AD	1 to 40 years	Distal weakness
CMT2F	7q11-21 HSPB1	AD	2nd decade	Difficulty walking
CMT2G	12q12	AD	15 to 25 years	Distal weakness
CMT2K	8q21 GDAP1	AD	Infant	Distal weakness Vocal cord
CMT2L	12q24 HSPB8	AD	15 to 33 years	Distal weakness
CMT2M	19p13 DNM2	AD	0 to 50 years	Distal weakness, legs > arms, ophthalmoparesis
CMT2N	16q22 AARS	AD	6 to 54 years	Distal leg weakness Asymmetric
	CMT4: RECESSIVE; DEMYELINATING			
CMT4A	8q13-q21.1 Ganglioside-induced differentiation protein-1 (GDAP1)	AR	Childhood	Distal weakness Vocal cord
CMT4B1	11q22 Myotubularin-related protein 2 (MTMR2)	AR	2 to 4 yrs	Distal and proximal weakness
CMT4B2	11p15 Set binding factor 2 (SBF2)	AR	1st 2 decades	Distal weakness Sensory loss Glaucoma
CMT4C	5q32 SH3TC2	AR	5 to 15 yrs	Delayed walking
CMT4D	8q24.3 NDRG1 protein	AR	1 to 10 yrs	Gait disorder Hearing loss
CMT4E	10q21.1-q22.1 Early growth response protein 2 (EGR2)	AR	Birth	Infant hypotonia Arthrogryposis Respiratory failure
CMT4F	19q13.1-q13.2 Periaxin (PRX)	AR	1 to 3 yrs	Motor delay Sensory loss
CMT4H	12p11.21 FGD4	AR	10 to 24 mo	Walking delay Scoliosis
CMT4J	6q21 FIG4	AR	Congenital to adult	Asymmetric proximal and distal weakness
CMT3 (Dejerine-Sottas)	P0, PMP-22, EGR2, Periaxin	AR	2 years	Severe weakness