

Table 3: Syndromic neuropathies that can present with neuropathy.

The table lists syndromic inherited neuropathies that can be diagnosed as non-syndromic neuropathies; **dominant diseases are bolded**. The genes are named according to the HUGO gene nomenclature (<http://www.genenames.org/>), and hyperlinked to this database. The diseases, their chromosomal locus, and genes are hyperlinked to OMIM (<http://www.ncbi.nlm.nih.gov/Omim/>).

disease (OMIM)	affected gene (OMIM)
CMT1-like (dominant demyelinating neuropathy)	
CMT1-like(614434)	FBLN5 (604580)
CMT2-like (dominant axonal neuropathy)	
FAP-1 (105210)	TTR (176300)
no OMIM	PRNP (176640)
CFEOM3A (600638)	TUBB3 (602661)
SPG10 (604187)	KIF5A (602809)
no OMIM	CRYAB (123590)
no OMIM	TARDBP (605078)
no OMIM	BAG3 (603883)
optic atrophy 3, with cataract (165300)	OPA3 (606580)
globoid cell leukodystrophy (245200)	GALC (606890)
metachromatic leukodystrophy (250100)	ARSA (607574)
ACCPN (218000)	SLC12A6 (604878)
Leigh syndrome (256000)	SURF1 (185620)
MTDPS1 (603041)	TYMP (131222)
CCFDN (604168)	CTDPI (604927)
ARCMT-like (recessive, axonal neuropathy)	
AOA4 (616267)	PNKP (605610)
GAN1 (256850)	GAN (605379)
MNGIE (603041)	TYMP (131222)
SANDO (607459)	POLG (174763)
APBD (263570)	GBE1 (607839)
SPAX6/SACS (270550)	SACS (604490)

DSMA1/SMARD1 (604320)	<i>IGHMBP2</i> (600502)
MTPD (609015)	<i>HADHB</i> (143450)
ARCMT (616668)	<i>SPG11</i> (610844)
CEMCOX1 (604377)	<i>SCO2</i> (604272)
Friedreich ataxia (229300)	<i>FXN</i> (606829)
Tangier disease (205400)	<i>ABCA1</i> (600046)
MTDPS6 (256810)	<i>MPV17</i> (137960)
LBSL (611105)	<i>DARS2</i> (610956)
SPG55 (615035)	<i>CI2orf65</i> (613541)
AAAS (231550)	<i>AAAS</i> (605378)
no OMIM	<i>REEP1</i> (609139)
HMSN6B (616505)	<i>SLC25A46</i> (610826)
X-linked axonal neuropathy	
ALD (300100)	<i>ABCD1</i> (300371)
Fabry disease (301500)	<i>GLA</i> (300644)
Cowchock syndrome (310490)	<i>AIFM1</i> (300169)
McLeod syndrome (300842)	<i>XK</i> (314850)
no OMIM	<i>MT-ATP6</i> (516060) m9185T>C, m9176T>C
hereditary motor neuropathy (HMN)	
HNARMD (608895)	<i>FBLN5</i> (604580)
SPG4 (182601)	<i>SPAST</i> (604277)
SPG3A (182600)	<i>ATLI</i> (606439)
no OMIM	<i>MT-ATP6</i> (516060)
ALS 1 (105400)	<i>SOD1</i> (147450)
“CMT2X” (616668)	<i>SPG11</i> (610844)
HMN-like (no OMIM)	<i>HNRNPA1</i> (164017)

AAAS: achalasia-addisonianism-alacrima syndrome;
ACCPN: genesis of the corpus callosum with peripheral neuropathy;
ALD: adrenoleukodystrophy;
AOA4: Ataxia-oculomotor apraxia 4;

CCFDN: congenital cataracts, facial dysmorphism, and neuropathy;
CEMCOX1: cardioencephalomyopathy, fatal infantile, due to cytochrome C oxidase deficiency 1;
CFEOM: congenital fibrosis of extraocular muscles;
FAP: familial amyloidotic polyneuropathy;
GAN1: giant axonal neuropathy-1;
HNARMD: hereditary neuropathy with or without age-related macular degeneration;
LBSL: leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation;
MNGIE: mitochondrial neurogastrointestinal encephalopathy syndrome;
MTDPS: mitochondria DNA depletion syndrome;
MTPD: mitochondrial trifunctional protein deficiency;
SACS: spastic ataxia, Charlevoix-Saguenay type;
SANDO: sensory ataxic neuropathy, dysarthria, and ophthalmoparesis;
SMARD: spinal muscular atrophy with respiratory distress;
SPG: spastic paraplegia;