Table 2: Genetic classification of the syndromic inherited neuropathies.
The table lists neuropathies that have been found in various syndromes. Neuropathies are grouped by their pattern of inheritance and phenotypes, some diseases are thus listed more than once. 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)	gene (OMIM)	clinical
syndromic neuropathies with slow conduction velocities and/or dysmyelinating/demyelinating neuropathies		
Potocki-Lupski syndrome (610883)	Contiguous gene duplication syndrome including *PMP22*	developmental hypotonia, failure to thrive, mental retardation, pervasive developmental disorders, congenital anomalies
Waardenburg syndrome type 2E (611584)	*SOX10* (602229)	hypopigmentation,deafness, , hypogonadotropic hypogonadism, anosmia, agenesis of the olfactory bulbs
PCWH (609136)	*SOX10* (602229)	CNS and PNS, Hirschsprung disease
Cowden syndrome 1 (158350)	*PTEN* (601728)	a man with multifocal motor neuropathy since childhood, learning difficulties, cranial neuropathies had a de novo *PTEN* mutation
metachromatic leukodystrophy (250100)	*ARSA* (607574)	optic atrophy, mental retardation, hypotonia; possibly treatable with bone marrow transplant
globoid cell leukodystrophy (245200)	*GALC* (606890)	spasticity, optic atrophy, mental retardation; possibly treatable with bone marrow transplant
Refsum Disease (266500)	*PHYH* (602026)	deafness, ataxia, retinitis pigmentosa, ichthyosis, heart failure; treatable with dietary restriction
PBD9B (614879)	*PEX7* (601757)	infantile (more severe) Refsum-like disease, with dysmorphism, developmental delay
PHARC (612674)	*ABHD12* (613599)	demyelinating Polyneuropathy, Hearing loss, Ataxia, Retinitis pigmentosa, and Cataract
gonadal dysgenesis with minifascicular neuropathy (607080)	*DHH* (605423)	mental retardation, hypogonadism
CDG1A (212065)	*PMM2* (601785)	leukodystrophy, abnormal serum glycoproteins, static encephalopathy, hypotonia, ataxia, retinitis pigmentosa
MDC1A (607855)	*LAMA2* (156225)	congenital muscular dystrophy, mildly slowed PNS conduction, abnormal T2 MRI signal white matter
Duchenne muscular dystrophy (310200)	*DMD* (300377)	muscular dystrophy
hypomyelinating leukodystrophy 5 (610532)	*FAM126A* (610531)	congenital cataracts, abnormal MRI signal in CNS white matter

SPG75 (616680)	*MAG* (159460)	infantile-onset Pelizaeus-Merzbacher disease-like phenotype that slowly evolves into complicated HSP with mental retardation, dysarthria, optic atrophy
ACPHD (616192)	*DNAJC3* (601184)	juvenile-onset diabetes, early-onset ataxia, spasticity, hearing loss, cerebral atrophy
ACCPN (218000)	*SLC12A6* (604878)	seizures, malformed corpus callosum, mental retardation
AMRF (254900)	*SCARB2* (602257)	progressive myoclonic epilepsy, sometimes associated with renal failure, and less commonly with neuropathy with slow conduction velocity
PEPNS (616113)	*DMXL2* (612186)	hypogonadism, mental retardation
APECED (240300)	*AIRE* (607358)	2 of 3 major clinical symptoms: Addison disease, and/or hypoparathyroidism, and/or chronic mucocutaneous candidiasis, CIDP-like neuropathy
CCFDN (604168)	*CTDP1* (604927)	Rudari Gypsies, congenital cataracts and microcornea, delayed psychomotor development, skeletal anomalies, hypogonadism, facial dysmorphism, marked hyperCKemia after viral infections
LCCS2 (607598)	*ERBB3* (190151)	hydramnios, fetal akinesia, limb contractures, atrophy, distended urinary bladder, high myopia, and degenerative vitreoretinopathy
LCCS5 (615368)	*DNM2* (602378)	akinesia, joint contractures, hypotonia, skeletal abnormalities, brain and retinal hemorrhages
LCCS7 (616286)	*CNTNAP1* (602346)	polyhydramnios, distal arthrogryposis, hypotonia, respiratory distress, facial diplegia, abnormally myelinated axons
LCCS8 (616287)	*ADCY6* (600294)	distal arthrogryposis, hypotonia, respiratory distress, facial diplegia, no motor responses, no myelinated axons
LCSS9 (616503)	*ADGRG6* (612243)	arthrogryposis, atrophy, lack of myelinated axons
LCSS11 (617194)	*GLDN* (608603)	arthrogryposis, widened nodes of Ranvier
AMCNMY (617468)	*LGI4* (608303)	arthrogryposis, severe lack of myelinated axons
Cockayne syndrome A (216400)	*ERCC8* (609412)	dwarfism, optic atrophy, mental retardation
Cockayne syndrome B (133540)	*ERCC6* (609413)	dwarfism, optic atrophy, mental retardation
MTDPS1 (603041)	*TYMP* (131222)	MNGIE phenotype
Leigh syndrome variant (256000)	*SURF1* (185620)	Leigh syndrome
no OMIM	*COX10* (602125)	myopathy, premature ovarian failure, hearing loss, pigmentary maculopathy,

		renal tubular dysfunction
axonal neuropathies associated with ataxia		
NARP (551500)	*MT-ATP6* (516060)	Neuropathy, Ataxia, Retinitis Pigmentosa, seizures
SCA1 (164400)	*ATXN1* (601556)	adult-onset, gaze paresis, slow/absent saccades, spasticity, neuropathy in 40%
SCA2 (183090)	*ATXN2* (601517)	slow saccades, Parkinsonism, myoclonus, neuropathy in 80%
SCA3/MJD (109150)	*ATXN3* (607047)	adult-onset, gaze paresis, extrapyramidal, bulging eyes, neuropathy in 50%
SCA4 (600223)		sensory neuropathy
SCA7 (164500)	*ATXN7* (607640)	adult-onset, retinopathy and neuropathy
SCA10 (603516)	*ATXN10* (611150)	neuropathy in some families
SCA12 (604326)	*PPP2R2B* (604325)	adult-onset, subclinical neuropathy
SCA18 (607458)		sensory and motor neuropathy with ataxia
SCA23 (610245)	*PDYN* (131340)	sensory neuropathy
SCA25 (608703)	*IFRD1*	sensory neuropathy
SCA27 (609307)	*FGF14* (601515)	cerebellar findings, mild neuropathy
SCA36 (614153)	*NOP56* (614154)	distal motor neuropathy
SCA43 (617018)	*MME* (120520)	adult-onset gait and limb ataxia
PEOA3 (609286)	*TWNK* (606075)	PEO, myopathy, sensory neuropathy, hearing loss, depression, dysphagia, cardiomyopathy, multiple mDNA deletions
SCAR1/AOA2 (606002)	*SETX* (608465)	oculomotor apraxia, juvenile onset ataxia, increased alpha fetoprotein, nystagmus, cerebellar and pontine atrophy
SCAR3 (271250)		blindness and deafness
SCAR4 (607317)	*VPS13D* (608877)	saccadic intrusions, acanthocytosis, dyskinesias, psychiatric issues
SCAR8 (6q25.2)	*SYNE1* (608441)	cerebellar atrophy, motor neuropathy in some patients
SCAR21 (616719)	*SCYL1* (607982)	recurrent episodes of liver failure, neuropathy
SCAR26 (617633)	*XRCC1* (194360)	progressive cerebellar atrophy
SANDO (607459)	*POLG* (174763)	Sensory Ataxic Neuropathy, Dysarthria, Ophthalmoplegia
EAOH/AOA1 (208920)	*APTX* (606350)	cerebellar atrophy, hypoalbuminemia
AOA3 (615217)	*PIK3R5* (611317)	oculomotor apraxia
AOA4 (616267)	*PNKP* (605610)	dystonia and ataxia, oculomotor apraxia; onset in the first decade and rapid progression to wheelchair-bound in the second or third decade; allelic to ARCMT2B2 (605589)
SCAN1 (607250)	*TDP1* (607198)	cerebellar ataxia
SCAN3 (no OMIM)	*COA7* (615623)	cerebellar ataxia and atrophy

MCSZ (613402)	*PNKP* (605610)	microcephaly, seizures, and developmental delay; allelic to ARCMT2B2 (605589) and AOA4 (616267)
AXPC1 (609033)	*FLVCR1* (609144)	childhood-onset retinitis pigmentosa, later onset of gait ataxia due to sensory loss, abnormal posterior columns on MRI
DBP deficiency (261515)	*HSD17B4* (601860)	neonatal encephalopathy, retinopathy, hepatomegaly, dysmorphic features, death in infancy
Perrault syndrome 1 (233400)	*HSD17B4* (601860)	hearing loss, hypogonadism, hyperreflexia
Perrault syndrome 5 (616138)	*TWNK* (606075)	Onset second or third decades, ataxia, nystagmus, sensory neuropathy, hearing loss, ovarian dysgenesis; allelic to MTDPS7 (271245), which is more severe
MTDPS7 (271245)	*TWNK* (606075)	ataxia, ophthalmoplegia, ptosis, encephalopathy, seizures; allelic to Perrault syndrome 5 (616138)
LBSL (611105)	*DARS2* (610956)	ataxia, myelopathy, characteristic brain MRI findings, elevated CSF lactate
abetalipoproteinemia (200100)	*MTTP* (157147)	fat malabsorption, vitamin E deficiency, retinitis pigmentosa, acanthocytosis; treatable with vitamin E
VED (277460)	*TTPA* (600415)	similar to Friedreich ataxia, pyramidal syndrome, skeletal deformities
Friedreich ataxia (229300)	*FXN* (606829)	ataxia, cardiomyopathy
FXTAS (300623)	*FMR1* (309550)	tremor, cognitive changes, parkinsonian, MRI changes in cerebellum
ataxia-telangectasia (208900)	*ATM* (607585)	oculomotor apraxia, telangiectasias
galactosialidosis (256540)	*CTSA* (613111)	coarse facies, cerebellar findings
CANVAS (614575)	*RFC1* (102579)	Cerebellar Ataxia, Neuropathy, Vestibular Areflexia Syndrome
no OMIM	*PEX10* (602859)	progressive ataxia, motor neuropathy
axonal neuropathies associated with spastic paraparesis		
SPG2 (312920)	*PLP1* (300401)	delayed motor and cognitive development, optic pallor, limb spasticity, mild atrophy, decreased proprioception
SPG3A (182600)	*ATL1* (606439)	early onset, neuropathy is not associated with most mutations; allelic to HSN1D (613708)
SPG4 (182601)	*SPAST* (604277)	neuropathy infrequently reported
SPG5 (270800)	*CYP7B1* (603711)	some have neuropathy
SPG6 (600363)	*NIPA1* (608145)	neuropathy infrequently reported
SPG7 (607259)	*SPG7* (602783)	optic atrophy, neuropathy reported in some patients

SPG9A (601162)	*ALDH18A1* (138250)	short stature, cataracts, motor neuropathy
SPG10 (604187)	*KIF5A* (602821)	adult onset, neuropathy in some patients, can mimic CMT2
SPG11 (604360)	*SPG11* (610844)	thin corpus callosum, intellectual disability, parkinsonism, dystonia
SPG14 (605229)		mild mental retardation, distal motor neuropathy, sural nerve biopsy normal!
SPG15 (270700)	*ZFYVE26* (612012)	mental retardation, retinal degeneration
SPG17 (270685)	*BSCL2* (606158)	Silver syndrome, spasticity, motor neuropathy in arms > legs, allelic to HMN5A (600794)
SPG20 (275900)	*SPG20* (275900)	mental retardation, pseudobulbar affect
SPG23 (270750)	*DSTYK* (612666)	pigmentation changes, scoliosis
SPG26 (609195)	*B4GALNT1* (601873)	intellectual disability, ataxia, peripheral neuropathy
SPG28 (609340)	*DDHD1* (14603)	subclinical axonal neuropathy
SPG30 (610357)	*KIF1A* (601255)	HSP with sensory motor axonal neuropathy, optic neuropathy; allelic to HSN2C (614213) and MRD9 (614255)
SPG35 (612319)	*FA2H* (611026)	minimal to mild neuropathy
SPG36 (613096)		axonal neuropathy
SPG38 (612335)		Silver syndrome
SPG39 (612020)	*PNPLA6* (603197)	childhood onset, slowly progressive spastic paraplegia and distal motor neuropathy, allelic to Boucher-Neuhauser Syndrome (215470) and Gordon-Holmes syndrome (212840)
SPG43 (615043)	*C19orf12* (614297)	childhood onset spastic paraplegia and axonal neuropathy, brain iron accumulation, optic atrophy, extrapyramidal signs; allelic to NBIA4 (614298)
SPG46 (614409)	*GBA2* (609471)	mental impairment, thin corpus callosum, ataxia, cataract
SPG48 (613647)	*AP5Z1* (613653)	ataxia, dystonia, myoclonus, and parkinsonism
SPG49 (615031)	*TECPR2* (615000)	dysmorphic features, developmental delay, spasticity, ataxia
SPG55 (615035)	*C12orf65* (613541)	early onset, optic atrophy, intellectual impairment, mild neuropathy; allelic to SPG65 (613559)
SPG56 (615030)	*CYP2U1* (610670)	Mild axonal neuropathy
SPG57 (615658)	*TFG* (602498)	optic atrophy and neuropathy, R106C allelic to HMSNO (604484)
SPG61 (615685)	*ARL6IP1* (607669)	onset at birth, plus sensory > motor neuropathy, self-mutilation
SPG65 (613559)	*C12orf65* (613541)	Leigh-like presentation; allelic to SPG55 (615035)

SPG76 (616907)	*CAPN1* (114220)	onset 3rd decade, spasticity, dysarthria and ataxia, some have neuropathy
SPG78 (617225)	*ATP13A2* (610513)	adult-onset spasticity and neuropathy, resulting in gait difficulties, also dysarthria, oculomotor disturbances, limb and gait ataxia, cognitive impairment, parkinsonism; allelic to Kufor-Rakeb syndrome (606693)
SPAX2 (611302)	*KIF1C* (603060)	spasticity
SPAX5 (614487)	*AFG3L2* (604581)	early onset, oculomotor apraxia, dystonia, and myoclonic epilepsy; allelic to SCA28 (610246)
SACS/SPAX6 (270550)	*SACS* (604490)	childhood onset, progressive myelopathy and ataxia, myelinated retinal axons, intermediate conduction velocity
no OMIM	*TUBB2A* (615101)	Progressive, infantile onset spasticity, ataxia, optic atrophy, polyneuropathy with intermediate slowing
HSN with spastic paraplegia (256840)	*CCT5* (610150)	severe mutilating sensory neuropathy
SPOAN (609541)	*KLC2* (611729)	Spastic Paraplegia, Optic Atrophy, and Neuropathy, early onset, Brazil
AMACRD (614307)	*AMACR (*604489*)*	retinopathy, myelopathy, neuropathy, elevated phytanic and pristanic acids
no OMIM	*SELENOI* (607915)	severe spastic paraplegia, sensorineural-deafness, blindness, seizures; one patient said to have a demyelinating neuropathy
adrenoleukodystrophy (300100)	*ABCD1* (300371)	spastic paraparesis, adrenal insufficiency; possibly treatable with bone marrow transplant
APBD (263570)	*GBE1* (607839)	axonal neuropathy, cognitive impairment, MRI changes in white matter, myelopathy
other syndromic axonal neuropathies		
deafness, aminoglycoside-induced (580000)	*MT-RNR1* (561000)	deafness, sensitivity to aminoglycosides, parkinsonism
Leber hereditary optic neuropathy (53500)	mitochondrial many alleles	optic neuropathy, rarely peripheral neuropathy
Kearns-Sayre syndrome (530000)	mitochondrial deletions	PEO, retinitis pigmentosa, heart block, ptosis
MELAS (540000)	*MT-TL1* (590050)	mitochondrial Myopathy, Encephalopathy, lactic Acidosis, and Stroke-like episodes
MERFF (545000)	*MT-TK* (590060)	Myoclonic Epilepsy associated with Ragged Red Fibers, lipomas, lipomas, sensory neuropathy
mitochondrial complex V deficiency (516070)	*MT-ATP8* (516070)	dysarthria, ataxic gait, external ophthalmoparesis, cardiomyopathy
porphyria, acute intermittent (176000)	*HMBS* (609806)	abdominal pain, psychosis, depression, dementia, seizures; treatable with hematin

coproporphyria (121300)	*CPOX* (612732)	skin photosensitivity; treatable with hematin
porphyria, variegata (176200)	*PPOX* (600923)	skin photosensitivity (mainly in South Africa) ; treatable with hematin
FAP-1 (105210) **FAP-2** (115430)	*TTR* (176300)	dysautonomia, cardiac disease, type 2 has carpal tunnel syndrome; treatable with liver transplanation and ASOs
FAP-3 (105200)	*APOA1* (107680)	nephropathy, liver disease
FAP-4 (105120)	*GSN* (137350)	corneal lattice dystrophy, cranial neuropathies
familial visceral amyloidosis (105200)	*B2M* (109700)	orthostatic hypotension, diarrhea
no OMIM	*PRNP* (176640)	autonomic/sensory neuropathy precedes cognitive decline
CFEOM1 (600638)	*TUBB3* (602661)	congenital fibrosis of extraocular muscles
optic neuropathy plus (125250)	*OPA1* (605290)	early onset deafness, optic atrophy, spasticity, ataxia, PEO, myopathy, neuropathy
optic atrophy 3, with cataract (165300**)**	*OPA3* (606580)	sensory and motor neuropathy, optic neuropathy, cataracts, GI dysmotility
no OMIM	*LMNA* (150330)	myopathy, cardiomyopathy, leuconychia
MFM2 (608810)	*CRYAB* (123590)	myofibrillary myopathy, cardiomyopathy
MFM3 (609200)	*MYOT* (604103)	myofibrillary myopathy, cardiomyopathy
MFM4 (609452)	*LDB3* (605906)	myofibrillary myopathy, cardiomyopathy
MFM5 (609524)	*FLNC* (102565)	myofibrillary myopathy, cardiomyopathy
MFM6 (612954)	*BAG3* (603883)	giant axons, myofibrillary myopathy cardiomyopathy, scoliosis
DM1 (160900)	*DMPK* (605377)	myopathy, myotonia, encephalopathy, mild neuropathy
MRD9 (614255)	*KIF1A* (601255)	global developmental delay and intellectual disability, optic nerve atrophy, microcephaly, seizures, progressive spastic paraparesis, and cerebral and/or cerebellar atrophy
NF2 (101000)	*NF2* (607379)	Schwannomas
LEOPARD syndrome 1 (151100)	*PTPN11* (176876)	massive burden of paraspinal tumors, progressive neuropathy
Marfan syndrome (154700)	*FBN1* (134797)	disproportionately long limbs and digits, dilatation of the aortic root
CLIFAHDD (616266)	*NALCN* (611549)	congenital contractures of the limbs and face with hypotonia and developmental delay
no OMIM	*EMILIN1*	one family

CTRCT21 (610202)	*MAF* (177075)	reduced threshold for vibration in a person with R288P mutation, which also causes cataract, microcornea and iris coloboma
McLeod syndrome (300842)	*XK* (314850)	onset 25-60, Huntington-like syndrome, epilepsy, cardiomyopathy, acanthocytes
Fabry disease (301500)	*GLA* (300644)	angiokeratoma, painful neuropathy, renal failure, cardiomyopathy; , treated with enzyme replacement therapy or migalastat
adrenoleukodystrophy (300100)	*ABCD1* (300371)	spastic paraparesis, adrenal insufficiency
PDHAD (312170)	*PDHA1* (300502)	hypotonia, lethargy, mental retardation, episodic lactic acidosis
SBMA (313200)	*AR* (313700)	motor neuron disease and sensory neuropathy, androgen insensitivity
Cowchock syndrome (310490)	*AIFM1* (300169)	mental retardation, deafness; possibly treatable with riboflavin
PGK1 deficiency (300653)	*PGK1* (311800)	hemolytic anemia, myopathy, myoglobinuria, neurologic involvement
Menkes disease (09400)	*ATP7A* (300011)	motor neuropathy, spasticity, ataxia
Wilson disease (277900)	*ATP7B* (606882)	neuropsychiatric and/or hepatic manifestations owing ot copper accumulation in basal ganglia and liver, treatable
IHPRF1 (615419)	*NALCN* (611549)	infantile hypotonia with psychomotor retardation and characteristic facies; nerve biopsy was said to show neuraxonal dystrophy
EMPF1 (614388)	*DNM1L* (603850)	born with profound hypotonia, no spontaneous movements, absent deep tendon reflexes, decreased numbers of myelinated axons, giant mitochondria
EMPF2 (617086)	*MFF* (614785)	Leigh-like syndrome, delayed psychomotor development, severe hypotonia, inability to walk, microcephaly, optic atrophy
Harel-Yoon syndrome (617183)	*ATAD3A* (612316)	global developmental delay, hypotonia, optic atrophy, axonal neuropathy, hypertrophic cardiomyopathy
PNRIID (618124)	*MCM3AP* (603294)	early childhood-onset neurologic disorder characterized by slowly progressive distal motor impairment resulting in gait difficulties, often with loss of ambulation, and difficulties using the hands in most patients.
MTDPS3 (251880)	*DGUOK* (601465)	progressive liver failure, neuropathy infrequently reported
MTDPS4B (613662)	*POLG* (174763)	MNGIE phenotype
MTDPS5 (612073)	*SUCLA2* (603921)	Leigh-like syndrome, hypotonia, external ophthalmoplegia, deafness, dystonia
MTDPS6 (256810)	*MPV17* (137960)	progressive liver failure, often leading to death in the first year of life, survivors develop progressive neurologic involvement, including ataxia, neuropathy, dystonia, psychomotor regression, corneal ulcers, vomiting
MTDPS8B (612075)	*RRM2B* (604712)	MNGIE phenotype
IMNEPD (616263)	*PTRH2* (608625)	microcephaly, failure to thrive, progressive ataxia, distal muscle weakness,

		demyelinating sensorimotor neuropathy, and sensorineural deafness.
glutaric acidemia IIC (231680)	*ETFDH* (231675)	lipid storage myopathy, increased acylcarnitine levels, responsive to Co-Q and/or riboflavin
CAGSSS (616007)	*IARS2* (612801)	cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, skeletal dysplasia
distal arthrogryposis, 5D (615065)	*ECEL1* (605896)	congenital contractures
giant axonal neuropathy-1 (256850)	*GAN* (605379)	mental retardation, spasticity, kinky or curly hair, slowed NCVs
hypomyelinating leukodystrophy 3 (260600)	*AIMP1* (603605)	global developmental delay, spasticity
CONDSIAS (618170)	*ADPRHL2* (610624)	stress-induced childhood-onset neurodegeneration, variable ataxia, seizures
no OMIM	*CLTCL1* (601273)	unable to sense pain, unresponsive to soft touch, developmentally delayed, brain MRI
IND1 (256600)	*PLA2G6* (603604)	infantile onset, neuroaxonal dystrophy of PNS and CNS, progressive motor and mental deterioration
NBIA2B (610217)	*PLA2G6* (603604)	allelic to INAD, progressive quadriparesis and mental deterioration, optic atrophy, iron accumulation in the globus pallidus
NBIA4 (614298)	*C19orf12* (614297)	allelic to SPG43, progressive spasticity, psychiatric features, Parkinsonism, iron accumulation in the globus pallidus
Schindler disease, type I (609241)	*NAGA* (104170)	infantile onset, neuroaxonal dystrophy, severe, progressive psychomotor retardation, optic atrophy, blindness, spasticity
Schindler disease, type III/Kanzaki disease (609242)	*NAGA* (104170)	adult onset - angiokeratoma, sensorineural hearing loss, vertigo
Tay-Sachs disease (272800)	*HEXA (606869)*	onset in infancy, developmental retardation, followed by paralysis, dementia and blindness, accumulation of GM2; neuropathy rarely reported
Sandhoff disease (268800)	*HEXB* (606873)	similar to Tay-Sachs disease
CDDG (615273)	*NGLY1* (610661)	decreased tearing, choreoathetosis, liver disease, global developmental delay, microcephaly, hypotonia
Chediak-Higashi syndrome (214500)	*LYST* (606897)	partial albinism, immunodeficiency
HP1 (259900)	*AGXT* (604285)	calcium oxalate deposits in various bodily tissues, especially the kidney, resulting in renal failure, treatable with pyridoxine
PCH1A (607596)	*VRK1* (602168)	delayed motor development, microcephaly
PCH2D (613811)	*SEPSECS* (613009)	progressive microcephaly, atrophy of the cerebrum and cerebellum, profound mental retardation, spasticity, seizures

PCH9 (615809)	*AMPD2* (102771)	progressive postnatal microcephaly, absent corpus callosum
PCH10 (615803)	*CLP1* (615803	failure to develop motor skills, absent/delayed speech, progressive spasticity, epilepsy
NEDHND (617519)	*SPTBN4* (606214)	severe encephalopathy, congenital myopathy, neuropathy, deafness
AAAS (231550)	*AAAS* (605378)	achalasia, addisonianism, alacrima
Tangier disease (205400)	*ABCA1* (600046)	orange tonsils, organomegaly, pain, paresthesias, anesthesia, cranial neuropathies, multifocal neuropathies, low HDL
THMD4 (613710)	*SLC25A19* (606521)	childhood onset of episodic encephalopathy, often associated with a febrile illness, bilateral striatal necrosis; possibly treatable with thiamine
tyrosinemia type 1 (276700)	*FAH* (613871)	neuropathy similar to porphyria, progressive liver disease, secondary renal tubular, rickets; hematin may be therapeutic in acute crises
MMACHC (277400)	*MMACHC* (609831)	onset infancy to adulthood, thrombotic thrombocytopenia, retinitis pigmentosa, myelopathy, methylmalonic aciduria, and homocystinuria, high dose; parenteral hydroxycobalamin is a possible treatment
MTPD (609015)	*HADHA* (600890) *HADHB* (143450)	most early onset and lethal (cardiac and liver involvement), adolescent onset have axonal neuropathy and progressive myopathy with recurrent rhabdomyolysis
MSUDIb (248600)	*CKDHB* (248611)	progressive infantile encephalopathy to childhood onset, dietary restriction, may respond to thiamine
SMA1 (253300)	*SMN1* (600354)	infantile onset severe weakness and atrophy
BVVL1 (211530)	*SLC52A3* (613350)	infantile onset of hearing loss, hearing loss, bulbar dysfunction
BVVL2 (614707)	*SLC52A2* (607882)	infantile onset of vision and hearing loss, bulbar dysfunction
LKDMN (613724)	*SCP2* (184755)	motor neuropathy, myelopathy, dystonia, hypogonadism, cerebellar findings
PEAMO (617207)	*TBCE* (604934)	distal SMA, spasticity, encephalopathy
PEBAT (617193)	*TBCD* (604649)	motor neuropathy vs motoneuron loss
Martsolf syndrome (212720)	*RAB3GAP2* (611663)	microcephaly, mental retardation, cataracts, and hypogonadism; milder and allelic to WARBM2 (614225)
WARBM1 (600118)	*RAB3GAP1* (602536)	microcephaly, microphthalmia, microcornea, congenital cataracts, optic atrophy, cortical dysplasia, in particular corpus callosum hypoplasia, severe mental retardation, spastic diplegia, hypogonadism
WARBM2 (614225)	*RAB3GAP2* (609275)	congenital cataracts, microphthalmia, microcephaly, polymicrogyria, hypoplasia of the corpus callosum, severe developmental delay; allelic to Martsolf syndrome (212720)
WARBM3 (614222)	*RAB18* (602207)	microcephaly, microphthalmia, microcornea, congenital cataracts, optic

		atrophy, cortical dysplasia, in particular corpus callosum hypoplasia, severe mental retardation, spastic diplegia, and hypogonadism
GACR (258870)	*OAT* (613349)	chorioretinal degeneration, cataracts, and type II fiber atrophy
Marinesco-Sjögren syndrome (248800)	*SIL1* (608005)	congenital cataracts, cerebellar ataxia, progressive muscle weakness
cerebrotendinous xanthomatosis (213700)	*CYP27A1* (606530)	mild to intermediate slowing of conduction velocity, neonatal jaundice, ataxia, myelopathy, dementia, cataracts, low cholesterol, xanthomas, atherosclerosis; treatable with chenodeoxycholic acid
CEDNIK syndrome (609528)	*SNAP29* (604202)	CErebral Dysgenesis, Neuropathy, Ichthyosis, palmoplantar Keratoderma
MEDNIK (609313)	*AP1S1* (603531)	Mental retardation, Enteropathy, Deafness, Neuropathy, Keratoderma
CD59 deficiency (612300)	*CD59* (107271)	early-onset, immune-mediated motor axonal neuropathy, strokes, and chronic Coombs-negative hemolysis
xeroderma pigmentosum (278700)	*XPA* (611153)	cutaneous lesions, increased risk of skin cancers
EDS6 (225400)	*PLOD1* (153454)	skin hyperextensibility, articular hypermobility, tissue fragility
PNMHH (614369)	*MYH14* (608568)	myopathy, hoarseness, hearing loss
COXPD3 (610505)	*TSFM* (604723)	infantile-onset mitochondrial cardiomyopathy, progressing to Leigh syndrome, neuropathy, and optic atrophy
CEMCOX1 (604377)	*SCO2* (604272)	Infantile onset encephalopathy, heart failure, hypotonia
CTRCT21 (610202)	*MAF* (177075)	cataracts, diminished vibration sensation
glycogen storage disease III (232400)	*AGL* (610860)	hepatic and muscle involvement

AAAS: achalasia-addisonianism-alacrima syndrome;
ACCPN: genesis of the corpus callosum with peripheral neuropathy;
ACPHD: ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus;
AMACRD: alpha-methylacyl-CoA racemase deficiency;
AMCNMY: arthrogryposis multiplex congenita, neurogenic, with myelin defect;
AMRF: action myoclonus-renal failure syndrome;
APBD: polyglucosan body neuropathy, adult form;
APECED: autoimmune polyendocrinopathy-candidiasis-ectodermal-dystrophy;
AOA: Ataxia Oculomotor Apraxia;
AXPC1: posterior column ataxia with retinitis pigmentosa;
BVVL: Brown-Vialetto-Van Laere syndrome;
CAGSSS: Cataracts, Growth hormone deficiency, Sensory neuropathy, Sensorineural hearing loss and Skeletal dysplasia;
CANVAS: Cerebellar Ataxia, Neuropathy, Vestibular Areflexia Syndrome;

CCFDN: congenital cataracts, facial dysmorphism, and neuropathy;
CDDG: congenital disorder of deglycosylation;
CDG1A: congenital disorder of glycosylation type Ia;
CEDNIK syndrome: CErebral Dysgenesis Neuropathy, Ichthyosis, and palmoplantar Keratoderma;
CEMCOX1: cardioencephalomyopathy, fatal infantile, due to cytochrome C oxidase deficiency 1;
CFEOM: Congenital Fibrosis Extraocular Muscles;
CLIFAHDD: congenital contractures of the limbs and face, hypotonia, and developmental delay;
COXPD3: combined oxidative phosphorylation deficiency-3;
CTRCT21: cataract 21, multiple types;
DBP: (D-bifunctional protein) deficiency;
DM1: myotonic dystrophy type 1;
EAOH: ataxia, early onset, with oculomotor apraxia and hypoalbuminemia;
EDS6: Ehlers-Danlos type VI;
EMPF: encephalopathy due to defective mitochondrial and peroxisomal fission;
FAP: familial amyloidotic polyneuropathy;
FRDA: Friedreich ataxia;
FXTAS: fragile X tremor/ataxia syndrome;
GACR: gyrate atrophy of choroid and retina;
HP1: hyperoxaluria, primary type 1;
IHPRF: hypotonia, infantile, with psychomotor retardation and characteristic facies;
IMNEPD: infantile-onset multisystem neurologic, endocrine, and pancreatic disease;
IND1: infantile neuroaxonal dystrophy 1;
LBSL: leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation;
LCCS: lethal congenital contracture syndrome;
LKDMN: leukoencephalopathy with dystonia and motor neuropathy;
MADD: multiple acyl-CoA dehydrogenase deficiency;
MCSZ: microcephaly, seizures, and developmental delay;
MDC1A muscular dystrophy, congenital merosin-deficient, 1A;
MEDNIK: mental retardation enteropathy, deafness, peripheral neuropathy, keratoderma;
MELAS: mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes;
MERRF: myoclonic epilepsy associated with ragged-red fibers;
MFM: myofibrillar myopathy;
MJD: Machado-Joseph disease;
MMACHC: early onset methylmalonic aciduria and homocystinuria;
MNGIE: mitochondrial neurogastrointestinal encephalopathy syndrome;
MRD9: mental retardation dominant 9;

MSUDIb: maple syrup urine disease;
MTDPS: mitochondria DNA depletion syndrome;
MTPD: Mitochondrial Trifunctional Protein Deficiency;
NARP: Neuropathy, Ataxia, Retinitis Pigmentosa,
NBIA: neurodegeneration with brain iron accumulation;
NEDHND: neurodevelopmental disorder with hyptonia, neuropathy, and deafness;
NF2: neurofibromatosis type 2;
PCH: pontocerebellar hypoplasia;
PCWH: peripheral demyelinating syndrome, central dysmyelinating leukodystrophy, Waardenburg syndrome, and Hirschprung disease;
PDB9B: peroxisome biogenesis disorder;
PDHAD: pyruvate dehydrogenase E1-alpha deficiency;
PEAMO: progressive encephalopathy with amyotrophy and optic atrophy;
PEBAT: progressive, early-onset encephalopathy with brain atrophy and thin corpus callosum;
PEO: progressive external ophthalmoplegia;
PEPNS: polyendocrine-polyneuropathy syndrome;
PGK1: Phosphoglycerate kinase 1;
PHARC: (demyelinating) Polyneuropathy, Hearing loss, Ataxia, Retinitis pigmentosa, and Cataract;
PKAN: pantothenate kinase-associated neurodegeneration;
PMD: Pelizaeus-Merzbacher disease;
PNMHH: peripheral neuropathy, myopathy, hoarseness, hearing loss;
PNRIID: peripheral neuropathy with or without impaired intellectual development;
TFP deficiency;
THMD4: thiamine metabolism dysfunction syndrome 4;
SACS: spastic ataxia, Charlevoix-Saguenay type;
SANDO: sensory ataxic neuropathy, dysarthria, and ophthalmoparesis;
SBMA: spinal and bulbar muscular atrophy, X-linked;
SCA: spinocerebellar ataxia;
SCAN: spinocerebellar ataxia, autosomal recessive, with axonal neuropathy;
SCAR: spinocerebellar ataxia, autosomal recessive;
SMA: Spinal Muscular Atrophy;
SMNA: autosomal dominant sensory/motor neuropathy with ataxia;
SPAX: spastic ataxia;
SPG: spastic paraplegia;
SPOAN: spastic paraplegia, optic atrophy, and neuropathy;
TFP: trifunctional protein deficiency;
THMD4: thiamine metabolism dysfunction syndrome, type 4;

VED: vitamin E, familial isolated deficiency;
WARBM: Warburg micro syndrome

References

Ahmed MY et al. (2017) A mutation of EPT1 (SELENOI) underlies a new disorder of Kennedy pathway phospholipid biosynthesis. Brain 140:547-554.

Ahola S, Isohanni P, Euro L, Brilhante V, Palotie A, Pihko H, Lonnqvist T, Lehtonen T, Laine J, Tyynismaa H, Suomalainen A (2014) Mitochondrial EFTs defects in juvenile-onset Leigh disease, ataxia, neuropathy, and optic atrophy. Neurology 83:743-751.

Al Tassan N, Khalil D, Shinwari J, Al Sharif L, Bavi P, Abduljaleel Z, Abu Dhaim N, Magrashi A, Bobis S, Ahmed H, Alahmed S, Bohlega S (2012) A missense mutation in PIK3R5 gene in a family with ataxia and oculomotor apraxia. Hum Mutat 33:351-354.

Al-Sayed MD, Al-Zaidan H, Albakheet A, Hakami H, Kenana R, Al-Yafee Y, Al-Dosary M, Qari A, Al-Sheddi T, Al-Muheiza M, Al-Qubbaj W, Lakmache Y, Al-Hindi H, Ghaziuddin M, Colak D, Kaya N (2013) Mutations in NALCN cause an autosomal-recessive syndrome with severe hypotonia, speech impairment, and cognitive delay. Am J Hum Genet 93:721-726.

Alazami AM et al. (2015) Accelerating novel candidate gene discovery in neurogenetic disorders via whole-exome sequencing of prescreened multiplex consanguineous families. Cell Rep 10:148-161.

Albers JW, Fink JK (2004) Porphyric neuropathy. Muscle Nerve 30:410-422.

Alonso I, Jardim LB, Artigalas O, Saraiva-Pereira ML, Matsuura T, Ashizawa T, Sequeiros J, Silveira I (2006) Reduced penetrance of intermediate size alleles in spinocerebellar ataxia type 10. Neurology 66:1602-1604.

Angelicheva D (1999) Congenital cataracts facial dysmorphism neuropathy (CCFDN) syndrome: a novel developmental disorder in gypsies maps to 18qter. Eur J Human Genet 7:560-566.

Anheim M, Fleury MC, Franques J, Moreira MC, Delaunoy JP, StoppaLyonnet D, Koenig M, Tranchant C (2008) Clinical and molecular findings of ataxia with oculomotor apraxia type 2 in 4 families. Arch Neurol 65:958-962.

Antonicka H, Ostergaard E, Sasarman F, Weraarpachai W, Wibrand F, Pedersen AM, Rodenburg RJ, van der Knaap MS, Smeitink JA, Chrzanowska-Lightowlers ZM, Shoubridge EA (2010) Mutations in C12orf65 in patients with encephalomyopathy and a mitochondrial translation defect. Am J Hum Genet 87:115-122.

Anttonen A-K, al. e (2015) Selenoprotein biosynthesis defect causes progressive encephalopathy with elevated lactate. Neurology 85:306-315.

Apartis E et al. (2012) FXTAS: New insights and the need for revised diagnostic criteria. Neurology 79:1898-1907.

Appenzeller O (1976) Acromutilating, paralyzing neuropathy with corneal ulceration in navajo children. Arch Neurol 33:733-738.

Argov Z, Soffer D, Eisenberg S, Zimmerman Y (1986) Chronic demyelinating peripheral neuropathy in cerebrotendinous xanthomatosis. Ann Neurol 20:89-91.

Arnoldi A, Crimella C, Tenderini E, Martinuzzi A, D'Angelo MG, Musumeci O, Toscano A, Scarlato M, Fantin M, Bresolin N, Bassi MT (2012) Clinical phenotype variability in patients with hereditary spastic paraplegia type 5 associated with CYP7B1 mutations. Clin Genet 81:150-157.

Atkinson D, al. e (2016) Sphingosine 1-phosphate lyase deficiency causes Charcot-Marie-Tooth neuropathy. Neurology 88:533-542.

Baets J, Deconinck T, Smets K, Goossens D, Van den Bergh P, Dahan K, Schmedding E, Santens P, Rasic VM, Van Damme P, Robberecht W, De Meirleir L, Michielsens B, Del-Favero J, Jordanova A, De Jonghe P (2010) Mutations in SACS cause atypical and late-onset forms of ARSACS. Neurology 75:1181-1188.

Bakalkin G, Watanabe H, Jezierska J, Depoorter C, Verschuuren-Bemelmans C, Bazov I, Artemenko KA, Yakovleva T, Dooijes D, Van de Warrenburg BP, Zubarev RA, Kremer B, Knapp PE, Hauser KF, Wijmenga C, Nyberg F, Sinke RJ, Verbeek DS (2010) Prodynorphin mutations cause the neurodegenerative disorder spinocerebellar ataxia type 23. Am J Hum Genet 87:593-603.

Bansagi B, Lewis-Smith DJ, Pal E, Duff J, Griffin J, Pyle A, Muller JS, Rudas G, Aranyi Z, Lochmuller H, Chinnery PF, Horvath R (2016) Phenotypic convergence of Menkes and Wilson disease. Neurol Genet.

Bansagi B, Phan V, Baker MR, O'Sullivan J, Jennings MJ, Whittaker RG, Muller JS, Duff J, Griffin H, Miller JAL, Gorman GS, Lochmuller H, Chinnery PF, Roos A, Swan LE, Horvath R (2018) Multifocal demyelinating motor neuropathy and hamartoma syndrome associated with a de novo PTEN mutation. Neurology 90:e1842-e1848.

Bassen FA, Kornzweig AL (1950) Malformations of the erythrocytes in a case of atypical retinitis pigmentosa. Blood 5:381-387.

Baumer P, Mautner VF, Baumer T, Schuhmann MU, Tatagiba M, Heiland S, Kaestel T, Bendszus M, Pham M (2013) Accumulation of non-compressive fascicular lesions underlies NF2 polyneuropathy. J Neurol 260:38-46.

Bedlack RS, Vu T, Hammans S, Sparr SA, Myers B, Morgenlander J, Hirano M (2004) MNGIE neuropathy: Five cases mimicking chronic inflammatory demyelinating polyneuropathy. Muscle Nerve 29:364-368.

Beetz C, Koch N, Khundadze M, Zimmer G, Nietzsche S, Hertel N, Huebner AK, Mumtaz R, Schweizer M, Dirren E, Karle KN, Irintchev A, Alvarez V, Redies C, Westermann M, Kurth I, Deufel T, Kessels MM, Qualmann B, Hubner CA (2013) A spastic paraplegia mouse model reveals REEP1-dependent ER shaping. J Clin Invest 123:4273-4282.

Bem D et al. (2011) Loss-of-function mutations in RAB18 cause Warburg micro syndrome. Am J Hum Genet 88:499-507.

Bend EG, al. e (2016) NALCN channelopathies - Distinguishing gain-of-function and loss-of-function mutations. Neurology 87:1131-1139.

Benedetti S, Bertini E, Iannaccone S, Angelini C, Trisciani M, Toniolo D, Sferrazza B, Carrera P, Comi G, Ferrari M, Quattrini A, Previtali SC (2005) Dominant LMNA mutations can cause combined muscular dystrophy and peripheral neuropathy. J Neurol Neurosurg Psychiat 76:1019-1021.

Berini SE, Tracy JA, Engelstad JK, Lorenz EC, Milliner DS, Dyck PJ (2015) Progressive polyradiculoneuropathy due to intraneural oxalate deposition in type 1 primary hyperoxaluria. Muscle Nerve 51:449-454.

Blumen SC, al. e (2003) A locus for complicated hereditary paraplegia maps to chromosome 1q24-q25. Ann Neurol 54:796-803.

Bomont P, al. e (2000) Homozygosity mapping of spinocerebellar ataxia with cerebellar atrophy and peripheral neuropathy to 9q33–34, and with hearing impairment and optic atrophy to 6p21–23. Eur J Hum Genet 8:986-990.

Bomont P, Cavalier L, Blondeau F, Hamida CB, Belal S, Tazir M, Demir E, Topaloglu H, Korinthenberg R, Tuysuz B, Landrieu P, Hentati F, Koenig M (2000) The gene encoding gigaxonin, a new member of the cytoskeletal BTB/kelch repeat family, is mutated in giant axonal neuropathy. Nat Genet 26:370-374.

Boquist L, Lindqvist B, O'stberg Y, Steen L (1973) Primary oxalosis. Am J Med 54:673-681.

Bouhouche A, Benomar A, Bouslam N, al. e (2006a) Mutation in the epsilon subunit of the cytosolic chaperonin-containing t-complex peptide-1 (Cct5) gene causes autosomal recessive mutilating sensory neuropathy with spastic paraplegia. . J Med Genet 43:441–443.

Bouhouche A, Benomar A, Bouslam N, Ouazzani R, Chkili T, Yahyaoui M (2006b) Autosomal recessive mutilating sensory neuropathy with spastic paraplegia maps to chromosome 5p15.31-14.1. Eur J Human Genet 14:249-252.

Boukhris A, Stevanin G, Feki I, Denis E, Elleuch N, Miladi MI, Truchetto J, Denora P, Belal S, Mhiri C, Brice A (2008) Hereditary spastic paraplegia with mental impairment and thin corpus callosum in Tunisia: SPG11, SPG15, and further genetic heterogeneity. . Arch Neurol 65:393-402.

Boukhris A et al. (2013) Alteration of ganglioside biosynthesis responsible for complex hereditary spastic paraplegia. Am J Hum Genet 93:118-123.
Bouslam N, Benomar A, Azzedine H, Bouhouche A, Namekawa M, Klebe S, Charon C, Durr A, Ruberg M, Brice A, Yahyaoui M, Stevanin G (2005) Mapping of a new form of pure autosomal recessive spastic paraplegia (SPG28). Ann Neurol 57:567-571.
Bras J, Alonso I, Barbot C, Costa MM, Darwent L, Orme T, Sequeiros J, Hardy J, Coutinho P, Guerreiro R (2015) Mutations in PNKP cause recessive ataxia with oculomotor apraxia type 4. Am J Hum Genet 96:474-479.
Brkanac Z, Fernandez M, Matsushita M, Lipe H, Wolff J, Bird TD, Raskind WH (2002) Autosomal dominant sensory/motor neuropathy with Ataxia (SMNA): Linkage to chromosome 7q22-q32. Am J Med Genet 114:450-457.
Brkanac Z, Spencer D, Shendure J, Robertson PD, Matsushita M, Vu T, Bird TD, Olson MV, Raskind WH (2009) IFRD1 is a candidate gene for SMNA on chromosome 7q22-q23. Am J Hum Genet 84:692-697.
Capuano A, Bucciotti F, Farwell KD, Tippin Davis B, Mroske C, Hulick PJ, Weissman SM, Gao Q, Spessotto P, Colombatti A, Doliana R (2016) Diagnostic Exome Sequencing Identifies a Novel Gene, EMILIN1, Associated with Autosomal-Dominant Hereditary Connective Tissue Disease. Hum Mutat 37:84-97.
Carrozzo R, Dionisi-Vici C, Steuerwald U, Lucioli S, Deodato F, Di Giandomenico S, Bertini E, Franke B, Kluijtmans LA, Meschini MC, Rizzo C, Piemonte F, Rodenburg R, Santer R, Santorelli FM, van Rooij A, Vermunt-de Koning D, Morava E, Wevers RA (2007) SUCLA2 mutations are associated with mild methylmalonic aciduria, Leigh-like encephalomyopathy, dystonia and deafness. Brain 130:862-874.
Choi BO, Kang SH, Hyun YS, Kanwal S, Park SW, Koo H, Kim SB, Choi YC, Yoo JH, Kim JW, Park KD, Choi KG, Kim SJ, Züchner S, Chung KW (2011) A complex phenotype of peripheral neuropathy, myopathy, hoarseness, and hearing loss is linked to an autosomal dominant mutation in MYH14. Hum Mutat 32:669-677.
Christodoulou K, Zamba E, Tsingis M, Mubaidin A, Horani K, AbuSheik S, El Khateeb M, Kyriacou K, Kyriakides T, Al Qudah AK, Middleton L (2000) A novel form of distal hereditary motor neuronopathy maps to chromosome 9p21.1-p12. Ann Neurol 48:877-884.
Collongues N, Depienne C, Boehm N, Echaniz-Laguna A, Samama B, Durr A, Stevanin G, Leguern E, Brice A, Labauge P, de Seze J (2012) Novel SPG10 mutation associated with dysautonomia, spinal cord atrophy, and skin biopsy abnormality. Eur J Neurol.
Comi G, Ciafaloni E, de Silva HAR, Prelle A, Bardoni A, Rigoletto C, Robotti M, Bresolin N, Moggio M, Fortunato F, Ciscato P, Turconi A, Roses AD, Scarlato G (1995) A G^{+1}-> A transversion at the 5' splice site of intron 69 of the dystrophin gene causing the absence of peripheral nerve Dp116 and severe clinical involvement in a DMD patient. Hum Mol Genet 4:2171-2174.
Conboy E, Dhamija R, Wang M, Xie J, Dyck PJ, Bridges AG, Spinner RJ, Clayton AC, Watson RE, Messiaen L, Babovic-Vuksanovic D (2016) Paraspinal neurofibromas and hypertrophic neuropathy in Noonan syndrome with multiple lentigines. J Med Genet 53:123-126.
Coutelier M et al. (2015) Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. Brain 138:2191-2205.
Dalski A, Atici J, Kreuz FR, Hellenbroich Y, Schwinger E, Zuhlke C (2005) Mutation analysis in the *fibroblast growth factor 14* gene: frameshift mutation and polymorphisms in patients with inherited ataxias. . Eur J Human Genet 13:118-120.
Danhauser K et al. (2018) Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. Am J Hum Genet 103:817-825.
de la Chapelle A, al. e (1992) Gelsolin-derived familial amyloidosis caused by asparagine or tyrosine substitution for aspartic acid at residue 187. Nature Genet 2:157-160.
De Michele G, De Fusco M, Cavalcanti F, Filla A, Marconi R, Volpe G, Monticelli A, Ballabio A, Casari G, Cocozza S (1998) A new locus for autosomal recessive hereditary spastic paraplegia maps to chromosome 16q24.3. Am J Hum Genet 63:135-139.

den Boer M, Dionisi-Vici C, Chakrapani A, van Thuijl A, Wanders R, Wijburg F (2003) Mitochondrial trifunctional protein deficiency: a severe fatty acid oxidation disorder with cardiac and neurologic involvement. J Pediatr 142:684–689.

Deodato F, Sabatelli M, Ricci E, Mercuri E, Muntoni F, Sewry C, Naom I, Tonali P, Guzzetta F (2002) Hypermyelinating neuropathy, mental retardation and epilepsy in a case of merosin deficiency. Neuromuscular Disorders 12:392-298.

Dibbens LM, Karakis I, Bayly MA, Costello DJ, Cole AJ, Berkovic SF (2012) Mutation of *SCARB2* in a patient with progressive myoclonus epilepsy and demyelinating peripheral neuropathy. Arch Neurol 68:812-813.

Dimmock DP et al. (2008) Clinical and molecular features of mitochondrial DNA depletion due to mutations in deoxyguanosine kinase. Hum Mutat 29:330-331.

Duman O, Uysal H, Skjei KL, Kizilay F, Karauzum S, Haspolat S (2013) Sensorimotor polyneuropathy in patients with SMA type-1: electroneuromyographic findings. Muscle Nerve 48:117-121.

Echaniz-Laguna A, Ghezzi D, Chassagne M, Mayencon M, Padet S, Melchionda L, Rouvet I, Lannes B, Bozon D, Latour P, Zeviani M, Mousson de Camaret B (2013) *SURF1* deficiency causes demyelinating Charcot-Marie-Tooth disease. Neurology 81:1-8.

El Euch-Fayache G, Bouhlal Y, Amouri R, Feki M, Hentati F (2014) Molecular, clinical and peripheral neuropathy study of Tunisian patients with ataxia with vitamin E deficiency. Brain 137:402-410.

El-Khamisy SF, Saifi GM, Weinfeld M, Helleday T, Lupski JR, Caldecott KW (2005) Defective DNA single-strand break repair in spinocerebellar ataxia with axonal neuropathy-2. Nature 434.

Enns GM et al. (2014) Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulum-associated degradation pathway. Genet Med 16:751-758.

Fadic R, Russell JA, Vedanarayanan VV, Lehar M, Kuncl RW, Johns DR (1997) Sensory ataxic neuropathy as the presenting feature of a novel mitochondrial disease. Neurology 49:239-245.

Feinstein M, al. e (2010) Pelizaeus-Merzbacher-like Disease Caused by AIMP1/p43 Homozygous Mutation. Am J Hum Genet 87:820-828.

Ferdinandusse S, Denis S, Clayton PT, Graham A, Rees JE, Allen JT, McLean BN, Brown AY, Vreken P, Waterham HR, Wanders RJA (2000) Mutations in the gene encoding peroxisomal a-methylacyl-CoA racemase cause adult-onset sensory motor neuropathy. Nat Genet 24:188-191.

Ferdinandusse S, Kostopoulos P, Denis S, Rusch H, Overmars H, Dillmann U, Reith W, Haas D, Wanders RJA, Duran M, Marziniak M (2006) Mutations in the gene encoding peroxisomal sterol carrier protein X (SCPx) cause leukencephalopathy with dystonia and motor neuropathy. Am J Hum Genet 78:1046-1052.

Fiskerstrand T, H'Mida-Ben Brahim D, Johansson S, M'Zahem A, Haukanes BI, Drouot N, Zimmermann J, Cole AJ, Vedeler C, Bredrup C, Assoum M, Tazir M, Klockgether T, Hamri A, Steen VM, Boman H, Bindoff LA, Koenig M, Knappskog PM (2010) Mutations in ABHD12 cause the neurodegenerative disease PHARC: An inborn error of endocannabinoid metabolism. Am J Hum Genet 87:410-417.

Flex E et al. (2016) Biallelic Mutations in TBCD, Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. Am J Hum Genet 99:962-973.

Foley AR et al. (2014) Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2 Brain 137:44-56.

Foubert-Samier A, Kazadi A, Rouanet M, Vital A, Lagueny A, Tison F, Meissner W (2009) Axonal sensory motor neuropathy in copper-deficient Wilson's disease. Muscle Nerve 40:294-296.

Frattini D, Fusco C, Ucchino V, Tavazzi B, DellaGiustina E (2010) Early Onset Methylmalonic Aciduria and Homocystinuria cblC Type With Demyelinating Neuropathy. Pediatr Neurol 43:135-138.
Funfschilling U, Supplie LM, Mahad D, Boretius S, Saab AS, Edgar J, Brinkmann BG, Kassmann CM, Tzvetanova ID, Mobius W, Diaz F, Meijer D, Suter U, Hamprecht B, Sereda MW, Moraes CT, Frahm J, Goebbels S, Nave KA (2012) Glycolytic oligodendrocytes maintain myelin and long-term axonal integrity. Nature 485:517-521.
Furiya Y, Hirano M, Nomura M, Asai H, Kiriyama T, Ueno S (2007) Peripheral neuropathy in chromosome 16q22.1 linked autosomal dominant cerebellar ataxia. J Neurol Neurosurg Psychiat 78:1009-1011.
Gan-Or Z et al. (2016) Mutations in CAPN1 Cause Autosomal-Recessive Hereditary Spastic Paraplegia. Am J Hum Genet 98:1038-1046.
Garbern JY, Cambi F, Tang XM, Sima AAF, Vallat JM, Bosch EP, Lewis R, Shy M, Sohi J, Kraft G, Chen KL, Joshi I, Leonard DGB, Johnson W, Raskind W, Dlouhy SR, Pratt V, Hodes ME, Bird T, Kamholz J (1997) Proteolipid protein is necessary in peripheral as well as central myelin. Neuron 19:205-218.
Garcia-Murias M, Quintans B, Arias M, Seixas AI, Cacheiro P, Tarrio R, Pardo J, Millan MJ, Arias-Rivas S, Blanco-Arias P, Dapena D, Moreira R, Rodriguez-Trelles F, Sequeiros J, Carracedo A, Silveira I, Sobrido MJ (2012) 'Costa da Morte' ataxia is spinocerebellar ataxia 36: clinical and genetic characterization. Brain 135:1423-1435.
Ghezzi D, Sevrioukova I, Invernizzi F, Lamperti C, Mora M, D'Adamo P, Novara F, Zuffardi O, Uziel G, Zeviani M (2010) Severe X-linked mitochondrial encephalomyopathy associated with a mutation in apoptosis-inducing factor. Am J Hum Genet 86:639-649.
Goizet C (2008) Complicated forms of autosomal dominant hereditary spastic paraplegia are frequent in SPG10. Hum Mutat 30:E376-E385.
Goizet C (2009) SPG15 is the second most common cause of hereditary spastic paraplegia with thin corpus callosum. Neurology 73:1111-1119.
Goizet C, Yaou RB, Demay L, Richard P, Bouillot S, Rouanet M, Hermosilla E, G Le Masson, Lagueny A, Bonne G, Ferrer X (2004) A new mutation of the lamin A/C gene leading to autosomal dominant axonal neuropathy, muscular dystrophy, cardiac disease, and leuconychia. J Med Genet 41:e29.
Gonzaga-Jauregui C, Lotze T, Jamal L, Penney S, Campbell IM, Pehlivan D, Hunter JV, Woodbury SL, Raymond G, Adesina AM, Jhangiani SN, Reid JG, Muzny DM, Boerwinkle E, Lupski JR, Gibbs RA, Wiszniewski W (2013) Mutations in VRK1 associated with complex motor and sensory axonal neuropathy plus microcephaly. JAMA neurology 70:1491-1498.
Gonzaga-Jauregui C et al. (2015) Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. Cell reports 12:1169-1183.
Gregory A (2008) Neurodegeneration associated with genetic defects in phospholipase A2. Neurology 71:1402-1479.
Hakamada S, Sobue G, Watanabe K, Kumagai T, Hara K, Miyazaki S (1981) Peripheral neuropathy in Marinesco-Sjögren syndrome. Brain and Development 3:403-406.
Haliloglu G, al. e (2015) Early-onset chronic axonal neuropathy, strokes, and hemolysis - Inherited CD59 deficiency. Neurology 84:1220-1224.
Hall BM, Walsh JC, Horvath JS, Lytton DG (1976) Peripheral neuropathy complicating primary hyperoxaluria. J Neurol Sci 29:343-349.
Hamdan FF et al. (2011) Excess of de novo deleterious mutations in genes associated with glutamatergic systems in nonsyndromic intellectual disability. Am J Hum Genet 88:306-316.
Hammer MB, Eleuch-Fayache G, Schottlaender LV, Nehdi H, Gibbs JR, Arepalli SK, Chong SB, Hernandez DG, Sailer A, Liu G, Mistry PK, Cai H, Shrader G, Sassi C, Bouhlal Y, Houlden H, Hentati F, Amouri R, Singleton AB (2013) Mutations in GBA2 cause autosomal-recessive cerebellar ataxia with spasticity. Am J Hum Genet 92:245-251.

Handley MT et al. (2013) Mutation spectrum in RAB3GAP1, RAB3GAP2, and RAB18 and genotype-phenotype correlations in warburg micro syndrome and Martsolf syndrome. Hum Mutat 34:686-696.

Hanein S, Martin E, Boukhris A, Byrne P, Goizet C, Hamri A, al. e (2008) Identification of the SPG15 gene, encoding spastizin, as a frequent cause of complicated autosomal-recessive spastic paraplegia, including Kjellin syndrome. . Am J Hum Genet 82:992–1002.

Harel T et al. (2016) Recurrent de novo and biallelic variation of *ATAD3A*, encoding a mitochondrial membrane protein, results in distinct neurological syndromes. Am J Hum Genet 99:831-845.

Hartig MB et al. (2011) Absence of an orphan mitochondrial protein, c19orf12, causes a distinct clinical subtype of neurodegeneration with brain iron accumulation. Am J Hum Genet 89:543-550.

Hermans MCE, al. e (2011) Peripheral neuropathy in myotonic dystrophy type 1. J Peripher Nerv Syst 16:24-29.

Higuchi Y et al. (2018) Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. Brain 141:1622-1636.

Hirst J, Madeo M, Smets K, Edgar JR, Schols L, Li J, Yarrow A, Deconinck T, Baets J, Van Aken E, De Bleecker J, Datiles MB, 3rd, Roda RH, Liepert J, Züchner S, Mariotti C, De Jonghe P, Blackstone C, Kruer MC (2016) Complicated spastic paraplegia in patients with AP5Z1 mutations (SPG48). Neurol Genet 2:e98.

Hoch NC, Hanzlikova H, Rulten SL, Tetreault M, Komulainen E, Ju L, Hornyak P, Zeng Z, Gittens W, Rey SA, Staras K, Mancini GM, McKinnon PJ, Wang ZQ, Wagner JD, Care4Rare Canada C, Yoon G, Caldecott KW (2017) XRCC1 mutation is associated with PARP1 hyperactivation and cerebellar ataxia. Nature 541:87-91.

Horga A, Pitceathly RD, Blake JC, Woodward CE, Zapater P, Fratter C, Mudanohwo EE, Plant GT, Houlden H, Sweeney MG, Hanna MG, Reilly MM (2014) Peripheral neuropathy predicts nuclear gene defect in patients with mitochondrial ophthalmoplegia. Brain 137:3200-3212.

Horga A, Bugiardini E, Manole A, Bremner F, Jaunmuktane Z, Dankwa L, Rebelo AP, Woodward CE, Hargreaves IP, Cortese A, Pittman AM, Brandner S, Polke JM, Pitceathly RDS, Züchner S, Hanna MG, Scherer SS, Houlden H, Reilly MM (2019) Autosomal dominant optic atrophy and cataract “plus” phenotype including axonal neuropathy. Neurol Genet (in press).

Horibata Y, Elpeleg O, Eran A, Hirabayashi Y, Savitzki D, Tal G, Mandel H, Sugimoto H (2018) EPT1 (selenoprotein I) is critical for the neural development and maintenance of plasmalogen in humans. J Lipid Res 59:1015-1026.

Horn MA, al. e (2007) Phenotype of adult Refsum disease due to a defect in peroxin 7. Neurology 68:698-700.

Howard HC et al. (2002) The K-Cl cotransporter KCC3 is mutant in a severe peripheral neuropathy associated with agenesis of the corpus callosum. Nat Genet 32:384-392.

Hu H et al. (2014) Mutations in PTRH2 cause novel infantile-onset multisystem disease with intellectual disability, microcephaly, progressive ataxia, and muscle weakness. Ann Clin Transl Neurol 1:1024-1035.

Hudson G, al. e (2008) Mutation of OPA1 causes dominant optic atrophy with external ophthalmoplegia, ataxia, deafness and multiple mitochondrial DNA deletions: a novel disorder of mtDNA maintenance. Brain 131:329–337.

Ibdah JA, Tein I, DionisiVici C, Bennett MJ, Ijlst L, Gibson B, Wanders RJA, Strauss AW (1998) Mild trifunctional protein deficiency is associated with progressive neuropathy and myopathy and suggests a novel genotype-phenotype correlation. J Clin Invest 102:1193-1199.

Ikeda Y, Ohta Y, Kobayashi H, Okamoto M, Takamatsu K, Ota T, Manabe Y, Okamoto K, Koizumi A, Abe K (2012) Clinical features of SCA36: a novel spinocerebellar ataxia with motor neuron involvement (Asidan). Neurology 79:333-341.

Inoue K, Khajavi M, Ohyama T, Hirabayashi S-i, Wilson J, Reggin JD, Mancias P, Butler IJ, Wilkinson MF, Wegner M, Lupski JR (2004) Molecular mechanism for distinct neurological phenotypes conveyed by allelic truncating mutations. Nat Genet 36:361-369.

Ivanova N et al. (2007) Hereditary spastic paraplegia 3A associated with axonal neuropathy. Arch Neurol 64:706-713.
Izumi Y, al. e (2013) Cerebellar ataxia with SYNE1 mutation accompanying motor neuron disease. Neurology 80:600-601.
Jacobs JM, Wilson J (1992) An unusual demyelinating neuropathy in a patient with Waardenburg's syndrome. Acta Neuropathol 83:670-674.
Jacquemont S, al. e (2003) Fragile X premutation tremor/ataxia syndrome: molecular, clinical, and neuroimaging correlates. Am J Hum Genet 72:869-878.
Jaffer F, al. e (2012) BAG3 mutations: another cause of giant axonal neuropathy. J Periph Nerv Syst 17:210-216.
Jain A, Kohli A, Sachan D (2010) Infantile Sandhoff's Disease With Peripheral Neuropathy. Pediatr Neurol 42:459-461.
Jaksch M, Horvath R, Horn N, Auer DP, Macmillan C, Peters J, Gerbitz KD, KraegelohMann I, Muntau A, Karcagi V, Kalmanchey R, Lochmuller H, Shoubridge EA, Freisinger P (2001) Homozygosity (E140K) in SCO2 causes delayed infantile onset of cardiomyopathy and neuropathy. Neurology 57:1440-1446.
Jamieson RV, Perveen R, Kerr B, Carette M, Yardley J, Heon E, Wirth MG, van Heyningen V, Donnai D, Munier F, Black GCM (2002) Domain disruption and mutation of the bZIP transcription factor, *MAF*, assocated with cataract, ocular anteriour segment dysgenesis and coloboma. Hum Mol Genet 11:33-42.
Jansen GA, Hogenhout EM, Ferdinandusse S, Waterham HR, Ofman R, Jakobs C, Skjeldal OH, Wanders RJA (1997) Human phytanoyl-CoA hydroxylase: resolution of the gene structure and the molecular basis of Refsum's disease. Nat Genet 17:190-193.
Jonckheere AI, Hogeveen M, Nijtmans LGJ, vandenBrand MAM, Janssen AJM, Diepstra JHS, vandenBrandt FCA, vandenHeuvel LP, Hol FA, Hofste TGJ, Kapusta L, Dillmann U, Shamdeen MG, Smeitink JAM, Rodenburg RJT (2008) A novel mitochondrial ATP8 gene mutation in a patient with apical hypertrophic cardiomyopathy and neuropathy. J Med Genet 45:129-133.
Jung KH, Ahn TB, Jeon BS (2005) Wilson disease with an initial manifestation of polyneuropathy. Arch Neurol 62:1628-1631.
Kabzińska D, Mierzewska H, Senderek J, Kochański A (2016) Warburg micro syndrome type 1 associated with peripheral neuropathy and cardiomyopathy. Folia Neuropathologica 3:273-281.
Kanda T, Oda M, Yonezawa M, Tamagawa K, Isa F, Hanakago R, Tsukagoshi H (1990) Peripheral neuropathy in xeroderma pigmentosum. Brain 113:1025-1044.
Karaca E et al. (2014) Human CLP1 mutations alter tRNA biogenesis, affecting both peripheral and central nervous system function. Cell 157:636-650.
Karadimas CL, Vu TH, Holve SA, Chronopoulou P, Quinzii C, Johnsen SD, Kurth J, Eggers E, Palenzuela L, Tanji K, Bonilla E, DeVivo DC, DiMauro S, Hirano M (2006) Navajo neurohepatopathy is caused by a mutation in the MPV17 gene. Am J Hum Genet 79:544-548.
Klebe S, Azzedine H, Durr A, Bastien P, Bouslam N, Elleuch N, Forlani S, Charon C, Koenig M, Melki J, Brice A, Stevanin G (2006) Autosomal recessive spastic paraplegia (SPG30) with mild ataxia and sensory neuropathy maps to chromosome 2q37.3. Brain 129:1456-1462.
Klebe S et al. (2012) KIF1A missense mutations in SPG30, an autosomal recessive spastic paraplegia: distinct phenotypes according to the nature of the mutations. Eur J Hum Genet 20:645-649.
Klein CJ, Boes CJ, Chapin JE, Lynch CD, Campeau NG, Dyck PJB, Dyck PJ (2004) Adult polyglucosan body disease: Case description of an expanding genetic and clinical syndrome. Muscle Nerve 29:323-328.
Kleopa KA, Raizen DM, Friedrich CA, Brown MJ, Bird SJ (2001) Acute axonal neuropathy in maple syrup urine disease. Muscle Nerve 24:284-287.
Knierim E, Gill E, Seifert F, Morales-Gonzalez S, Unudurthi SD, Hund TJ, Stenzel W, Schuelke M (2017) A recessive mutation in beta-IV-spectrin (SPTBN4) associates with congenital myopathy, neuropathy, and central deafness. Hum Genet 136:903-910.

Kobayashi H, Abe K, Matsuura T, Ikeda Y, Hitomi T, Akechi Y, Habu T, Liu W, Okuda H, Koizumi A (2011) Expansion of intronic GGCCTG hexanucleotide repeat in NOP56 causes SCA36, a type of spinocerebellar ataxia accompanied by motor neuron involvement. Am J Hum Genet 89:121-130.

Koch J, Feichtinger RG, Freisinger P, Pies M, Schrodl F, Iuso A, Sperl W, Mayr JA, Prokisch H, Haack TB (2016) Disturbed mitochondrial and peroxisomal dynamics due to loss of MFF causes Leigh-like encephalopathy, optic atrophy and peripheral neuropathy. J Med Genet 53:270-278.

Koutsopoulos OS, Kretz C, Weller CM, Roux A, Mojzisova H, Bohm J, Koch C, Toussaint A, Heckel E, Stemkens D, Ter Horst SA, Thibault C, Koch M, Mehdi SQ, Bijlsma EK, Mandel JL, Vermot J, Laporte J (2013) Dynamin 2 homozygous mutation in humans with a lethal congenital syndrome. Eur J Hum Genet 21:637-642.

Kuhlenbaumer G, Young P, Oberwittler C, Hunermund G, Schirmacher A, Domschke K, Ringelstein B, Stogbauer F (2002) Giant axonal neuropathy (GAN): Case report and two novel mutations in the gigaxonin gene. Neurology 58:1273-1276.

Laquerriere A et al. (2014) Mutations in CNTNAP1 and ADCY6 are responsible for severe arthrogryposis multiplex congenita with axoglial defects. Hum Mol Genet 23:2279-2289.

Lee JR et al. (2015) De novo mutations in the motor domain of KIF1A cause cognitive impairment, spastic paraparesis, axonal neuropathy, and cerebellar atrophy. Hum Mutat 36:69-78.

Lee JYW et al. (2017) Large Intragenic Deletion in DSTYK Underlies Autosomal-Recessive Complicated Spastic Paraparesis, SPG23. Am J Hum Genet 100:364-370.

Lehmann D, al. e (2016) Peripheral neuropathy in patients with CPEO associated with singe and multiple mtDNA deletions. Neurol Genet 2:e113.

Liberfarb RM, Jackson AH, Eavey RD, Robb RM (1993) Unique hereditary sensory and autonomic neuropathy with growth hormone deficiency. J Child Neurol 8:271-276.

Lines MA, al. e (2014) Peroxisomal D-bifunctional protein deficiency: three adults diagnosed by whole-exome sequencing. Neurology 82:963-968.

Liu YT, Laura M, Hersheson J, Horga A, Jaunmuktane Z, Brandner S, Pittman A, Hughes D, Polke JM, Sweeney MG, Proukakis C, Janssen JC, Auer-Grumbach M, Zuchner S, Shields KG, Reilly MM, Houlden H (2014) Extended phenotypic spectrum of KIF5A mutations: From spastic paraplegia to axonal neuropathy. Neurology 83:612-619.

Lossos A et al. (2015) Myelin-associated glycoprotein gene mutation causes Pelizaeus-Merzbacher disease-like disorder. Brain 138:2521-2536.

Macedo-Souza LI, Kok F, Santos S, Amorim SC, Starling A, Nishimura A, Lezirovitz K, Lino AMM, Zatz M (2005) Spastic paraplegia, optic atrophy, and neuropathy is linked to chromosome 11q13. Ann Neurol 57:730-737.

Macedo-Souza LI, Kok F, Santos S, Licinio L, Lezirovitz K, Cavacana N, Bueno C, Amorim S, Pessoa A, Graciani Z, Ferreira A, Prazeres A, de Melo AN, Otto PA, Zatz M (2009) Spastic paraplegia, optic atrophy, and neuropathy: new observations, locus refinement, and exclusion of candidate genes. Annals of Human Genetics 73:382-387.

Malandrini A, Fabrizi GM, Truschi F, Di Pietro G, Moschini F, Bartalucci P, Berti G, Salvadori C, Bucalossi A, Guazzi G (1994) Atypical McLeod syndrome manifested as X-linked chorea-acanthocytosis, neuromyopathy and dilated cardiomyopathy: report of a family. J Neurol Sci 124:89-94.

Maluenda J, Manso C, Quevarec L, Vivanti A, Marguet F, Gonzales M, Guimiot F, Petit F, Toutain A, Whalen S, Grigorescu R, Coeslier AD, Gut M, Gut I, Laquerriere A, Devaux J, Melki J (2016) Mutations in GLDN, Encoding Gliomedin, a Critical Component of the Nodes of Ranvier, Are Responsible for Lethal Arthrogryposis. Am J Hum Genet.

Marsh APL, Lukic V, Pope K, Bromhead C, Tankard R, Ryan MM, Yiu EM, Sim JCH, Delatycki MB, Amor DJ, McGillivray G, Sherr EH, Bahlo M, Leventer RJ, Lockhart PJ (2015) Complete callosal agenesis, pontocerebellar hypoplasia, and axonal neuropathy due to AMPD2 loss. Neurology: Genetics 1:e16-e16.

Marti R, al. e (2005) Late-Onset MNGIE due to partial loss thymidine phosphorylase activity. . Ann Neurol 58:649-652.

Martin E et al. (2013) Loss of function of glucocerebrosidase GBA2 is responsible for motor neuron defects in hereditary spastic paraplegia. Am J Hum Genet 92:238-244.

Martinelli D, Travaglini L, Drouin CA, Ceballos-Picot I, Rizza T, Bertini E, Carrozzo R, Petrini S, de Lonlay P, El Hachem M, Hubert L, Montpetit A, Torre G, Dionisi-Vici C (2013) MEDNIK syndrome: a novel defect of copper metabolism treatable by zinc acetate therapy. Brain 136:872-881.

McDermott CJ, Dayaratne RK, Tomkins J, Lusher ME, Lindsey JC, Johnson MA, Casari G, Turnbull DM, Bushby K, Shaw PJ (2001) Paraplegin gene analysis in hereditary spastic paraparesis (HSP) pedigrees in northeast England. Neurology 56:467-471.

McMillan HJ, al. e (2012) Specific combination of compound heterozygous mutations in 17b-hydroxysteroid dehydrogenase type 4 (HSD17B4) defines a new subtype of C-bifunctional protein deficiency. Orphanet J Rare Dis 7:90.

McMillin MJ et al. (2013) Mutations in ECEL1 cause distal arthrogryposis type 5D. Am J Hum Genet 92:150-156.

Mead S et al. (2013) A Novel Prion Disease Associated with Diarrhea and Autonomic Neuropathy. N Engl J Med 369:1904-1914.

Mier M, Schwartz SO, Boshes B (1960) Acanthrocytosis, Pigmentary Degeneration of the Retina and Ataxic Neuropathy: A Genetically Determined Syndrome with Associated Metabolic Disorder. Blood 16:1586-1608.

Migliaccio AA, Halmagyi GM, McGarvie LA, Cremer PD (2004) Cerebellar ataxia with bilateral vestibulopathy: description of a syndrome and its characteristic clinical sign. Brain 127:280-293.

Mihalik SJ, Morrell JCD, Sacksteder KA, Watkins PA, Gould SJ (1997) Identification of PAHX, a Refsum disease gene. Nat Genet 17:185-189.

Mitsumoto H, al. e (1985) Motor neuron disease and adult hexosaminase A deficiency in tow familes: evidence for multisystem degeneration. Ann Neurol 17:378-385.

Miyake N et al. (2016) Biallelic TBCD Mutations Cause Early-Onset Neurodegenerative Encephalopathy. Am J Hum Genet 99:950-961.

Mochizuki A, Motoyoshi Y, Takeuchi M, Sonoo M, Shimizu T (2000) A case of adult type galactosialidosis with involvement of peripheral nerves. J Neurol 247:708-710.

Moorhead PJ, Cooper DJ, Timperley WR (1975) Progressive peripheral neuropathy in patient with priary hyperoxaluria. Br Med J 2.

Moreira M-C, al. e (2004) Senataxin, the orthologue of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. Nat Genet 36:225-227.

Morgan NV, al. e (2006) PLA2G6, encoding a phospholipase A2, is mutated in neurodegenerative disorders with high brain iron. Nat Genet 38:752-754.

Morino H, al. e (2014) Mutations in Twinkle primase-helicase cause Perrault syndrome with neurologic features. Neurology 83:2054-2061.

Mukamel M, Weitz R, Metzker A, Varsano I (1985) Spastic paraparesis, mental retardation, and cutaneous pigmentation disorder. Arch Pediatr 139:1090-1092.

Nahorski MS, Al-Gazali L, Hertecant J, Owen DJ, Borner GH, Chen YC, Benn CL, Carvalho OP, Shaikh SS, Phelan A, Robinson MS, Royle SJ, Woods CG (2015) A novel disorder reveals clathrin heavy chain-22 is essential for human pain and touch development. Brain 138:2147-2160.

Narkis G, Ofir R, Landau D, Manor E, Volokita M, Hershkowitz R, Elbedour K, Birk OS (2007) Lethal contractural syndrome type 3 (LCCS3) is caused by a mutation
in PIP5K1C, which encodes PIPKIg of the phophatidylinsitol pathway. Am J Hum Genet 81:530-539.
Nasca A, Legati A, Baruffini E, Nolli C, Moroni I, Ardissone A, Goffrini P, Ghezzi D (2016) Biallelic Mutations in DNM1L are Associated with a Slowly Progressive Infantile Encephalopathy. Hum Mutat 37:898-903.
Nichols WC, Benson MD (1990) Hereditary amyloidosis: detection of variant prealbumin genes by restriction enzyme analysis of amplified genomic DNA sequences. Clin Genet 37:44-53.
Nizon M, Kury S, Pereon Y, Besnard T, Quinquis D, Boisseau P, Marsaud T, Magot A, Mussini JM, Mayrargue E, Barbarot S, Bezieau S, Isidor B (2017) ARL6IP1 mutation causes congenital insensivity to pain, self-mutilation and spastic paraplegia. Clin Genet.
Novarino G et al. (2014) Exome sequencing links corticospinal motor neuron disease to common neurodegenerative disorders. Science 343:506-511.
Orlacchio A, Patrono C, Gaudiello F, Rocchi c, Moschella V, Floris R, Bernardi G, Kawarai T (2008) Silver syndrome variant of hereditary spastic paraplegia. Neurology 70.
Panza E, Escamilla-Honrubia JM, Marco-Marin C, Gougeard N, De Michele G, Morra VB, Liguori R, Salviati L, Donati MA, Cusano R, Pippucci T, Ravazzolo R, Nemeth AH, Smithson S, Davies S, Hurst JA, Bordo D, Rubio V, Seri M (2016) ALDH18A1 gene mutations cause dominant spastic paraplegia SPG9: loss of function effect and plausibility of a dominant negative mechanism. Brain 139:e3.
Park MH, Woo HM, Hong YB, Park JH, Yoon BR, Park JM, Yoo JH, Koo H, Chae JH, Chung KW, Choi BO, Koo SK (2014) Recessive C10orf2 mutations in a family with infantile-onset spinocerebellar ataxia, sensorimotor polyneuropathy, and myopathy. Neurogenetics 15:171-182.
Patel H, al. e (2002) SPG20 is mutated in Troyer syndrome, an hereditary spastic paraplegia. Nature Genet 31:347-348.
Patel KP, O'Brien TW, Subramony SH, Shuster J, Stacpoole PW (2012) The spectrum of pyruvate dehydrogenase complex deficiency: clinical, biochemical and genetic features in 371 patients. Molecular Genetics and Metabolism 105:34-43.
Peltola KE, Jaaskelainen S, Heinonen OJ, Falck B, Nanto-Salonen K, Heinanen K, Simell O (2002) Peripheral nervous system in gyrate atrophy of the choroid and retina with hyperornithinemia. Neurology 59:735-740.
Pierson TM, Simeonov DR, Sincan M, Adams DA, Markello T, Golas G, Fuentes-Fajardo K, Hansen NF, Cherukuri PF, Cruz P, Mullikin JC, Blackstone C, Tifft C, Boerkoel CF, Gahl WA, Program NCS (2012) Exome sequencing and SNP analysis detect novel compound heterozygosity in fatty acid hydroxylase-associated neurodegeneration. Eur J Hum Genet 20:476-479.
Pierson TM et al. (2011) Whole-exome sequencing identifies homozygous AFG3L2 mutations in a spastic ataxia-neuropathy syndrome linked to mitochondrial m-AAA proteases. PLoS Genet 7:e1002325.
Pitceathly RD et al. (2013) COX10 Mutations Resulting in Complex Multisystem Mitochondrial Disease That Remains Stable Into Adulthood. JAMA neurology.
Pitceathly RD et al. (2012) Genetic dysfunction of MT-ATP6 causes axonal Charcot-Marie-Tooth disease. Neurology 79:1145-1154.
Pollock M, Nukada H, Frith RW, Simcock JP, Allpress S (1983) Peripheral neuropathy in Tangier disease. Brain 106:911-928.
Potocki L, Chen K-S, Koeuth T, Killian J, Iannaccone ST, Shapira SK, Kashork CD, Spikes AS, Shaffer LG, Lupski JR (1999) DNA rearrangements on both homologues of chromosome 17 in a mildly delayed individual with a family history of autosomal dominant carpal tunnel syndrome. Am J Hum Genet 64:471-478.

Potocki L, Chen K-S, Park S-S, Osterhom DE, Withers MA, Kimonis V, Summers, Meschino WS, Anyane-Yeboa K, Kashork CD, Shaffer LG, Lupski JR (2000) Molecular mechanism for duplication 17p11.2 - the homologous recombination reciprocal of the Smith-Magenis microdeletion. Nat Genet 24:84-87.

Poulton C, Oegema R, Heijsman D, Hoogeboom J, Schot R, Stroink H, Willemsen MA, Verheijen FW, van de Spek P, Kremer A, Mancini GM (2013) Progressive cerebellar atrophy and polyneuropathy: expanding the spectrum of PNKP mutations. Neurogenetics.

Powell HC, al. e (1985) Peripheral nerve in type III glycogenosis: selective involvement of unmyelinated fiber Schwann cells. Muscle Nerve 8:667-671.

Rainier S, Albers JW, Dyck PJ, Eldevik OP, Wilcock S, Richardson RJ, Fink JK (2011) Motor neuron disease due to neuropathy target esterase gene mutation: Clinical features of the index families Muscle Nerve 43:19-25.

Rainier S, Bui M, Mark E, Thornas D, Tokarz D, Ming L, Delaney C, Richardson RJ, Albers JW, Matsunami N, Stevens J, Coon H, Leppert M, Fink JK (2008) Neuropathy target esterase gene mutations cause motor neuron disease. Am J Hum Genet 82:780-785.

Rajadhyaksha AM, al. e (2010) Mutations in FLVCR1 cause posterior column ataxia and retinitis pigmentosa. Am J Hum Genet 87:643-654.

Rasmussen A, al. e (2001) Clinical and Genetic Analysis of Four Mexican Families With Spinocerebellar Ataxia Type 10. Ann Neurol 50:234-239.

Ravenscroft G et al. (2015) Mutations of GPR126 Are Responsible for Severe Arthrogryposis Multiplex Congenita. Am J Hum Genet 96:955-961.

Regal L, al. e (2010) Mutations in PEX10 Are a Cause of Autosomal Recessive Ataxia. Ann Neurol 68:259-263.

Reid E, Kloos M, Ashley-Koch A, Hughes L, Bevan S, Svenson IK, Lennon F, Gaskell PC, Dearlove A, Pericak-Vance MA, Rubinsztein DC, Marchuk DA (2002) A kinesin heavy chain (KIF5A) mutation in hereditary spastic paraplegia (SPG10). Am J Hum Genet 71:1189-1194.

Remaley AT, Rust S, Rosier M, Knapper C, Naudin L, Broccardo C, Peterson KM, Koch C, Arnould I, Prades C, Duverger N, Funke H, Assman G, Dinger M, Dean M, Chimini G, Santamarina-Fojo S, Fredrickson DS, Denefle P, Brewer HB, Jr. (1999) Human ATP-binding cassette transporter 1 (ABC1): Genomic organization and identification of the genetic defect in the original Tangier disease kindred. Proc Natl Acad Sci USA 96:12685-12690.

Reuber BE, Germain-Lee E, Collins CS, Morrell JC, Ameritunga R, Moser HW, Valle D, Gould SJ (1997) Mutations in PEX1 are the most common cause of peroxisome biogenesis disorders. Nat Genet 17:445-448.

Ries M, Ramaswami U, Parini R, Lindblad B, Whybra C, Willers I, Gal A, Beck M (2003) The early clinical phenotype of Fabry disease: a study on 35 European children and adolescents. European Journal of Pediatrics 162:767-772.

Rinaldi C, Grunseich C, Sevrioukova IF, Schindler A, Horkayne-Szakaly I, Lamperti C, Landoure G, Kennerson ML, Burnett BG, Bonnemann C, Biesecker LG, Ghezzi D, Zeviani M, Fischbeck KH (2012) Cowchock syndrome is associated with a mutation in apoptosis-inducing factor. Am J Hum Genet 91:1095-1102.

Roda RH, FitzGibbon EJ, Boucekkine H, Schindler AB, Blackstone C (2016) Neurologic syndrome associated with homozygous mutation at MAG sialic acid binding site. Ann Clin Transl Neurol 3:650-654.

Roze E, al. e (2003) Neuropsychiatric disturbance in presumed late-onset cobalamin C disease. Arch Neurol 60:1457-1462.

Rudnick-Schoneborn S, Goebel HH, Schlote W, Molaian S, Omran H, Keletsen U, Korinthenberg R, Wenzel D, Lauffer H, Kreib-Nachtsheim M, Wirth B, Zerres K (2003) Classical infantile spinal muscular atrophy with SMN deficiency causes sensory neuronopathy. Neurology 60:983-987.

Saba TG, al. e (2005) An atypical form of erythrokeratodermia variabilis maps to chromoseome 7q22. Hum Genet 116:167-171.

Said G, Lacroix C, Plante-Bordeneuve V, Messing B, Slama A, Crenn P, Nivelon-Chevallier A, Bedenne L, Soichot P, Manceau E, Rigaud D, Guiochon-Mantel A, Matuchansky C (2005) Clinicopathological aspects of the neuropathy of neurogastrointestinal encephalomyopathy (MNGIE) in four patients including two with a Charcot-Marie-Tooth presentation. J Neurol 252:655-662.

Santoro L, Carrozzo R, Malandrini A, Piemonte F, Patrono C, Villanova M, Tessa A, Palmeri S, Bertini E, Santorelli FM (2000) A novel SURF1 mutation results in Leigh syndrome with peripheral neuropathy caused by cytochrome c oxidase deficiency. Neuromuscul Disord 10:450–453.

Scaioli V, DArrigo S, Pantaleoni C (2004) Unusual neurophysiological features in Cockayne's syndrome: a report of two cases as a contribution to diagnosis and classification. Brain Develop 26:273-280.

Scarano V, Mancini P, Criscuolo C, DeMichele G, Rinaldi C, Tucci T, Tessa A, Santorelli FM, Perretti A, Santoro L, Filla A (2005) The R495W mutation in SPG3A causes spastic paraplegia associated with axonal neuropathy. J Neurol 252:901-903.

Schaffer AE et al. (2014) CLP1 founder mutation links tRNA splicing and maturation to cerebellar development and neurodegeneration. Cell 157:651-663.

Schmidt WM, Rutledge SL, Schule R, Mayerhofer B, Züchner S, Boltshauser E, Bittner RE (2015) Disruptive SCYL1 Mutations Underlie a Syndrome Characterized by Recurrent Episodes of Liver Failure, Peripheral Neuropathy, Cerebellar Atrophy, and Ataxia. Am J Hum Genet 97:855-861.

Schottmann G, Stenzel W, Lutzkendorf S, Schuelke M, Knierim E (2014) A novel frameshift mutation of C19ORF12 causes NBIA4 with cerebellar atrophy and manifests with severe peripheral motor axonal neuropathy. Clin Genet 85:290-292.

Schule R, Bonin M, Durr A, Forlani S, Sperfeld A-D, Klimpe S, Mueller JC, seibel A, van de Warrenburg BPC, Bauer P, Schols L (2009) Autosomal dominant spastic paraplegia with peripheral neuropathy maps to chr12q23-24. Neurology 72:1893-1898.

Schwartzentruber J, Buhas D, Majewski J, Sasarman F, Papillon-Cavanagh S, Thiffault I, Sheldon KM, Massicotte C, Patry L, Simon M, Zare AS, McKernan KJ, Consortium FC, Michaud J, Boles RG, Deal CL, Desilets V, Shoubridge EA, Samuels ME (2014) Mutation in the nuclear-encoded mitochondrial isoleucyl-tRNA synthetase IARS2 in patients with cataracts, growth hormone deficiency with short stature, partial sensorineural deafness, and peripheral neuropathy or with Leigh syndrome. Hum Mutat 35:1285-1289.

Selcen D, Engel AG (2003) Myofibrillar myopathy caused by novel dominant negative ab-crystallin mutations. Ann Neurol 54:804-810.

Selcen D, Engel AG (2004) Mutations in myotilin cause myofibrillar myopathy. Neurology 62:1363-1371.

Selcen D, Engel AG (2005) Mutations in ZASP define a novel form of muscular dystrophy in humans. Ann Neurol 57:269-276.

Selcen D, Muntoni F, Burton BK, Pegoraro E, Sewry C, Bite AV, Engel AG (2009) Mutation in BAG3 causes severe dominant childhood muscular dystrophy. Ann Neurol 65:83-89.

Semmler AL et al. (2014) Unusual multisystemic involvement and a novel BAG3 mutation revealed by NGS screening in a large cohort of myofibrillar myopathies. Orphanet journal of rare diseases 9:121.

Seri M, Cusano R, Forabosco P, Cinti R, Caroli F, Picco P, Bini R, Morra VB, De Michele G, Lerone M, Silengo M, Pela I, Borrone C, Romeo G, Devoto M (1999) Genetic mapping to 10q23.3-q24.2, in a large Italian pedigree, of a new syndrome showing bilateral cataracts, gastroesophageal reflux, and spastic paraparesis with amyotrophy. Am J Hum Genet 64:586-593.

Sferra A, Fattori F, Rizza T, Flex E, Bellacchio E, Bruselles A, Petrini S, Cecchetti S, Teson M, Restaldi F, Ciolfi A, Santorelli FM, Zanni G, Barresi S, Castiglioni C, Tartaglia M, Bertini E (2018) Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. Hum Mol Genet 27:1892-1904.

Sferra A et al. (2016) *TBCE* mutations cause early-onset progressive encephalopathy with distal spinal muscular atrophy. Am J Hum Genet 99:974-983.

Shaibani A, al. e (2009) Mitochondrial Neurogastrointestinal Encephalopathy Due to Mutations in RRM2B. Arch Neurol 66:1028-1032.

Shimazaki H, Takiyama Y, Ishiura H, Sakai C, Matsushima Y, Hatakeyama H, Honda J, Sakoe K, Naoi T, Namekawa M, Fukuda Y, Takahashi Y, Goto J, Tsuji S, Goto Y, Nakano I (2012) A homozygous mutation of C12orf65 causes spastic paraplegia with optic atrophy and neuropathy (SPG55). J Med Genet 49:777-784.

Slavotinek AM, Pike M, Mills K, Hurst JA (1996) Cataracts, motor system disorder, short stature, learning difficulties, and skeletal abnormalities: a new syndrome? . Am J Med Genet 62:42-47.

Sperfeld A-D, Hein C, Schroder JM, Ludolph AC, Hanemann CO (2002) Occurrence and characterization of peripheral nerve involvement in neurofibromatosis type 2. Brain 125:996-1004.

Spiegel R, Shaag A, Edvardson S, Mandel H, Stepensky P, Shalev SA, Horovitz Y, Pines O, Elpeleg O (2009) SLC25A19 mutation as a cause of neuropathy and bilateral striatal necrosis. Ann Neurol 66:419-424.

Sprecher E, IshidaYamamoto A, MizrahiKoren M, Rapaport D, Goldsher D, Indelman M, Topaz O, Chefetz I, Keren H, OBrien TJ, Bercovich D, Shalev S, Geiger D, Bergman R, Horowitz M, Mandel H (2005) A mutation in SNAP29, coding for a SNARE protein involved in intracellular trafficking, causes a novel neurocutaneous syndrome characterized by cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma. Am J Hum Genet 77:242-251.

Stevanin G, al. e (2007) Mutations in SPG11, encoding spatacsin, are a major cause of spastic paraplegia with thin corpus callosum. Nat Genet 39:366-.

Stevanin G, Bouslam N, Thobois S, Azzedine H, Ravaux L, Boland A, Schalling M, Broussolle E, Durr A, Brice A (2004) Spinocerebellar ataxia with sensory neuropathy (SCA25) maps to chromosome 2p. Ann Neurol 55:97-104.

Sunada Y (1993) Inherited amyloid polyneuropathy type IV (gelsolin variant) in a Japanese family Ann Neurol 33:57-62.

Swartz BE, Li S, Bespalova I, Burmeister M, Dulaney E, Robinson FR, Leight RJ (2003) Pathogenesis of cliinical signs in recessive ataxia with saccadic intrusions. Ann Neurol 54:824-828.

Synofzik M, Haack TB, Kopajtich R, Gorza M, Rapaport D, Greiner M, Schonfeld C, Freiberg C, Schorr S, Holl RW, Gonzalez MA, Fritsche A, Fallier-Becker P, Zimmermann R, Strom TM, Meitinger T, Zuchner S, Schule R, Schols L, Prokisch H (2014) Absence of BiP Co-chaperone DNAJC3 Causes Diabetes Mellitus and Multisystemic Neurodegeneration. Am J Hum Genet 95:689-697.

Szmulewicz DJ, Waterston JA, Halmagyi GM, Mossman S, Chancellor AM, McLean CA, Storey E (2011a) Sensory neuropathy as part of the cerebellar ataxia neuropathy vestibular areflexia syndrome. Neurology 76:1903-1910.

Szmulewicz DJ, Waterston JA, MacDougall HG, Mossman S, Chancellor AM, McLean CA, Merchant S, Patrikios P, Halmagyi GM, Storey E (2011b) Cerebellar ataxia, neuropathy, vestibular areflexia syndrome (CANVAS): a review of the clinical features and video-oculographic diagnosis. Annals of the New York Academy of Sciences 1233:139-147.

Takashima H, Boerkoel CF, John J, Saifi GM, Salih MAM, Armstrong D, Mao Y, Quiocho FA, Roa BB, Nakagawa M, Stockton DW, Lupski JR (2002) Mutation of TDP1, encoding a topoisomerase 1-dependent DNA damage repair enzyme, in spinocerebellar ataxia with axonal neuropathy. Nature Genet 32:267-272.

Tan EK, Wong MC, Ng I, Teo SH, Lo YL, Cho MM (1998) Unusual pure motor axonal neuropathy in a Burmese family with galactosialidosis. J Inherit Metab Dis 21:869-870.

Tardieu M, Lacroix C, Neven B, Bordigoni P, de Saint Basile G, Blanche S, Fischer A (2005) Progressive neurologic dysfunctions 20 years after allogeneic bone marrow transplantation for Chediak-Higashi syndrome. Blood 106:40-42.

Tata B, Huijbregts L, Jacquier S, Csaba Z, Genin E, Meyer V, Leka S, Dupont J, Charles P, Chevenne D, Carel JC, Leger J, de Roux N (2014) Haploinsufficiency of Dmxl2, encoding a synaptic protein, causes infertility associated with a loss of GnRH neurons in mouse. PLoS Biol 12:e1001952.

Tesson C et al. (2012a) Alteration of fatty-acid-metabolizing enzymes affects mitochondrial form and function in hereditary spastic paraplegia. Am J Hum Genet 91:1051-1064.

Tesson C et al. (2012b) Alteration of fatty-acid-metabolizing enzymes affects mitochondrial form and function in hereditary spastic paraplegia. Am J Hum Genet 91:1051-1064.

Thyagarajan D, Bressman S, Bruno C, Przedborski S, Shanske S, Lynch T, Fahn S, DiMauro S (2000) A novel mitochondrial 12SrRNA point mutation in parkinsonism, deafness, and neuropathy. Ann Neurol 48:730-736.

Tournev I et al. (1999) Congenital cataracts facial dysmorphism neuropathy syndrome, a novel complex genetic disease in Balkan gypsies: Clinical and electrophysiological observations. Ann Neurol 45:742-750.

Tucci A, Liu YT, Preza E, Pitceathly RD, Chalasani A, Plagnol V, Land JM, Trabzuni D, Ryten M, Ukbec, Jaunmuktane Z, Reilly MM, Brandner S, Hargreaves I, Hardy J, Singleton AB, Abramov AY, Houlden H (2014) Novel C12orf65 mutations in patients with axonal neuropathy and optic atrophy. J Neurol Neurosurg Psychiatry 85:486-492.

Ugawa Y, Inoue K, Takemura T, Iwamasa T (1986) Accumulation of glycogen in sural nerve axons in adult-onset type III glycogenosis. Ann Neurol 19:294-297.

Umehara F, Matsumuro K, Kurono Y, Arimura K, Osame M, Kanzaki T (2004) Neurologic manifestations of Kanzaki disease. Neurology 62:1604-1606.

Umehara F, Tate G, Itoh K, Yamaguchi N, Douchi T, Mitsuya T, Osame M (2000) A novel mutation of desert hedgehog in a patient with 46,XY partial gonadal dysgenesis accompanied by minifascicular neuropathy. Am J Hum Genet 67:1302-1305.

Valenzise M, Aversa T, Salzano G, Zirilli G, De Luca F, Su M (2017) Novel insight into Chronic Inflammatory Demyelinating Polineuropathy in APECED syndrome: molecular mechanisms and clinical implications in children. Ital J Pediatr 43:11.

Vallat JM, Nizon M, Magee A, Isidor B, Magy L, Pereon Y, Richard L, Ouvrier R, Cogne B, Devaux J, Züchner S, Mathis S (2016) Contactin-Associated Protein 1 (CNTNAP1) Mutations Induce Characteristic Lesions of the Paranodal Region. J Neuropathol Exp Neurol.

Valleix S, al. e (2012) Hereditary Systemic Amyloidosis Due to Asp76Asn Variant β2-Microglobulin. N Engl J Med 366:2276-2283.

van de Warrenburg BPC, Notermans NC, Schelhaas HJ, van Alfen N, Sinke RJ, Knoers NVAM, Zwarts MJ, Kremer BPH (2004) Peripheral nerve involvement in spinocerebellar ataxias. Arch Neurol 61:257-261.

Van Swieten JC, al. e (2003) A mutation in the *fibroblast growth factor 14* gene is associated with autosomal dominant cerebellar ataxia. Am J Hum Genet 72:191-199.

Varon R et al. (2003) Partial deficiency of the C-terminal-domain phosphatase of RNA polymerase II is associated with congenital cataracts facial dysmorphism neuropathy syndrome. Nat Genet 35:185-189.

Vazza G, Zortea M, Boaretto F, Micaglio GF, Sartori V, Mostacciuolo ML (2000) A new locus for autosomal recessive spastic paraplegia associated with mental retardation and distal motor neuropathy, SPG14, maps to chromosome 3q27-q28. Am J Hum Genet 67:504-509.

Verbeek DS, van de Warrenburg BP, Wesseling P, Pearson PL, Kremer HP, Sinke RJ (2004) Mapping of the SCA23 locus involved in autosomal dominant cerebellar ataxia to chromosome region 20p13-12.3. Brain 127:2551-2557.

Verheij JB, Sival DA, van der Hoeven JH, Vos YJ, Meiners LC, Brouwer OF, van Essen AJ (2006) Shah-Waardenburg syndrome and PCWH associated with SOX10 mutations: a case report and review of the literature. Eur J Paediatr Neurol 10:11-17.

Voermans NC, Bonnemann CG, Lammens M, vanEngelen BG, Hamel BCJ (2009) Myopathy and Polyneuropathy in an Adolescent With the Kyphoscoliotic Type of Ehlers-Danlos Syndrome. Am J Med Genet 149A:2311-2316.

Vorgerd M, van der Ven PF, Bruchertseifer V, Lowe T, Kley RA, Schroder R, Lochmuller H, Himmel M, Koehler K, Furst DO, Huebner A (2005) A mutation in the dimerization domain of filamin c causes a novel type of autosomal dominant myofibrillar myopathy. Am J Hum Genet 77:297-304.

Wada M, Kimura M, Daimon M, Kurita K, Kato T, Johmura Y, Johkura K, Kuroiwa Y, Sobue G (2003) An unusual phenotype of McLeod syndrome with late onset axonal neuropathy. J Neurol Neurosurg Psychiat 74:1697-1698.

Walter MC, Bernert G, Zimmermann U, Mullner-Eidenbock A, Moser E, Kalaydjieva L, Lochmuller H, Muller-Felber W (2014) Long-term follow-up in patients with CCFDN syndrome. Neurology 83:1337-1344.

Wang AM, Schindler D, Desnick R (1990) Schindler disease: the molecular lesion in the alpha-N-acetylgalactosaminidase gene that causes an infantile neuroaxonal dystrophy. J Clin Invest 86:1752-1756.

Wang Z, Hong D, Zhang W, Li W, Shi X, Zhao D, Yang X, Lv H, Yuan Y (2016) Severe sensory neuropathy in patients with adult-onset multiple acyl-CoA dehydrogenase deficiency. Neuromuscul Disord 26:170-175.

Wende H, Lechner SG, Cheret C, Bourane S, Kolanczyk ME, Pattyn A, Reuter K, Munier FL, Carroll P, Lewin GR, Birchmeier C (2012) The transcription factor c-Maf controls touch receptor development and function. Science 335:1373-1376.

Windpassinger C, Auer-Grumbach M, Irobi J, Patel H, Petek E, Horl G, Malli R, Reed JA, Dierick I, Verpoorten N, Warner TT, Proukakis C, Van den Bergh P, Verellen C, Van Maldergem L, Merlini L, De Jonghe P, Timmerman V, Crosby AH, Wagner K (2004) Heterozygous missense mutations in BSCL2 are associated with distal hereditary motor neuropathy and Silver syndrome. Nat Genet 36:271-276.

Winner B, Uyanik G, Gross C, Lange M, SchulteMattler W, Schuierer G, Marienhagen J, Hehr U, Winkler J (2004) Clinical progression and genetic analysis in hereditary spastic paraplegia with thin corpus callosum in spastic gait gene 11 (SPG11). Arch Neurol 61:117-121.

Xue S et al. (2017) Loss-of-Function Mutations in LGI4, a Secreted Ligand Involved in Schwann Cell Myelination, Are Responsible for Arthrogryposis Multiplex Congenita. Am J Hum Genet 100:659-665.

Ylikallio E et al. (2017) MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain.

Yoon G, Malam Z, Paton T, Marshall CR, Hyatt E, Ivakine Z, Scherer SW, Lee KS, Hawkins C, Cohn RD, Finding of Rare Disease Genes in Canada Consortium Steering C (2016) Lethal Disorder of Mitochondrial Fission Caused by Mutations in DNM1L. J Pediatr 171:313-316 e311-312.

Yu-Wai-Man P, Griffiths PG, Hudson G, Chinnery PF (2009) Inherited mitochondrial optic neuropathies. J Med Genet 46:145-158.

Yu-Wai-Man P et al. (2010) Multi-system neurological disease is common in patients with OPA1 mutations. Brain 133:771-786.

Zanssen S, Molnar M, Schroder JM (1998) Mitochondrial cytochrome b gene deletion in Kearns-Sayre syndrome associated with a subclnical type of peripheral neuropathy. Clin Neuropathol 17:291-296.

Zara F, Biancheri R, Bruno C, Bordo L, Assereto S, Gazzerro E, Sotgia F, Wang XB, Gianotti S, Stringara S, Pedemonte M, Uziel G, Rossi A, Schenone A, TortoriDonati P, vanderKnaap MS, Lisanti MP, Minetti C (2006) Deficiency of hyccin, a newly identified membrane protein, causes hypomyelination and congenital cataract. Nat Genet 38:1111-1113.