Updated complete list of hereditary ataxia

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| **Autosomal dominant forms** | | | | |
| 1. **Disorders that present with ataxia as a predominant or consistent feature** | | | | |
| **Designation** | **Less common movement phenotype** | **Clinical features** | **OMIM** | **MOI** |
| 1. **Pure or relatively pure ataxias** | | | | |
| ATX-*ATXN8OS*467 |  | Relatively pure; pyramidal signs, neuropsychiatric features | 608768 (SCA8) | AD |
| ATX-*CACNA1AA,* 468 |  | Pure ataxia. | 183086 (SCA6) | AD |
| ATX-*DAB1469-471* |  | Adult-onset, slowly progressive, relatively pure cerebellar ataxia with gait instability, frequent falls, dysarthria, and ocular abnormalities | 615945 (SCA37) | AD |
| ATX-*ELOVL4472, 473* |  | Relatively pure ataxia, slowly progressive, usually young adult onset, less common additional signs including ocular abnormalities, pyramidal tract signs, or autonomic symptoms, one family with skin abnormalities (erythrokeratodermia) | 133190 (SCA34) | AD |
| ATX-*ELOVL5*474 |  | Relatively pure ataxia; eventually neuropathy | 615957 (SCA38) | AD |
| ATX-*FGF14*475 |  | Relatively pure ataxia; less common additional signs including early-onset hand tremor, orofacial dyskinesia, and behavioural problems | 609307 (SCA27) | AD |
| ATX-*ITPR1*476, 477 |  | Relatively pure ataxia; less common additional signs including myoclonus and dystonia | 606658 (SCA15/16) | AD |
| ATX-*KCNC3478* |  | Slowly progressive cerebellar ataxia with variable age at onset and variable additional features including cognitive impairment and developmental delay | 605259 (SCA13) | AD |
| ATX-*KCND3*479 |  | Relatively pure ataxia; less common additional signs including hand tremor, peripheral neuropathy, and cognitive disturbances | 607346 (SCA19/22) | AD |
| ATX-*PDYN*480 |  | Pure ataxia | 610245 (SCA23) | AD |
| ATX-*PPP2R2B*481 |  | Relatively pure ataxia; less common additional signs including head and hand tremor | 604326 (SCA12) | AD |
| ATX-*PRKCG*482 |  | Relatively pure ataxia; sometimes other movement disorders (dystonia, myoclonus) | 605361 (SCA14) | AD |
| ATX-*SPTBN2*483 |  | Pure ataxia | 600224 (SCA5) | AD |
| ATX-*TGM6*484 |  | Relatively pure ataxia; less common additional signs including pyramidal features and cervical dystonia | 613908 (SCA35) | AD |
| ATX-*TTBK2*485 |  | Pure ataxia | 604432 (SCA11) | AD |
| 1. **Complex ataxias** | | | | |
| ATX-*AFG3L2*486 |  | Ophthalmoparesis | 610246 (SCA28) | AD |
| ATX-*ATN1*487 | Chorea488 | Dentatorubropallidoluysian atrophy (DRPLA): Myoclonus, chorea, parkinsonism, dementia, supranuclear gaze palsy, seizures (particularly in young patients) | 607462 | AD |
| ATX-*ATXN1*485 |  | Marked non-ataxia features; can have dominant chorea, pyramidal features, peripheral neuropathy, ophthalmoplegia | 164400 (SCA1) | AD |
| ATX-*ATXN2*348 | Parkinsonism349 | Marked non-ataxia features, can have predominant parkinsonism or chorea; neuronopathy, dementia, myoclonus | 183090 (SCA2) | AD |
| ATX-*ATXN3*460 | Spasticity, dystonia461, 462 | Marked non-ataxia features; can have predominant parkinsonism, spasticity, dystonia, chorea, neuropathy, lower motor neuron involvement | 109150 (SCA3) | AD |
| ATX-*ATXN7*489 |  | Retinitis pigmentosa with marked visual loss | 164500 (SCA7) | AD |
| ATX-*ATXN10*490 |  | Seizures | 603516 (SCA10) | AD |
| ATX-*BEAN1*491 |  | Hearing loss, vertigo | 117210 (SCA31) | AD |
| ATX-*CACNA1G492, 493* | Spasticity | Ataxia with gait instability, variable age at onset, additional signs including dysarthria, nystagmus, and less commonly spasticity, pyramidal signs and cognitive impairment;  Phenotype can also be much more severe with neurodevelopmental deficits and early-onset ataxia and (OMIM 618087)494 | 604065 (SCA42) | AD |
| ATX-*CCDC88C495, 496* | Tremor, parkinsonism | Adult-onset cerebellar ataxia associated with action tremor, parkinsonism, pyramidal signs and less frequently with impaired vertical gaze and cognitive impairment | 616053 (SCA40) | AD |
| ATX-*DNMT1*497 |  | Sensorineural deafness, narcolepsy, dementia | 126375 | AD |
| ATX-*EBF3498-500* |  | Hypotonia, ataxia, and delayed development syndrome (HADDS); neurodevelopmental syndrome characterized by congenital hypotonia, delayed psychomotor development, variable intellectual disability with speech delay, variable dysmorphic facial features, and ataxia (often associated with cerebellar hypoplasia) | 617330 | AD |
| ATX-*LMNB1501, 502* |  | Autosomal dominant, adult-onset demyelinating leukodystrophy (ADLD); slowly progressive and fatal disorder characterized clinically by early autonomic abnormalities, pyramidal and cerebellar dysfunction, and symmetric demyelination of the central nervous system | 169500 | AD |
| ATX-*NOP56*503 |  | Motor neuron involvement | 614153 (SCA36) | AD |
| ATX-*PUM1504, 505* | Chorea, spasticity | Variable phenotypic presentation ranging from adult-onset, slowly progressive cerebellar ataxia without additional signs to early-onset ataxia with variable additional signs including developmental delay, chorea, spasticity, seizures, and dysmorphic facial features | 617931 (SCA47) | AD |
| ATX-*SAMD9L506, 507* |  | Ataxia-pancytopenia syndrome (ATXPC); cerebellar ataxia, variable hematologic cytopenias, and predisposition to bone marrow failure and myeloid leukemia | 159550 | AD |
| ATX-*SNAP25b508-510* | Tremor | Early-onset fatigable muscle weakness associated with ataxia, developmental delay, intellectual disability, seizures, craniofacial dysmorphism and rarely resting and intention tremor | 616330 | AD |
| ATX-*TBP*511 | Chorea512 | Marked non-ataxia features, can present with predominant chorea, may be HD-like | 607136 (SCA17) | AD |
| ATX-*TMEM240*513 |  | Cognitive impairment/mental retardation | 607454 (SCA21) | AD |
| ATX-*TUBB2AB,514, 515* | Spasticity | Broad phenotypic spectrum including ataxia, spasticity, developmental delay, seizures, distal amyotrophy, and rarely optic atrophy |  | AD |
| 1. **Combined ataxias (disorders where ataxia frequently coexists with other predominant or consistent movement disorders)** | | | | |
| **Designation** | **Alternate name** | **Clinical features** | **OMIM** | **MOI** |
| ATX/HSP-*KCNA2253, 516-518* | Developmental and epileptic encephalopathy-32 (DEE32) | Variable phenotypic spectrum including (myoclonic) seizures, (episodic) ataxia, spasticity, action tremor, myoclonus, dystonia, chorea, dysarthria, developmental delay, and intellectual disability | 616366 | AD |
| ATX/HSP-*VAMP1*519 |  | Spastic ataxia, supranuclear upgaze limitation | 108600 (SPAX1) | AD |
| ATX/MYC-*NUS1C,520-522* | Mental retardation 55 with seizures (MRD55) | Broad phenotypic spectrum including developmental delay, intellectual disability, ataxia, myoclonus, (myoclonic) seizures, resting and intention tremor, and rarely parkinsonism | 617831 | AD |
| **Autosomal recessive forms** | | | | |
| 1. **Disorders that present with ataxia as a predominant or consistent feature** | | | | |
| **Designation** | **Alternate name** | **Clinical features** | **OMIM** | **MOI** |
| 1. **Pure or relatively pure ataxias** | | | | |
| ATX-*ANO10*523 |  | Cognitive impairment, nystagmus, hypermetric saccades, tortuous conjunctival vessels, pyramidal signs, intention tremor, proximal lower limbs atrophy, fasciculations, seizures, pes cavus | 613728 | AR |
| ATX-*APTX*524, 525 | Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia (AOA1) | Hypometric saccades, oculomotor apraxia, nystagmus, ophthalmoplegia, peripheral neuropathy, scoliosis, pes cavus, choreoathetosis, tremor, dystonia, cognitive decline | 208920 (AOA1) | AR |
| ATX-*CWF19L1*526 |  | Developmental delay, intellectual disability, tremor, hyperreflexia in lower limbs | 616127 (SCAR17) | AR |
| ATX-*FXN*527, 528 | Friedreich ataxia (FRDA) | Nystagmus, square wave jerks, optic atrophy, hearing loss, peripheral sensory neuropathy, pes cavus, hammertoes, muscle weakness, amyotrophy, extensor plantar responses, spasticity, spastic ataxia, chorea, scoliosis, hypertrophic cardiomyopathy, diabetes | 229300 | AR |
| ATX-*GRID2*529 |  | Developmental delay, cognitive impairment, esotropia, nystagmus, oculomotor apraxia, tonic upgaze, pale optic discs, retinopathy, pyramidal signs, muscle atrophy, joint contractures, scoliosis | 616204 (SCAR18) | AR |
| ATX-*KIAA0226*530 | Salih ataxia | Developmental delay, mental retardation, nystagmus, abnormal saccadic eye movements, seizures | 615705 (SCAR15) | AR |
| ATX-*PMPCA*531 |  | Developmental delay, mental retardation, visuospatial defects, nystagmus, dysmetric saccades, pes cavus, hyperreflexia, spasticity, tremor, short stature | 213200 (SCAR2) | AR |
| ATX-*SETX*532 |  | Saccadic pursuit, oculomotor apraxia, nystagmus, strabismus, intention tremor, head tremor, dystonia, chorea, pyramidal signs, peripheral neuropathy, distal muscle atrophy and weakness, pes cavus, scoliosis | 606002 (SCAR1, AOA2) | AR |
| ATX-*SLC52A2D,* 533, 534 |  | Optic atrophy, blindness, cochlear degeneration, deafness | 271250 (SCAR3) | AR |
| ATX-*SNX14*535 |  | Developmental delay, mental retardation, autistic behavior, macrocephaly, sensorineural hearing loss, nystagmus, apraxia, spasticity, extensor plantar responses, hyporeflexia, seizures, hypertrichosis, scoliosis, distal skeletal deformities, brachycamptodactyly, facial dysmorphism | 616354 (SCAR20) | AR |
| ATX-*SPTBN2*536 |  | Developmental delay, cognitive impairment, speech delay, intention tremor, spasticity, hyperreflexia, hypometric saccades, nystagmus, abnormal eye movements with convergent squint | 615386 (SCAR14) | AR |
| ATX-*SYNE1*537 |  | Nystagmus, abnormal saccades and slow or jerky pursuit, hyperreflexia in lower limbs, motor neuron involvement, respiratory dysfunction due to multisystemic neuromuscular compromise, mental retardation | 610743 (SCAR8, ARCA1) | AR |
| ATX-*TTPA*538 | Ataxia with vitamin E deficiency | Nystagmus, retinopathy, propioception loss, areflexia in lower limbs, peripheral neuropathy, extensor plantar response, head titubation or tremor, dystonia, hypoacusia, tendon xanthomas, pes cavus, hammer toes, kyphoscoliosis | 277460 | AR |
| 1. **Complex ataxias** | | | | |
| ATX-*ABCA2539, 540* | Intellectual developmental disorder with poor growth and with or without seizures or ataxia (IDPOGSA) | Highly variable phenotype including developmental delay, intellectual disability, hypotonia, poor overall growth, intellectual disability, sometimes borderline microcephaly, and seizures. Cases have been reported with ataxia as the predominant manifestation. | 618808 | AR |
| ATX-*ABHD12*541 | Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract (PHARC) | Retinopathy, cataract, hearing loss, intention tremor, pyramidal signs, peripheral neuropathy, pes cavus | 612674 | AR |
| ATX-*ADCK3*542, 543 |  | Developmental delay, muscle weakness, pes cavus, exercise intolerance, myoclonus, dystonia, headache, stroke-like episodes, seizures | 612016 (SCAR9, ARCA2) | AR |
| ATX-*ADPRHL2544, 545* | Stress-induced childhood-onset neurodegeneration with variable ataxia and seizures (CONDSIAS) | Highly variable phenotype including cyclic episodic deterioration in response to stress, developmental delay, intellectual disability, ataxia, muscle weakness, seizures, neuropathy, and rarely tremor, dystonia, strabismus, nystagmus, hearing loss, and microcephaly | 618170 | AR |
| ATX-*AHI1*546, 547 | Joubert syndrome 3 (JBTS3) | Developmental delay, morphological abnormalities, oculomotor apraxia, nystagmus, retinopathy, spasticity, scoliosis, seizures, renal failure, respiratory dysfunction | 608629 | AR |
| ATX-*ALDH5A1*548 | Succinic semialdehyde dehydrogenase deficiency | Developmental delay, mental retardation, hyperkinesis, hyporeflexia, psychiatric symptoms, abnormal eye movements, seizures | 271980 | AR |
| ATX-*ALG6*549 |  | Developmental delay, psychiatric symptoms, nystagmus, strabismus, peripheral neuropathy, muscle weakness, seizures, skeletal deformities, coagulation anomalies | 603147 | AR |
| ATX-*ARL13B*550 | Joubert syndrome 8 (JBTS8) | Developmental delay, oculomotor apraxia, retinopathy, respiratory dysfunction | 612291 | AR |
| ATX-*ATM*551 | Ataxia-telangiectasia (including variant ataxia-telangiectasia) | Telangiectases and other skin alterations, oculomotor apraxia, dystonia, chorea, myoclonus, tremor, seizures, peripheral neuropathy, distal muscular atrophy, short stature, hypogonadism, respiratory dysfunction, immunodeficiency, predisposition to neoplasia, glucose intolerance | 208900 | AR |
| ATX-*BCKDHB*552 | Maple syrup urine disease | Maple syrup urine odor, life-threatening metabolic decompensation, lethargy, coma, hypoglycemia, ketosis, lactic acidosis, hallucinations, seizures, mental retardation if untreated, vomiting, pancreatitis | 248600 | AR |
| ATX-*BRAT1E,228, 553* | Neurodevelopmental disorder with cerebellar atrophy and with or without seizures (NEDCAS) | Hypotonia, developmental delay, intellectual disability, oculomotor apraxia, saccadic smooth pursuit, gaze-evoked nystagmus. Cases have been reported with ataxia as the predominant manifestation. | 618056 | AR |
| ATX-*BTD*554 | Biotinidase deficiency | Developmental delay, optic atrophy, vision and hearing loss, seizures, metabolic ketoacidosis, organic aciduria, skin problems, alopecia, hepatosplenomegaly, breathing problems | 253260 | AR |
| ATX-*C10orf2*555 | Hepatocerebral type of Mitochondrial DNA depletion syndrome | Psychomotor retardation, psychiatric symptoms, ophthalmoplegia, nystagmus, optic atrophy, hearing loss, peripheral neuropathy, myopathy, status epilepticus, epileptic encephalopathy, headaches, liver disease, hypergonadotrophic hypogonadism | 271245 | AR |
| ATX-*CA8*556 | Cerebellar ataxia and mental retardation with or without quadrupedal locomotion type 3 | Mental retardation, dysarthria, quadrupedal gait, tremor | 613227 | AR |
| ATX-*CACNA2D2228, 557, 558* | Cerebellar atrophy with seizures and variable developmental delay (CASVDD) | Ataxia with variable seizures and/or developmental delay (epileptic encephalopathy), tremor, and also myoclonus and choreic movements in some patients | 618501 | AR |
| ATX-*CEP290*559, 560 | Joubert syndrome 5 (JBTS5) | Mental retardation, congenital amaurosis, oculomotor apraxia, retinopathy, retinal coloboma, nystagmus, nephronophthisis, neonatal breathing dysregulation | 610188 | AR |
| ATX-*COA7561, 562* |  | Ataxia, distal muscle weakness and atrophy, peripheral neuropathy, tremor, intellectual disability and developmental delay | 618387 (SCAN3) | AR |
| ATX-*COG5563, 564* | Congenital disorder of glycosylation, type IIi (CDG IIi) | Variable phenotype including developmental delay, intellectual disability, hypotonia, seizures, microcephaly, and hypotonia. Cases have been reported with ataxia as the predominant manifestation. | 613612 | AR |
| ATX-*COX20*565 | Mitochondrial complex IV deficiency or cytochrome c oxidase deficiency | Developmental delay, mental retardation, pyramidal signs, peripheral neuropathy, dystonia, lactic acidosis retinopathy, optic atrophy, respiratory insufficiency | 220110 | AR |
| ATX-*CYP27A1*566 | Cerebrotendinous xanthomatosis (CTX) | Tuberous skin and tendon xantomas, xanthelasmas, cataracts, chronic diarrhea, cognitive decline, psychiatric symptoms, peripheral neuropathy, parkinsonism, dystonia, myoclonus, spastic paraplegia, pseudobulbar palsy, seizures | 213700 | AR |
| ATX-*DNAJC19*567 | 3-methylglutaconic aciduria, type V | Developmental delay, mental retardation, growth retardation, optic atrophy, muscle weakness, dilated cardiomyopathy, long QT syndrome, genitourinary deformities | 610198 | AR |
| ATX- *DOCK3568-570* |  | Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia | 618292 | AR |
| ATX-*ERCC4571-574* | XFE progeroid syndrome and the allelic entity Xeroderma pigmentosum complementation group F/Cockayne syndrome | Dwarfism, cachexia, skin photosensitivity, wizened appearance, scoliosis, delayed sexual maturity, intellectual disability, mental retardation, cognitive decline, nystagmus, astigmatism, deep-set eyes, hearing impairment, short stature, seborrheic keratosis-like papules, abnormal pigmentation, skin cancer susceptibility, plantar warts, microcephaly, and in some patients chorea and tremor. Cases have been reported with ataxia as the predominant manifestation. | 610965, 278760 | AR |
| ATX-*GDAP2575-577* |  | Adult-onset cerebellar ataxia, dysarthria, and cognitive impairment associated with pyramidal signs, including spasticity. Cervical dystonia was reported in one patient. | 618369 (SCAR27) | AR |
| ATX-*GRN*578 | Neuronal ceroid lipofuscinosis 11 (CLN11) | Dementia, myoclonic retinopathy, optic atrophy, seizures | 614706 | AR |
| ATX-*ITPR1*579 | Gillespie syndrome | Developmental delay, mental retardation, aniridia or iris hypoplasia, scalloped pupillary margins of iris, nystagmus, visual impairments, postural tremor | 206700 | AR |
| ATX-*KCNJ10*580 | Seizures, sensorineural deafness, ataxia, mental retardation, and electrolyte imbalance (SESAME syndrome) | Developmental delay, mental retardation, sensorineural deafness, intention tremor, peripheral neuropathy, seizures, short stature, salt craving, enuresis, polydipsia, polyuria, electrolyte imbalance | 612780 | AR |
| ATX-*L2HGDH*581 | L-2-hydroxyglutaric aciduria or academia | Psychomotor regression, mental retardation, cognitive impairment, hearing loss, strabismus, optic atrophy, nystagmus, spastic tetraparesis, facial dyskinesia, rigidity, dystonia, intention tremor, action-induced negative myoclonus, pyramidal signs, seizures, macrocephaly | 236792 | AR |
| ATX-*MAN2B1*582 | Alpha-mannosidosis | Developmental delay, mental retardation, growth retardation, sensorineural deafness, nystagmus, pyramidal signs, macrocephaly, facial dysmorphism, skeletal deformities, hepatosplenomegaly | 248500 | AR |
| ATX-*MTCL1\*,583, 584* |  | Slowly progressive cerebellar ataxia, developmental delay, intellectual disability, seizures, nystagmus, slow saccadic eye movements, dysarthria, hyperreflexia, spasticity, and tremor | 615766 | AR |
| ATX-*MRE11A*585 | Ataxia-telangiectasia-like disorder type 1 | Hypometric saccades, oculomotor apraxia, nystagmus, chorea, dystonia, myoclonus, tremor, hyporeflexia, distal muscle atrophy | 604391 | AR |
| ATX-*MSTO1*586 |  | Myopathy, developmental delay, growth impairment, pigmentary retinopathy with  papillary pallor, tremor, skeletal abnormalities, pes cavus, dysmoprhism | 617619 | AR |
| ATX-*NFASC587-589* | Neurodevelopmental disorder with central and peripheral motor dysfunction (NEDCPMD) | Highly variable severity and phenotypic spectrum including hypotonia, developmental delay, ataxia, pyramidal signs, and demyelinating peripheral neuropathy; tremor and myoclonus in some patients | 618356 | AR |
| ATX-*NPC1*590 | Niemann-Pick disease type C1 | Developmental regression, cognitive impairment, psychiatric symptoms, loss of speech, vertical supranuclear gaze palsy, dystonia, intention tremor, spasticity, seizures, hepatosplenomegaly, cholestatic jaundice, gelastic cataplexy | 257220 | AR |
| ATX-*NPC2*591 | Niemann-Pick disease type C2 | Similar to ATX-*NPC1* with severe pulmonary involvement and respiratory failure | 607625 | AR |
| ATX-*PEX7*592 | Peroxisome biogenesis disorder 9B or Zellweger spectrum disorder | Developmental delay, cognitive impairment, cataracts, retinopathy, anosmia, hearing loss, muscle weakness, pes cavus, peripheral neuropathy | 614879 | AR |
| ATX-*PEX10*593 | Peroxisome biogenesis disorder 6B or Zellweger spectrum disorder | Mental retardation, intention tremor, peripheral neuropathy, pyramidal signs, distal muscle atrophy, pes cavus, dysmetric saccades, impaired smooth pursuit, nystagmus, diabetes | 614871 | AR |
| ATX-*PHYH*594 | Refsum disease or or hereditary motor and sensory neuropathy type IV | Muscle weakness and atrophy, peripheral neuropathy, sensory impairment, pes cavus, anosmia, sensorineural deafness, retinopathy, ichthyosis, shortening of the metacarpals and metatarsals, multiple epiphyseal dysplasia, cardiomyopathy, sudden death | 266500 | AR |
| ATX-*PIBF1595-597* | Joubert syndrome 33 (JBTS33) | Hypotonia, ataxia, and developmental delay, additional features including retinal dystrophy, cystic kidney disease, liver fibrosis, and dysmorphism in a subset of patients; spastic tetraparesis reported in one patient | 617767 | AR |
| ATX-*PMM2*598 | Congenital disorder of glycosylation type Ia (CDGIa) or Jaeken syndrome | Developmental delay, psychomotor retardation, cognitive impairment, strabismus, nystagmus, retinopathy, peripheral neuropathy, stroke-like episodes, seizures, microcephaly, morphological abnormalities, abnormal subcutaneous fat tissue distribution, pericardial effusion, hepatomegaly, liver steatosis, diarrhea, renal cysts, nephrotic syndrome, thrombotic events, hypothyroidism, hypergonadotropic hypogonadism, scoliosis, osteoporosis | 212065 | AR |
| ATX-*PNKF, 599-603* | Ataxia-oculomotor apraxia type 4 (AOA4) | Early-onset progressive ataxia, dystonia, oculomotor apraxia, peripheral neuropathy, and cognitive impairment | 616267 (AOA4) | AR |
| ATX-*PNPLA6*604 | Boucher-Neuhauser syndrome | Visual impairment due to chorioretinal dystrophy, distal muscle atrophy, intention tremor, spasticity, cognitive impairment, peripheral neuropathy, hypogonadism | 215470 | AR |
| ATX-*POLR3B*605 | Hypomyelinating leukodystrophy type 8 with or without oligodontia and/or hypogonadotropic hypogonadism | Developmental delay, cognitive decline, mental retardation, nystagmus, abnormal saccades, vertical gaze palsy, myopia, spasticity, tremor, oligodontia, hypodontia, delayed dentition, short stature, hypogonadism | 614381 | AR |
| ATX-*PRKCG*606 |  | Peripheral neuropathy, pyramidal signs, cognitive impairment, depression, myoclonus, tremor | 605361 | AR |
| ATX-*PTRH2*607 | Infantile-onset multisystem neurologic, endocrine, and pancreatic disease (IMNEPD) | Developmental delay, failure to thrive, poor postnatal growth, poor expressive speech, peripheral neuropathy, distal muscle weakness, foot and hand deformities, hypothyroidism, pancreatic endocrine insufficiency, facial dysmorphism, brachycephaly, short stature | 616263 | AR |
| ATX-*RNF168*608 | RIDDLE syndrome | Learning difficulties, immunodeficiency, dry skin, progressive pulmonary fibrosis and failure, ocular telangiectasia, short stature, microcephaly, dysmorphic features | 611943 | AR |
| ATX-*RNF216*609 | Gordon Holmes syndrome | Mental retardation, dementia, psychiatric symptoms, chorea, sensorineural deafness, hypogonadism | 212840 | AR |
| ATX-*RFC1610-614* | Cerebellar ataxia, neuropathy and vestibular areflexia syndrome (CANVAS) | Adult onset, slowly progressive, other features include autonomic dysfunction, chronic spasmodic dry cough, and action tremor. More rarely: bradykinesia, orofacial dyskinesia or dystonia and limb chorea | 614575 | AR |
| ATX-*SIL1*615 | Marinesco-Sjögren syndrome | Developmental delay or regression, mental retardation, growth retardation, microcephaly, facial dysmorphism, short stature, congenital cataracts, nystagmus, strabismus, spasticity, muscle weakness and atrophy, peripheral neuropathy, scoliosis, skeletal deformities, hypogonadism | 248800 | AR |
| ATX-*SLC17A5*616 | Salla disease or Finnish type sialuria and the variant syndrome of infantile form of sialic acid storage disorder: | Developmental delay, mental retardation, rigidity, spasticity, seizures, visceromegaly, facial dysmorphism, hypopigmented skin | 604369, 269920 | AR |
| ATX-*SLC33A1*617 | Congenital cataracts, hearing loss, and neurodegeneration (CCHLND) | Psychomotor retardation, nystagmus, congenital cataracts, hearing loss, seizures | 614482 | AR |
| ATX-*SRD5A3*618 | Congenital disorder of glycosylation, type Iq | Developmental delay, mental retardation, coloboma, nystagmus, facial dysmorphism, hypertrichosis, skin abnormalities, coagulation defects, microcytic anemia | 612379 | AR |
| ATX-*TANGO2619-621* | Recurrent metabolic encephalomyopathic crises with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration (MECRCN) | Developmental delay followed by acute encephalomyopathic features, including rhabdomyolysis, hypotonia, and neurologic regression; during disease course progressive neurodegeneration with seizures, intellectual disability, pyramidal signs, ataxia, spasticity, loss of expressive language, as well as cardiac involvement with severe arrhythmias | 616878 | AR |
| ATX-*TBC1D23622-624* | Pontocerebellar hypoplasia type 11 (PCH11) | Neurodevelopmental disorder with severe developmental delay, intellectual disability, ataxia, hypotonia, behavioral abnormalities, microcephaly, dysmorphic features, and recurrent respiratory infections. Stereotypies and spasticity were reported in some patients. | 617695 | AR |
| ATX-*TMEM216*625 | Joubert syndrome 2 (JBTS2) | Developmental delay, failure to thrive, mental retardation, impaired saccades, oculomotor apraxia, nystagmus, optic nerve coloboma, chorioretinal coloboma, retinopathy, esotropia, polydactyly, nephronophthisis, renal cysts, hypoplastic genitalia, episodic hyperpnea or apnea, facial dysmorphism, macrocephaly | 608091 | AR |
| ATX-*TMEM67*626, 627,  ATX-*RPGRIP1L*628-630, ATX-CC2D2A631 | COACH syndrome (cerebellar vermis hypo/aplasia, oligophrenia, ataxia, ocular coloboma, and hepatic fibrosis) and allelic disorders;  Joubert syndrome 6 and 7 (JBTS6 and JBTS7) | Developmental delay, mental retardation, oculomotor apraxia, ocular coloboma, retinopathy, nystagmus, facial dysmorphism, polydactyly, pyramidal signs, seizures, splenomegaly, renal failure, liver disease, breathing dysregulation | 216360, 611560, 610688 | AR |
| ATX-*TMEM231*632 | Joubert syndrome 20 (JBTS20) | Developmental delay, oculomotor apraxia, psychiatric symptoms, polydactyly, syndactyly, renal cysts, retinopathy | 614970 | AR |
| ATX-*TSEN54633, 634* | Pontocerebellar hypoplasia types 5, 2A and 4 | Ataxia, dysarthria, intellectual disability, peripheral neuropathy, and pyramidal signs | 610204, 277470, 225753 | AR |
| ATX-*TTC19*635 | Mitochondrial complex III deficiency nuclear type 2 | Developmental delay, cognitive impairment, apraxia, psychiatric symptoms, dysphonia, nystagmus, bradykinesia, dystonia, muscle atrophy and weakness, pyramidal signs | 615157 | AR |
| ATX-*VLDLR*636 | Cerebellar ataxia, mental retardation, and dysequilibrium syndrome type 1 | Developmental delay, mental retardation, lack of speech development, strabismus, postnatal cataracts, nystagmus, saccadic visual pursuit, quadrupedal gait, intention tremor, hyperrefexia, seizures, pes planus, short stature | 224050 | AR |
| ATX-*WDR73*637 | Galloway-Mowat syndrome | Delayed psychomotor development, mental retardation, oculomotor apraxia, optic atrophy, retinopathy, seizures, spastic quadriplegia, dystonia, hyperreflexia, skin abnormalities (osmiophilic skin vessels), skeletal deformities, genitourinary affectation, facial dysmorphias, microcephaly, short stature, intrauterine growth retardation | 251300 (SCAR5) | AR |
| ATX-*WDR81*638 | Cerebellar ataxia, mental retardation, and dysequilibrium syndrome type 2 | Developmental delay, mental retardation, strabismus, facial dysmorphism, quadrupedal locomotion, poor or absence language development, tremor, hyporeflexia, hirsutism, small hands and feet, thoracic kyphosis, short stature | 610185 | AR |
| ATX-*XRCC1639, 640* |  | Ataxia associated with dysarthria, intellectual disability, slow and hypometric saccadic eye movements, nystagmus, oculomotor apraxia, and peripheral neuropathy | 617633 (SCAR26) | AR |
| 1. **Combined ataxias (disorders where ataxia frequently coexists with other predominant or consistent movement disorders)** | | | | |
| ATX/HSP-*AFG3L2*641 |  | Spastic paraparesis, oculomotor apraxia, dystonia, myoclonus, myoclonic epilepsy, generalized tonic-clonic seizures, distal muscle atrophy, peripheral neuropathy | 614487 (SPAX5) | AR |
| ATX/HSP-*DARS2*642 | Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation | Spastic paraparesis, developmental delay, cognitive decline, nystagmus, tremor, peripheral neuropathy, sensation deficits, muscle weakness and atrophy, joint contractures | 611105 | AR |
| ATX/HSP-*FOLR1*643 | Neurodegeneration due to cerebral folate transport deficiency | Spastic paraparesis, developmental regression, mental retardation, visual disturbances, sensorineural hearing loss, chorea, generalized tonic-clonic, atonic and myoclonic seizures | 613068 | AR |
| ATX/HSP-*GJC2*644 | Hypomyelinating leukodystrophy-2 or Pelizaeus-Merzbacher-like disease | Spastic paraparesis, developmental delay, mental retardation, lack of independent ambulation, poor head and trunk control in infancy, optic atrophy, rotary nystagmus, myopia, facial weakness, tremor, head titubation, dystonia, spasticity, seizures, peripheral neuropathy | 608804 | AR |
| ATX/HSP-*HEXA*645 | Tay-Sachs disease or GM2-gangliosidosis type I | Spastic paraparesis, cognitive decline, psychiatric symptoms, late spasticity, dystonia, peripheral neuropathy, macular pallor with prominence of fovea centralis (cherry red spot), blindness, muscular weakness and atrophy, seizures | 272800 | AR |
| ATX/HSP-*HEXB*646 | Sandhoff disease or GM2-Gangliosidosis type II | Spastic paraparesis, progressive mental and motor deterioration, macrocephaly, macular pallor with prominence of fovea centralis (cherry red spot), blindness, dysmorphic features, startle reaction, hyperreflexia, muscular atrophy, fasciculations, cardiomegaly, episodic abdominal pain, chronic diarrhea, hepatosplenomegaly, macroglossia, high lumbar gibbus | 268800 | AR |
| ATX/HSP-*POLR3A*647 | Hypomyelinating leukodystrophy type 7 with or without oligodontia and/or hypogonadotropic hypogonadism or 4H syndrome | Spastic paraparesis, developmental delay, cognitive decline, mental retardation, optic atrophy, nystagmus, abnormal saccades, vertical gaze palsy, pyramidal signs, postural tremor, dystonia, seizures, peripheral neuropathy, oligodontia, hypodontia, delayed dentition, hypogonadotropic hypogonadism, short stature | 607694 | AR |
| ATX/HSP-*SACS*648 | Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) or autosomal recessive spastic ataxia type 6 | Spastic paraparesis, delayed walking development, retinal striation, nystagmus, impaired smooth pursuit, pes cavus, hammertoes, finger deformities, hyperreflexia, ankle areflexia, spasticity, extensor plantar responses, scoliosis, distal muscle weakness and atrophy, peripheral neuropathy, dystonia, erectile dysfunction | 270550 (SPAX6) | AR |
| ATX/HSP-*VPS13D649-652* |  | Variable phenotype including ataxia, spasticity, other pyramidal signs, dystonia, myoclonus, chorea, tremor, dysarthria, oculomotor abnormalities, distal sensory impairment, hypotonia, sometimes global developmental delay or mild intellectual disability | 607317 (SCAR4) | AR |
| HSP/ATX-*B4GALNT1*653 | Spastic paraplegia | Spastic paraparesis, distal amyotrophy, nonprogressive cognitive impairment, sensory polyneuropathy, pes cavus, stereotypies, emotional lability, psychiatric symptoms, seizures | 609195 | AR |
| HSP/ATX-*CAPN1654, 655,* 656 | Spastic paraplegia | Pure or complex; spasticity in lower and upper limbs, other pyramidal signs, cerebellar ataxia, dysarthria, foot deformities, ocular movement abnormalities, amyotrophy peripheral neuropathy, pes cavus, pes valgus, nystagmus | 616907 | AR |
| HSP/ATX-*CLCN2*657 | Leukoencephalopathy with ataxia | Spastic paraparesis, learning disabilities, headache, optic neuropathy, chorioretinopathy, visual field defects | 615651 | AR |
| HSP/ATX-*CYP7B1*658 | Spastic paraplegia | Spastic paraparesis, other pyramidal signs besides spasticity, cognitive impairment, nystagmus, optic atrophy, cataracts, altered saccadic eye movements, pes cavus, sensation deficits | 270800 | AR |
| HSP/ATX-*GAN*659 |  | Spastic paraparesis, distal limb muscle weakness and atrophy due to peripheral neuropathy, distal sensory impairment, kinky or curly hair, foot or hand deformities, scoliosis, nystagmus, facial weakness | 256850 | AR |
| HSP/ATX-*GBA2*660 | Spastic paraplegia | Spastic paraparesis, other pyramidal signs, muscle weakness, pseudobulbar dysarthria, cognitive impairment, mental retardation, congenital cataracts, nystagmus, hearing loss, head tremor, peripheral neuropathy, pes cavus, scoliosis, infertility, small testicles, hypogonadism in males | 614409 | AR |
| HSP/ATX-*KIF1CG,* 463, 464 | Spastic ataxia, Spastic paraplegia | Spastic paraparesis, chorea, myoclonus, dystonia, developmental delay, mild mental retardation, hypodontia, ptosis, short stature, sensorineural deafness, pes planus | 611302 | AR |
| HSP/ATX-*MLC1*661 | Megalencephalic leukoencephalopathy with subcortical cysts | Spastic paraparesis, developmental delay, mental retardation, seizures, macrocephaly, spasticity | 604004 | AR |
| HSP/ATX-*UCHL1*662 | Childhood-onset neurodegeneration with optic atrophy | Spastic paraparesis, other pyramidal signs, myotonia, myokymia, head titubation, intellectual impairment, impaired distal sensation to vibration and position, optic atrophy, nystagmus | 615491 | AR |
| HSP/ATX-*FA2H*-(NBIA)362 | Fatty acid hydroxylase-associated neurodegeneration (FAHN); Spastic paraplegia | Spastic paraparesis, cognitive decline, optic nerve atrophy, seizures, dystonia, parkinsonism | 612319 | AR |
| ATX/MYC-*TPP1H,* 663,664 |  | Myoclonus, developmental regression, speech and language difficulties, nystagmus, diplopia, hypermetric saccades, progressive vision loss, retinopathy, postural tremor, pyramidal signs, spastic paraparesis, decreased vibration sense, fasciculations, seizures | 609270 (SCAR7) | AR |
| MYC/ATX-*CSTB*665 | Myoclonic epilepsy of Unverricht and Lundborg | Myoclonic epilepsy, stimulus sensitive segmental or generalized myoclonus, action myoclonus, generalized tonic-clonic or absence seizures, mental and motor deterioration | 254800 | AR |
| MYC/ATX-  *EPM2A364, 365* | Progressive myoclonus epilepsy (Lafora disease) | Myoclonic or other types of seizures, focal visual seizures, drop attacks cognitive decline, psychosis, myoclonus | 607566 | AR |
| MYC/ATX-*GOSR2*666 |  | Myoclonic, absence and tonic-clonic seizures, drop attacks, action myoclonus, tremor, areflexia, scoliosis, pes cavus, syndactyly | 614018 | AR |
| MYC/ATX-*KCTD7*667 | Progressive myoclonic epilepsy type 3 with or without intracellular inclusions | Myoclonic epilepsy, secondary generalization seizures, neurologic regression following seizure onset, mental retardation, hyperreflexia, opsoclonus, optic atrophy, visual loss, microcephaly | 611726 | AR |
| MYC/ATX-*NEU1*668 | Neuraminidase deficiency or sialidosis type I and II | Myoclonus, mental retardation, seizures, hyperreflexia, muscle atrophy, skeletal malformations, hepatosplenomegaly, cardiomyopathy, progressive vision loss, cherry-red spots, cataracts, nystagmus, sensorineural hearing loss, short stature, dysmorphic features | 256550 | AR |
| MYC/ATX-*NHLRC1*669 | Lafora disease | Myoclonic or other types of seizures, focal visual seizures, drop attacks cognitive decline, psychosis, myoclonus | 608072 | AR |
| ATX/DYT-*SQSTM1458, 459, 670, 671* | Neurodegeneration with ataxia, dystonia, and gaze palsy (NADGP) | Ataxia, dystonia, chorea, gaze palsy, cognitive decline, nystagmus, pyramidal signs, and dysarthria | 617145 | AR |
| DYT/ATX-*ATP7B*407 | Wilson disease | Dystonia, occasionally parkinsonism, chorea, flapping tremor, rest, action, and intention tremor, orofacial dyskinesias, liver disease, Kayser Fleischer rings, psychiatric symptoms | 277900 | AR |
| **Autosomal dominant or recessive forms** | | | | |
| **Designation** | **Alternate name** | **Clinical features** | **OMIM** | **MOI** |
| 1. **Disorders that present with ataxia as a predominant or consistent feature** | | | | |
| ATX-*MSTO1586, 672, 673* |  | Mitochondrial myopathy and ataxia (MMYAT); complex neurologic disorder with variable manifestation including early-onset global developmental delay, mitochondrial myopathy, ataxia and variable additional features like growth impairment, cognitive impairment, muscle weakness, elevated creatine kinase, and psychiatric comorbidities | 617675 | AR (AD) |
| ATX-*STUB1*674*,675-682* |  | Ataxia with cognitive-affective symptoms, such as depression, anxiety, or apathy, and variable additional features like parkinsonism, tremor, chorea, dystonia, myoclonus, dysarthria, dysphagia, pyramidal signs, peripheral neuropathy, reduced vibration sense in lower limbs, nystagmus, external ophthalmoplegia, and hypogonadism | 618093 (SCA48), 615768 (SCAR16) | AD or AR |
| 1. **Combined ataxias (disorders where ataxia frequently coexists with other predominant or consistent movement disorders)** | | | | |
| HSP/ATX-*SPG7*683 | Spastic paraplegia | Spastic paraparesis, optic atrophy, chronic external ophthalmoplegia-like phenotype, nystagmus, decreased vibratory sense in the lower limbs, pes cavus, scoliosis | 607259 | AR or AD |
| **X-linked forms** | | | | |
| **Designation** | **Alternate name** | **Clinical features** | **OMIM** | **MOI** |
| 1. **Disorders that present with ataxia as a predominant or consistent feature** | | | | |
| ATX-*ABCB7*684 | Sideroblastic anemia with spinocerebellar ataxia | Intention tremor, pyramidal signs, hypochromic, microcytic anemia, abnormal pigmentation, skin atrophy | 301310 (SCAR10, ARCA3) | XLR |
| ATX-*AIFM1685-688* |  | Ataxia, peripheral neuropathy, hearing loss, pyramidal signs, behavioral disorder, and intellectual disability |  | XL |
| ATX-OFD1689 | Joubert syndrome 10 (JBTS10) | Developmental delay, mental retardation, recurrent infections, hirsutism, postaxial polydactyly, cystic renal disease, facial dysmorphism, macrocephaly | 300804 | XLR |
| ATX-*OPHN1*690 | X-linked mental retardation with cerebellar hypoplasia and distinctive facial appearance | Developmental delay, mental retardation, spasticity, psychiatric symptoms, seizures, macrocephaly, facial dysmorphism, microphaly, hypoplastic scrotum, cryptorchidism, strabismus | 300486 | XLR |
| ATX-*OTC*691  (Heterozygous females) | Ornithine transcarbamylase deficiency | Episodic extreme irritability, episodic vomiting and lethargy, protein avoidance, coma, delayed growth, developmental delay, seizures | 311250 | XLR |
| 1. **Combined ataxias (disorders where ataxia frequently coexists with other predominant or consistent movement disorders)** | | | | |
| HSP/ATX-*PLP1*692 |  | Spastic paraparesis, lower limb weakness, other pyramidal signs, mental retardation, upper limb spasticity, pes cavus, joint contractures, nystagmus, optic atrophy, tremor | 312920 | XLR |
| **Mitochondrial inheritance** | | | | |
| **Designation** | **Alternate name** | **Clinical features** | **OMIM** | **MOI** |
| ATX-*MT-ATP6693-696* | MT-ATP6-mitochondrial disease: neuropathy, ataxia, and retinitis pigmentosa (NARP); Leigh syndrome; mitochondrial encephalomyopathy | Variable phenotype including ataxia, less commonly myoclonus, cognitive dysfunction, neuropathy, seizures, and retinopathy | 551500 | mt |
| **Disorders that usually present with other phenotypes but can occasionally include ataxia (no ATX prefix)** | | | | |
| **Designation or *Gene*** | **Associated disease/Alternate name** | **Main clinical features** | **OMIM** | **MOI** |
| HSP-*ACP33*697 | Spastic paraplegia | Spastic paraparesis, other pyramidal signs, apraxia, bulbar dysfunction, developmental delay, cognitive impairment, akinetic mutism, dykinesias, peripheral neuropathy | 248900 | AR |
| HSP-*DDHD2*698 | Spastic paraplegia | Spastic paraparesis, other pyramidal signs, developmental delay, mental retardation, lower limbs weakness, strabismus, facial dysmorphism, short stature, optic nerve hypoplasia | 615033 | AR |
| HSP-*KIAA0415*699 | Spastic paraplegia | Spastic paraparesis, lower limb weakness, dystonia, myoclonus, parkinsonism, peripheral neuropathy | 613647 | AR |
| HSP- *KIAA1840*700 | Spastic paraplegia | Spastic paraparesis, lower limb atrophy, weakness, peripheral neuropathy, pes cavus, parkinsonism, cognitive impairment, mental retardation, retinopathy, pigmented macular degeneration, nystagmus | 604360 | AR |
| HSP-*KIF1A*701 | Spastic paraplegia | Spastic paraparesis, lower limb atrophy and weakness, peripheral neuropathy, saccadic ocular pursuit | 610357 | AR |
| HSP-*PNPLA6/NT*702 | Spastic paraplegia | Spastic paraparesis, axonal peripheral neuropathy, distal muscle atrophy, cognitive impairment, speech impairment | 612020 | AR |
| HSP-*REEP1*703 | Spastic paraplegia | Pure or complex; cerebellar ataxia less common704, additional clinical features include distal motor neuronopathy, axonal peripheral neuropathy, Silver-like syndrome, tremor, and dementia | 610250 | AD |
| HSP-*SPARTIN*700 | Spastic paraplegia | Spastic paraplegia and upper limb spasticity, distal muscle atrophy, pes cavus, hammertoes, short stature, dysmorphism, developmental delay | 275900 | AR |
| HSP-*ZFYVE26*360 | Spastic paraplegia | Spastic paraparesis, thin corpus callosum, mental retardation, cognitive impairment, psychiatric symptoms, parkinsonism, distal amyotrophy, pes cavus, peripheral neuropathy, retinopathy, hearing loss, pigmentary maculopathy | 270700 | AR |
| MYC-*CLN5*705 | Myoclonus, Neuronal ceroid lipofuscinosis 5 (CLN5) | Myoclonus, tremor, mental retardation, cognitive decline, visual loss, glaucoma, retinopathy, nystagmus, hyperreflexia, seizures | 256731 | AR |
| MYC-*CLN6*706 | Myoclonus, Neuronal ceroid lipofuscinosis 6 (CLN6) | Myoclonus, dystonia, bradykinesia, myoclonic epilepsy, dementia, mental retardation, psychiatric symptoms, blindness | 204300 | AR |
| MYC-*SCARB2*707 | Progressive myoclonic epilepsy type 4 with or without renal failure | Action and resting myoclonus, intention and postural tremor, horizontal saccades, seizures, nephrotic syndrome, renal failure | 254900 | AR |
| DYT-*PANK2-*(NBIA)352 | Pantothenate kinase-associated neurodegeneration (PKAN) | Dystonia, parkinsonism, chorea, tremor, spasticity, cognitive decline, apraxia of eyelid opening, retinopathy, optic atrophy, psychiatric symptoms, muscle atrophy | 234200 | AR |
| DYT/PARK-*GLB1*708 | Dystonia-parkinsonism | Dystonia, parkinsonism, pyramidal signs, cognitive deficits, skeletal abnormalities, short stature, corneal clouding, cardiomyopathy | 230600 | AR |
| DYT/PARK-*SPR*342 | Sepiapterin reductase deficiency | Dystonia, parkinsonism, motor and speech delay, truncal hypotonia, limb hypertonia and hyperreflexia, oculogyric crises, psychiatric symptoms, autonomic dysfunction, diurnal fluctuation | 612716 | AR |
| DYT/PARK-*CP-*(NBIA)327 | Aceruloplasminemia | Dystonia, parkinsonism, chorea, cognitive impairment, retinopathy, blepharospasm, systemic hemosiderosis, diabetes, anemia | 604290 | AR |
| DYT/PARK-*PLA2G6-*(NBIA)*I,* 335 | PLA2G6-associated neurodegeneration (PLAN) | *PLA2G6*-associated neurodegeneration (PLAN): dystonia, parkinsonism, cognitive decline, pyramidal signs, psychiatric symptoms (adult phenotype), ataxia (childhood phenotype)  Iron accumulation: GP, SN in some; adults may have striatal involvement; about half of INAD and the majority of adult-onset cases lack brain iron accumulation on MRI | 612953 | AR |
| PARK-*GBA709* | Gaucher disease type III or subacute neuronopathic type | Myoclonus, developmental delay, dementia, psychiatric symptoms, spastic paraparesis, horizontal supranuclear gaze palsy, abnormal saccade eye movements, strabismus, seizures, short stature, hepatosplenomegaly, pancytopenia | 231000 | AR |
| *AAAS*710 | Achalasia-addisonianism-alacrimia syndrome or Triple-A syndrome or Allgrove syndrome | Developmental delay, mental retardation, pyramidal signs, distal muscle weakness and atrophy, achalasia, autonomic dysfunction, anisocoria, peripheral neuropathy, adrenal insufficiency, alacrimia, optic atrophy, short stature, hyperpigmentation, hyperkeratosis of the palms and soles | 231550 | AR |
| *AARS2*711 | Progressive leukoencephalopathy with ovarian failure | Dystonia, tremor, developmental delay, cognitive decline, apraxia, psychiatric symptoms, nystagmus, spasticity, premature ovarian failure | 615889 | AR |
| *ABCD1*712 | X-linked adrenoleukodystrophy | Visual disturbances, sensation deficits, spastic paraplegia, autonomic failure, adrenal dysfunction | 300100 | XLR |
| *ARX*713 | Partington syndrome | Dystonia, mental retardation, spasticity, seizures, morphological abnormalities | 309510 | XLR |
| *ATAD3A*714, 715 | Harel-Yoon syndrome | Developmental delay, intellectual disability, optic atrophy, nystagmus, spasticity, distal limb muscle atrophy, peripheral neuropathy, scoliosis, dysmorphism, hypertrophic cardiomyopathy | 617183 | AR or AD |
| *AUH*716 | 3-methylglutaconic aciduria type I | Dystonia, developmental delay, failure to thrive, mental retardation, spastic quadriplegia, cognitive impairment, hyperreflexia, metabolic acidosis, febrile seizures, optic atrophy | 250950 | AR |
| *BCS1L, COX10, COX15, FOXRED1, NDUFAF2, NDUFS3, NDUFS4, NDUFAF6, NDUFS7, NDUFS8, NDUFA10, SDHA, SURF1*717 | Leigh syndrome | Dystonia, failure to thrive, psychomotor retardation, mental retardation, pyramidal signs, seizures, psychiatric symptoms, lactic acidosis, hypertrochosis, respiratory failure, pigmentary retinopathy, ptosis, strabismus, nystagmus, optic atrophy, ophthalmoplegia | 256000 | Mt/AR |
| *C5orf42718* | Joubert syndrome 17 (JBTS17) | Developmental delay, oculomotor apraxia, polydactyly, syndactyly, abnormal breathing pattern | 614615 | AR |
| *C9orf72* | Frontotemporal dementia (FTD) and/or Amyotrophic Lateral Sclerosis (ALS) | Broad phenotypic spectrum including frontotemporal dementia and features of motor neuron disease, parkinsonism (mostly atypical, e.g., PSP-like, MSA or CBS), and dystonia, cerebellar signs, or chorea | 105550 | AD, repeat expansion |
| *CTC1719* | Cerebroretinal microangiopathy with calcifications and cysts or Coats plus syndrome | Dystonia, tremor, intrauterine growth retardation, growth retardation, seizures, spasticity, hemiplegia, cognitive decline, pyramidal signs, bone marrow failure, skin and hair abnormalities, intracranial calcifications, skeletal deformities, intestinal bleeding, retinopathy, optic atrophy, blindness | 612199 | AR |
| *CTDP1720* | Congenital cataracts with facial dysmorphism and neuropathy | Chorea, developmental delay, congenital cataracts, nystagmus, cognitive impairment, pyramidal signs, peripheral neuropathy, hypo- or hypergonadotrophic hypogonadism, acute rhabdomyolysis, pes cavus, talipes equinovarus, skeletal deformities, facial dysmorphism | 604168 | AR |
| *CTSA721* | Galactosialidosis | Myoclonus, mental retardation, seizures, angiokeratoma, facial dysmorphism, conjunctival telangiectases, macular cherry red spot, hearing loss, hemangiomas, hepatosplenomegaly, dysostosis multiplex, cardiac valvular disease | 256540 | AR |
| *CTSF722* | Neuronal ceroid lipofuscinosis 13 (CLN13) | Myoclonus, tremor, dementia, perioral dyskinesias, pyramidal signs, seizures, psychiatric symptoms | 615362 | AR |
| *CUL4B723* | Cabezas type of X-linked syndromic mental retardation | Tremor, mental retardation, speech delay, hypogonadism, short stature, facial dysmorphism, skeletal abnormalities, seizures, psychiatric symptoms, central obesity, macrocephaly | 300354 | XLR |
| *DKC1724* | X-linked dyskeratosis congenital (ataxia reported in the severe variant: Hoyeraal-Hreidarsson syndrome) | Intrauterine growth retardation, developmental delay, mental retardation, microcephaly, multisystem involvement, bone marrow failure resulting in immunodeficiency, enteropathy, strabismus, cataracts, optic atrophy, sparse eyelashes, conjunctival leukoplakia, short stature, pulmonary fibrosis, liver failure, skin atrophy, nail dystrophy, carcinomas, leukemia | 305000 | XLR |
| *DLD725* | Dihydrolipoamide dehydrogenase deficiency or Maple syrup urine disease type II | Dystonia, developmental delay, episodic encephalopathy, seizures, lactic or metabolic acidosis, recurrent vomiting, hepatomegaly, liver dysfunction, hypertrophic cardiomyopathy, microcephaly | 246900 | AR |
| *EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5726* | Leukoencephalopathies with vanishing white matter | Mental and motor retardation or regression, cognitive impairment, psychiatric symptoms, optic atrophy, pyramidal signs, seizures, ovarian failure, clinical features worsened by head trauma or fever, macrocephaly, lethargy | 603896 | AR |
| *EXOSC3727* | Pontocerebellar hypoplasia type 1B | Tremor, developmental delay, poor growth, axial hypotonia, spasticity, hyperreflexia, lack of speech, seizures, peripheral neuropathy, muscle atrophy and weakness, tongue atrophy and fasciculations, foot deformities, joint contractures, respiratory insufficiency, oculomotor apraxia, nystagmus, strabismus, retinopathy, microcephaly | 614678 | AR |
| *GALC728* | Krabbe disease or galacto-cerebrosidase deficiency | Failure to thrive, developmental delay or regression, deafness, blindness, optic atrophy, nystagmus, hypersensitive to stimuli, irritability, spastic tetraparesis, seizures, muscular, decerebrate posturing, peripheral neuropathy, pes cavus, tongue atrophy, episodic fever | 245200 | AR |
| *GFAP*729 | Spastic ataxia730 | Usually presenting with infantile onset megalencephaly, (pseudo)bulbar signs, spasticity, cognitive deficits, developmental delay, white matter changes (Alexander disease) | 137780 | AD |
| *GJC2731* |  | Tremor, slow saccades, sensorineural hearing loss, strabismus, spastic paraparesis, other pyramidal signs, pes cavus, scoliosis, seizures, cognitive impairment | 613206 | AR |
| *GPR56 or ADGRG1732* | Bilateral frontoparietal polymicrogyria | Developmental delay, mental retardation, pyramidal signs, esotropia, exotropia, strabismus, nystagmus, seizures | 606854 | AR |
| *HEPACAM733* | Autosomal recessive megalencephalic leukoencephalopathy with subcortical cysts type 2A | Developmental delay, mental retardation, cognitive decline, spasticity, seizures, macrocephaly | 613925 | AR |
| *HIBCH734* | 3-hydroxyisobutyrl-CoA hydrolase deficiency | Dystonia, myoclonus, developmental delay or regression, seizures, nystagmus, strabismus, facial dysmorphism, head titubation, persistent vomiting | 250620 | AR |
| *HSD17B4J, 735* | Perrault syndrome type 1 | Sensorineural deafness, ovarian dysgenesis, primary amenorrhea, developmental delay, mental retardation, spastic diplegia, pes cavus, pes equinovarus, peripheral neuropathy, nystagmus, short stature, scoliosis | 233400 | AR |
| *LRPPRC736* | French Canadian type of Leigh syndrome | Tremor, developmental delay, mental retardation, failure to thrive, language delay, seizures, lactic acidosis, metabolic crises, strabismus, facial dysmorphism, liver dysfunction | 220111 | AR |
| *LYST737* | Chediak-Higashi syndrome | Parkinsonism, tremor, mental retardation, cranial nerve palsies, spastic paraparesis, peripheral neuropathy, foot drop, seizures, anemia, recurrent cutaneous and systemic pyogenic infections, severe immune deficiency, hair hypopigmentation, reduced visual acuity, nystagmus, strabismus, reduced iris pigmentation, macular hypoplasia, hepatosplenomegaly, jaundice | 214500 | AR |
| *MAG284* |  | Developmental delay, cognitive impairment, impaired distal vibration sense, peripheral neuropathy, distal muscle atrophy in lower limbs, spastic paraplegia, optic atrophy, nystagmus, visual impairment | 616680 | AR |
| *MECP2738* | Lubs X-linked mental retardation syndrome | Chorea, psychomotor retardation, macro- or microcephaly, facial dysmoprhism, seizures, spasticity, recurrent respiratory infections, cryptorchidism, asymmetric skull, stereotypic hand movements, autistic features, depression, compulsions, psychosis | 300260 | XLR |
| *MFSD8739* | Neuronal ceroid lipofuscinosis 7 (CLN7) | Developmental regression, mental retardation, cognitive impairment, optic atrophy, retinopathy, blindness, seizures, myoclonus | 610951 | AR |
| *MKS1740* | Joubert syndrome 28 (JBTS28) | Developmental delay, intellectual disability, nystagmus, oculomotor apraxia, retinopathy | 617121 | AR |
| *MMACHC741* | cblC type of combined methylmalonic aciduria and homocystinuria | Tremor, failure to thrive, developmental delay, mental retardation, dementia, retinopathy, visual deficits, nystagmus, facial dysmorphism, seizures, hypergonadotropic hypogonadism, anemia, renal failure, microcephaly | 277400 | AR |
| *MPV17742* | Mitochondrial DNA depletion syndrome-6 or Navajo neurohepatopathy | Dystonia, neonatal jaundice, failure to thrive, developmental delay, peripheral neuropathy, hypo-areflexia, pain insensitivity, acral ulceration and osteomyelitis leading to autoamputation, painless fractures due to injury, distal muscle weakness, lactic acidosis, systemic infections, liver dysfunction, Reye syndrome-like episodes, short stature | 256810 | AR |
| *MTFMT743* | Combined oxidative phosphorylation deficiency type 15 | Tremor, developmental delay, cognitive impairment, pyramidal signs, seizures, strabismus, nystagmus, optic atrophy, short stature, obesity, cardiopathy | 614947 | AR |
| *MTTP744* | Abetalipoproteinemia | Peripheral neuropathy, retinopathy, acanthocytosis, steatorrhea (celiac-like syndrome), hepatic steatosis | 200100 | AR |
| *MVK745* | Mevalonic aciduria | Developmental delay, psychomotor retardation, failure to thrive, recurrent febrile crises with lymphadenopathy, hepatosplenomegaly, anemia, morbilliform rash, kyphoscoliosis, arthralgias, facial dysmorphism, nystagmus, central cataracts, retinal dystrophy, microcephaly | 610377 | AR |
| *NPHP1746* | Joubert syndrome 4 (JBTS4) | Developmental delay, mental retardation, congenital head tilt, abnormal eye movements, nystagmus, oculomotor apraxia, hypometric saccades, tubulointerstitial medullary cystic kidney disease, nephronophthisis, renal failures | 609583 | AR |
| *NUBPL747* | Mitochondrial complex I deficiency | Developmental delay, strabismus, nystagmus, contractures, spasticity, cognitive decline | 252010 | AR/Mt/AD |
| *OPA1748* | Behr syndrome or infantile hereditary optic atrophy with neurologic abnormalities | Tremor, developmental delay, mental retardation, optic atrophy, progressive visual loss, nystagmus, spasticity, pyramidal signs, myopathy, posterior column sensory loss, peripheral neuropathy, tendon and muscular contractures | 210000 | AR |
| *PDHX or PDX1749* | Lactic acidemia due to PDX1 deficiency | Dystonia, developmental delay, mental retardation, microcephaly, optic atrophy, hypertelorism, facial dysmorphism, spastic quadriplegia, seizures, lactic or metabolic acidosis | 245349 | AR |
| *PEX2750* | Peroxisome biogenesis disorder 5B or Zellweger spectrum disorders | Tremor, developmental delay, peripheral neuropathy, pes cavus, hypoacusia, slow saccades, oculomotor apraxia, nystagmus, retinopathy, strabismus | 614867 | AR |
| *PEX6534* |  | Optic atrophy, blindness, cochlear degeneration, deafness | 271250 (SCAR3) | AR |
| *PNKP599,751,752* | Ataxia with oculomotor apraxia 4 (AOA4), Microcephaly, seizures, and developmental delay, and Charcot-Marie-Tooth disease type 2B2 | Dystonia, developmental delay, mental retardation, cognitive impairment, oculomotor apraxia, tetraplegia, impaired vibration sense, peripheral neuropathy, pes cavus, hammertoes, distal muscle weakness and atrophy, seizures, microcephaly | 616267, 613402, 605589 | AR |
| *PNP753* | Purine nucleoside phosphorylase deficiency or Nucleoside phosphorylase deficiency | Tremor, developmental delay, failure to thrive, mental retardation, spastic diplegia, tetraparesis, behavioral disorder, autoimmune hemolytic anemia, frequent infections, splenomegaly, pneumonia | 613179 | AR |
| *PNPLA6754,755,756* | Pure cerebellar ataxia with homozygous mutations in the PNPLA6 gene and allelic disorders: Laurence-Moon syndrome and Oliver-McFarlane syndrome | Pure cerebellar ataxia  Laurence-Moon syndrome: mental retardation, growth retardation, retinopathy, choroidal atrophy, nystagmus, peripheral neuropathy, spastic paraplegia, short stature, pituitary dysfunction, micropenis, hypoplastic scrotum  Oliver-McFarlane syndrome: trichomegaly, chorioretinal dystrophy, nystagmus, ring iris heterochromia, short stature, mental retardation, hypogonadotropic hypogonadism, peripheral neuropathy, spastic paraplegia, frontal alopecia | 215470, 245800, 275400 | AR and isolated cases |
| *POLR1CK, 757* | Hypomyelinating leukodystrophy type 11 | Tremor, developmental delay, intellectual disability, spasticity, myopia, dental abnormalities, head titubation | 616494 | AR |
| *PRF1758,759* | Familial hemophagocytic lymphohistiocytosis type 2 and the allelic disorder of recurrent immune-mediated neurodegeneration | Developmental delay, failure to thrive, meningitis, encephalitis, hemiplegia, tetraplegia, seizures, coma, pancytopenia, coagulation abnormalities, lymphadenopathy, fever, edema, liver dysfunction  Neurodegeneration triggered by infections, recurrent subacute post-viral onset of ataxia, primary immunodeficiency | 603553, 170280 | AR |
| *PRPS1760, 761, 762* | Allelic disorders or continuum with Arts syndrome, X-linked recessive Charcot-Marie-Tooth disease-5 or Rosenberg-Chutorian syndrome and Hyperuricemia, mental retardation and sensorineural deafness with PRPS1 superactivity. | Developmental delay, mental retardation, poor growth, sensorineural hearing loss, optic atrophy, retinopathy, nystagmus, muscle weakness, hyperreflexia, peripheral neuropathy, distal muscle weakness and atrophy, distal sensory impairment, pes cavus, flaccid tetraplegia, immune deficiency, recurrent respiratory tract infections, uric acid urolithiasis, secondary renal insufficiency, gout, gouty arthritis | 301835, 300661, 311070 | XLR |
| *PRX763* |  | Distal and proximal lower limb muscle weakness and atrophy, peripheral neuropathy, pes cavus, scoliosis, delayed motor development | 614895 | AR |
| *PSEN1764-767* | Alzheimer’s disease | Gene is linked to Alzheimer’s disease; a few cases with prominent (spastic) ataxia have been described. | 607822 | AD |
| *RARS768* |  | Tremor, developmental delay, mental retardation, pyramidal signs, nystagmus, altered smooth pursuit, microcephaly | 616140 | AR |
| *RELN769* | Norman-Roberts type of Lissencephaly | Microcephaly, facial dysmorphism, mental retardation, nystagmus, seizures, congenital lymphedema | 257320 | AR |
| *ROGDI770* | Kohlschutter-Tonz syndrome | Developmental delay, mental retardation, cognitive impairment, spasticity, seizures, amelogenesis imperfecta, enamel hypoplasia, discolored teeth | 226750 | AR |
| *RRM2B771* | Mitochondrial DNA depletion syndrome 8B (MNGIE type) | Failure to thrive, lactic acidosis, proximal renal tubulopathy, seizures, external ophthalmoplegia, ptosis, gastrointestinal dysmotility, cachexia, peripheral neuropathy | 612075 | AR |
| *RTN4IP1772* | Optic atrophy type 10 with or without ataxia, mental retardation, and seizures | Mental retardation, photophobia, nystagmus, reduced visual acuity, color vision impairment of red/green axis, optic atrophy, central scotoma seizures | 616732 | AR |
| *SLC2A1773* |  | Dystonia, developmental delay, seizures, myoclonic epilepsy, spasticity | 606777 | AR or AD |
| *SLC6A19774* | Hartnup disease | Delayed cognitive development, psychiatric symptoms, seizures, hypertonia, light-sensitive dermatitis, atrophic glossitis | 234500 | AR |
| *SLC16A2775* | Allan-Herndon-Dudley syndrome or monocarboxylate transporter type 8 deficiency | Dystonia, developmental delay, mental retardation, pyramidal signs, amyotrophy, behavior disorders, scoliosis, nystagmus, facial dysmorphism, microcephaly, pectus excavatum | 300523 | XLR |
| *SLC19A3776* | Thiamine metabolism dysfunction syndrome type 2 or biotin-thiamine-responsive basal ganglia disease | Dystonia, psychomotor retardation, encephalopathy, coma, psychiatric symptoms, external ophthalmoplegia, nystagmus, ptosis, gaze palsy, seizures, pyramidal signs, paraparesis, rigidity | 607483 | AR |
| *SLC25A15777* | Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome | Psychomotor retardation, failures to thrive, mental retardation, lethargy, episodic confusion, acute encephalopathy, coma, pyramidal signs, myoclonic epilepsy, decreased vibration sense, coagulopathy due to liver dysfunction | 238970 | AR |
| *SLC25A46778* | Hereditary motor and sensory neuropathy type VIB or Charcot-Marie-Tooth disease type 6B | Myoclonus, delayed development, optic atrophy, pyramidal signs, peripheral neuropathy, distal sensory impairment, pes cavus, morphological abnormalities | 616505 | AR |
| *SLC52A2779* | Brown-Vialetto-Van Laere syndrome type 2 | Cranial nerve palsies, bulbar palsy, optic atrophy, nystagmus, visual loss, absent pupillary reflex, sensorineural hearing loss, peripheral neuropathy, muscle weakness and atrophy, tongue fasciculations, psychiatric symptoms, claw hands, scoliosis, respiratory insufficiency | 614707 | AR |
| *SNORD118780* | Leukoencephalopathy, brain calcifications, and cysts | Dystonia, tremor, seizures, spasticity, hemiplegia, cognitive decline, pyramidal signs | 614561 | AR |
| *SUOX781* | Sulfocysteinuria or sulfite oxidase deficiency | Dystonia, developmental delay, infantile hemiplegia, seizures, behavior disorders, fine hair, eczema, delayed teething, ectopia lentis | 272300 | AR |
| *SURF1782* |  | Peripheral neuropathy, distal muscle weakness and atrophy, kyphoscoliosis, nystagmus, sensorineural hearing loss | 616684 | AR |
| *TCTN1783* | Joubert syndrome 13 (JBTS13) | Cognitive impairment, limb abnormalities | 614173 | AR |
| *TCTN2784* | Joubert syndrome 24 (JBTS24) | Developmental delay, absent speech, pyramidal signs, nystagmus, hyperopia, polydactyly, talipes equinovarus | 616654 | AR |
| *TCTN3785* | Joubert syndrome 18 (JBTS18) | Mental retardation, abnormal eye movements, facial dysmorphism, scoliosis, polydactyly, camptodactyly, breathing anomalies, ventricular septal defect, horseshoe kidney | 614815 | AR |
| *TRAPPC11786* | Limb-girdle muscular dystrophy type 2S | Dystonia, chorea, tremor, developmental delay, proximal muscle weakness, muscle cramps, scapular winging, scoliosis, hip dysplasia, cataracts, strabismus, myopia, microcephaly, short stature | 615356 | AR |
| *TRNT1787* | Congenital sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay | Severe sideroblastic anemia, developmental delay, growth retardation, lactic acidosis, recurrent fevers, brittle hair, nephrocalcinosis, cardiomyopathy, retinopathy, sensorineural hearing loss seizures | 616084 | AR |
| *VARS2788* | Combined oxidative phosphorylation deficiency type 20 | Developmental delay, ptosis, progressive external ophthalmoplegia, seizures, facial dysmorphism, microcephaly | 615917 | AR |
| *VRK1789* |  | Psychomotor retardation, mental retardation, microcephaly, nystagmus, muscle weakness, distal spinal muscular atrophy, fasciculations, peripheral neuropathy, hyperreflexia, foot deformities, skeletal contractures, arthrogryposis, scoliosis, respiratory insufficiency | 607596 | AR |
| *WFS1790* | Wolfram syndrome-1 or Diabetes insipidus and mellitus with optic atrophy and deafness | Parkinsonism, tremor, myoclonus, mental retardation or dementia, poor growth, optic atrophy, retinopathy, ptosis, nystagmus, sensorineural hearing loss, hyposmia, seizures, peripheral neuropathy, stroke-like episodes, psychiatric symptoms, diabetes mellitus and diabetes insipidus, hypothyroidism, hydronephrosis, testicular atrophy, cardiomyopathy | 222300 | AR |
| *XRCC4791* | Short stature, microcephaly, and endocrine dysfunction syndrome | Intrauterine growth failure, developmental delay, cognitive impairment, apraxia, pyramidal signs, peripheral neuropathy, dyslipidemia, diabetes mellitus, hypothyroidism, anemia, acanthosis nigricans, cryptorchidism, renal dysgenesis, malpositioned teeth, facial dysmorphism, short stature, microcephaly | 616541 | AR |

AD = autosomal dominant, AOA = Ataxia with oculomotor apraxia, AR = autosomal recessive, ARCA = autosomal recessive cerebellar ataxia, GP = Globus pallidus, HD = Huntington’s disease, MOI = Mode of inheritance, OMIM = Online Mendelian Inheritance in Man (<https://www.omim.org/about>), SCA = Spinocerebellar ataxia, SCAN = Spinocerebellar ataxia with axonal neuropathy, SCAR = Recessive spinocerebellar ataxia, SN = Subthalamic nucleus, SPAX = Spastic ataxia, SPG = Spastic paraplegia, XLR = x-linked recessive

\* Comment: Evidence is limited as only two patients in total were reported in two independent publications.

A Allelic with episodic ataxia type 2 and familial hemiplegic migraine type 1.

B Gene mutations can also cause complex cortical dysplasia with other brain malformations 5 (OMIM: 615763).

C Gene mutations can also cause congenital disorder of glycosylation, type 1AA (OMIM: 617082).

D Allelic with Brown-Vialetto-Van Laere syndrome type 2.

E Gene mutations can also cause the lethal neonatal rigidity and multifocal seizure syndrome (OMIM: 614498).

F Gene mutations can also cause autosomal recessive microcephaly, seizures, and developmental delay (OMIM: 613402).

G Allelic with autosomal recessive spastic ataxia at the SAX2 locus.

H Allelic with neuronal ceroid lipofuscinosis type 2 (CLN2, OMIM: 204500).

I Gene mutations more commonly cause infantile neuroaxonal dystrophy (INAD) with developmental delay/regression, hypotonia, spasticity/pyramidal signs, optic nerve atrophy, sensorimotor neuropathy, and seizures.

J Allelic with Peroxisomal D-bifunctional protein deficiency.

K Allelic with Treacher Collins syndrome type 3.

**References**

228. Valence S, Cochet E, Rougeot C, et al. Exome sequencing in congenital ataxia identifies two new candidate genes and highlights a pathophysiological link between some congenital ataxias and early infantile epileptic encephalopathies. Genet Med 2019;21(3):553-563.

253. Masnada S, Hedrich UBS, Gardella E, et al. Clinical spectrum and genotype-phenotype associations of KCNA2-related encephalopathies. Brain 2017;140(9):2337-2354.

327. Harris ZL, Takahashi Y, Miyajima H, Serizawa M, MacGillivray RT, Gitlin JD. Aceruloplasminemia: molecular characterization of this disorder of iron metabolism. Proceedings of the National Academy of Sciences of the United States of America 1995;92(7):2539-2543.

335. Morgan NV, Westaway SK, Morton JE, et al. PLA2G6, encoding a phospholipase A2, is mutated in neurodegenerative disorders with high brain iron. Nat Genet 2006;38(7):752-754.

342. Bonafe L, Thony B, Penzien JM, Czarnecki B, Blau N. Mutations in the sepiapterin reductase gene cause a novel tetrahydrobiopterin-dependent monoamine-neurotransmitter deficiency without hyperphenylalaninemia. Am J Hum Genet 2001;69(2):269-277.

348. Pulst SM, Nechiporuk A, Nechiporuk T, et al. Moderate expansion of a normally biallelic trinucleotide repeat in spinocerebellar ataxia type 2. Nat Genet 1996;14(3):269-276.

349. Shan DE, Soong BW, Sun CM, Lee SJ, Liao KK, Liu RS. Spinocerebellar ataxia type 2 presenting as familial levodopa-responsive parkinsonism. Annals of neurology 2001;50(6):812-815.

352. Zhou B, Westaway SK, Levinson B, Johnson MA, Gitschier J, Hayflick SJ. A novel pantothenate kinase gene (PANK2) is defective in Hallervorden-Spatz syndrome. Nat Genet 2001;28(4):345-349.

362. Edvardson S, Hama H, Shaag A, et al. Mutations in the fatty acid 2-hydroxylase gene are associated with leukodystrophy with spastic paraparesis and dystonia. Am J Hum Genet 2008;83(5):643-648.

364. Lynch DS, Wood NW, Houlden H. Late-onset Lafora disease with prominent parkinsonism due to a rare mutation in EPM2A. Neurol Genet 2016;2(5):e101.

365. Yildiz EP, Yesil G, Ozkan MU, Bektas G, Caliskan M, Ozmen M. A novel EPM2A mutation in a patient with Lafora disease presenting with early parkinsonism symptoms in childhood. Seizure 2017;51:77-79.

407. Bull PC, Thomas GR, Rommens JM, Forbes JR, Cox DW. The Wilson disease gene is a putative copper transporting P-type ATPase similar to the Menkes gene. Nature genetics 1993;5(4):327-337.

458. Muto V, Flex E, Kupchinsky Z, et al. Biallelic SQSTM1 mutations in early-onset, variably progressive neurodegeneration. Neurology 2018;91(4):e319-e330.

459. Haack TB, Ignatius E, Calvo-Garrido J, et al. Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. Am J Hum Genet 2016;99(3):735-743.

463. Bouslam N, Bouhouche A, Benomar A, et al. A novel locus for autosomal recessive spastic ataxia on chromosome 17p. Human genetics 2007;121(3-4):413-420.

464. Dor T, Cinnamon Y, Raymond L, et al. KIF1C mutations in two families with hereditary spastic paraparesis and cerebellar dysfunction. J Med Genet 2014;51(2):137-142.

467. Koob MD, Moseley ML, Schut LJ, et al. An untranslated CTG expansion causes a novel form of spinocerebellar ataxia (SCA8). Nat Genet 1999;21(4):379-384.

468. Zhuchenko O, Bailey J, Bonnen P, et al. Autosomal dominant cerebellar ataxia (SCA6) associated with small polyglutamine expansions in the alpha 1A-voltage-dependent calcium channel. Nat Genet 1997;15(1):62-69.

469. Serrano-Munuera C, Corral-Juan M, Stevanin G, et al. New subtype of spinocerebellar ataxia with altered vertical eye movements mapping to chromosome 1p32. JAMA Neurol 2013;70(6):764-771.

470. Seixas AI, Loureiro JR, Costa C, et al. A Pentanucleotide ATTTC Repeat Insertion in the Non-coding Region of DAB1, Mapping to SCA37, Causes Spinocerebellar Ataxia. Am J Hum Genet 2017;101(1):87-103.

471. Corral-Juan M, Serrano-Munuera C, Rabano A, et al. Clinical, genetic and neuropathological characterization of spinocerebellar ataxia type 37. Brain 2018;141(7):1981-1997.

472. Cadieux-Dion M, Turcotte-Gauthier M, Noreau A, et al. Expanding the clinical phenotype associated with ELOVL4 mutation: study of a large French-Canadian family with autosomal dominant spinocerebellar ataxia and erythrokeratodermia. JAMA Neurol 2014;71(4):470-475.

473. Ozaki K, Doi H, Mitsui J, et al. A Novel Mutation in ELOVL4 Leading to Spinocerebellar Ataxia (SCA) With the Hot Cross Bun Sign but Lacking Erythrokeratodermia: A Broadened Spectrum of SCA34. JAMA Neurol 2015;72(7):797-805.

474. Di Gregorio E, Borroni B, Giorgio E, et al. ELOVL5 mutations cause spinocerebellar ataxia 38. American Journal of Human Genetics 2014;95(2):209-217.

475. van Swieten JC, Brusse E, de Graaf BM, et al. A mutation in the fibroblast growth factor 14 gene is associated with autosomal dominant cerebellar ataxia [corrected]. Am J Hum Genet 2003;72(1):191-199.

476. Iwaki A, Kawano Y, Miura S, et al. Heterozygous deletion of ITPR1, but not SUMF1, in spinocerebellar ataxia type 16. Journal of medical genetics 2008;45(1):32-35.

477. van de Leemput J, Chandran J, Knight MA, et al. Deletion at ITPR1 underlies ataxia in mice and spinocerebellar ataxia 15 in humans. PLoS Genet 2007;3(6):e108.

478. Waters MF, Minassian NA, Stevanin G, et al. Mutations in voltage-gated potassium channel KCNC3 cause degenerative and developmental central nervous system phenotypes. Nat Genet 2006;38(4):447-451.

479. Lee YC, Durr A, Majczenko K, et al. Mutations in KCND3 cause spinocerebellar ataxia type 22. Annals of neurology 2012;72(6):859-869.

480. Bakalkin G, Watanabe H, Jezierska J, et al. Prodynorphin mutations cause the neurodegenerative disorder spinocerebellar ataxia type 23. Am J Hum Genet 2010;87(5):593-603.

481. Holmes SE, O'Hearn EE, McInnis MG, et al. Expansion of a novel CAG trinucleotide repeat in the 5' region of PPP2R2B is associated with SCA12. Nat Genet 1999;23(4):391-392.

482. Chen DH, Brkanac Z, Verlinde CL, et al. Missense mutations in the regulatory domain of PKC gamma: a new mechanism for dominant nonepisodic cerebellar ataxia. Am J Hum Genet 2003;72(4):839-849.

483. Ikeda Y, Dick KA, Weatherspoon MR, et al. Spectrin mutations cause spinocerebellar ataxia type 5. Nat Genet 2006;38(2):184-190.

484. Wang JL, Yang X, Xia K, et al. TGM6 identified as a novel causative gene of spinocerebellar ataxias using exome sequencing. Brain : a journal of neurology 2010;133(Pt 12):3510-3518.

485. Banfi S, Servadio A, Chung MY, et al. Identification and characterization of the gene causing type 1 spinocerebellar ataxia. Nat Genet 1994;7(4):513-520.

486. Di Bella D, Lazzaro F, Brusco A, et al. Mutations in the mitochondrial protease gene AFG3L2 cause dominant hereditary ataxia SCA28. Nat Genet 2010;42(4):313-321.

487. Koide R, Ikeuchi T, Onodera O, et al. Unstable expansion of CAG repeat in hereditary dentatorubral-pallidoluysian atrophy (DRPLA). Nat Genet 1994;6(1):9-13.

488. Nørremølle A, Nielsen JE, Sørensen SA, Hasholt L. Elongated CAG repeats of the B37 gene in a Danish family with dentato-rubro-pallido-luysian atrophy. Human genetics 1995;95(3):313-318.

489. Trottier Y, Lutz Y, Stevanin G, et al. Polyglutamine expansion as a pathological epitope in Huntington's disease and four dominant cerebellar ataxias. Nature 1995;378(6555):403-406.

490. Matsuura T, Yamagata T, Burgess DL, et al. Large expansion of the ATTCT pentanucleotide repeat in spinocerebellar ataxia type 10. Nat Genet 2000;26(2):191-194.

491. Sato N, Amino T, Kobayashi K, et al. Spinocerebellar ataxia type 31 is associated with "inserted" penta-nucleotide repeats containing (TGGAA)n. Am J Hum Genet 2009;85(5):544-557.

492. Coutelier M, Blesneac I, Monteil A, et al. A Recurrent Mutation in CACNA1G Alters Cav3.1 T-Type Calcium-Channel Conduction and Causes Autosomal-Dominant Cerebellar Ataxia. Am J Hum Genet 2015;97(5):726-737.

493. Morino H, Matsuda Y, Muguruma K, et al. A mutation in the low voltage-gated calcium channel CACNA1G alters the physiological properties of the channel, causing spinocerebellar ataxia. Mol Brain 2015;8:89.

494. Chemin J, Siquier-Pernet K, Nicouleau M, et al. De novo mutation screening in childhood-onset cerebellar atrophy identifies gain-of-function mutations in the CACNA1G calcium channel gene. Brain 2018;141(7):1998-2013.

495. Tsoi H, Yu AC, Chen ZS, et al. A novel missense mutation in CCDC88C activates the JNK pathway and causes a dominant form of spinocerebellar ataxia. J Med Genet 2014;51(9):590-595.

496. Lenska-Mieciek M, Charzewska A, Krolicki L, et al. Familial ataxia, tremor, and dementia in a polish family with a novel mutation in the CCDC88C gene. Mov Disord 2019;34(1):142-144.

497. Winkelmann J, Lin L, Schormair B, et al. Mutations in DNMT1 cause autosomal dominant cerebellar ataxia, deafness and narcolepsy. Hum Mol Genet 2012;21(10):2205-2210.

498. Chao HT, Davids M, Burke E, et al. A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. Am J Hum Genet 2017;100(1):128-137.

499. Harms FL, Girisha KM, Hardigan AA, et al. Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. Am J Hum Genet 2017;100(1):117-127.

500. Sleven H, Welsh SJ, Yu J, et al. De Novo Mutations in EBF3 Cause a Neurodevelopmental Syndrome. Am J Hum Genet 2017;100(1):138-150.

501. Finnsson J, Sundblom J, Dahl N, Melberg A, Raininko R. LMNB1-related autosomal-dominant leukodystrophy: Clinical and radiological course. Ann Neurol 2015;78(3):412-425.

502. Zhang Y, Li J, Bai R, et al. LMNB1-Related Adult-Onset Autosomal Dominant Leukodystrophy Presenting as Movement Disorder: A Case Report and Review of the Literature. Front Neurosci 2019;13:1030.

503. Kobayashi H, Abe K, Matsuura T, et al. Expansion of intronic GGCCTG hexanucleotide repeat in NOP56 causes SCA36, a type of spinocerebellar ataxia accompanied by motor neuron involvement. Am J Hum Genet 2011;89(1):121-130.

504. Gennarino VA, Palmer EE, McDonell LM, et al. A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. Cell 2018;172(5):924-936 e911.

505. Lai KL, Liao YC, Tsai PC, Hsiao CT, Soong BW, Lee YC. Investigating PUM1 mutations in a Taiwanese cohort with cerebellar ataxia. Parkinsonism Relat Disord 2019;66:220-223.

506. Chen DH, Below JE, Shimamura A, et al. Ataxia-Pancytopenia Syndrome Is Caused by Missense Mutations in SAMD9L. Am J Hum Genet 2016;98(6):1146-1158.

507. Tesi B, Davidsson J, Voss M, et al. Gain-of-function SAMD9L mutations cause a syndrome of cytopenia, immunodeficiency, MDS, and neurological symptoms. Blood 2017;129(16):2266-2279.

508. Shen XM, Selcen D, Brengman J, Engel AG. Mutant SNAP25B causes myasthenia, cortical hyperexcitability, ataxia, and intellectual disability. Neurology 2014;83(24):2247-2255.

509. Fukuda H, Imagawa E, Hamanaka K, et al. A novel missense SNAP25b mutation in two affected siblings from an Israeli family showing seizures and cerebellar ataxia. J Hum Genet 2018;63(5):673-676.

510. Heyne HO, Singh T, Stamberger H, et al. De novo variants in neurodevelopmental disorders with epilepsy. Nat Genet 2018;50(7):1048-1053.

511. Koide R, Kobayashi S, Shimohata T, et al. A neurological disease caused by an expanded CAG trinucleotide repeat in the TATA-binding protein gene: a new polyglutamine disease? Hum Mol Genet 1999;8(11):2047-2053.

512. Schneider SA, van de Warrenburg BP, Hughes TD, et al. Phenotypic homogeneity of the Huntington disease-like presentation in a SCA17 family. Neurology 2006;67(9):1701-1703.

513. Delplanque J, Devos D, Huin V, et al. TMEM240 mutations cause spinocerebellar ataxia 21 with mental retardation and severe cognitive impairment. Brain : a journal of neurology 2014;137(Pt 10):2657-2663.

514. Sferra A, Fattori F, Rizza T, et al. Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. Hum Mol Genet 2018;27(11):1892-1904.

515. Cai S, Li J, Wu Y, Jiang Y. De novo mutations of TUBB2A cause infantile-onset epilepsy and developmental delay. J Hum Genet 2020;65(7):601-608.

516. Syrbe S, Hedrich UBS, Riesch E, et al. De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. Nat Genet 2015;47(4):393-399.

517. Helbig KL, Hedrich UB, Shinde DN, et al. A recurrent mutation in KCNA2 as a novel cause of hereditary spastic paraplegia and ataxia. Ann Neurol 2016;80(4).

518. Corbett MA, Bellows ST, Li M, et al. Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. Neurology 2016;87(19):1975-1984.

519. Bourassa CV, Meijer IA, Merner ND, et al. VAMP1 mutation causes dominant hereditary spastic ataxia in Newfoundland families. Am J Hum Genet 2012;91(3):548-552.

520. Hamdan FF, Myers CT, Cossette P, et al. High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. Am J Hum Genet 2017;101(5):664-685.

521. Den K, Kudo Y, Kato M, et al. Recurrent NUS1 canonical splice donor site mutation in two unrelated individuals with epilepsy, myoclonus, ataxia and scoliosis - a case report. BMC Neurol 2019;19(1):253.

522. Araki K, Nakamura R, Ito D, et al. NUS1 mutation in a family with epilepsy, cerebellar ataxia, and tremor. Epilepsy Res 2020;164:106371.

523. Vermeer S, Hoischen A, Meijer RP, et al. Targeted next-generation sequencing of a 12.5 Mb homozygous region reveals ANO10 mutations in patients with autosomal-recessive cerebellar ataxia. American journal of human genetics 2010;87(6):813-819.

524. Date H, Onodera O, Tanaka H, et al. Early-onset ataxia with ocular motor apraxia and hypoalbuminemia is caused by mutations in a new HIT superfamily gene. Nature genetics 2001;29(2):184-188.

525. Moreira MC, Barbot C, Tachi N, et al. The gene mutated in ataxia-ocular apraxia 1 encodes the new HIT/Zn-finger protein aprataxin. Nature genetics 2001;29(2):189-193.

526. Burns R, Majczenko K, Xu J, et al. Homozygous splice mutation in CWF19L1 in a Turkish family with recessive ataxia syndrome. Neurology 2014;83(23):2175-2182.

527. Delatycki MB, Knight M, Koenig M, Cossee M, Williamson R, Forrest SM. G130V, a common FRDA point mutation, appears to have arisen from a common founder. Human genetics 1999;105(4):343-346.

528. Lodi R, Cooper JM, Bradley JL, et al. Deficit of in vivo mitochondrial ATP production in patients with Friedreich ataxia. Proc Natl Acad Sci U S A 1999;96(20):11492-11495.

529. Utine GE, Haliloglu G, Salanci B, et al. A homozygous deletion in GRID2 causes a human phenotype with cerebellar ataxia and atrophy. J Child Neurol 2013;28(7):926-932.

530. Assoum M, Salih MA, Drouot N, et al. Rundataxin, a novel protein with RUN and diacylglycerol binding domains, is mutant in a new recessive ataxia. Brain 2010;133(Pt 8):2439-2447.

531. Jobling RK, Assoum M, Gakh O, et al. PMPCA mutations cause abnormal mitochondrial protein processing in patients with non-progressive cerebellar ataxia. Brain 2015;138(Pt 6):1505-1517.

532. Moreira MC, Klur S, Watanabe M, et al. Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. Nature genetics 2004;36(3):225-227.

533. Foley AR, Menezes MP, Pandraud A, et al. Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. Brain 2014;137(Pt 1):44-56.

534. Guissart C, Drouot N, Oncel I, et al. Genes for spinocerebellar ataxia with blindness and deafness (SCABD/SCAR3, MIM# 271250 and SCABD2). Eur J Hum Genet 2016;24(8):1154-1159.

535. Thomas AC, Williams H, Seto-Salvia N, et al. Mutations in SNX14 cause a distinctive autosomal-recessive cerebellar ataxia and intellectual disability syndrome. American journal of human genetics 2014;95(5):611-621.

536. Lise S, Clarkson Y, Perkins E, et al. Recessive mutations in SPTBN2 implicate beta-III spectrin in both cognitive and motor development. PLoS Genet 2012;8(12):e1003074.

537. Gros-Louis F, Dupre N, Dion P, et al. Mutations in SYNE1 lead to a newly discovered form of autosomal recessive cerebellar ataxia. Nature genetics 2007;39(1):80-85.

538. Ouahchi K, Arita M, Kayden H, et al. Ataxia with isolated vitamin E deficiency is caused by mutations in the alpha-tocopherol transfer protein. Nature genetics 1995;9(2):141-145.

539. Hu H, Kahrizi K, Musante L, et al. Genetics of intellectual disability in consanguineous families. Mol Psychiatry 2019;24(7):1027-1039.

540. Aslam F, Naz S. Ataxia and dysarthria due to an ABCA2 variant: Extension of the phenotypic spectrum. Parkinsonism Relat Disord 2019;64:328-331.

541. Fiskerstrand T, H'Mida-Ben Brahim D, Johansson S, et al. Mutations in ABHD12 cause the neurodegenerative disease PHARC: An inborn error of endocannabinoid metabolism. American journal of human genetics 2010;87(3):410-417.

542. Lagier-Tourenne C, Tazir M, Lopez LC, et al. ADCK3, an ancestral kinase, is mutated in a form of recessive ataxia associated with coenzyme Q10 deficiency. American journal of human genetics 2008;82(3):661-672.

543. Mollet J, Delahodde A, Serre V, et al. CABC1 gene mutations cause ubiquinone deficiency with cerebellar ataxia and seizures. American journal of human genetics 2008;82(3):623-630.

544. Ghosh SG, Becker K, Huang H, et al. Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. Am J Hum Genet 2018;103(3):431-439.

545. Danhauser K, Alhaddad B, Makowski C, et al. Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. Am J Hum Genet 2018;103(5):817-825.

546. Ferland RJ, Eyaid W, Collura RV, et al. Abnormal cerebellar development and axonal decussation due to mutations in AHI1 in Joubert syndrome. Nature genetics 2004;36(9):1008-1013.

547. Dixon-Salazar T, Silhavy JL, Marsh SE, et al. Mutations in the AHI1 gene, encoding jouberin, cause Joubert syndrome with cortical polymicrogyria. American journal of human genetics 2004;75(6):979-987.

548. Chambliss KL, Hinson DD, Trettel F, et al. Two exon-skipping mutations as the molecular basis of succinic semialdehyde dehydrogenase deficiency (4-hydroxybutyric aciduria). American journal of human genetics 1998;63(2):399-408.

549. Imbach T, Burda P, Kuhnert P, et al. A mutation in the human ortholog of the Saccharomyces cerevisiae ALG6 gene causes carbohydrate-deficient glycoprotein syndrome type-Ic. Proc Natl Acad Sci U S A 1999;96(12):6982-6987.

550. Cantagrel V, Silhavy JL, Bielas SL, et al. Mutations in the cilia gene ARL13B lead to the classical form of Joubert syndrome. American journal of human genetics 2008;83(2):170-179.

551. Savitsky K, Bar-Shira A, Gilad S, et al. A single ataxia telangiectasia gene with a product similar to PI-3 kinase. Science 1995;268(5218):1749-1753.

552. Nobukuni Y, Mitsubuchi H, Akaboshi I, et al. Maple syrup urine disease. Complete defect of the E1 beta subunit of the branched chain alpha-ketoacid dehydrogenase complex due to a deletion of an 11-bp repeat sequence which encodes a mitochondrial targeting leader peptide in a family with the disease. J Clin Invest 1991;87(5):1862-1866.

553. Mahjoub A, Cihlarova Z, Tetreault M, et al. Homozygous pathogenic variant in BRAT1 associated with nonprogressive cerebellar ataxia. Neurol Genet 2019;5(5):e359.

554. Pomponio RJ, Reynolds TR, Cole H, Buck GA, Wolf B. Mutational hotspot in the human biotinidase gene causes profound biotinidase deficiency. Nature genetics 1995;11(1):96-98.

555. Nikali K, Suomalainen A, Saharinen J, et al. Infantile onset spinocerebellar ataxia is caused by recessive mutations in mitochondrial proteins Twinkle and Twinky. Human molecular genetics 2005;14(20):2981-2990.

556. Turkmen S, Guo G, Garshasbi M, et al. CA8 mutations cause a novel syndrome characterized by ataxia and mild mental retardation with predisposition to quadrupedal gait. PLoS Genet 2009;5(5):e1000487.

557. Butler KM, Holt PJ, Milla SS, da Silva C, Alexander JJ, Escayg A. Epileptic Encephalopathy and Cerebellar Atrophy Resulting from Compound Heterozygous CACNA2D2 Variants. Case Rep Genet 2018;2018:6308283.

558. Punetha J, Karaca E, Gezdirici A, et al. Biallelic CACNA2D2 variants in epileptic encephalopathy and cerebellar atrophy. Ann Clin Transl Neurol 2019;6(8):1395-1406.

559. Sayer JA, Otto EA, O'Toole JF, et al. The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF4. Nature genetics 2006;38(6):674-681.

560. Valente EM, Silhavy JL, Brancati F, et al. Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature genetics 2006;38(6):623-625.

561. Martinez Lyons A, Ardissone A, Reyes A, et al. COA7 (C1orf163/RESA1) mutations associated with mitochondrial leukoencephalopathy and cytochrome c oxidase deficiency. J Med Genet 2016;53(12):846-849.

562. Higuchi Y, Okunushi R, Hara T, et al. Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. Brain 2018;141(6):1622-1636.

563. Paesold-Burda P, Maag C, Troxler H, et al. Deficiency in COG5 causes a moderate form of congenital disorders of glycosylation. Hum Mol Genet 2009;18(22):4350-4356.

564. Wang X, Han L, Wang XY, et al. Identification of Two Novel Mutations in COG5 Causing Congenital Disorder of Glycosylation. Front Genet 2020;11:168.

565. Szklarczyk R, Wanschers BF, Nijtmans LG, et al. A mutation in the FAM36A gene, the human ortholog of COX20, impairs cytochrome c oxidase assembly and is associated with ataxia and muscle hypotonia. Human molecular genetics 2013;22(4):656-667.

566. Cali JJ, Hsieh CL, Francke U, Russell DW. Mutations in the bile acid biosynthetic enzyme sterol 27-hydroxylase underlie cerebrotendinous xanthomatosis. The Journal of biological chemistry 1991;266(12):7779-7783.

567. Davey KM, Parboosingh JS, McLeod DR, et al. Mutation of DNAJC19, a human homologue of yeast inner mitochondrial membrane co-chaperones, causes DCMA syndrome, a novel autosomal recessive Barth syndrome-like condition. J Med Genet 2006;43(5):385-393.

568. Wiltrout K, Ferrer A, van de Laar I, et al. Variants in DOCK3 cause developmental delay and hypotonia. Eur J Hum Genet 2019;27(8):1225-1234.

569. Helbig KL, Mroske C, Moorthy D, Sajan SA, Velinov M. Biallelic loss-of-function variants in DOCK3 cause muscle hypotonia, ataxia, and intellectual disability. Clin Genet 2017;92(4):430-433.

570. Iwata-Otsubo A, Ritter AL, Weckselbatt B, et al. DOCK3-related neurodevelopmental syndrome: Biallelic intragenic deletion of DOCK3 in a boy with developmental delay and hypotonia. Am J Med Genet A 2018;176(1):241-245.

571. Marelli C, Guissart C, Hubsch C, et al. Mini-Exome Coupled to Read-Depth Based Copy Number Variation Analysis in Patients with Inherited Ataxias. Hum Mutat 2016;37(12):1340-1353.

572. Carre G, Marelli C, Anheim M, et al. Xeroderma pigmentosum complementation group F: A rare cause of cerebellar ataxia with chorea. J Neurol Sci 2017;376:198-201.

573. Doi H, Koyano S, Miyatake S, et al. Cerebellar ataxia-dominant phenotype in patients with ERCC4 mutations. J Hum Genet 2018;63(4):417-423.

574. Shanbhag NM, Geschwind MD, DiGiovanna JJ, et al. Neurodegeneration as the presenting symptom in 2 adults with xeroderma pigmentosum complementation group F. Neurol Genet 2018;4(3):e240.

575. Eidhof I, Baets J, Kamsteeg EJ, et al. GDAP2 mutations implicate susceptibility to cellular stress in a new form of cerebellar ataxia. Brain 2018;141(9):2592-2604.

576. Breza M, Bourinaris T, Efthymiou S, et al. A homozygous GDAP2 loss-of-function variant in a patient with adult-onset cerebellar ataxia. Brain 2020;143(6):e49.

577. Dong HL, Cheng HL, Bai G, Shen Y, Wu ZY. Novel GDAP2 pathogenic variants cause autosomal recessive spinocerebellar ataxia-27 (SCAR27) in a Chinese family. Brain 2020;143(6):e50.

578. Smith KR, Damiano J, Franceschetti S, et al. Strikingly different clinicopathological phenotypes determined by progranulin-mutation dosage. American journal of human genetics 2012;90(6):1102-1107.

579. Gerber S, Alzayady KJ, Burglen L, et al. Recessive and Dominant De Novo ITPR1 Mutations Cause Gillespie Syndrome. American journal of human genetics 2016;98(5):971-980.

580. Scholl UI, Choi M, Liu T, et al. Seizures, sensorineural deafness, ataxia, mental retardation, and electrolyte imbalance (SeSAME syndrome) caused by mutations in KCNJ10. Proc Natl Acad Sci U S A 2009;106(14):5842-5847.

581. Topcu M, Jobard F, Halliez S, et al. L-2-Hydroxyglutaric aciduria: identification of a mutant gene C14orf160, localized on chromosome 14q22.1. Human molecular genetics 2004;13(22):2803-2811.

582. Nilssen O, Berg T, Riise HM, et al. alpha-Mannosidosis: functional cloning of the lysosomal alpha-mannosidase cDNA and identification of a mutation in two affected siblings. Human molecular genetics 1997;6(5):717-726.

583. Krygier M, Kwarciany M, Wasilewska K, et al. A study in a Polish ataxia cohort indicates genetic heterogeneity and points to MTCL1 as a novel candidate gene. Clin Genet 2019;95(3):415-419.

584. Jiao B, Zhou Z, Hu Z, et al. Homozygosity mapping and next generation sequencing for the genetic diagnosis of hereditary ataxia and spastic paraplegia in consanguineous families. Parkinsonism Relat Disord 2020;80:65-72.

585. Stewart GS, Maser RS, Stankovic T, et al. The DNA double-strand break repair gene hMRE11 is mutated in individuals with an ataxia-telangiectasia-like disorder. Cell 1999;99(6):577-587.

586. Nasca A, Scotton C, Zaharieva I, et al. Recessive mutations in MSTO1 cause mitochondrial dynamics impairment, leading to myopathy and ataxia. Hum Mutat 2017;38(8):970-977.

587. Anazi S, Maddirevula S, Salpietro V, et al. Expanding the genetic heterogeneity of intellectual disability. Hum Genet 2017;136(11-12):1419-1429.

588. Monfrini E, Straniero L, Bonato S, et al. Neurofascin (NFASC) gene mutation causes autosomal recessive ataxia with demyelinating neuropathy. Parkinsonism Relat Disord 2019;63:66-72.

589. Kvarnung M, Shahsavani M, Taylan F, et al. Ataxia in Patients With Bi-Allelic NFASC Mutations and Absence of Full-Length NF186. Front Genet 2019;10:896.

590. Carstea ED, Morris JA, Coleman KG, et al. Niemann-Pick C1 disease gene: homology to mediators of cholesterol homeostasis. Science 1997;277(5323):228-231.

591. Naureckiene S, Sleat DE, Lackland H, et al. Identification of HE1 as the second gene of Niemann-Pick C disease. Science 2000;290(5500):2298-2301.

592. Braverman N, Chen L, Lin P, et al. Mutation analysis of PEX7 in 60 probands with rhizomelic chondrodysplasia punctata and functional correlations of genotype with phenotype. Human mutation 2002;20(4):284-297.

593. Warren DS, Morrell JC, Moser HW, Valle D, Gould SJ. Identification of PEX10, the gene defective in complementation group 7 of the peroxisome-biogenesis disorders. American journal of human genetics 1998;63(2):347-359.

594. Mihalik SJ, Morrell JC, Kim D, Sacksteder KA, Watkins PA, Gould SJ. Identification of PAHX, a Refsum disease gene. Nature genetics 1997;17(2):185-189.

595. Wheway G, Schmidts M, Mans DA, et al. An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy genes. Nat Cell Biol 2015;17(8):1074-1087.

596. Hebbar M, Kanthi A, Shukla A, Bielas S, Girisha KM. A biallelic 36-bp insertion in PIBF1 is associated with Joubert syndrome. J Hum Genet 2018;63(8):935-939.

597. Ott T, Kaufmann L, Granzow M, et al. The Frog Xenopus as a Model to Study Joubert Syndrome: The Case of a Human Patient With Compound Heterozygous Variants in PIBF1. Front Physiol 2019;10:134.

598. Matthijs G, Schollen E, Pardon E, et al. Mutations in PMM2, a phosphomannomutase gene on chromosome 16p13, in carbohydrate-deficient glycoprotein type I syndrome (Jaeken syndrome). Nature genetics 1997;16(1):88-92.

599. Bras J, Alonso I, Barbot C, et al. Mutations in PNKP cause recessive ataxia with oculomotor apraxia type 4. Am J Hum Genet 2015;96(3):474-479.

600. Tzoulis C, Sztromwasser P, Johansson S, Gjerde IO, Knappskog P, Bindoff LA. PNKP Mutations Identified by Whole-Exome Sequencing in a Norwegian Patient with Sporadic Ataxia and Edema. Cerebellum 2017;16(1):272-275.

601. Schiess N, Zee DS, Siddiqui KA, Szolics M, El-Hattab AW. Novel PNKP mutation in siblings with ataxia-oculomotor apraxia type 4. J Neurogenet 2017;31(1-2):23-25.

602. Rudenskaya GE, Marakhonov AV, Shchagina OA, et al. Ataxia with Oculomotor Apraxia Type 4 with PNKP Common "Portuguese" and Novel Mutations in Two Belarusian Families. J Pediatr Genet 2019;8(2):58-62.

603. Gatti M, Magri S, Nanetti L, et al. From congenital microcephaly to adult onset cerebellar ataxia: Distinct and overlapping phenotypes in patients with PNKP gene mutations. Am J Med Genet A 2019;179(11):2277-2283.

604. Synofzik M, Gonzalez MA, Lourenco CM, et al. PNPLA6 mutations cause Boucher-Neuhauser and Gordon Holmes syndromes as part of a broad neurodegenerative spectrum. Brain 2014;137(Pt 1):69-77.

605. Saitsu H, Osaka H, Sasaki M, et al. Mutations in POLR3A and POLR3B encoding RNA Polymerase III subunits cause an autosomal-recessive hypomyelinating leukoencephalopathy. American journal of human genetics 2011;89(5):644-651.

606. Najmabadi H, Hu H, Garshasbi M, et al. Deep sequencing reveals 50 novel genes for recessive cognitive disorders. Nature 2011;478(7367):57-63.

607. Hu H, Matter ML, Issa-Jahns L, et al. Mutations in PTRH2 cause novel infantile-onset multisystem disease with intellectual disability, microcephaly, progressive ataxia, and muscle weakness. Ann Clin Transl Neurol 2014;1(12):1024-1035.

608. Stewart GS, Panier S, Townsend K, et al. The RIDDLE syndrome protein mediates a ubiquitin-dependent signaling cascade at sites of DNA damage. Cell 2009;136(3):420-434.

609. Margolin DH, Kousi M, Chan YM, et al. Ataxia, dementia, and hypogonadotropism caused by disordered ubiquitination. N Engl J Med 2013;368(21):1992-2003.

610. Cortese A, Simone R, Sullivan R, et al. Biallelic expansion of an intronic repeat in RFC1 is a common cause of late-onset ataxia. Nat Genet 2019;51(4):649-658.

611. Rafehi H, Szmulewicz DJ, Bennett MF, et al. Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS. Am J Hum Genet 2019;105(1):151-165.

612. Akcimen F, Ross JP, Bourassa CV, et al. Investigation of the RFC1 Repeat Expansion in a Canadian and a Brazilian Ataxia Cohort: Identification of Novel Conformations. Front Genet 2019;10:1219.

613. Cortese A, Tozza S, Yau WY, et al. Cerebellar ataxia, neuropathy, vestibular areflexia syndrome due to RFC1 repeat expansion. Brain 2020;143(2):480-490.

614. Traschutz A, Cortese A, Reich S, et al. Natural History, Phenotypic Spectrum, and Discriminative Features of Multisystemic RFC1 Disease. Neurology 2021;96(9):e1369-e1382.

615. Anttonen AK, Mahjneh I, Hamalainen RH, et al. The gene disrupted in Marinesco-Sjogren syndrome encodes SIL1, an HSPA5 cochaperone. Nature genetics 2005;37(12):1309-1311.

616. Verheijen FW, Verbeek E, Aula N, et al. A new gene, encoding an anion transporter, is mutated in sialic acid storage diseases. Nature genetics 1999;23(4):462-465.

617. Huppke P, Brendel C, Kalscheuer V, et al. Mutations in SLC33A1 cause a lethal autosomal-recessive disorder with congenital cataracts, hearing loss, and low serum copper and ceruloplasmin. American journal of human genetics 2012;90(1):61-68.

618. Cantagrel V, Lefeber DJ, Ng BG, et al. SRD5A3 is required for converting polyprenol to dolichol and is mutated in a congenital glycosylation disorder. Cell 2010;142(2):203-217.

619. Lalani SR, Liu P, Rosenfeld JA, et al. Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. Am J Hum Genet 2016;98(2):347-357.

620. Kremer LS, Distelmaier F, Alhaddad B, et al. Bi-allelic Truncating Mutations in TANGO2 Cause Infancy-Onset Recurrent Metabolic Crises with Encephalocardiomyopathy. Am J Hum Genet 2016;98(2):358-362.

621. Jennions E, Hedberg-Oldfors C, Berglund AK, et al. TANGO2 deficiency as a cause of neurodevelopmental delay with indirect effects on mitochondrial energy metabolism. J Inherit Metab Dis 2019;42(5):898-908.

622. Marin-Valencia I, Gerondopoulos A, Zaki MS, et al. Homozygous Mutations in TBC1D23 Lead to a Non-degenerative Form of Pontocerebellar Hypoplasia. Am J Hum Genet 2017;101(3):441-450.

623. Ivanova EL, Mau-Them FT, Riazuddin S, et al. Homozygous Truncating Variants in TBC1D23 Cause Pontocerebellar Hypoplasia and Alter Cortical Development. Am J Hum Genet 2017;101(3):428-440.

624. Laugwitz L, Buchert R, Groeschel S, et al. Pontocerebellar hypoplasia type 11: Does the genetic defect determine timing of cerebellar pathology? Eur J Med Genet 2020;63(7):103938.

625. Edvardson S, Shaag A, Zenvirt S, et al. Joubert syndrome 2 (JBTS2) in Ashkenazi Jews is associated with a TMEM216 mutation. American journal of human genetics 2010;86(1):93-97.

626. Brancati F, Iannicelli M, Travaglini L, et al. MKS3/TMEM67 mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. Human mutation 2009;30(2):E432-442.

627. Baala L, Romano S, Khaddour R, et al. The Meckel-Gruber syndrome gene, MKS3, is mutated in Joubert syndrome. American journal of human genetics 2007;80(1):186-194.

628. Doherty D, Parisi MA, Finn LS, et al. Mutations in 3 genes (MKS3, CC2D2A and RPGRIP1L) cause COACH syndrome (Joubert syndrome with congenital hepatic fibrosis). J Med Genet 2010;47(1):8-21.

629. Arts HH, Doherty D, van Beersum SE, et al. Mutations in the gene encoding the basal body protein RPGRIP1L, a nephrocystin-4 interactor, cause Joubert syndrome. Nature genetics 2007;39(7):882-888.

630. Delous M, Baala L, Salomon R, et al. The ciliary gene RPGRIP1L is mutated in cerebello-oculo-renal syndrome (Joubert syndrome type B) and Meckel syndrome. Nature genetics 2007;39(7):875-881.

631. Gorden NT, Arts HH, Parisi MA, et al. CC2D2A is mutated in Joubert syndrome and interacts with the ciliopathy-associated basal body protein CEP290. American journal of human genetics 2008;83(5):559-571.

632. Srour M, Hamdan FF, Schwartzentruber JA, et al. Mutations in TMEM231 cause Joubert syndrome in French Canadians. J Med Genet 2012;49(10):636-641.

633. Qian Y, Wang H, Jin T, et al. A familial lateonset hereditary ataxia mimicking pontocerebellar hypoplasia caused by a novel TSEN54 mutation. Mol Med Rep 2014;10(3):1423-1425.

634. Arslan EA, Oncel I, Ceylan AC, Topcu M, Topaloglu H. Genetic and phenotypic features of patients with childhood ataxias diagnosed by next-generation sequencing gene panel. Brain Dev 2020;42(1):6-18.

635. Ghezzi D, Arzuffi P, Zordan M, et al. Mutations in TTC19 cause mitochondrial complex III deficiency and neurological impairment in humans and flies. Nature genetics 2011;43(3):259-263.

636. Boycott KM, Flavelle S, Bureau A, et al. Homozygous deletion of the very low density lipoprotein receptor gene causes autosomal recessive cerebellar hypoplasia with cerebral gyral simplification. American journal of human genetics 2005;77(3):477-483.

637. Colin E, Huynh Cong E, Mollet G, et al. Loss-of-function mutations in WDR73 are responsible for microcephaly and steroid-resistant nephrotic syndrome: Galloway-Mowat syndrome. American journal of human genetics 2014;95(6):637-648.

638. Gulsuner S, Tekinay AB, Doerschner K, et al. Homozygosity mapping and targeted genomic sequencing reveal the gene responsible for cerebellar hypoplasia and quadrupedal locomotion in a consanguineous kindred. Genome Res 2011;21(12):1995-2003.

639. Hoch NC, Hanzlikova H, Rulten SL, et al. XRCC1 mutation is associated with PARP1 hyperactivation and cerebellar ataxia. Nature 2017;541(7635):87-91.

640. O'Connor E, Vandrovcova J, Bugiardini E, et al. Mutations in XRCC1 cause cerebellar ataxia and peripheral neuropathy. J Neurol Neurosurg Psychiatry 2018;89(11):1230-1232.

641. Pierson TM, Adams D, Bonn F, et al. Whole-exome sequencing identifies homozygous AFG3L2 mutations in a spastic ataxia-neuropathy syndrome linked to mitochondrial m-AAA proteases. PLoS Genet 2011;7(10):e1002325.

642. Scheper GC, van der Klok T, van Andel RJ, et al. Mitochondrial aspartyl-tRNA synthetase deficiency causes leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation. Nature genetics 2007;39(4):534-539.

643. Steinfeld R, Grapp M, Kraetzner R, et al. Folate receptor alpha defect causes cerebral folate transport deficiency: a treatable neurodegenerative disorder associated with disturbed myelin metabolism. Am J Hum Genet 2009;85(3):354-363.

644. Uhlenberg B, Schuelke M, Ruschendorf F, et al. Mutations in the gene encoding gap junction protein alpha 12 (connexin 46.6) cause Pelizaeus-Merzbacher-like disease. American journal of human genetics 2004;75(2):251-260.

645. Myerowitz R, Costigan FC. The major defect in Ashkenazi Jews with Tay-Sachs disease is an insertion in the gene for the alpha-chain of beta-hexosaminidase. The Journal of biological chemistry 1988;263(35):18587-18589.

646. O'Dowd BF, Klavins MH, Willard HF, Gravel R, Lowden JA, Mahuran DJ. Molecular heterogeneity in the infantile and juvenile forms of Sandhoff disease (O-variant GM2 gangliosidosis). The Journal of biological chemistry 1986;261(27):12680-12685.

647. Bernard G, Chouery E, Putorti ML, et al. Mutations of POLR3A encoding a catalytic subunit of RNA polymerase Pol III cause a recessive hypomyelinating leukodystrophy. American journal of human genetics 2011;89(3):415-423.

648. Engert JC, Berube P, Mercier J, et al. ARSACS, a spastic ataxia common in northeastern Quebec, is caused by mutations in a new gene encoding an 11.5-kb ORF. Nature genetics 2000;24(2):120-125.

649. Seong E, Insolera R, Dulovic M, et al. Mutations in VPS13D lead to a new recessive ataxia with spasticity and mitochondrial defects. Ann Neurol 2018;83(6):1075-1088.

650. Gauthier J, Meijer IA, Lessel D, et al. Recessive mutations in VPS13D cause childhood onset movement disorders. Ann Neurol 2018;83(6):1089-1095.

651. Koh K, Ishiura H, Shimazaki H, et al. VPS13D-related disorders presenting as a pure and complicated form of hereditary spastic paraplegia. Mol Genet Genomic Med 2020;8(3):e1108.

652. Dziurdzik SK, Bean BDM, Davey M, Conibear E. A VPS13D spastic ataxia mutation disrupts the conserved adaptor-binding site in yeast Vps13. Hum Mol Genet 2020;29(4):635-648.

653. Boukhris A, Schule R, Loureiro JL, et al. Alteration of ganglioside biosynthesis responsible for complex hereditary spastic paraplegia. American journal of human genetics 2013;93(1):118-123.

654. Gan-Or Z, Bouslam N, Birouk N, et al. Mutations in CAPN1 Cause Autosomal-Recessive Hereditary Spastic Paraplegia. Am J Hum Genet 2016;98(5):1038-1046.

655. Wang Y, Hersheson J, Lopez D, et al. Defects in the CAPN1 Gene Result in Alterations in Cerebellar Development and Cerebellar Ataxia in Mice and Humans. Cell Rep 2016;16(1):79-91.

656. Gan-Or Z, Bouslam N, Birouk N, et al. Mutations in CAPN1 Cause Autosomal-Recessive Hereditary Spastic Paraplegia. American journal of human genetics 2016;98(5):1038-1046.

657. Depienne C, Bugiani M, Dupuits C, et al. Brain white matter oedema due to ClC-2 chloride channel deficiency: an observational analytical study. Lancet Neurol 2013;12(7):659-668.

658. Tsaousidou MK, Ouahchi K, Warner TT, et al. Sequence alterations within CYP7B1 implicate defective cholesterol homeostasis in motor-neuron degeneration. Am J Hum Genet 2008;82(2):510-515.

659. Bomont P, Cavalier L, Blondeau F, et al. The gene encoding gigaxonin, a new member of the cytoskeletal BTB/kelch repeat family, is mutated in giant axonal neuropathy. Nature genetics 2000;26(3):370-374.

660. Martin E, Schule R, Smets K, et al. Loss of function of glucocerebrosidase GBA2 is responsible for motor neuron defects in hereditary spastic paraplegia. American journal of human genetics 2013;92(2):238-244.

661. Leegwater PA, Yuan BQ, van der Steen J, et al. Mutations of MLC1 (KIAA0027), encoding a putative membrane protein, cause megalencephalic leukoencephalopathy with subcortical cysts. American journal of human genetics 2001;68(4):831-838.

662. Bilguvar K, Tyagi NK, Ozkara C, et al. Recessive loss of function of the neuronal ubiquitin hydrolase UCHL1 leads to early-onset progressive neurodegeneration. Proc Natl Acad Sci U S A 2013;110(9):3489-3494.

663. Sun Y, Almomani R, Breedveld GJ, et al. Autosomal recessive spinocerebellar ataxia 7 (SCAR7) is caused by variants in TPP1, the gene involved in classic late-infantile neuronal ceroid lipofuscinosis 2 disease (CLN2 disease). Human mutation 2013;34(5):706-713.

664. Sleat DE, Donnelly RJ, Lackland H, et al. Association of mutations in a lysosomal protein with classical late-infantile neuronal ceroid lipofuscinosis. Science 1997;277(5333):1802-1805.

665. Pennacchio LA, Lehesjoki AE, Stone NE, et al. Mutations in the gene encoding cystatin B in progressive myoclonus epilepsy (EPM1). Science 1996;271(5256):1731-1734.

666. Corbett MA, Schwake M, Bahlo M, et al. A mutation in the Golgi Qb-SNARE gene GOSR2 causes progressive myoclonus epilepsy with early ataxia. Am J Hum Genet 2011;88(5):657-663.

667. Van Bogaert P, Azizieh R, Desir J, et al. Mutation of a potassium channel-related gene in progressive myoclonic epilepsy. Ann Neurol 2007;61(6):579-586.

668. Bonten E, van der Spoel A, Fornerod M, Grosveld G, d'Azzo A. Characterization of human lysosomal neuraminidase defines the molecular basis of the metabolic storage disorder sialidosis. Genes Dev 1996;10(24):3156-3169.

669. Chan EM, Young EJ, Ianzano L, et al. Mutations in NHLRC1 cause progressive myoclonus epilepsy. Nat Genet 2003;35(2):125-127.

670. Zuniga-Ramirez C, de Oliveira LM, Kramis-Hollands M, et al. Beyond dystonia and ataxia: Expanding the phenotype of SQSTM1 mutations. Parkinsonism Relat Disord 2019;62:192-195.

671. Vedartham V, Sundaram S, Nair SS, Ganapathy A, Mannan A, Menon R. Homozygous sequestosome 1 (SQSTM1) mutation: a rare cause for childhood-onset progressive cerebellar ataxia with vertical gaze palsy. Ophthalmic Genet 2019;40(4):376-379.

672. Gal A, Balicza P, Weaver D, et al. MSTO1 is a cytoplasmic pro-mitochondrial fusion protein, whose mutation induces myopathy and ataxia in humans. EMBO Mol Med 2017;9(7):967-984.

673. Li K, Jin R, Wu X. Whole-exome sequencing identifies rare compound heterozygous mutations in the MSTO1 gene associated with cerebellar ataxia and myopathy. Eur J Med Genet 2020;63(1):103623.

674. Shi Y, Wang J, Li JD, et al. Identification of CHIP as a novel causative gene for autosomal recessive cerebellar ataxia. PLoS One 2013;8(12):e81884.

675. Genis D, Ortega-Cubero S, San Nicolas H, et al. Heterozygous STUB1 mutation causes familial ataxia with cognitive affective syndrome (SCA48). Neurology 2018;91(21):e1988-e1998.

676. De Michele G, Lieto M, Galatolo D, et al. Spinocerebellar ataxia 48 presenting with ataxia associated with cognitive, psychiatric, and extrapyramidal features: A report of two Italian families. Parkinsonism Relat Disord 2019;65:91-96.

677. Lieto M, Riso V, Galatolo D, et al. The complex phenotype of spinocerebellar ataxia type 48 in eight unrelated Italian families. Eur J Neurol 2020;27(3):498-505.

678. Chen DH, Latimer C, Yagi M, et al. Heterozygous STUB1 missense variants cause ataxia, cognitive decline, and STUB1 mislocalization. Neurol Genet 2020;6(2):1-13.

679. Capitanio GL, Leone M, Croce S, Schiapparelli P. [Evaluation of progesterone receptors in epithelial culture of human endometrium using a cytofluorescence method]. Boll Soc Ital Biol Sper 1988;64(7):671-676.

680. Mol MO, van Rooij JGJ, Brusse E, et al. Clinical and pathologic phenotype of a large family with heterozygous STUB1 mutation. Neurol Genet 2020;6(3):e417.

681. Roux T, Barbier M, Papin M, et al. Clinical, neuropathological, and genetic characterization of STUB1 variants in cerebellar ataxias: a frequent cause of predominant cognitive impairment. Genet Med 2020;22(11):1851-1862.

682. Ravel JM, Benkirane M, Calmels N, et al. Expanding the clinical spectrum of STIP1 homology and U-box containing protein 1-associated ataxia. J Neurol 2021;268(5):1927-1937.

683. Casari G, De Fusco M, Ciarmatori S, et al. Spastic paraplegia and OXPHOS impairment caused by mutations in paraplegin, a nuclear-encoded mitochondrial metalloprotease. Cell 1998;93(6):973-983.

684. Allikmets R, Raskind WH, Hutchinson A, Schueck ND, Dean M, Koeller DM. Mutation of a putative mitochondrial iron transporter gene (ABC7) in X-linked sideroblastic anemia and ataxia (XLSA/A). Human molecular genetics 1999;8(5):743-749.

685. Pandolfo M, Rai M, Remiche G, Desmyter L, Vandernoot I. Cerebellar ataxia, neuropathy, hearing loss, and intellectual disability due to AIFM1 mutation. Neurol Genet 2020;6(3):e420.

686. Bogdanova-Mihaylova P, Alexander MD, Murphy RP, et al. Clinical spectrum of AIFM1-associated disease in an Irish family, from mild neuropathy to severe cerebellar ataxia with colour blindness. J Peripher Nerv Syst 2019;24(4):348-353.

687. Kawarai T, Yamazaki H, Yamakami K, et al. A novel AIFM1 missense mutation in a Japanese patient with ataxic sensory neuronopathy and hearing impairment. J Neurol Sci 2020;409:116584.

688. Wang Q, Xingxing L, Ding Z, Qi Y, Liu Y. Whole exome sequencing identifies a novel variant in an apoptosis-inducing factor gene associated with X-linked recessive hearing loss in a Chinese family. Genet Mol Biol 2019;42(3):543-548.

689. Coene KL, Roepman R, Doherty D, et al. OFD1 is mutated in X-linked Joubert syndrome and interacts with LCA5-encoded lebercilin. American journal of human genetics 2009;85(4):465-481.

690. Billuart P, Bienvenu T, Ronce N, et al. Oligophrenin-1 encodes a rhoGAP protein involved in X-linked mental retardation. Nature 1998;392(6679):923-926.

691. Tuchman M, Jaleel N, Morizono H, Sheehy L, Lynch MG. Mutations and polymorphisms in the human ornithine transcarbamylase gene. Human mutation 2002;19(2):93-107.

692. Saugier-Veber P, Munnich A, Bonneau D, et al. X-linked spastic paraplegia and Pelizaeus-Merzbacher disease are allelic disorders at the proteolipid protein locus. Nat Genet 1994;6(3):257-262.

693. Ganetzky RD, Stendel C, McCormick EM, et al. MT-ATP6 mitochondrial disease variants: Phenotypic and biochemical features analysis in 218 published cases and cohort of 14 new cases. Hum Mutat 2019;40(5):499-515.

694. Ng YS, Martikainen MH, Gorman GS, et al. Pathogenic variants in MT-ATP6: A United Kingdom-based mitochondrial disease cohort study. Ann Neurol 2019;86(2):310-315.

695. Bugiardini E, Bottani E, Marchet S, et al. Expanding the molecular and phenotypic spectrum of truncating MT-ATP6 mutations. Neurol Genet 2020;6(1):e381.

696. Stendel C, Neuhofer C, Floride E, et al. Delineating MT-ATP6-associated disease: From isolated neuropathy to early onset neurodegeneration. Neurol Genet 2020;6(1):e393.

697. Simpson MA, Cross H, Proukakis C, et al. Maspardin is mutated in mast syndrome, a complicated form of hereditary spastic paraplegia associated with dementia. American journal of human genetics 2003;73(5):1147-1156.

698. Schuurs-Hoeijmakers JH, Geraghty MT, Kamsteeg EJ, et al. Mutations in DDHD2, encoding an intracellular phospholipase A(1), cause a recessive form of complex hereditary spastic paraplegia. American journal of human genetics 2012;91(6):1073-1081.

699. Slabicki M, Theis M, Krastev DB, et al. A genome-scale DNA repair RNAi screen identifies SPG48 as a novel gene associated with hereditary spastic paraplegia. PLoS Biol 2010;8(6):e1000408.

700. Stevanin G, Santorelli FM, Azzedine H, et al. Mutations in SPG11, encoding spatacsin, are a major cause of spastic paraplegia with thin corpus callosum. Nature genetics 2007;39(3):366-372.

701. Erlich Y, Edvardson S, Hodges E, et al. Exome sequencing and disease-network analysis of a single family implicate a mutation in KIF1A in hereditary spastic paraparesis. Genome Res 2011;21(5):658-664.

702. Rainier S, Bui M, Mark E, et al. Neuropathy target esterase gene mutations cause motor neuron disease. Am J Hum Genet 2008;82(3):780-785.

703. Zuchner S, Wang G, Tran-Viet KN, et al. Mutations in the novel mitochondrial protein REEP1 cause hereditary spastic paraplegia type 31. Am J Hum Genet 2006;79(2):365-369.

704. Goizet C, Depienne C, Benard G, et al. REEP1 mutations in SPG31: frequency, mutational spectrum, and potential association with mitochondrial morpho-functional dysfunction. Hum Mutat 2011;32(10):1118-1127.

705. Savukoski M, Klockars T, Holmberg V, Santavuori P, Lander ES, Peltonen L. CLN5, a novel gene encoding a putative transmembrane protein mutated in Finnish variant late infantile neuronal ceroid lipofuscinosis. Nat Genet 1998;19(3):286-288.

706. Arsov T, Smith KR, Damiano J, et al. Kufs disease, the major adult form of neuronal ceroid lipofuscinosis, caused by mutations in CLN6. Am J Hum Genet 2011;88(5):566-573.

707. Berkovic SF, Dibbens LM, Oshlack A, et al. Array-based gene discovery with three unrelated subjects shows SCARB2/LIMP-2 deficiency causes myoclonus epilepsy and glomerulosclerosis. Am J Hum Genet 2008;82(3):673-684.

708. Nishimoto J, Nanba E, Inui K, Okada S, Suzuki K. GM1-gangliosidosis (genetic beta-galactosidase deficiency): identification of four mutations in different clinical phenotypes among Japanese patients. American journal of human genetics 1991;49(3):566-574.

709. Dahl N, Lagerstrom M, Erikson A, Pettersson U. Gaucher disease type III (Norrbottnian type) is caused by a single mutation in exon 10 of the glucocerebrosidase gene. American journal of human genetics 1990;47(2):275-278.

710. Tullio-Pelet A, Salomon R, Hadj-Rabia S, et al. Mutant WD-repeat protein in triple-A syndrome. Nature genetics 2000;26(3):332-335.

711. Dallabona C, Diodato D, Kevelam SH, et al. Novel (ovario) leukodystrophy related to AARS2 mutations. Neurology 2014;82(23):2063-2071.

712. Kemp S, Pujol A, Waterham HR, et al. ABCD1 mutations and the X-linked adrenoleukodystrophy mutation database: role in diagnosis and clinical correlations. Human mutation 2001;18(6):499-515.

713. Stromme P, Mangelsdorf ME, Shaw MA, et al. Mutations in the human ortholog of Aristaless cause X-linked mental retardation and epilepsy. Nature genetics 2002;30(4):441-445.

714. Desai R, Frazier AE, Durigon R, et al. ATAD3 gene cluster deletions cause cerebellar dysfunction associated with altered mitochondrial DNA and cholesterol metabolism. Brain : a journal of neurology 2017;140(6):1595-1610.

715. Harel T, Yoon WH, Garone C, et al. Recurrent De Novo and Biallelic Variation of ATAD3A, Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American journal of human genetics 2016;99(4):831-845.

716. L IJ, Loupatty FJ, Ruiter JP, Duran M, Lehnert W, Wanders RJ. 3-Methylglutaconic aciduria type I is caused by mutations in AUH. American journal of human genetics 2002;71(6):1463-1466.

717. DiMauro S, De Vivo DC. Genetic heterogeneity in Leigh syndrome. Annals of neurology 1996;40(1):5-7.

718. Srour M, Schwartzentruber J, Hamdan FF, et al. Mutations in C5ORF42 cause Joubert syndrome in the French Canadian population. American journal of human genetics 2012;90(4):693-700.

719. Anderson BH, Kasher PR, Mayer J, et al. Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature genetics 2012;44(3):338-342.

720. Varon R, Gooding R, Steglich C, et al. Partial deficiency of the C-terminal-domain phosphatase of RNA polymerase II is associated with congenital cataracts facial dysmorphism neuropathy syndrome. Nature genetics 2003;35(2):185-189.

721. Zhou XY, van der Spoel A, Rottier R, et al. Molecular and biochemical analysis of protective protein/cathepsin A mutations: correlation with clinical severity in galactosialidosis. Human molecular genetics 1996;5(12):1977-1987.

722. Smith KR, Dahl HH, Canafoglia L, et al. Cathepsin F mutations cause Type B Kufs disease, an adult-onset neuronal ceroid lipofuscinosis. Human molecular genetics 2013;22(7):1417-1423.

723. Tarpey PS, Raymond FL, O'Meara S, et al. Mutations in CUL4B, which encodes a ubiquitin E3 ligase subunit, cause an X-linked mental retardation syndrome associated with aggressive outbursts, seizures, relative macrocephaly, central obesity, hypogonadism, pes cavus, and tremor. American journal of human genetics 2007;80(2):345-352.

724. Heiss NS, Knight SW, Vulliamy TJ, et al. X-linked dyskeratosis congenita is caused by mutations in a highly conserved gene with putative nucleolar functions. Nature genetics 1998;19(1):32-38.

725. Liu TC, Kim H, Arizmendi C, Kitano A, Patel MS. Identification of two missense mutations in a dihydrolipoamide dehydrogenase-deficient patient. Proc Natl Acad Sci U S A 1993;90(11):5186-5190.

726. Leegwater PA, Vermeulen G, Konst AA, et al. Subunits of the translation initiation factor eIF2B are mutant in leukoencephalopathy with vanishing white matter. Nature genetics 2001;29(4):383-388.

727. Wan J, Yourshaw M, Mamsa H, et al. Mutations in the RNA exosome component gene EXOSC3 cause pontocerebellar hypoplasia and spinal motor neuron degeneration. Nature genetics 2012;44(6):704-708.

728. Sakai N, Inui K, Fujii N, et al. Krabbe disease: isolation and characterization of a full-length cDNA for human galactocerebrosidase. Biochem Biophys Res Commun 1994;198(2):485-491.

729. Brenner M, Johnson AB, Boespflug-Tanguy O, Rodriguez D, Goldman JE, Messing A. Mutations in GFAP, encoding glial fibrillary acidic protein, are associated with Alexander disease. Nat Genet 2001;27(1):117-120.

730. Kaneko H, Hirose M, Katada S, et al. Novel GFAP mutation in patient with adult-onset Alexander disease presenting with spastic ataxia. Movement Disorders 2009;24(9):1393-1395.

731. Orthmann-Murphy JL, Salsano E, Abrams CK, et al. Hereditary spastic paraplegia is a novel phenotype for GJA12/GJC2 mutations. Brain 2009;132(Pt 2):426-438.

732. Piao X, Hill RS, Bodell A, et al. G protein-coupled receptor-dependent development of human frontal cortex. Science 2004;303(5666):2033-2036.

733. Lopez-Hernandez T, Ridder MC, Montolio M, et al. Mutant GlialCAM causes megalencephalic leukoencephalopathy with subcortical cysts, benign familial macrocephaly, and macrocephaly with retardation and autism. American journal of human genetics 2011;88(4):422-432.

734. Loupatty FJ, Clayton PT, Ruiter JP, et al. Mutations in the gene encoding 3-hydroxyisobutyryl-CoA hydrolase results in progressive infantile neurodegeneration. American journal of human genetics 2007;80(1):195-199.

735. Pierce SB, Walsh T, Chisholm KM, et al. Mutations in the DBP-deficiency protein HSD17B4 cause ovarian dysgenesis, hearing loss, and ataxia of Perrault Syndrome. American journal of human genetics 2010;87(2):282-288.

736. Mootha VK, Lepage P, Miller K, et al. Identification of a gene causing human cytochrome c oxidase deficiency by integrative genomics. Proc Natl Acad Sci U S A 2003;100(2):605-610.

737. Barbosa MD, Barrat FJ, Tchernev VT, et al. Identification of mutations in two major mRNA isoforms of the Chediak-Higashi syndrome gene in human and mouse. Human molecular genetics 1997;6(7):1091-1098.

738. Meins M, Lehmann J, Gerresheim F, et al. Submicroscopic duplication in Xq28 causes increased expression of the MECP2 gene in a boy with severe mental retardation and features of Rett syndrome. J Med Genet 2005;42(2):e12.

739. Siintola E, Topcu M, Aula N, et al. The novel neuronal ceroid lipofuscinosis gene MFSD8 encodes a putative lysosomal transporter. American journal of human genetics 2007;81(1):136-146.

740. Romani M, Micalizzi A, Kraoua I, et al. Mutations in B9D1 and MKS1 cause mild Joubert syndrome: expanding the genetic overlap with the lethal ciliopathy Meckel syndrome. Orphanet J Rare Dis 2014;9:72.

741. Lerner-Ellis JP, Tirone JC, Pawelek PD, et al. Identification of the gene responsible for methylmalonic aciduria and homocystinuria, cblC type. Nature genetics 2006;38(1):93-100.

742. Spinazzola A, Santer R, Akman OH, et al. Hepatocerebral form of mitochondrial DNA depletion syndrome: novel MPV17 mutations. Arch Neurol 2008;65(8):1108-1113.

743. Tucker EJ, Hershman SG, Kohrer C, et al. Mutations in MTFMT underlie a human disorder of formylation causing impaired mitochondrial translation. Cell Metab 2011;14(3):428-434.

744. Shoulders CC, Brett DJ, Bayliss JD, et al. Abetalipoproteinemia is caused by defects of the gene encoding the 97 kDa subunit of a microsomal triglyceride transfer protein. Human molecular genetics 1993;2(12):2109-2116.

745. Prietsch V, Mayatepek E, Krastel H, et al. Mevalonate kinase deficiency: enlarging the clinical and biochemical spectrum. Pediatrics 2003;111(2):258-261.

746. Parisi MA, Bennett CL, Eckert ML, et al. The NPHP1 gene deletion associated with juvenile nephronophthisis is present in a subset of individuals with Joubert syndrome. American journal of human genetics 2004;75(1):82-91.

747. Calvo SE, Tucker EJ, Compton AG, et al. High-throughput, pooled sequencing identifies mutations in NUBPL and FOXRED1 in human complex I deficiency. Nature genetics 2010;42(10):851-858.

748. Bonneau D, Colin E, Oca F, et al. Early-onset Behr syndrome due to compound heterozygous mutations in OPA1. Brain 2014;137(Pt 10):e301.

749. Aral B, Benelli C, Ait-Ghezala G, et al. Mutations in PDX1, the human lipoyl-containing component X of the pyruvate dehydrogenase-complex gene on chromosome 11p1, in congenital lactic acidosis. American journal of human genetics 1997;61(6):1318-1326.

750. Shimozawa N, Imamura A, Zhang Z, et al. Defective PEX gene products correlate with the protein import, biochemical abnormalities, and phenotypic heterogeneity in peroxisome biogenesis disorders. J Med Genet 1999;36(10):779-781.

751. Shen J, Gilmore EC, Marshall CA, et al. Mutations in PNKP cause microcephaly, seizures and defects in DNA repair. Nature genetics 2010;42(3):245-249.

752. Pedroso JL, Rocha CR, Macedo-Souza LI, et al. Mutation in PNKP presenting initially as axonal Charcot-Marie-Tooth disease. Neurol Genet 2015;1(4):e30.

753. Williams SR, Gekeler V, McIvor RS, Martin DW, Jr. A human purine nucleoside phosphorylase deficiency caused by a single base change. The Journal of biological chemistry 1987;262(5):2332-2338.

754. Wiethoff S, Bettencourt C, Paudel R, et al. Pure Cerebellar Ataxia with Homozygous Mutations in the PNPLA6 Gene. Cerebellum 2016.

755. Hufnagel RB, Arno G, Hein ND, et al. Neuropathy target esterase impairments cause Oliver-McFarlane and Laurence-Moon syndromes. J Med Genet 2015;52(2):85-94.

756. Kmoch S, Majewski J, Ramamurthy V, et al. Mutations in PNPLA6 are linked to photoreceptor degeneration and various forms of childhood blindness. Nat Commun 2015;6:5614.

757. Thiffault I, Wolf NI, Forget D, et al. Recessive mutations in POLR1C cause a leukodystrophy by impairing biogenesis of RNA polymerase III. Nat Commun 2015;6:7623.

758. Stepp SE, Dufourcq-Lagelouse R, Le Deist F, et al. Perforin gene defects in familial hemophagocytic lymphohistiocytosis. Science 1999;286(5446):1957-1959.

759. Dias C, McDonald A, Sincan M, et al. Recurrent subacute post-viral onset of ataxia associated with a PRF1 mutation. Eur J Hum Genet 2013;21(11):1232-1239.

760. de Brouwer AP, Williams KL, Duley JA, et al. Arts syndrome is caused by loss-of-function mutations in PRPS1. American journal of human genetics 2007;81(3):507-518.

761. Roessler BJ, Nosal JM, Smith PR, et al. Human X-linked phosphoribosylpyrophosphate synthetase superactivity is associated with distinct point mutations in the PRPS1 gene. The Journal of biological chemistry 1993;268(35):26476-26481.

762. Kim HJ, Sohn KM, Shy ME, et al. Mutations in PRPS1, which encodes the phosphoribosyl pyrophosphate synthetase enzyme critical for nucleotide biosynthesis, cause hereditary peripheral neuropathy with hearing loss and optic neuropathy (cmtx5). American journal of human genetics 2007;81(3):552-558.

763. Guilbot A, Williams A, Ravise N, et al. A mutation in periaxin is responsible for CMT4F, an autosomal recessive form of Charcot-Marie-Tooth disease. Human molecular genetics 2001;10(4):415-421.

764. Appel-Cresswell S, Guella I, Lehman A, Foti D, Farrer MJ. PSEN1 p.Met233Val in a Complex Neurodegenerative Movement and Neuropsychiatric Disorder. J Mov Disord 2018;11(1):45-48.

765. Ryan NS, Nicholas JM, Weston PSJ, et al. Clinical phenotype and genetic associations in autosomal dominant familial Alzheimer's disease: a case series. Lancet Neurol 2016;15(13):1326-1335.

766. Seliverstov Y, Kanivets I, Illarioshkin S. Spinocerebellar Ataxia-Like Presentation of the M233V PSEN1 Mutation. Cerebellum 2020;19(5):744-747.

767. Testi S, Peluso S, Fabrizi GM, et al. A novel PSEN1 mutation in a patient with sporadic early-onset Alzheimer's disease and prominent cerebellar ataxia. J Alzheimers Dis 2014;41(3):709-714.

768. Wolf NI, Salomons GS, Rodenburg RJ, et al. Mutations in RARS cause hypomyelination. Annals of neurology 2014;76(1):134-139.

769. Hong SE, Shugart YY, Huang DT, et al. Autosomal recessive lissencephaly with cerebellar hypoplasia is associated with human RELN mutations. Nature genetics 2000;26(1):93-96.

770. Schossig A, Wolf NI, Fischer C, et al. Mutations in ROGDI Cause Kohlschutter-Tonz Syndrome. American journal of human genetics 2012;90(4):701-707.

771. Bourdon A, Minai L, Serre V, et al. Mutation of RRM2B, encoding p53-controlled ribonucleotide reductase (p53R2), causes severe mitochondrial DNA depletion. Nature genetics 2007;39(6):776-780.

772. Angebault C, Guichet PO, Talmat-Amar Y, et al. Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. American journal of human genetics 2015;97(5):754-760.

773. Seidner G, Alvarez MG, Yeh JI, et al. GLUT-1 deficiency syndrome caused by haploinsufficiency of the blood-brain barrier hexose carrier. Nat Genet 1998;18(2):188-191.

774. Kleta R, Romeo E, Ristic Z, et al. Mutations in SLC6A19, encoding B0AT1, cause Hartnup disorder. Nature genetics 2004;36(9):999-1002.

775. Dumitrescu AM, Liao XH, Best TB, Brockmann K, Refetoff S. A novel syndrome combining thyroid and neurological abnormalities is associated with mutations in a monocarboxylate transporter gene. Am J Hum Genet 2004;74(1):168-175.

776. Gerards M, Kamps R, van Oevelen J, et al. Exome sequencing reveals a novel Moroccan founder mutation in SLC19A3 as a new cause of early-childhood fatal Leigh syndrome. Brain 2013;136(Pt 3):882-890.

777. Miyamoto T, Kanazawa N, Kato S, et al. Diagnosis of Japanese patients with HHH syndrome by molecular genetic analysis: a common mutation, R179X. J Hum Genet 2001;46(5):260-262.

778. Abrams AJ, Hufnagel RB, Rebelo A, et al. Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. Nature genetics 2015;47(8):926-932.

779. Johnson JO, Gibbs JR, Megarbane A, et al. Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. Brain 2012;135(Pt 9):2875-2882.

780. Jenkinson EM, Rodero MP, Kasher PR, et al. Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature genetics 2016;48(10):1185-1192.

781. Kisker C, Schindelin H, Pacheco A, et al. Molecular basis of sulfite oxidase deficiency from the structure of sulfite oxidase. Cell 1997;91(7):973-983.

782. Echaniz-Laguna A, Ghezzi D, Chassagne M, et al. SURF1 deficiency causes demyelinating Charcot-Marie-Tooth disease. Neurology 2013;81(17):1523-1530.

783. Garcia-Gonzalo FR, Corbit KC, Sirerol-Piquer MS, et al. A transition zone complex regulates mammalian ciliogenesis and ciliary membrane composition. Nature genetics 2011;43(8):776-784.

784. Sang L, Miller JJ, Corbit KC, et al. Mapping the NPHP-JBTS-MKS protein network reveals ciliopathy disease genes and pathways. Cell 2011;145(4):513-528.

785. Thomas S, Legendre M, Saunier S, et al. TCTN3 mutations cause Mohr-Majewski syndrome. American journal of human genetics 2012;91(2):372-378.

786. Bogershausen N, Shahrzad N, Chong JX, et al. Recessive TRAPPC11 mutations cause a disease spectrum of limb girdle muscular dystrophy and myopathy with movement disorder and intellectual disability. American journal of human genetics 2013;93(1):181-190.

787. Chakraborty PK, Schmitz-Abe K, Kennedy EK, et al. Mutations in TRNT1 cause congenital sideroblastic anemia with immunodeficiency, fevers, and developmental delay (SIFD). Blood 2014;124(18):2867-2871.

788. Taylor RW, Pyle A, Griffin H, et al. Use of whole-exome sequencing to determine the genetic basis of multiple mitochondrial respiratory chain complex deficiencies. JAMA 2014;312(1):68-77.

789. Renbaum P, Kellerman E, Jaron R, et al. Spinal muscular atrophy with pontocerebellar hypoplasia is caused by a mutation in the VRK1 gene. American journal of human genetics 2009;85(2):281-289.

790. Strom TM, Hortnagel K, Hofmann S, et al. Diabetes insipidus, diabetes mellitus, optic atrophy and deafness (DIDMOAD) caused by mutations in a novel gene (wolframin) coding for a predicted transmembrane protein. Human molecular genetics 1998;7(13):2021-2028.

791. Shaheen R, Faqeih E, Ansari S, et al. Genomic analysis of primordial dwarfism reveals novel disease genes. Genome Res 2014;24(2):291-299.