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 withpapillary 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 ataxiaLaurence-Moon syndrome: mental retardation, growth retardation, retinopathy, choroidal atrophy, nystagmus, peripheral neuropathy, spastic paraplegia, short stature, pituitary dysfunction, micropenis, hypoplastic scrotumOliver-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 dysfunctionNeurodegeneration 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.

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