Updated complete list of hereditary dystonia

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| **Designation** | **Less common movement phenotype** | **Clinical clues** | | **OMIM** | **MOI** |
| **Isolated dystonia** | | | | | |
| DYT-*ANO3132, 377* | (Head) Tremor, myoclonus | Cranial-cervical dystonia, variable age at onset | | 615034 | AD |
| DYT-*EIF2AK2171, 172, 174* |  | Early onset, mostly generalized dystonia including laryngeal involvement, may be accompanied by leukoencephalopathy, spasticity, and developmental delay | | 618877 | AD, AR |
| DYT-*GNAL*132, 378 |  | Adult onset cranial-cervical dystonia | | 615073 | AD |
| DYT-*HPCA132, 379, 380* |  | Childhood-onset generalized dystonia and adolescence-onset segmental dystonia; first affecting the distal limbs and later involving neck, orofacial and craniocervical regions, dysarthria, febrile seizures and developmental delay in one case | | [**224500**](https://www.omim.org/entry/224500) | AR |
| DYT-*KMT2B132, 381, 382* |  | Childhood-onset, generalized dystonia, usually first affecting the lower limbs, variable additional signs including developmental delay, microcephaly, intellectual disability, facial dysmorphia | | 617284 | AD |
| DYT-*PRKRA*383 |  | Rare form of usually generalized dystonia, parkinsonism inconsistent | | 612067 | AR |
| DYT-*THAP1*131, 132 |  | Adolescent-onset dystonia of mixed type | | 602629 | AD |
| DYT-*TOR1A*132, 384 |  | Early-onset generalized dystonia | | 128100 | AD |
| DYT-*VPS16112, 385-387* |  | Early-onset generalized dystonia, mild to moderate intellectual disability and neuropsychiatric symptoms in a subset of patients | | 619291 | AD |
| **Combined dystonias (disorders where dystonia frequently coexists with other movement disorders)** | | | | | |
| DYT-*COX20388-390* | Ataxia | Mitochondrial complex IV deficiency nuclear type 11;  hypotonia, gait ataxia, dysarthria, and sensory neuropathy | | 619054 | AR |
| DYT-*DNAJC12350, 391, 392* | Parkinsonism | Hyperphenylalaninemia, developmental delay, phenotype can also include non-progressive or mild levodopa-responsive parkinsonism | | 617384 | AR |
| DYT-*SLC39A14393-396* | Parkinsonism | Hypermagnesemia, dysarthria, and generalized dystonia, MR imaging: T1 hyperintense, diffuse, non-enhancing signal of basal ganglia | | 617013 | AR |
| DYT/PARK-*ATP1A3A*,326 |  | Rapid-onset dystonia-parkinsonism, chorea in later life | | 128235 | AD |
| DYT/PARK-*GCH1*329 |  | GTP cyclohydrolase I deficiency (mild form)330: childhood-onset dopa-responsive dystonia, adult-onset dystonia-parkinsonism  Additional clinical manifestations: diurnal fluctuation, pyramidal signs | | 128230 | AD |
| GTP cyclohydrolase I deficiency (severe form)331, 332: dystonia, parkinsonism  Additional clinical manifestations: developmental delay, truncal hypotonia, spasticity, oculogyric crises, seizures, with or without hyperphenylalaninemia332 | | 605407 | AR |
| DYT/PARK- *TAF1\*,*135 |  | Dystonia-parkinsonism | | 314250 | XL |
| DYT/PARK-*TH344* |  | Tyrosine hydroxylase deficiency345 | | 605407 |  |
|  | Mild form: dopa-responsive infantile to early childhood onset dystonia | |  | AR |
|  | Severe form: infantile-onset dystonia and parkinsonism, truncal hypotonia, global developmental delay | |  | AR |
|  | Very severe form: infantile-onset dystonia and parkinsonism, oculogyric crises, severe global developmental delay, truncal hypotonia, limb spasticity, autonomic dysfunction | |  | AR |
| DYT/CHOR-*GNAO1105, 397* | Myoclonus | Hypotonia and motor delay, exacerbated by febrile illness, stress, high ambient temperature | | 617493 | AD |
| MYC/DYT-*KCTD17398-401* |  | Onset of mild myoclonic symptoms in the first or second decade of life, followed by later onset of progressive dystonia with predominant involvement of the cranial and laryngeal muscles; dystonia dominates the clinical picture | | 616398 | AD |
| MYC/DYT-*SGCE*402 |  | Myoclonus-dystonia | | 159900 | AD |
| **Complex dystonias** **(where dystonia dominates the clinical picture but this occurs in the context of a complex phenotype including symptoms other than movement disorders)** | | | | | |
| DYT-*ACTBB,403-406* |  | Sensorineural hearing loss, generalized dystonia, skeletal abnormalities | | 607371 | AD |
| DYT-*ATP7B*407 |  | Wilson’s disease: Dystonia, occasionally parkinsonism and/or chorea  Additional clinical features: flapping tremor, rest-, action- and intention tremor, orofacial dyskinesias, dysarthria, liver disease, Kayser-Fleischer rings, psychiatric symptoms | | 277900 | AR |
| DYT-*BCAP31408-414* |  | Deafness, central hypomyelination, microcephaly, ophthalmoplegia, intellectual disability | | 300475 | XLD |
| DYT- *DCAF17*-(NBIA)*415* | Chorea416 | Woodhouse-Sakati syndrome417: Iron accumulation: GP, SN, other BG (variable)  Additional clinical features: Dysarthria, deafness, seizures, cognitive impairment, hypogonadism, alopecia, diabetes mellitus, thyroid dysfunction, acanthosis nigrans, keratoconus, camptodactyly | | 241080 | AR |
| DYT-*DDC*418 |  | Aromatic l-amino acid decarboxylase deficiency419: Dystonia, occasionally chorea, hypokinesia  Additional clinical features: Developmental delay, truncal hypotonia, oculogyric crises, ptosis, autonomic symptoms, sleep disorder, diurnal fluctuations with sleep benefit | | 608643 | AR |
| DYT-*FITM2420-422* |  | Global developmental delay, sensorineural hearing loss, poor growth, and low body mass index | | 618635 | AR |
| DYT-*IRF2BPL124-127* |  | Developmental delay, hypotonia, seizures, pyramidal signs, dysarthria | | 618088 | AD |
| DYT-*MECR423, 424* |  | Childhood-onset dystonia with optic atrophy and basal ganglia abnormalities (DYTOBAG);  optic atrophy, MR imaging: basal ganglia signal abnormalities, T2 hyperintense signal in putamen and globus pallidus, cystic changes in putamen | | 617282 | AR |
| DYT-*mt-ND6425* |  | Leber’s hereditary optic neuropathy/dystonia (G14459A mutation): dystonia  Additional clinical features: juvenile-onset subacute vision loss (Leber hereditary optic neuropathy), encephalopathy, spasticity, bulbar dysfunction, cognitive impairment | | 516006 | mt |
| DYT-*OPA1426, 427* | Ataxia | Optic atrophy, peripheral neuropathy, myopathy, and progressive external ophthalmoplegia | | 616896 | AD |
| DYT-*PANK2-*(NBIA)352 | Parkinsonism, chorea | Pantothenate kinase-associated neurodegeneration (PKAN):  Iron accumulation: GP – eye of the tiger sign  Additional clinical features: Spasticity, dysarthria, cognitive decline, gaze palsy, psychiatric symptoms, pigmentary retinopathy | | 234200 | AR |
| DYT-*SERAC1428-430* |  | 3-Methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome (MEGDEL);  sensorineural hearing loss, delayed psychomotor development, increased excretion of 3-methylglutaconic acid, transient liver dysfunction in the neonatal period, MR imaging: bilateral basal ganglia hyperintensities | | 614739 | AR |
| DYT- *SLC19A3*431 |  | Biotin-responsive basal ganglia disease (within the thiamine transporter–2 (hTHTR2) deficiency spectrum)432, 433: dystonia, parkinsonism (mainly rigidity), occasionally ataxia, chorea  Additional clinical features: subacute encephalopathy/coma (often triggered by febrile illness), cranial nerve palsy, pyramidal signs, cerebellar signs, dysphagia, intellectual disability, epilepsy, responsive to thiamine and/or biotin therapy. | | 606152 | AR |
| DYT-*SUCLA2*128, 129 |  | SUCLA2-related mitochondrial DNA (mtDNA) depletion syndrome, encephalomyopathic form, with mild methylmalonic aciduria128, 130: dystonia  Additional clinical features: severe hypotonia, developmental delay, seizures, progressive spasticity, cerebral atrophy, sensorineural hearing loss, ophthalmoplegia, feeding problems and postnatal growth retardation, ptosis | | 603921 | AR |
| DYT-*TIMM8A*434 |  | Mohr-Tranebjaerg syndrome435: dystonia  Additional clinical features: sensorineural deafness, visual impairment, cognitive impairment, behavioral problems, pyramidal signs | | 304700 | XL |
| DYT-*TUBB4AC,* 436 | HSP437, 438 | Spasmodic dysphonia is most common dystonic presentation, alternative phenotype: hypomyelinating leukodystrophy (see footnote) | | 128101 | AD |
| DYT-*VAC14439-442* |  | Neurodegeneration, ataxia, dysarthria, hypotonia | | 617054 | AR |
| DYT/CHOR-*ACAT1*443 |  | Mitochondrial acetoacetyl-CoA thiolase deficiency: metabolic decompensation and basal ganglia injury during acute stress resulting in dystonia and chorea444 | | 203750 | AR |
| DYT/CHOR-*ADAR1445, 446* | Spasticity | Aicardi-Goutières syndrome, includes dystonia and spastic paraparesis. MRI may reveal isolated bilateral striatal necrosis, adult-onset psychological difficulties, linked to characteristic interferon signature (upregulation of interferon-stimulated genes) | | 615010 | AR, rarely AD |
| DYT/CHOR-*FOXG1447-449* | Dyskinesia | Rett-like phenotype (with congenital encephalopathy) | | 613454 | AD |
| DYT/CHOR-*GCDH*450 |  | Glutaric aciduria type I451, 452: dystonia, chorea (usually following acute metabolic crises), parkinsonism (later)  Additional clinical features: acute metabolic crises with basal ganglia injury (predominantly putamen and caudate nucleus), severe truncal hypotonia, macrocephaly, orofacial dyskinesias, spasticity, cognitive impairment (variable), enlarged subdural space, subdural hygroma/hemorrhages, headaches, seizures453 | | 231670 | AR |
| DYT/CHOR-*HPRT*454 |  | Lesch-Nyhan syndrome: dystonia, chorea, occasionally ballism  Additional clinical features: hyperuricemia, crystalluria, developmental delay/intellectual disability, eye movement abnormalities, spasticity, compulsive self-injurious behavior, gouty arthritis, nephrolithiasis, renal failure | | 300322 | XL |
| DYT/CHOR-*MUT*455 |  | Methylmalonic aciduria456: dystonia, chorea, occasionally ataxia  Additional clinical features: Neonatal-onset vomiting, seizures, lethargy and hypotonia, ketoacidosis, hyperammonemia, developmental delay, spasticity, pancreatitis, nephritis, growth failure, acute metabolic crises with confusion/encephalopathy, basal ganglia injury (predominantly globus pallidus) | | 251000 | AR |
| DYT/CHOR-*PCCA/PCCB*457 |  | Propionic aciduria456: dystonia, occasionally chorea  Additional clinical features: neonatal-onset vomiting, seizures, lethargy and hypotonia, ketoacidosis, hyperammonemia, developmental delay, spasticity, cardiomyopathy, acute metabolic crises with confusion/encephalopathy, basal ganglia injury (predominantly putamen and caudate nucleus) | | 606054 | AR |
| DYT/PARK-*CP-*(NBIA)*327* | Chorea | Aceruloplasminemia328: dystonia, ataxia, chorea, parkinsonism, tremors  Iron accumulation: more homogeneous involvement of primarily caudate, putamen, thalamus, dentate  Additional clinical features: cognitive impairment, psychiatric symptoms, diabetes mellitus, retinal degeneration, anemia, liver iron storage | | 604290 | AR |
| DYT/PARK-*GLB1*333, 334 |  | GM1 gangliosidosis (type III, chronic/adult form): Dystonia, parkinsonism  Additional clinical features: pyramidal signs, dysarthria, cognitive deficits (often mild initially), skeletal abnormalities and short statue, corneal clouding, vacuolated cells, cardiomyopathy, progressive disease | | 230650 | AR |
| DYT/PARK-*PLA2G6-*(NBIA)*D,335-337* | Ataxia | *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 |
| DYT/PARK-*PTS338* |  | 6-pyruvoyl-tetrahydropterin synthase deficiency: dystonia, parkinsonism  Additional clinical features: neonatal irritability, truncal hypotonia, developmental delay, seizures, oculogyric crises, autonomic dysfunction, hyperphenylalaninemia | | 612719 | AR |
| DYT/PARK-*QDPR338* |  | Dihydropteridine reductase deficiency: dystonia, parkinsonism  Additional clinical features: developmental delay, truncal hypotonia, seizures, autonomic dysfunction, hyperphenylalaninemia | | 612676 | AR |
| DYT/PARK-*SLC6A3*339, 340 |  | Dopamine transporter deficiency syndrome: dystonia and parkinsonism (typically infantile-onset, atypical cases with juvenile-onset exist), occasionally chorea in infancy  Additional clinical features: mild developmental delay, truncal hypotonia, ocular flutter / oculogyric crises, saccade initiation failure, bulbar dysfunction | | 126455 | AR |
| DYT/PARK-*SLC30A10*133 |  | Hypermanganesemia with dystonia, polycythemia, and liver cirrhosis, parkinsonism  Additional clinical features: hypermanganesemia, polycythemia, chronic liver disease, dysarthria | | 611146 | AR |
| DYT/PARK-*SPR*342 |  | Sepiapterin reductase deficiency: dystonia, parkinsonism  Additional clinical features: motor and speech delay, truncal hypotonia, limb hypertonia and hyperreflexia, oculogyric crises, psychiatric symptoms, autonomic dysfunction, diurnal fluctuation and sleep benefit, no hyperphenylalaninemia | | 612716 | AR |
| ATX/DYT-*SQSTM1458, 459* |  | Childhood-onset neurodegeneration, gait ataxia, cognitive decline, oculomotor abnormalities including vertical gaze palsy and nystagmus, and hypergonadotropic hypogonadism | | 617145 | AR |
| **Disorders that usually present with other phenotypes but can have predominant dystonia** | | | | | |
| ***Gene*** | **Associated disease** | **OMIM** | **Clinical phenotype** | | **MOI** |
| ATX-*ATXN3460-462* | Machado-Joseph Disease (Spinocerebellar ataxia) | 109150 | Marked non-ataxia features; can have predominant parkinsonism, dystonia, chorea, spasticity, neuropathy, lower motor neuron involvement | | AD |
| HSP-*C19orf12*-(NBIA)356 | Neuro-degeneration with Brain Iron Accumulation 4 | 614298 | Mitochondrial membrane protein-associated neurodegeneration (MPAN)357; progressive spastic paresis, dystonia, parkinsonism, and variable additional clinical features: dysarthria, dysphagia, cognitive decline/dementia, motor axonal neuropathy, optic nerve atrophy, psychiatric symptoms, bowel/bladder incontinence  Iron accumulation: GP (hyperintense streaking of medial medullary lamina between GPi and GPe), SN | | AR or AD |
| HSP/ATX-*FA2H*-(NBIA)362 | Spastic paraplegia | 612319 | Complex SPG; Fatty Acid Hydroxylase-associated Neurodegeneration (FAHN)363; variable additional clinical features: spastic tetraparesis, cognitive decline, cerebellar and brainstem atrophy, dystonia, parkinsonism, ataxia, dysarthria, dysphagia, optic nerve atrophy, seizures  Iron accumulation: GP (more subtle than other NBIAs) | | AR |
| HSP/ATX-*KIF1CE,* 463, 464 | Spastic ataxia, Spastic paraplegia | 611302 | Pure and complicated; variable additional features including dystonia, ataxia, chorea, myoclonus, dysarthria, developmental delay, mild mental retardation, hypodontia, ptosis, short stature, sensorineural deafness, pes planus, white matter lesions | | AR |
| CHOR-*FTL*-(NBIA)354 | Neuro-degeneration with Brain Iron Accumulation 3 | 606159 | Neuroferritinopathy355: dystonia, chorea, parkinsonism, and variable additional clinical features including oromandibular dyskinesia, dysphagia, cognitive impairment, behavioral symptoms, low serum ferritin  Iron accumulation: GP, caudate, putamen, SN, red nucleus; cystic BG changes – pallidal necrosis | | AD |
| PARK-*DNAJC6465, 466* | Atypical parkinsonism | 615528 | Progressive parkinsonism in first decade, neurological regression, loss of ambulation mid-adolescence, epilepsy and neuropsychiatric features also present | | AR |
| PARK- *WDR45*-(NBIA)324 | Beta-propeller protein-associated neurodegeneration (BPAN, previously SENDA syndrome) 325 | 300894 | Predominant parkinsonism, dystonia less common, variable additional clinical features: developmental delay/intellectual disability, progressive cognitive decline, seizures, spasticity, Rett-like stereotypies, autistic-features, neuropsychiatric symptoms, sleep disorders, bowel/bladder incontinence, infantile epileptic encephalopathy  Iron accumulation: SN > GP Halo of hyperintensity surrounding linear hypointensity in SN on T1 scans | | XL |

AD = autosomal dominant, AR = autosomal recessive, BG = Basal ganglia, GP = Globus pallidus, HSP = Hereditary Spastic Paraplegia, INAD = infantile neuroaxonal dystrophy, MOI = Mode of inheritance, mt = mitochondrial, OMIM = Online Mendelian Inheritance in Man (<https://www.omim.org/about>), SENDA = static encephalopathy of childhood with neurodegeneration in adulthood, SN = Subthalamic nucleus, XL = x-linked, XLD = x-linked dominant

\*Due to a founder effect, genetic testing is possible. The pathogenicity of the *TAF1* gene is not absolutely confirmed, however testing of selected variants in this gene is sufficient for the diagnosis.

A Gene mutations can also cause autosomal dominant alternating hemiplegia of childhood (OMIM: 614820), CAPOS syndrome (Cerebellar ataxia, pes cavus, optic atrophy and sensorineural hearing loss; OMIM: 601338), as well as CAOS syndrome (Episodic Cerebellar Ataxia, Areflexia, Optic Atrophy, and Sensorineural Hearing Loss).

B This gene has also been associated with Baraitser-Winter syndrome 1 (OMIM [**243310**](https://www.omim.org/entry/243310)).

C Gene mutations more commonly cause a hypomyelinating leukodystrophy with developmental delay, dystonia, choreoathetosis, rigidity, opisthotonus, and oculogyric crises, progressive spastic tetraplegia, ataxia, and, more rarely, seizures.

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

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

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