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Recently identified or confirmed forms of hereditary dystonia

Designation	Less common movement phenotype	Clinical clues	OMIM	MOI
Isolated dystonia				
DYT-*ANO3*[1, 2]	(head) tremor, myoclonus	Cranial-cervical dystonia, variable age at onset	615034	AD
DYT-*EIF2AK2*[3-5]		Early onset, mostly generalized dystonia including laryngeal involvement, may be accompanied by leukoencephalopathy, spasticity, and developmental delay	618877	AD, AR
DYT-*HPCA*[2, 6, 7]		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	AR
DYT-*KMT2B*[2, 8, 9]		Childhood-onset, generalized dystonia, usually first affecting the lower limbs, variable additional signs including developmental delay, microcephaly, intellectual disability, facial dysmorphia	617284	AD
DYT-*VPS16*[10-13]		Early-onset generalized dystonia, mild to moderate intellectual disability and neuropsychiatric symptoms in a subset of patients	619291	AD
Combined dystonia				
DYT-*COX20*[14-16]	Ataxia	Mitochondrial complex IV deficiency nuclear type 11; hypotonia, gait ataxia, dysarthria, and sensory neuropathy	619054	AR
DYT-*DNAJC12*[17-19]	Parkinsonism	Hyperphenylalaninemia and developmental delay. Phenotype can also include non-progressive or mild levodopa-responsive parkinsonism	617384	AR
DYT-*SLC39A14*[20-23]	Parkinsonism	Hypermagnesemia, dysarthria, and generalized dystonia, MR imaging: T1 hyperintense, diffuse, non-enhancing signal of basal ganglia	617013	AR
DYT/CHOR-*GNAO1*[24, 25]	Myoclonus	Hypotonia and motor delay, exacerbated by febrile illness, stress, high ambient temperature	617493	AD
MYC/DYT-*KCTD17*[26-29]		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
Complex dystonia				
DYT-*MECR*[30, 31]		Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities (DYTOABG); MR imaging: basal ganglia signal abnormalities, T2 hyperintense signal in putamen and globus pallidus, cystic changes in putamen	617282	AR
DYT-*OPA1*[32, 33]	Ataxia	Optic atrophy, peripheral neuropathy, myopathy, and progressive external ophthalmoplegia		AD

DYT/CHOR-*ADAR*[34, 35]	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
ATX/DYT-*SQSTM1*[36, 37]	Chorea	Neurodegeneration with ataxia, dystonia, and gaze palsy (NADGP): gait ataxia, cognitive decline, oculomotor abnormalities including vertical gaze palsy and nystagmus, dysarthria and hypergonadotropic hypogonadism	617145	AR
Dystonia presenting with deafness				
DYT-*ACTB*[A,38-41]		Sensorineural hearing loss, generalized dystonia, skeletal abnormalities	607371	AD
DYT-*BCAP31*[42-48]		Deafness, central hypomyelination, microcephaly, ophthalmoplegia, intellectual disability	300475	XLD
DYT-*FITM2*[49-51]		Global developmental delay, sensorineural hearing loss, poor growth, and low body mass index	618635	AR
DYT-*SERAC1*[52-54]		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
Dystonia presenting with developmental delay				
DYT-*IRF2BPL*[55-58]		Developmental delay, hypotonia, seizures, pyramidal signs, dysarthria	618088	AD
DYT-*VAC14*[59-62]	Ataxia	Neurodegeneration, ataxia, dysarthria, hypotonia	617054	AR
DYT/CHOR-*FOXG1*[63-65]	Dyskinesia	Rett-like phenotype (with congenital encephalopathy)	613454	AD

AD = autosomal dominant, AR = autosomal recessive, MOI = mode of inheritance, OMIM = Online Mendelian Inheritance in Man (https://www.omim.org/about), XLD = x-linked dominant

[A] This gene has also been linked to Baraitser-Winter syndrome 1 (OMIM 243310).

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