Title: Hereditary Dystonia Overview *GeneReview* Table 2. Previous Nomenclature System
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Table 2. Previous Nomenclature System for Inherited (Monogenic) Forms of Isolated or Combined Dystonia/Dyskinesias (DYTs)

Locus	Disorder	MOI	Gene / Chromosome Locus [1]	Status & Remarks re Gene / Chromosome Locus
DYT1	Early-onset generalized dystonia	AD	*TOR1A*	Confirmed
DYT2	Autosomal recessive dystonia	AR	*HPCA*	Unconfirmed
DYT3	X-linked dystonia parkinsonism; "lubag"	XL	Xq13.1	Unconfirmed [2]; molecular genetic testing for founder haplotype linked to disease is possible & can be used for diagnostic purposes [3]
DYT4	"Non-DYT1" dystonia; whispering dysphonia [4]	AD	*TUBB4A*	Discovered independently in the same family by 2 different groups
DYT5	Dopa-responsive dystonia; Segawa syndrome	AD	*GCH1*	Confirmed
DYT6	Adolescent-onset dystonia of mixed type	AD	*THAP1*	Confirmed
DYT7	Adult-onset focal dystonia	AD	18p	Unconfirmed (not replicated since first described in 1996) [5]
DYT8	Paroxysmal nonkinesigenic dyskinesia 1 (PNKD1)	AD	*PNKD* (previously known as *MR-1*)	Confirmed
DYT9	Paroxysmal choreoathetosis w/episodic ataxia & spasticity	AD	*SLC2A1* (also known as *GLUT1*)	Identical to DYT18
DYT10	Paroxysmal kinesigenic choreoathetosis (PKD1) & infantile convulsions	AD	*PRRT2*	Confirmed
DYT11	Myoclonus-dystonia	AD	*SGCE*	Confirmed
DYT12	Rapid-onset dystonia-parkinsonism [6]	AD	*ATP1A3*	Confirmed
DYT13	Multifocal/segmental dystonia	AD	1p36	Unconfirmed (not replicated since first described in 2001)
DYT14	Dopa-responsive dystonia, Segawa syndrome	AD	*GCH1*	Withdrawn; erroneous locus (identical to DYT5a)
DYT15	Myoclonus-dystonia	AD	18p11	Unconfirmed (not replicated since first described in 2002)
DYT16	Young-onset dystonia-(parkinsonism)	AR	*PRKRA*	Confirmed (one pathogenic variant identified)
DYT17	Autosomal recessive primary dystonia	AR	20p11.22-q13.12	Unconfirmed (not replicated since symbol in 2008)
DYT18	Paroxysmal exertion-induced dyskinesia 2	AD	*SLC2A1*	Confirmed

Locus	Disorder	MOI	Gene / Chromosome Locus [1]	Status & Remarks re Gene / Chromosome Locus
DYT19	Episodic kinesigenic dyskinesia 2 (PKD2)	AD	16q	Unconfirmed (clinical overlap w/PKD1; locus very close to DYT10)
DYT20	Paroxysmal nonkinesigenic dyskinesia 2 (PNKD2)	AD	2q	Unconfirmed (clinical overlap w/PNKD1; locus very close to DYT8)
DYT21	Late-onset pure dystonia	AD	2q14.3-q21.3	Unconfirmed [7]
DYT22			Not listed in OMIM or PubMed	Undescribed form of dystonia; designation may have been 'reserved'
DYT23	Adult onset cranial-cervical dystonia	AD	*CIZ1*	Unconfirmed
DYT24	Adult onset cranial-cervical dystonia	AD	*ANO3*	Confirmed
DYT25	Adult onset cranial-cervical dystonia	AD	*GNAL*	Confirmed
DYT26	Myoclonic dystonia	AD	*KCTD17*	Unconfirmed
DYT27	Adolescent-onset segmental dystonia	AR	*COL6A3*	Unconfirmed
DYT28	Childhood-onset generalized dystonia	AD	*KMT2B*	Confirmed
DYT29	Childhood-onset mixed movement disorder w/optic atrophy & basal ganglia abnormalities	AR	*MECR*	Unconfirmed & complex phenotype

Data are compiled from the following standard references: gene from HGNC; chromosome locus and locus from OMIM.

MOI = mode of inheritance

AD = autosomal dominant

AR = autosomal recessive

XL = X-linked

1. Chromosome locus included only if gene has not been identified

2. The most likely candidate gene is *TAF1*

3. Domingo et al [2015]

4. Pathogenic variants in *TUBB4A* may cause a broader phenotype including leukoencephalopathy.

5. Unconfirmed but supported by the description of several individuals with an 18p deletion syndrome and co-occurrence of dystonia

6. Alternating hemiplegia of childhood is the phenotype in some families.

7. Norgren et al [2011]

References

Domingo A, Westenberger A, Lee LV, Brænne I, Liu T, Vater I, Rosales R, Jamora RD, Pasco PM, Cutiongco-Dela Paz EM, Freimann K, Schmidt TG, Dressler D, Kaiser FJ, Bertram L, Erdmann J, Lohmann K, Klein C. New insights into the genetics of X-linked dystonia-parkinsonism (XDP, DYT3). Eur J Hum Genet. 2015;23:1334-40.

Norgren N, Mattson E, Forsgren L, Holmberg M. A high-penetrance form of late-onset torsion dystonia maps to a novel locus (DYT21) on chromosome 2q14.3-q21.3. Neurogenetics. 2011;12:137-43.