Dystonia Classification

The dystonias can be divided into three groups: idiopathic, genetic, and acquired.

Genetic or Primary dystonia

Classification is based on two main axes: clinical features and etiology. Currently, genes have been reported for 14 types of monogenic isolated and combined dystonia. Isolated dystonia (with dystonic tremor) can be caused by mutations in TOR1A (DYT1), TUBB4 (DYT4), THAP1 (DYT6), PRKRA (DYT16), CIZ1 (DYT23), ANO3 (DYT24), and GNAL (DYT25). Combined dystonias (with parkinsonism or myoclonus) are further subdivided into persistent (GCHI [DYT5], SGCE [DYT11], and ATP1A3 [DYT12], with TAF1 most likely but not yet proven to be linked to DYT3) and paroxysmal (PNKD [DYT8], PRRT2 [DYT10], and SLC2A1 [DYT18]). Recent insights from neurophysiologic studies identified functional abnormalities in two networks in dystonia: the basal ganglia-sensorimotor network and, more recently, the cerebellothalamocortical pathway. Besides the well-known lack of inhibition at different CNS levels, dystonia is specifically characterized by maladaptive plasticity in the sensorimotor cortex and loss of cortical surround inhibition. The exact role (modulatory or compensatory) of the cerebellar-cortical pathways still has to be further elucidated. In addition to botulinum toxin for focal forms, deep brain stimulation of the globus pallidus internus is increasingly recognized as an effective treatment for generalized and segmental dystonia.

Summary: The revised classification and identification of new genes for different forms of dystonia, including adult-onset segmental dystonia, enable an improved diagnostic approach. Recent pathophysiologic insights have fundamentally contributed to a better understanding of the disease mechanisms and impact on treatment, such as functional neurosurgery and nonpharmacologic treatment options [1)].

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see Familial dystonia

see Primary dystonia.

Here are some common classifications of genetic dystonias:

Primary (Idiopathic) Dystonias:

Early-Onset Primary Dystonia (DYT1): This is one of the most well-known genetic dystonias. It typically begins in childhood or adolescence and is associated with a mutation in the TOR1A gene. This form of dystonia often affects the limbs and torso.

DYT6 (THAP1): DYT6 is another genetic form of primary dystonia associated with mutations in the THAP1 gene. It can present with focal or generalized dystonia and usually has an adult-onset.

DYT5 (GCH1): Also known as Segawa syndrome, DYT5 is caused by mutations in the GCH1 gene. It usually starts in childhood with diurnal fluctuations in symptoms and responds well to levodopa.

Dystonia-Plus Syndromes:

Some genetic dystonias are associated with additional neurological features, leading to the classification of dystonia-plus syndromes.

DYT11 (SGCE): This is associated with myoclonus and psychiatric symptoms and is known as myoclonus-dystonia syndrome.

Paroxysmal Dystonias:

Some forms of dystonia are paroxysmal, meaning they occur in episodes rather than being continuous. DYT8 (PNKD): This form is associated with paroxysmal nonkinesigenic dyskinesia and can include dystonic movements during episodes. Dystonia with Neurodegeneration:

Some genetic forms of dystonia are associated with neurodegenerative features. DYT3 (ATP1A3): Also known as rapid-onset dystonia-parkinsonism (RDP), this condition presents with abrupt onset dystonia and parkinsonism. It's important to note that genetic dystonias can be inherited in an autosomal dominant, autosomal recessive, or X-linked manner. Genetic testing is often employed to identify specific gene mutations associated with dystonia, providing valuable information for diagnosis and sometimes influencing treatment decisions

Acquired or Secondary dystonia

see Secondary dystonia.

Axial dystonia

Cervical dystonia.

Nocturnal paroxysmal dystonia.

Medically refractory upper extremity dystonia

Status Dystonicus is a severe form of dystonia, a movement disorder

1)
Morgante F, Klein C. Dystonia. Continuum (Minneap Minn). 2013 Oct;19(5 Movement Disorders):1225-41. doi: 10.1212/01.CON.0000436154.08791.67. PMID: 24092288.