Diagnosis and Treatment of Dystonia

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KEYWORDS

• Blepharospasm • Botulinum toxin • Cervical dystonia • Deep brain stimulation
• Focal hand dystonia • Meige syndrome • Oromandibular dystonia
• Spasmodic dysphonia

KEY POINTS

- The dystonias are a large group of heterogeneous conditions characterized by excessive muscle contractions leading to abnormal postures and/or repetitive movements.
- A careful evaluation is needed for all patients with dystonia to identify uncommon subtypes in which specific etiology-based treatments can dramatically alter the course of the disorder.
- Botulinum toxins are the treatment of first choice for most focal and segmental dystonias, and may be used also to target specific problematic regions in patients with broader involvement.
- Deep brain stimulation and other surgical procedures are available when oral medications and botulinum toxins provide inadequate relief of symptoms.

Videos of various dystonias accompany this article at http://www.neurologic.theclinics.com/

INTRODUCTION

The dystonias are a group of disorders defined by specific types of abnormal movements. The essential feature is overactivity of muscles needed for movement. This overactivity can be expressed as excessive force in the primary muscles used for a movement, overflow activation of additional muscles that are not required for a

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movement, or coactivation of muscles that antagonize the primary muscles. The clinical expression of dystonia is determined by the severity and distribution of muscles involved. In mild cases, dystonic movements appear merely as exaggerations of specific actions. In moderate cases, the movements are more clearly abnormal with a quality that is cramped, stiff, or twisting. In more severe cases, dystonic movements appear as persistent odd postures or fixed deformities.

Dystonic movements are often slow, but they sometimes may be rapid or jerky.[1,2] Sometimes the movements may resemble tremor.[3–5] They tend to be patterned or stereotyped in individual cases. A recent consensus work group provided the following formal definition for the dystonias[6]:

> *Dystonia is a movement disorder characterized by sustained or intermittent muscle contractions causing abnormal, often repetitive movements, postures, or both. Dystonic movements are typically patterned, twisting, and may be tremulous. Dystonia is often initiated or worsened by voluntary action and associated with overflow muscle activation.*

Virtually any region of the body may be affected, alone or in various combinations. The dystonias may emerge at any age; and once they begin, they rarely remit. Some remain relatively static, whereas others are progressive or intermittent. Dystonia may occur in isolation, or it may be combined with other clinical problems. The many different clinical manifestations are classified according to 4 dimensions (**Table 1**), including the region of the body affected, the age at onset, temporal aspects, and whether there are associated clinical problems.[6] Each of these dimensions has implications for diagnosis and treatment.

In addition to the widely varying clinical manifestations of the dystonias, there also are many different causes.[7] Some dystonias are inherited, others are acquired (**Table 2**). Some dystonias have no apparent pathology in the nervous system, whereas others are associated with defects that can be detected by neuroimaging or postmortem histopathological studies. At the molecular level, multiple genes have been discovered for rare subtypes of dystonia,[8,9] and they are involved in diverse biochemical processes.[9–11] At the anatomic level, several brain regions have been implicated, leading to the concept that dystonia does not arise from dysfunction of

Table 1
Classification of the dystonias according to clinical features

Dimension for Classification	Subgroups
Age at onset	Infancy (birth to 2 y) Childhood (3–12 y) Adolescence (13–20 y) Early adulthood (21–40 y) Late adulthood (40 y and older)
Body distribution	Focal (1 isolated region) Segmental (2 or more contiguous regions) Multifocal (2 or more noncontiguous regions) Hemidystonia (half the body) Generalized (trunk plus 2 other sites)
Temporal pattern	Disease course (static vs progressive) Short-term variation (persistent, action-specific, diurnal, paroxysmal)
Associated features	Isolated (with or without tremor) Combined (with other neurologic or systemic features)