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Dystonia: clinical approach

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Abstract

Dystonia refers to sustained and vigorous contractions forcing a body region into an abnormal position that is consistently present. Dystonic postures and movements can variably combine to produce a wide spectrum of clinical presentations. The movement can affect one, two or more body regions, as in focal, segmental or generalized dystonia. Dystonic movements display specific features that can be recognised by clinical observation, such as speed, consistency, predictability, variability and relationship with voluntary movement. Sensory tricks and *gestes antagonistes* are manoeuvres that specifically alleviate dystonic movements and postures, thereby providing diagnostic clues. The diagnosis of primary dystonia can be established by applying a simple diagnostic flow chart during neurological examination to guide further laboratory testing.

Keywords: Dystonia; Primary dystonia; Diagnostic algorithm; Clinical features

1. Introduction

Dystonia is characterized by sustained muscle contractions, frequently causing repetitive twisting movements or abnormal postures [1,2]. Although dystonia is thought to be rare, it is possibly underdiagnosed or misdiagnosed due to the lack of specific clinical criteria. The classification of dystonia is based on three axes: (a) etiology, (b) age at onset of symptoms, and (c) distribution of affected body regions (Table 1). The etiological axis discriminates primary (idiopathic) dystonia, in which dystonia is the only clinical sign without any identifiable exogenous cause or other inherited or degenerative disease, from non-primary forms in which dystonia is usually just one of several clinical signs. Dystonia-plus is characterized by dystonia in combination with other movement disorders such as myoclonus or parkinsonism. Primary dystonia and dystonia-plus, whether sporadic or familial, are thought to be of genetic origin in most cases. In addition, dystonia can coexist with other clinical features (heredodegenerative diseases with dystonia), can be secondary to specific causes or may have a paroxysmal appearance (Table 1). Primary dystonias and dystonia-plus syndromes are the most important and common forms.

The prevalence of dystonia is difficult to ascertain. On the basis of the best available estimates, the prevalence of primary dystonia may be 11.1 per 100,000 for early-onset cases in Ashkenazi Jews from the New York area, 60 per 100,000 for late-onset cases in Northern England, and 300 per 100,000 for late-onset cases in the Italian population over age 50 [3]. Primary dystonia and dystonia-plus are chronic and often disabling conditions with a broad clinical spectrum mainly in young people. Areas of specific concern include differential diagnosis with other movement disorders, etiological diagnosis, drug treatment, surgical interventions, and genetic counseling.

A task force appointed by the European Federation of Neurological Societies (EFNS) and by the European Section of the Movement Disorders Society (MDS-ES) performed a systematic review on the diagnosis and treatment of primary (idiopathic) dystonia and dystonia-plus syndromes [4]. Literature search on the diagnosis of dystonia identified no existing guidelines or systematic reviews, two consensus agreements [1,5], two reports of workshops or taskforces [6,7], 69 primary studies on clinically based diagnosis, and 292 primary studies on the diagnostic accuracy of different laboratory tests.

2. Clinical features of primary dystonia

The clinical features of dystonia encompass a combination of dystonic movements and postures to create a sustained postural twisting ("mobile" dystonia). Dystonic postures can precede the occurrence of dystonic movements and in rare cases can persist without the appearance of

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Table 1
Classification of dystonia based on three axes[a]

By cause (etiology)

- Primary (or idiopathic): dystonia is the only clinical sign and there is no identifiable exogenous cause or other inherited or degenerative disease. Example: DYT-1 dystonia.
- Dystonia-plus: dystonia is a prominent sign, but is associated with another movement disorder. There is no evidence of neurodegeneration. Example: Myoclonus-dystonia (DYT-11).
- Heredodegenerative: dystonia is a prominent sign, among other neurological features, of a heredodegenerative disorder. Example: Wilson's disease.
- Secondary: dystonia is a symptom of an identified neurological condition, such as a focal brain lesion, exposure to drugs or chemicals. Examples: dystonia due to a brain tumor, "off"-period dystonia in Parkinson's disease.
- Paroxysmal: dystonia occurs in brief episodes with normalcy in between. These disorders are classified as idiopathic (often familial although sporadic cases also occur) and symptomatic due to a variety of causes. Three main forms are known depending on the triggering factor. In paroxysmal kinesigenic dyskinesia (PKD; DYT-9) attacks are induced by sudden movement; in paroxysmal exercise-induced dystonia (PED) by exercise such as walking or swimming, and in the non-kinesigenic form (PNKD; DYT-8) by alcohol, coffee, tea, etc. A complex familial form with PNKD and spasticity (DYT-10) has also been described.

By age at onset

- Early onset (variably defined as $\leqslant$20–30 years): usually starts in a leg or arm and frequently progresses to involve other limbs and the trunk.
- Late onset: usually starts in the neck (including the larynx), the cranial muscles or one arm. Tends to remain localized with restricted progression to adjacent muscles.

By distribution

- Focal: single body region (e.g., writer's cramp, blepharospasm)
- Segmental: contiguous body regions (e.g., cranial and cervical, cervical and upper limb) Multifocal: non-contiguous body regions (e.g., upper and lower limb, cranial and upper limb)
- Generalized: both legs and at least one other body region (usually one or both arms)
- Hemidystonia: half of the body (usually secondary to a structural lesion in the contralateral basal ganglia)

[a] From Albanese et al., 2006 [4].

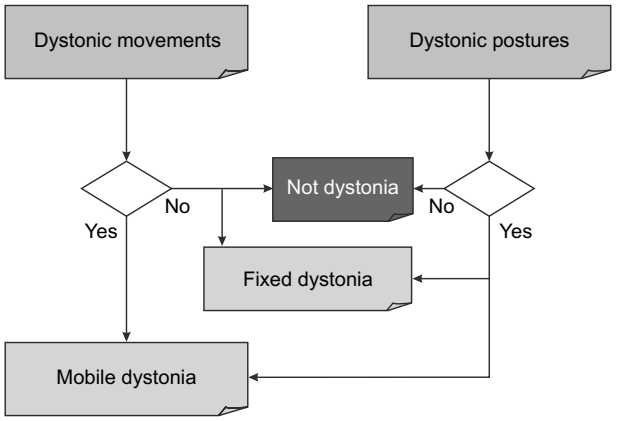

Fig. 1. Dystonia can be mobile or fixed based on the combination of postures and movements.

dystonic movements (called "fixed" dystonia; Figure 1) [8]. Sustained dystonic postures may be the presenting feature of torsion dystonia and may remain the only sign for many years before torsional movements become apparent. Dystonia has some specific features that can be recognized by clinical examination. Speed of contraction in dystonic movements may be slow or rapid, but at the peak of movement the contraction is sustained. The involuntary movement associated with dystonia is often variable over months or years and from one subject to the other. However, during a given period of observation, and within each affected individual, dystonia is distinctively consistent and predictable.

2.1. Basic features

Dystonic postures flex or twist a body part along its main axis, and are associated with a sensation of rigidity and traction. This is easy to observe on elongated body parts, such as the limbs or the trunk, but it is obviously less apparent in the cranial district. Postures are directional and force the involved body region into an abnormal position that is consistently present. In axial dystonia postural abnormalities are often a prominent feature, due to the rare occurrence of dystonic movements in the trunk. Predominantly postural forms of axial dystonia include scoliosis and camptocormia. Usually, pain is not a prominent feature of dystonia, except for cervical dystonia [9] and some secondary forms. Dystonic postures, rather than movements, may cause pain.

As a rule, dystonic movements have a twisting nature and a directional quality, are repetitive and patterned, consistent and predictable, and are sustained at their peak. The directional quality is sustained (if only for an instant), and consistency and predictability indicate that the same muscle groups are repeatedly involved. Movements are directional with variable speed. Dystonic neck movements have a directional preponderance, forcing the head to assume an abnormal position (e.g., horizontal rotation or lateral tilt), if only for a moment. Similarly, other focal forms of dystonia result in consistent directional or posturizing movements (e.g., ulnar deviation, plantar flexion, vocal cord adduction, eye closure).

Dystonic movements are occasionally rhythmic but most often arrhythmic. When rhythmic, they can be hard to differentiate from non-dystonic essential tremor [10]. Aside from their directional character, other clinical features that indicate rhythmic dystonia rather than essential tremor include: irregularity, the appearance or worsening of tremor when the affected body part is placed in a position opposite to the direction of pull, and activation of muscles not required for maintenance of the movement (overflow, as described below). By contrast, dystonic movements are easily distinguished from chorea: in dystonia there is no flowing of movement along the affected body parts, and muscle tone is not reduced. Dystonic movements may