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Focal Dystonias of the Hand and Upper Extremity

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Abstract

Hand dystonia can cause substantial functional disability and is frequently misdiagnosed. In general, dystonia is a disabling disorder of motor control characterized by excessive muscle contractions that can produce involuntary movements and abnormal postures. Prevalence of dystonia can be as high as 1 in 2500 people. Dystonias can be classified based on etiology, age of onset, anatomical part affected, and their tempo. Etiologically, they are classified as primary when the dystonia is the main sign and the cause is genetic or unknown and secondary when there are other disease manifestations and the cause may be identifiable. Dystonias that start before 27 years of age are called childhood-onset dystonia; they usually start in the lower limbs, trunk, or upper extremities and frequently spread to the rest of the body. Adult-onset dystonias usually begin in the upper half of the body, and the risk of progression to other body parts depends on the anatomic site of onset.¹ Anatomically, dystonias can be focal (1 body part), segmental (2 or more contiguous body parts), multifocal (2 noncontiguous areas), hemidystonia, or generalized. Based on their tempo, dystonias can be constant, intermittent, or situational, the latter including task-specific dystonias. The rest of the review focuses on the clinical manifestations, differential diagnosis, pathophysiology, and treatment of hand dystonia.

CLINICAL FEATURES OF UPPER EXTREMITY DYSTONIAS

Symptoms, signs, treatment, and prognosis of upper limb dystonias depend on their etiology.

Primary focal upper extremity dystonia

The initial presentation of hand dystonias includes excessive tightness in those muscles needed for a selected action with subsequent activation of surrounding muscles that produce abnormal movements or postures. The initial selected movements may be normal, but as the activity continues, dystonia may develop and worsen. Patients complain about lack of dexterity, discomfort, and fatigue of the involved hand. These abnormalities distort task performance. Primary dystonias of the upper extremity commonly begin as a task-specific dystonia occurring during selective, usually highly skilled and repetitive motor tasks, with no impairment of movement during other activities at onset.² Examples include writer's, typist's, or musician's cramp, or sport-related problems such as golfer's yips. As the condition progresses, dystonia may become less specific and impair other motor activities. About 16% of primary focal hand dystonias spread proximally or contralaterally or become generalized within 8 years (range = 2–32) of symptom onset.¹ Task-specific dystonias of the upper extremity should be differentiated from overuse syndromes because median or ulnar nerve entrapments are common in dystonia patients. Clinically apparent changes in sensation in the involved hand are more likely due to the associated peripheral nerve dysfunction rather than dystonia. It is unclear whether the repetitive tasks performed before the onset of dystonia leads to these

overuse syndromes or whether the excessive muscle activity from the dystonia also contributes to their development. Most important, treatment of the overuse syndrome will not alleviate the dystonia.

The mean age of onset of writer's cramp is 38 years and manifests as an abnormally tight grip while writing that may be associated with anomalous posture of the upper limb and impaired writing performance. The most common findings are excessive finger and wrist flexion, forearm pronation and humeral abduction, and in some cases, tremor. A "geste antagoniste" of rubbing the dorsal aspect of the hand can temporarily ameliorate symptoms. The majority of the patients have illegible penmanship; remissions are uncommon, and symptoms can progress to the contralateral hand.

Musician's cramp refers to a hand task-specific dystonia during instrumental performance that usually occurs at the peak professional stage of training and causes abnormal posturing with inability to perform specific passages.³ The affected hand is usually the one that performs the most demanding tasks, and the anomalous postural pattern depends on the instrument played. Some musicians can play a different instrument without difficulty. The prognosis of this task-specific dystonia is poor, as treatment remains suboptimal, and many artists lose the ability to perform at a professional level.

Generalized dystonias that present as focal dystonia

Primary childhood-onset dystonias may start as focal primary dystonias but then most commonly generalize. Examples of primary generalized dystonias include dopa-responsive dystonia and dystonia due to a genetic defect on chromosome 9, called DYT1. About half of those with DYT1 generalized dystonia begin with symptoms in the upper extremity.

Secondary dystonias

Patients with secondary dystonias usually have other abnormalities on physical examination, such as rigidity, poor balance, spasticity, weakness, dementia, altered reflexes, eye movement restrictions—all clues of other diseases such as Parkinson disease, stroke, or other neurodegenerative conditions.⁴ Acute or chronic exposure to antidopaminergic drugs such as metoclopramide, prochlorperazine, or neuroleptics may cause drug-induced dystonia. The treatment and prognosis for secondary dystonias differs for each diagnosis.

Differential diagnosis and evaluation

Focal hand dystonia is a clinical diagnosis. Electro-physiologic tests are not diagnostic. Differential diagnosis of focal hand dystonias includes neuropathies, radiculopathies, plexopathies, complex regional pain syndrome, repetitive stress injury, focal seizures, thoracic outlet syndrome, medications effects, and psychogenic movement disorder. A thorough history and examination will identify focal hand dystonia and whether these other conditions also contribute to patient's symptoms.

PATHOPHYSIOLOGY OF HAND DYSTONIA

Our current understanding of the pathophysiology of dystonia is limited. Structural and functional abnormalities of the basal ganglia and their pathways have been found. Dystonias may be associated with lack of inhibition at multiple levels of the nervous system, increased neural plasticity, and an abnormal sensory processing.⁵ Different types of dystonias may share common pathophysiological features but likely differ by etiology and specific anatomic sites of brain dysfunction.

TREATMENT

Oral medications like anticholinergics or baclofen provide modest, if any, benefit for primary hand dystonias. Judicious injections of botulinum toxin to chemically denervate muscles may reduce symptom severity, but excessive weakness with functional impairment must be avoided. Active research into approaches for retraining movement strategies may provide benefit in the future. Occupational therapists frequently suggest useful adaptive strategies. Finally, neurosurgical interventions, including deep brain stimulation, have potential to alleviate symptoms, but optimal specific brain targets remain to be determined. Treatment of secondary dystonias depends on the cause of the syndrome.

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References

1. Weiss EM, Hershey T, Karimi M, Racette B, Tabbal SD, Mink JW, et al. Relative risk of spread of symptoms among the focal onset primary dystonias. *Mov Disord* 2006;21:1175–1181. [PubMed: 16673404]
2. Torres-Russotto D, Perlmutter JS. Task-specific dystonias: a review. *Ann N Y Acad Sci* 2008;1142.
3. Conti AM, Pullman S, Frucht SJ. The hand that has forgotten its cunning—lessons from musicians' hand dystonia. *Mov Disord* 2008;23:1398–1406. [PubMed: 18398917]
4. Fahn S, Bressman SB, Marsden CD. Classification of dystonia. *Adv Neurol* 1998;78:1–10. [PubMed: 9750897]
5. Breakefield XO, Blood AJ, Li Y, Hallett M, Hanson PI, Standaert DG. The pathophysiological basis of dystonias. *Nat Rev Neurosci* 2008;9:222–234. [PubMed: 18285800]