



DEPARTMENT OF
NEUROLOGY

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Dystonia

Diagnosis

Dystonia is a neurologic movement disorder dominated by involuntary, sustained or repetitive, patterned muscle contractions or spasms, frequently causing squeezing, twisting, and other movements or abnormal postures. The diagnosis is based on clinical evaluation. Depending on the evaluation, additional testing such as brain MRI and genetic testing may be required. Typically, dystonia begins in a single body part (focal dystonia), such as the hand, neck or eyelids, and then it spreads to adjacent body parts (segmental dystonia). Blepharospasm, a focal dystonia manifested by an involuntary eye closure produced by spasmodic contractions of the eyelids and eyebrows, is often associated with dystonic movements of facial, jaw, laryngeal, and neck muscles. Oromandibular dystonia may be manifested by involuntary jaw opening or closing associated with clenching (trismus) and grinding of the teeth (bruxism), which may lead to secondary dental wear and temporal-mandibular joint (TMJ) syndrome.

In our Movement Disorders Clinic at Baylor College of Medicine, neck or cervical dystonia is the most frequent form of dystonia. This form of focal dystonia is characterized by patterned, repetitive, head movements or more sustained abnormal postures of the head. The term "spasmodic torticollis" is still occasionally used to describe this form of dystonia. However, since it is not always spasmodic and it does not always consist of head turning (torticollis), the term "cervical dystonia" is preferred as a generic descriptor of dystonic movements or postures involving the neck. In addition to torticollis (turning of the head), cervical dystonia may be manifested by neck flexion (anterocollis), extension (retrocollis), or head tilt (laterocollis). In approximately one third of all patients, cervical dystonia progresses to involve contiguous body parts such as the oromandibular region, shoulder, trunk,

and arm. Cervical dystonia is associated with pain in about 75% of patients and most patients have discovered certain alleviating maneuvers such as touching the chin or neck, to correct the abnormal postures. Segmental dystonia involving eyelids (blepharospasm) and other facial, jaw (oromandibular) and neck (cervical) muscles is referred to "cranial-cervical dystonia". Spasmodic dysphonia, a focal dystonia of the vocal cords and larynx, is characterized by strained, effortful voice interrupted by uncontrollable pitch breaks or voiceless pauses (adductor spasmodic dysphonia) or whispering, breathy voice (abductor spasmodic dysphonia). While cervical dystonia is the most common form of dystonia encountered in a specialized clinic, writer's cramp is probably more common in general population. In addition to this "task-specific" dystonia, there are many other examples of occupational dystonias affecting the performance of musicians, sportsmen, and others whose skills depend on finely coordinated movements. Dystonia, particularly of childhood onset, may spread to involve the legs, trunk and other body parts (generalized dystonia).

Unilateral dystonia, confined to only one half of the body, is referred to as hemidystonia. In contrast to focal and segmental dystonia, which is usually "idiopathic" or "primary" (no other neurological abnormality and no specific cause except possibly genetic), the majority of patients with hemidystonia have an identifiable etiology such as head trauma, stroke, arteriovenous malformation, tumor, encephalitis or other pathology affecting the opposite basal ganglia.

Cause

The cause of primary dystonias is not always apparent, but most are probably due to some genetic abnormalities (inherited). Although in most patients with dystonia no specific abnormality or lesion can be identified by neuroimaging or even autopsy studies, there is convincing evidence that this movement disorder is due to abnormal function of the deep portion of the brain, called basal ganglia. Currently, dystonia is considered the result of a combination of impaired modulation of the centers that control the movements and an aberrant plasticity of the neurons in that circuitry.

There are many genetic forms of dystonia, and new gene abnormalities are being added to the growing list every year. Mutation of the *DYT1* gene on chromosome 9 is the most studied genetic mutation among the inherited forms. The *DYT1* gene codes for the protein TorsinA, which accumulates abnormally in neurons. It is not yet known how this abnormality leads to dystonia. This dystonia, which occurs with particularly high frequency in Ashkenazi Jewish population, is manifested by onset of writer's cramp, foot inversion, or other dystonia of the limbs, typically before the age of 10, and progressing to a more generalized dystonia involving legs, arms and trunk. Although the progression may vary between individuals of the same or different families, many children with *DYT1* dystonia require assistance with ambulation by the time they reach early teens.

Not all dystonias, however, are of genetic origin. Some dystonias are secondary to specific causes such as metabolic and neurodegenerative disorders (e.g. Wilson's disease), brain injury or other lesions (e.g. cerebral palsy), certain drugs that block dopamine receptors, and many other causes. The dopamine receptor blocking drugs, also called neuroleptics, such as the major tranquilizers (e.g. haloperidol) and gastrointestinal drugs (e.g. metoclopramide) can cause not only an acute transient dystonic reaction, but also a persistent dystonic disorder (tardive dystonia). In addition, injuries to the nerves can result peripherally induced dystonia caused by an injury to the affected body part is being increasingly recognized as an important cause of focal and segmental dystonia. Finally, physical or emotional stress and a variety of other psychological factors may be associated with abnormal movements resembling dystonia, the so-called "psychogenic dystonia."

Treatment

Despite the paucity of knowledge about causes of dystonia, the treatment of this condition has markedly improved, largely as a result of application of botulinum toxin (BTX). Before contemplating BTX therapy, however, potentially curable causes of dystonia, such as certain drug-induced dystonias or Wilson's disease, should be considered. The treatment of dystonia should be individualized.

Involvement of rehabilitation team, such as physical therapist, and addressing potential depression or anxiety can improve the response to treatment. For those patients with a focal or segmental dystonia, BTX is the treatment of choice. However, the cases of generalized dystonia, may benefit from pharmacologic therapy using medications such as trihexyphenidyl, baclofen, tetrabenazine, and levodopa. Surgical techniques, such as local nerve or muscle excision and ablation or high frequency deep brain stimulation (DBS) have been used successfully in patients who continue to have disabling dystonia despite optimal medical or BTX therapies. DBS targeting the globus pallidus interna and the subthalamic nucleus have emerged as the surgical treatment of choice in patients with severe generalized dystonia and can be also used in segmental dystonia or hemidystonia.

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