



Dystonia:

A real pain in the neck

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What is dystonia?

Dystonia is a neurologic syndrome characterized by involuntary sustained muscle contractions of opposing muscles which cause twisting movements or abnormal postures. The dystonic movements are repetitive and patterned. Dystonia of the neck, the most common adult form, is often referred to as “spasmodic torticollis.”

How common is dystonia?

The prevalence of focal dystonia in the general population is 30/100,000 and generalized dystonia is 3.4/100,000. Childhood onset dystonia is more commonly inherited and generalized, whereas adult onset dystonia is more commonly focal and sporadic. The mean age of onset is 50 years. Once developed, the disease tends to be lifelong with spontaneous remissions occurring in only 10% of patients with focal dystonia.^{1,2}

What are the clinical features?

Focal dystonia (*i.e.*, involving one body part) includes cervical dystonia (torticollis), limb dystonia (writer’s cramp, foot dystonia), and cranial dystonia (blepharospasm). Focal dystonia occurs most commonly in adults and is generally idiopathic.

Cervical dystonia, or “torticollis,” is caused by spasmodic or sustained contractions of the neck muscles. It may involve neck rotation, side flexion, forward flexion or extension.

Apart from the obvious symptom of neck contracture, the condition is painful in three-quarters of patients. It is exacerbated by stress or fatigue and

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Practice Pointer

Dystonia can present in many ways:

- Focal dystonia involves one body part (*i.e.*, neck, limb, face).
- Segmental dystonia involves two contiguous body parts (*i.e.*, cranial-cervical dystonia).
- Multifocal dystonia involves two or more noncontiguous body parts (*i.e.*, neck and foot dystonia).
- Generalized dystonia involves the entire body with dystonia at rest.

associated with essential tremor in one-third of cases. It may be associated with cranial or facial dystonia. The dystonia is often relieved by a “sensory trick,” such as the patient touching the cheek or chin. The mechanism of this procedure is not known.^{1,2}

Idiopathic limb dystonia usually begins as action dystonia. Often associated with activities such as writing, typing or playing musical instruments, idiopathic limb dystonia can affect the arm or leg. As with cervical dystonia, it involves co-contraction of opposing muscles and occurs with action. In the resting state, the dystonia will often disappear. Limb dystonia

may also occur secondary to a degenerative disease, such as Parkinson’s. In this case, it occurs at rest and may be affected by medication.

Blepharospasm—involuntary bilateral eye closure—is caused by spasmodic contractions of orbicularis oculi and may be accompanied by the contraction of other facial muscles. These contractions impair vision and, in up to 15% of patients, may cause legal blindness. The majority of these patients are over 50 years of age. Blepharospasm is most commonly idiopathic.

Segmental dystonia (involving two contiguous body parts) includes combinations of the focal type, such as cranial-cervical dystonia. Multifocal dystonia involves two or more non-contiguous body parts, such as torticollis and foot dystonia.^{1,2}

Generalized dystonia most commonly begins in childhood and is hereditary. The two main diseases to consider are idiopathic torsion dystonia and dopa-responsive dystonia. Idiopathic torsion dystonia begins between nine and 12 years of age with arm or leg action dystonia and progresses over the next five to 10 years to involve the entire body with dystonia at rest. Pain is an uncommon feature. Because of common leg involvement, gait is affected and may be the presenting sign.

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Dopa-responsive dystonia begins between the ages of four and eight. It starts with a stiff-legged gait that worsens as the day progresses and improves with sleep. This type of dystonia may progress to involve the arms; Parkinsonian features may also occur. Dopa-responsive dystonia affects girls more than boys.

Hemidystonia, with dystonic posturing of the hemibody, is a distinct entity. Unlike the other types of dystonia, this is usually associated with a structural lesion in the contralateral basal ganglia and requires neuroimaging.¹

Practice Pointer

There are two types of dystonia, primary and secondary, and their presentations differ.

- Primary dystonia may be sporadic, as in torticollis and writer's cramp, or may be inherited, as in childhood idiopathic torsion dystonia.
- Secondary dystonia may be related to a structural lesion, metabolic disease, drugs or a neurodegenerative condition, such as Parkinson's disease.

What conditions are associated with dystonia?

Dystonia may be defined as primary (occurring in isolation) or secondary (as a symptom of an underlying neurological disease). Primary dystonia may be sporadic, as in torticollis and writer's cramp, or may be inherited, as in childhood idiopathic torsion dystonia. Secondary dystonia may be related to a structural lesion, metabolic disease, drugs or a neurodegenerative condition, such as Parkinson's disease.^{1,2}

The pathophysiology of dystonia is not well understood. In adult-onset dystonia, the etiology is generally unknown. Some cases may be linked to heredity or autoimmune diseases, but these findings are rare. Some cases have been linked with peripheral trauma or overuse (*i.e.*, writer's cramp), although this is controversial.² In idiopathic focal dystonias, neither pathologic studies nor neuroimaging reveal consistent abnormalities. In fact, these studies are usually normal. Functional imaging with positron emission tomography has demonstrated hypometabolism in the caudate and lentiform nucleus and frontal projections of the thalamus.^{1,2}

In children, genetic causes are more common. Idiopathic torsion dystonia is an autosomal dominant disease linked to the DYT1 gene on chromosome 9, with variable penetrance of 30% to 40%. It is more common in patients of Ashkenazi Jewish descent.² Pathologic studies have revealed no consistent structural abnormality. Changes have been noted in neurotransmitter levels, but it is unclear if these are related to the dystonia.^{1,2}

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Practice Pointer

Treatment options include pharmacologic therapy, chemodenervation therapy, and surgery.

- For focal dystonias, the treatment of choice is intramuscular botulinum toxin injections.
- For generalized dystonia or as a second-line treatment for focal dystonia, medications may be used.
- Surgery is used for severe intractable cases and includes peripheral surgery such as myotomy and ventral rhizotomy.

Dopa-responsive dystonia is another autosomal dominant trait with 30% to 40% penetrance. The pathogenesis is related to a defect in the synthesis of dopamine secondary to a genetic abnormality of the guanosine triphosphate gene on chromosome 14.^{1,2}

What is the etiology?

Although dystonia is most commonly idiopathic, it may be secondary to other medical conditions.

As stated above, hemidystonia can be related to a structural lesion (stroke, tumour, hemorrhage) of the basal ganglia or brainstem structures.

Degenerative conditions, such as Parkinson's disease, Wilson's disease and Huntington's disease can cause dystonia. Usually, in these cases there are other abnormalities which are discovered during the neurological examination and suggest other causes.

Dystonia may arise as result of a neurological insult, such as diffuse anoxia, encephalitis, head injury, neck trauma or peripheral injury. It can also occur with autoimmune diseases, such as antiphospholipid antibody syndrome, systemic lupus erythematosus or multiple sclerosis.

It is important to know that dystonia can occur with some medications, both acutely and as a tardive syndrome secondary to long-term neuroleptic use.^{1,2}

In the case of cervical dystonias, structural lesions at the base of the skull and cervical spine should be suspected. Cranial dystonia may be related to poorly fitting dentures, facial pain or eye problems.¹⁻³

How is a diagnosis made?

The diagnosis of dystonia is primarily clinical. The history, typical features and family history are important. A detailed history of current and past medications is vital. Despite the dystonia, the neurological examination and general examination should be completely normal.^{1,2}

Abnormalities of corticospinal, sensory, cerebellar or ocular systems imply a secondary cause of dystonia and warrant further investigations. These procedures may include neuroimaging of the head and spinal cord, blood tests for autoimmune diseases and serum vitamin E levels, as well as thick blood smear for neuroacanthocytosis.^{1,2}

All children and young adults presenting with dystonia should be screened for Wilson's disease, a degenerative disease that results in abnormal excretion of copper, causing liver, psychiatric and neurological symptoms. Wilson's disease is a devastating illness that is treatable if diagnosed early. Tests for Wilson's disease include serum ceruloplasmin and copper, 24-hour urine copper, and slit lamp ophthalmologic exam.^{1,2}

What are the options for treatment?

Treatment options include pharmacologic therapy, chemodeneration therapy, and surgery.

For focal dystonias, the treatment of choice is intramuscular botulinum toxin injections. This treatment works by blocking the release of acetylcholine at the presynaptic neuromuscular junction and causing temporary muscle weakness. The treatment is highly effective (up to 80% in randomized, controlled trials), targets only the affected muscles, and is generally well tolerated. The major disadvantages are that the medication is only effective for three to four months, requiring re-administration four times per year. The cost (\$350 to \$1,400 per treatment) is also prohibitive. Side effects include muscle weakness, local injection discomfort and, rarely, transient dysphagia.⁴

For generalized dystonia or as a second line treatment for focal dystonia, medications may be used. Oral medications include anticholinergic agents such as trihexiphenidyl, gamma-aminobutyric acid (GABA)-ergic medications such as baclofen or clonazepam, and dopamine-depleting agents such as tetrabenazine. These are modestly effective, but carry the risk of significant side effects.⁴

Surgery is used for severe, intractable cases and includes peripheral surgery such as myotomy and ventral rhizotomy. More recently, brain lesioning or deep brain stimulation of the thalamus or globus pallidus have been used for severe generalized dystonia. The results are variable and have not been subjected to controlled clinical trials.^{1,3,4}

CME



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