

What is Dystonia?

Dystonia is a very complex, highly variable neurological movement disorder characterized by involuntary muscle contractions or spasms. An estimated 500,000 people in the United States have been diagnosed with Dystonia with an estimated 500,000 yet to be diagnosed or misdiagnosed, making it the third most common movement disorder behind essential tremor and Parkinson's disease. It is a disorder that knows no age, ethnic, religious, sexual, or socioeconomic barriers – it can affect young children to older adults of all races, creeds, ethnicities, or social status.

Dystonia results from abnormal functioning of the basal ganglia, a deep part of the brain which helps control coordination of voluntary movement and inhibition of involuntary movements. This region of the brain controls the speed and fluidity of movement and prevents unwanted movements. Because this important basal ganglia function is altered in Dystonia patients, they experience simultaneous contractions of opposing muscle groups, resulting in abnormal postures, twisting or repetitive movements. These can affect any part of the body, including the hands, feet, arms, legs, trunk, neck, face, eyes and vocal cords. Symptoms may or not be obvious to the casual or even a trained observer.

Depending on the affected body part, Dystonia can seriously impact daily functions. For example, if neck muscles are affected, a patient may have difficulty with balance, posture, chewing, or swallowing. In some cases, Dystonia may also be quite painful. Though not life-threatening, the involuntary nature of the disorder may be embarrassing, causing emotional distress or depression in some individuals. The American Dystonia Society has an on-line Dystonia networking site to provide peer support at ADS Community Center that can help address some of these issues, but patients may need to be treated separately for mental health issues caused by the challenges of coping with this disorder.

Dystonia Classification

Dystonia is classified by three main factors: the age at which symptoms develop; the areas of the body affected; and the underlying cause.

The chance that Dystonia will affect multiple body parts is generally linked to the age of onset. The younger one is at onset, the greater the chance that symptoms will spread. Conversely, the older one is at onset, the more likely that the disorder will remain more moderate.

Dystonia Classification By Age

Early Onset – 0 to age 28

Adult onset – older than age 28

Dystonia Classification By Body Part

Focal Dystonia is limited to one area of the body, and can affect the neck (Cervical Dystonia or Spasmodic Torticollis, Retrocollis, Anterocollis, or Laterocollis), eyes (Blepharospasm), jaw/mouth/lower face (Oromandibular Dystonia), vocal cords (Laryngeal Dystonia or Spasmodic Dysphonia), or arms/legs (Limb Dystonia). Other less common types of focal Dystonias can cause unusual stretching, bending, or twisting of the trunk (Truncal Dystonia) or sustained contractions and involuntary, writhing movements of the abdominal wall (Abdominal Wall Dystonia).

Focal Dystonia more commonly affects people in their 40s and 50s and is frequently referred to as adult-onset Dystonia. Women are diagnosed about three times more frequently than men. In general, focal Dystonias are classified as primary (idiopathic) and are not hereditary.

Segmental Dystonia affects two or more parts of the body that are adjacent or close to one another. Up to 30 percent of people with focal Dystonia have spasms in areas adjacent to the primary site. A common form of segmental Dystonia affects the eyelids, jaw, mouth, and lower face and is called cranial Dystonia.

Other types of Dystonia include multifocal, which involves two or more body parts distant from one another; HemiDystonia, which affects half of the body; and generalized, which begins with leg involvement, but generally spreads to one or more additional regions of the body.

Dystonia Classification By Cause

Primary (idiopathic) – Dystonia is the only sign, and secondary causes have been ruled out. Most primary Dystonias are variable, have adult onset, and are focal or segmental in nature. However, there are specific primary Dystonias with childhood or adolescent onset that have been linked to genetic mutations.

The majority of early-onset primary Dystonias, which may appear during childhood or early adulthood, are due to mutations of a gene known as DYT1. This gene has been mapped to the long arm of chromosome 9 at 9q34.1. In about 90 to 95 percent of cases, symptoms begin in a limb and then spread to other regions of the body. This form of Dystonia has an average age of onset of 12 and seldom develops after age 29.

DYT6 Dystonia is an autosomal dominant primary Dystonia that has been mapped to chromosome 8 (8p21q22). It is rarer than DYT1 Dystonia and has been studied in two Mennonite families in the United States. In nearly all individuals with this form of Dystonia, the disorder begins at an initial site but spreads to multiple body regions, most commonly the limbs, head, or neck. Severe difficulties with speech articulation have been noted.

Other familial primary Dystonias identified are DYT2, DYT3, DYT4, and DYT7, all of which have been noted in specific ethnic groups, primarily of European descent. DYT3 is specific to a single family line in the Philippines.

Secondary (symptomatic) – Dystonia that results primarily from secondary causes. These include environmental, such as exposure to carbon monoxide, cyanide, manganese, or methanol; underlying conditions and diseases such as brain tumors, cerebral palsy, Parkinson’s disease, stroke, multiple sclerosis, hypo-parathyroidism, or vascular malformations; brain/spinal cord injuries; inflammatory, infectious, or post-infectious brain conditions; and specific anti-psychotic medications.

Dystonia-plus syndromes – Dystonia that results from non-degenerative, neurochemical disorders associated with other neurological conditions. Dystonia-plus syndromes include Dopa-Responsive Dystonia (DRD) or Segawa syndrome, rapid-onset Dystonia-Parkinsonism (RDP), and Myoclonus-Dystonia.

Hereditary degenerative Dystonia – Dystonia that generally results from neurodegenerative disorders in which other neurological symptoms are present and in which heredity plays a role. These include numerous disorders such as certain X-linked recessive, autosomal dominant, autosomal recessive, and/or parkinsonian syndromes. Included in this category: X-linked Dystonia-parkinsonism (Lubag), Huntington's disease, Wilson's disease, neuroacanthocytosis, Rett’s syndrome, Parkinson's disease, and juvenile parkinsonism.

Symptoms

Dystonia is sometimes misdiagnosed as stress-induced pains, a stiff neck, back pain, Temporomandibular Joint Disorder (TMJ), or a psychogenic disorder with psychological triggers. The intermittent character of the disorder may lead medical practitioners to conclude that a psychogenic disorder is either the primary cause or an underlying factor. Diagnosis is difficult because Dystonia symptoms are similar to those of many other conditions and are so variable in nature. Stress levels also affects the severity and frequency of the Dystonia symptoms but is not a primary cause of Dystonia.

Dystonia initially arises after specific movements or tasks, but in advanced stages it may occur at rest. It usually affects the same group of muscles, thus causing a pattern of movements over time. It generally develops gradually, with localized symptoms suggesting the presence of the disorder. Eye irritation, excessive sensitivity to bright light, or increased blinking may be an indication of Blepharospasm. Subtle facial spasms or tics, difficulty chewing or swallowing, or changes in speech cadence may indicate Oromandibular or Laryngeal Dystonia. Cramping of the hand during writing or performing a task specific motion like playing a musical instrument is associated with Writer's Cramp (Graphospasm) or Musician's Dystonia, while cramping or fatigue doing everyday tasks (preparing food, walking) may occur with other forms of Dystonia.

Dystonia is also variable in its progression. For some patients, the disorder steadily worsens; for others, it plateaus. For some, Dystonia stabilizes at a relatively minor stage and progresses no further. The advanced stage is marked by rapid and involuntary rhythmic movements, twisting postures, contortions of the torso, abnormal gait, or fixed postural deformities.

The disorder may lead to varying degrees of pain and dysfunction. Cervical and other Dystonias that affect the spine can be debilitating and painful due to degeneration of the spine and irritation of nerve roots. Headaches may also occur. Limb Dystonia may not cause pain initially but may become painful over time. Uncontrolled muscle movements may cause the joints to deteriorate, eventually leading to the onset of arthritis. Tendons and ligaments may also be affected, contributing to issues with tendonitis and tendonosis.

Treatment

There is a three-tiered approach to treating Dystonia: Botulinum Toxin (botox) injections, several types of medication, and surgery. These may be used alone or in combination. Medications and Botulinum Toxin (Botox, Dysport, Myobloc, or Xeomin) can both help block the communication between the nerve and the muscle and may alleviate abnormal movements and postures.

Botulinum toxin type A (Botox) was developed in the 1980s. The newer Type A toxins Dysport and Xeomin were developed in the 1990s. In 2001, the U.S. Food and Drug Administration approved Botulinum Toxin type B (Myobloc) for treatment of Cervical Dystonia. Researchers created the new drug after some patients began developing resistance to the type A form. Xeomin is a second generation Type A toxin that has a very low bacterial protein load that reduces the risk of developing resistance. The type B toxin (Myobloc) has mild to moderate side effects such as dry mouth, dysphagia (difficulty swallowing) and indigestion. Botulinum Toxin treatments are reversible as the effects wear off generally after 8-10 weeks.

Surgery is considered when other treatments have proven ineffective. The goal of surgery is to interrupt the pathways responsible for the abnormal movements at various levels of the nervous system. Some operations purposely damage small regions of the thalamus (thalamotomy), globus pallidus (pallidotomy), or other deep centers in the brain. These procedures are irreversible and must be considered carefully. Deep brain stimulation (DBS) has been tried recently with some success and is generally reversible. Other irreversible surgeries include cutting nerves leading to the nerve roots deep in the neck close to the spinal cord (anterior cervical rhizotomy) or removing the nerves at the point they enter the contracting muscles (selective peripheral denervation).

The benefits of surgery should always be weighed carefully against its risks. Although some Dystonia patients report minor to significant symptom reduction after surgery, there is no guarantee that surgery will help every individual. DBS has gained widespread acceptance and positive publicity lately but no data on success rates have yet to be published.