WHAT IS DYSTONIA?
Dystonia is a neurologic movement disorder that causes uncontrollable muscle contractions in one or more parts of the body. It may affect the face, neck, vocal cords, arms, legs or torso.

People living with dystonia are affected in different ways. The severity of the disease varies from person to person. Many people who have dystonia can maintain a relatively normal lifestyle. Others may need full-time assistance.

500,000 people are estimated to have dystonia in the United States and Canada. Men, women, and children of all racial and ethnic backgrounds can be affected.

WHAT ARE THE SYMPTOMS?
Dystonia causes involuntary repetitive twisting and sustained muscle contractions. These result in abnormal movements and postures. The symptoms usually begin in one body region, such as the neck, face, vocal cords, an arm or a leg, and then may spread to other parts of the body. When dystonia affects children, it often starts in a leg before spreading to other limbs and trunk. In adults, dystonia tends to remain localized to a body region and frequently affects first the upper body.

Dystonia usually occurs or worsens during voluntary movement. It may also happen with movement of other unaffected body regions or when the affected body part is at rest. Dystonia may lead to sustained fixed postures potentially causing permanent contractures. The symptoms of dystonia typically increase during stress, emotional upset or fatigue, and decrease during rest and sleep. Many people living with dystonia can temporarily suppress their symptoms by using “sensory tricks”. These are a unique feature of dystonia, consistent of touching the affected or an adjacent body part to decrease the pulling or posturing.

WHAT CAUSES IT?
Dystonia may be an inherited condition caused by genetic mutations. It can also result from exposure to certain drugs, traumas, or as a symptom of other neurological disorders. For many patients, however, the cause remains unknown. There are two main categories of causes of dystonia: primary and secondary (or non-primary).
Primary Dystonia
Primary dystonia is a condition in which dystonia is the only clinical feature. There is no evidence of cell death or a known cause. It is also known as idiopathic torsion dystonia. The primary dystonias are often inherited from a parent.

Researchers over the past twenty years have identified mutations in two genes (DYT1 and DYT6) as responsible for many cases of primary dystonia. Most genetic forms of dystonia start with symptoms in childhood or adolescence. Commercial tests are available to determine if these genes are affected in individuals. However, for the majority of people living with primary dystonia, the cause remains unknown.

Secondary Dystonia
In non-primary or secondary dystonia, an acquired or exogenous cause is identified. This can be a prior stroke, a birth injury or exposure to certain drugs. Secondary dystonia may also represent one symptom of other neurological disorders, such as Parkinson’s disease.

Some genetic causes include:
- Myoclonus Dystonia
- Dopa-responsive Dystonia
- Rapid-onset Dystonia Parkinsonism
- Wilson’s Disease
- Huntington’s Disease
- Spinocerebellar Ataxias
- Methyloacetic Aciduria
- Parkinson’s disease caused by Parkin mutations

Acquired causes include:
- Exposure to drugs such as certain antipsychotic and anti-nausea medications
- Multiple sclerosis
- Past history of stroke, birth injury, infections, or trauma

The basal ganglia, a part of the brain responsible for the control of normal movements, is believed to be involved in dystonia. An important neurotransmitter in the basal ganglia is dopamine. Inherited or acquired disturbances in dopamine receptors are responsible for some forms of dystonia. These forms include dopa-responsive dystonia, tardive dystonia, and dystonia related to Parkinson’s disease.
HOW IS DYSTONIA CLASSIFIED?
Dystonia is generally classified based on its cause, the age at which first symptoms occur, and the regions of the body it affects.

Age of onset
The age of onset is an important indicator of whether the dystonia is more likely to spread to other body regions. Early-onset dystonia refers to dystonia that develops before age 21. The younger the patient at dystonia onset, the higher the likelihood that the dystonia may involve other areas. Late-onset dystonia begins after age 21. In patients with primary late-onset dystonia, the dystonia often begins in the upper body, such as the neck, head, neck, or an arm.

Regions of the body
*Generalized Dystonia:* is the most widespread form of dystonia; it affects the legs or one leg and the trunk, plus other regions, most commonly the arms.

*Focal Dystonia:* involves only one region of the body, such as the neck, vocal cords or hand.

*Hemidystonia:* affects one half of the body.

*Segmental Dystonia:* affects two or more adjacent body regions, such as the neck and an arm.

*Multifocal Dystonia:* affects two or more distant regions of the body, such as the upper face and the hand.

WHERE CAN FOCAL DYSTONIA OCCUR?
Focal Dystonia is a form of dystonia that is limited to one area of the body. Commonly described forms of focal dystonia include:

*Blepharospasm:* is characterized by intermittent or sustained eyelid closure. It is caused by involuntary contractions of the muscles around the eyes. It leads to excessive blinking and spasms of eye closure.
**Oromandibular Dystonia:** or Meige’s syndrome, affects the lower facial and jaw muscles causing involuntary opening, closing or deviation of the jaw. The tongue may be also involved.

**Cervical Dystonia:** also called spasmodic torticollis or ST, affects the neck muscles leading to abnormal movements of the neck and head. It is the most common form of focal dystonia.

**Laryngeal Dystonia:** or spasmodic dysphonia (SD) affects the vocal cords. There are two forms of laryngeal dystonia. Adductor dysphonia, which is the most common form, causes the voice to have a restricted, strangled or hoarse quality. Abductor dysphonia leads to a breathy, whispering voice.

**Limb Dystonia:** affects the legs, feet, arms or hands. Upper limb dystonia often appears only when performing a specific task, such as writing (Writer’s Cramp). Other task-specific or occupational dystonias include musician’s dystonia and “the yips” that affect golfers.

**HOW IS DYSTONIA DIAGNOSED?**

Dystonia is usually diagnosed by a movement disorder neurologist. Proper diagnosis will be contingent on results from a thorough patient history, a careful family history, and complete physical and neurological examinations. Laboratory tests, imaging studies, and even genetic testing may be necessary to reach a diagnosis.

Unfortunately, it is common for dystonia to be misdiagnosed or to remain undiagnosed when symptoms are mild. Many physicians are still unfamiliar with the disease. More education is needed to better inform the medical community about the symptoms associated with dystonia, and how to provide proper treatment.
HOW IS DYSTONIA TREATED?
(treatment should be done under the guidance of a movement disorder specialist)

The treatment depends on the cause of the dystonia. Treating the underlying condition in cases of secondary dystonia may improve the symptoms. There are three approaches to the treatment of dystonia, depending on the region of the body affected and the severity of the symptoms: medications, botulinum toxin injections and surgery. Physical therapy may be helpful as a supplement to other therapies.

**Botulinum toxin injections:** are the treatments of choice for most forms of focal dystonia. The toxin is produced by the bacterium that causes botulism. When a small amount of commercially prepared toxin is selectively injected in the overactive muscles, it causes a change in the muscle firing, calming the abnormal movements for up to several months at a time.

**Medications:** Segmental, multifocal and generalized dystonia are usually treated with oral medications. These include anticholinergic drugs (trihexyphenidyl or Artane®, benztropine or Cogentin®) and muscle relaxants or antispastic agents (diazepam or Valium®, clonazepam or Klonopin®, baclofen or Lioresal®). In addition, specific forms of dystonia can respond to particular medications. For example, dopa-responsive dystonia is treated with levodopa (Sinemet®).

**Surgery:** Patients with widespread or severe debilitating dystonia can benefit from surgery if they are unresponsive to other treatments. The most widely used current surgical approach is called deep brain stimulation, or DBS. In this surgery, thin electrodes are implanted into a part of the basal ganglia called the globus pallidus and are attached to a pacemaker-like device implanted in the chest wall. These electrodes deliver controlled electrical pulses that can have a marked improvement of dystonia symptoms, especially for patients with generalized primary dystonia.
IS THERE A CURE?
Although remarkable progress has been made in the study of dystonia in the last few years, there is not a known cure for it yet. Dystonia affects more people than Muscular Dystrophy, Huntington’s disease and Lou Gehrig’s disease combined, yet awareness of dystonia is limited. More research, treatments and education about its diagnosis are needed before a cure can be found.

HOW IS DYSTONIA RELATED TO PARKINSON’S DISEASE (PD)?
Dystonia and PD are movement disorders that are closely related. First, both conditions can occur together in certain diseases. People living with PD may experience dystonia as an early symptom or as a complication of treatment. Dopa-responsive dystonia and rapid-onset-dystonia-parkinsonism are hereditary forms of dystonia in which PD is often also present. Other neurodegenerative disorders, such as Wilson’s disease, may have both dystonia and PD, in conjunction to other clinical features. Second, dystonia and PD share common treatments. Anticholinergic medications and levodopa may ameliorate both conditions, and DBS is a surgical alternative for both, although the final brain target may vary. Lastly, PD and dystonia are thought to result from dysfunction of the basal ganglia and their output, although the ultimate cause of the disorders is not known. Further research is necessary to determine the various underlying genetic, environmental, or other underlying mechanisms that may play a role in causing these two related disorders.
MAKING A DIFFERENCE

The Bachmann-Strauss Dystonia & Parkinson Foundation was established in 1995 to find better treatments and cures for the movement disorders dystonia and Parkinson’s disease, and to provide medical and patient information. Key among its efforts, the Foundation funds scientific and clinical research and helps to raise awareness of Parkinson’s disease and dystonia among the general public and the medical community.

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