

Dystonia

Questions & Answers



THE BACHMANN-STRAUSS
Dystonia & Parkinson Foundation, Inc.

What is Dystonia?

Dystonia is a neurological muscle disorder that causes uncontrollable, painful spasms in one or more parts of the body. Different forms of the disease affect the face, neck, throat, eyelids, arms, legs or torso. The severity of the disease varies. Some people who have dystonia can maintain a relatively normal life style, while others often need full-time assistance.

Striking men, women and children across racial and ethnic boundaries, it is estimated that 500,000 people in the United States and Canada have dystonia.

WHAT ARE THE SYMPTOMS?

Sustained muscle spasms, twitching, abnormal and painful postures or poses, as well as frequent twisting or other kinds of repetitive movements are common symptoms. Symptoms usually begin in one limb, spreading over time to others and may eventually affect the torso, neck and, less commonly, facial muscles. Muscle contractions may affect any voluntary muscle and any region of the body.

WHAT CAUSES IT?

The exact cause of dystonia is not known and numerous research studies are underway to try to discover why it occurs. Genetics, other neurological diseases, trauma, side effects from medications and environmental factors are among the suspected contributing causes. Scientists recently discovered that most people with early onset dystonia have a particular mutated or changed gene named DYT1. Investigations to see how other genes may be involved are currently ongoing.

The *basal ganglia*, the part of the brain that controls movement, is believed to be involved in dystonia. In addition, an increase or decrease in dopamine – a substance that occurs naturally in the brain and that affects movement, emotions and response to pleasure – can bring on dystonic symptoms.

Dystonia is considered to be primary, secondary or tardive:

Primary dystonia (also known as *generalized* or *early onset* dystonia) occurs when an abnormal DYT1 gene is inherited and is the most common cause of early onset primary dystonia. The gene defect is related to an abnormal protein called torsinA. Although the gene and abnormal protein have been identified, further research is still needed.

We do know that dystonia caused by the DYT1 gene is inherited in a *dominant* manner, which means that only one parent needs to pass on the abnormal gene in order for a child to be affected. Only approximately 30% of people who inherit the DYT1 gene will develop dystonia. When dystonia is genetically caused, symptoms generally begin in childhood, although its onset in adulthood is possible.

Secondary dystonia (also known as *symptomatic dystonia*) may be caused by a trauma such as a severe blow to the head, or by a neurological disease such as stroke, multiple sclerosis, or Wilson's disease.

Tardive dystonia, a form of secondary dystonia, is most often a side effect of some medications, particularly those used to treat schizophrenia and psychosis. Called *antipsychotics* or *neuroleptics*, these drugs can interfere with dopamine. Drugs used to treat nausea or other gastrointestinal symptoms can also cause dystonia.

HOW IS IT TREATED?

Treatment depends on the region of the body affected and the severity of the symptoms.

Focal dystonia is most often treated with injections of botulinum toxin (BOTOX®), while more widespread dystonia may require oral medications combined with BOTOX®. BOTOX® is a toxin produced by the bacterium that causes botulism and was originally developed to treat dystonia. When minute doses are injected into an overactive muscle, it weakens the muscle for up to several months at a time, allowing for some relief.

Anticholinergic drugs, which block the communication of brain cells, are sometimes effective in treating dystonia. These drugs include Cogentin (benztropine) and Artane (trihexyphenidyl).

SURGERY

Some patients with severe, generalized dystonia have undergone brain surgery to alleviate their symptoms, resulting in a range of improvements. These surgeries have been widely used in treating Parkinson's disease and include pallidotomy, thalamotomy, and deep brain stimulation. Deep brain stimulation, approved in 2003 by the U.S. Food & Drug Administration (FDA) for the treatment of dystonia, is performed by implanting an electrode into a deep nucleus in the brain which delivers controlled electrical pulses to targeted areas of the brain to block the signals that cause the loss of motor control.

HOW IS DYSTONIA CLASSIFIED?

Dystonia is generally classified in three different ways, based on its cause, the age at which it occurs and the region of the body that is affected.

Age of onset

Early onset dystonia begins in childhood to late adolescence.

Adult onset dystonia most commonly begins after age 26.

Regions of the body

Dystonia is often classified by the body region that is affected.

Focal dystonia involves only one region of the body, such as the neck, eyelids or hand.

Segmental dystonia affects adjacent body regions, such as the neck with the arm.

Multifocal dystonia affects two or more regions of the body that are not next to each other, such as the eyelids and the hand.

Generalized dystonia usually begins as a focal dystonia and then spreads to affect many regions of the body.

HOW IS IT DIAGNOSED?

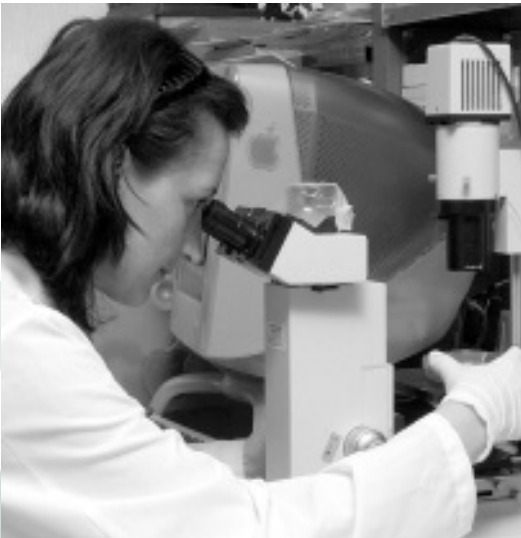
A neurologist, who is experienced in movement disorders, usually diagnoses dystonia. A physical and neurological examination, careful medical history and tests are generally taken to determine the form of the disease. A test that shows whether the DYT1 gene is present is now available.

When symptoms are mild, it is not uncommon for dystonia to be misdiagnosed or even undiagnosed. Many physicians are still unfamiliar with the disease and more must be done to better educate the medical community about the symptoms and treatment of this disease.

IS THERE A CURE?

Remarkable progress has occurred in the study of dystonia in the last few years, however right now there is no known cure. While dystonia affects more people than Muscular Dystrophy, Huntington's disease and Lou Gehrig's disease, awareness of dystonia remains limited both because it is under-researched and because more education about its diagnosis and treatment is needed.

The Bachmann-Strauss Dystonia & Parkinson Foundation is a leader in the field of dystonia research. It provides resources to educate the public and the medical community and funds research to uncover the causes, treatment and eventual cure of this debilitating disease.



WHERE DYSTONIA MAY OCCUR

TYPES OF FOCAL DYSTONIA

Excessive blinking (*Blepharospasm*) – affects muscles of the eyelids causing involuntary blinking and squeezing.

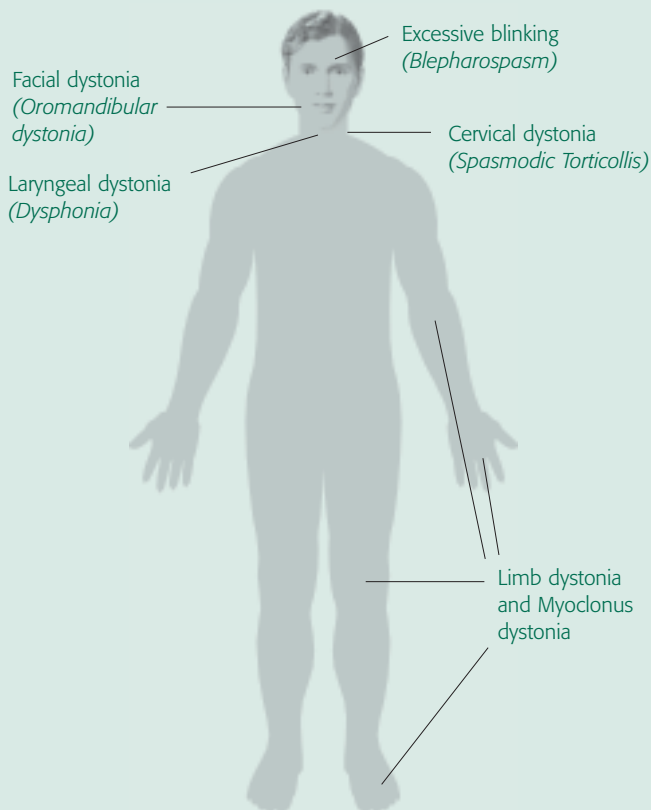
Cervical dystonia (*Spasmodic Torticollis*) – strikes neck muscles causing abnormal postures of the neck.

Facial dystonia (*Oromandibular dystonia*) – jaw muscles are affected causing involuntary opening, closing or deviation of the jaw.

Laryngeal dystonia (*Dysphonia*) – Strikes the muscles of the larynx or voice box and causes breathiness or vocal tightness, strain and strangulation.

Limb dystonia – Affects the legs, feet, arms or hands. This includes occupational dystonia such as writer's cramp.

Myoclonus dystonia – Affects the limbs and causes lightning-jerk movements and sustained postures.



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.

An independent, nonprofit, 501(c)3 organization, its funding is made possible through the generosity of individual and corporate contributors.



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