



# What is Dystonia?



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## What is Dystonia?

Dystonia is a complex disorder because of its causes, treatment, progression, and variability of symptoms.

Clinically, dystonia is a neurological disorder that is characterized by involuntary muscle contractions which force certain parts of the body into abnormal, sometimes painful, movements or positions.

Dystonia is the third most common movement disorder after Parkinson's disease and Tremor, affecting an estimated 300,000 persons in North America who suffer from some form of dystonia. Dystonia is often misunderstood by the public and misdiagnosed by medical doctors. It is not a psychological disorder nor does it affect intellect. Dystonia is not fatal, but, depending on the form, it can be debilitating.

Dystonia is not a single disease, but a syndrome: a set of symptoms that cannot be attributed to a single cause. Thus, both genetic and non-genetic events must be accounted for before we finally have a full understanding of the common elements – namely, the twisting, repetitive movements around an axis, such as an arm or the neck. Dystonia may occur in a generalized or focal form. It may result from hereditary condition or as a result of brain injury.

### IDENTIFIED INHERITED DYSTONIAS

**Early-Onset Childhood Dystonia** (Generalized Dystonia or Idiopathic Torsion Dystonia, ITD) usually starts in childhood or adolescence. Symptoms typically start in one part of the body, usually in an arm or leg and can eventually spread to the rest of the body, causing it to twist into unnatural positions. It is the most common hereditary form of dystonia, resulting, in most cases, from the *DYT1* gene.

**Dopa-Responsive Dystonia (DRD)** usually starts in childhood or adolescence with progressive difficulty in walking. It may be misdiagnosed since it mimics many of the symptoms of cerebral palsy or even parkinsonism. The affected gene, located on chromosome 14, is the gene for GTP cyclohydrolase, an enzyme that helps in the synthesis of dopamine. **DRD** symptoms are relieved with very low doses of levodopa.

**Paroxysmal Dystonia** refers to relatively brief attacks of dystonic movements and postures with a return to normal posture between episodes.

Paroxysmal dystonias take two main forms. Paroxysmal kinesiogenic dystonia refers to brief attacks triggered by sudden movement, occurring frequently many times a day. Paroxysmal dystonic choreoathetosis refers to attacks that may last for several hours and may occur three to four times a day. Thus far, one gene maps to chromosome 2q.

**X-linked Dystonia-Parkinsonism** (Lubag) is a form of dystonia found almost entirely among men from the Philippine Island of Panay. Females are believed to be the carriers of the gene, mapped to chromosome Xq13. It usually begins focally, often generalized, and can be replaced by parkinsonian features.

### FOCAL DYSTONIAS

**Blepharospasm** affects the muscles of the eyelids forcing them to close. The spasms may become sufficiently frequent to render the patient unable to see, although the eyes and vision are normal.

**Cervical Dystonia** (Spasmodic Torticollis) affects muscles in the neck and shoulders. The muscle spasms can be painful and cause the neck to twist to one side (torticollis), forward (*antecollis*), or backward (*retrocollis*). The neck may pull, turn, or jerk; it may be held persistently in one direction.

**Oromandibular Dystonia** is sometimes called Meige's syndrome. The muscles of the lower face irregularly pull or contract. Sometimes the jaw muscles that pull the mouth open or closed are involuntary. Usually blepharospasm is also present.

**Spasmodic Dysphonia** involves the muscles inside the larynx or voice box. In spasmodic dysphonia of the adductor muscles, the vocal cords are drawn tightly together, particularly when the person tries to talk. The voice typically has a strained, hoarse, choked quality. In spasmodic dysphonia, of the less common abductor type, the vocal cords are pulled apart and the voice has a low, breathy whispered quality; sometimes the person cannot speak at all.

**Writer's Cramp** is an occupational dystonia in which the hand and forearm muscles contract during the act of writing. The hand may be drawn up so tightly it cannot move. As soon as the writing instrument is removed from the hand, the hand relaxes.

A similar cramp may arise in a musician as the violin is bowed or certain fingers are moved in playing a flute or other instrument. Occupational cramps may occur in a wide variety of situations involving repetitive movements.

**Hemifacial spasm** is not strictly speaking a form of dystonia. In this disorder, the muscles on one side of the face irregularly contract. Sometimes this is secondary to inflammation or irritation of the facial nerve.

## SECONDARY DYSTONIA

**Secondary Dystonia** results from environmental or disease-related damage to the basal ganglia. Birth injury (particularly due to lack of oxygen), certain infections, reactions to certain drugs, trauma, or stroke can cause dystonic symptoms. Dystonia can also be secondary to other illnesses affecting the nervous system, including Wilson's disease.

### **What Goes Wrong in Dystonia?**

Researchers believe that some forms of dystonia may be caused by breakdown of the dopamine system in the basal ganglia, a collection of structures in the brain that control movement. Dopamine is a neurotransmitter that regulates neuronal communication within the basal ganglia.

A malfunctioning dopamine system in the basal ganglia is responsible for many movement disorders including Parkinson's disease; but in contrast to Parkinson's disease, in dystonia there is no visible evidence of damage to the brain.

When dystonia is secondary to certain injuries or small strokes, we often find lesions (areas of damage) in the putamen, one nucleus in the basal ganglia, as well as in certain nearby structures.

Even though we can see no microscopic abnormalities of the brain in the great majority of cases of dystonia, including those with generalized dystonia, the evidence is so clear in the secondary dystonias that we believe the same part of the brain is involved in all types.

### **Can One Predict the Course of Dystonia?**

Dystonia has a variable nature, therefore making it difficult to predict the prognosis of the disorder. As a general rule, the older one is when dystonia

develops, the more likely it plateaus and remains limited, or focal, to one part of the body.

The younger one is when dystonia develops, the more likely that it will progress over time, particularly if the dystonia begins in a leg.

## **Understanding the Genetics of Dystonia**

Great advancements have been made in the area of dystonia genetics, including the identification of the **DYT1** gene and Torsin A protein responsible for early-onset childhood dystonia.

Exactly how the abnormal gene causes the dystonia is presently unknown. Early-onset dystonia appears when a person has one copy of the mutated gene and one copy of the normal gene. This means that the disease is dominant, because only one copy of the mutated gene is needed to cause it, but fewer than half (30%) of the people who have the mutated gene will develop symptoms; therefore, 70% of the people who carry the gene will not develop symptoms. Geneticists call this phenomenon "**penetrance.**"

Another aspect to the inheritability is that the severity of the illness may differ markedly within a family. For example, the affected mother may have mild dystonia; one of her children may have severe generalized dystonia; while another may have a mild focal dystonia. And in the same family there may be still another child who is actually carrying the gene, has no symptoms at all, but who can pass the gene on to his/her children.

Each individual gene is responsible for production of a particular protein within the body. Even the alteration or omission of just one gene – and consequently of its associated product – results in a clinical disorder.

Scientists believe there must be other factors, environmental stresses or interacting genes, that influence the expression of the mutated gene and cause the disorder.

The discovery of the **DYT1** gene opens research to learn how the disease occurs and what treatments may then be possible. It also allows for better diagnostic testing, testing for confirmation of the diagnosis, and prenatal testing.

Studying the **DYT1** gene along with the other genes already identified may shed light on the mechanisms of the disease process itself or reveal a pattern which produces certain proteins.

This research has potential to help all. All dystonias have similar symptoms which involve the same area of the brain and the similar neurotransmitters. Until now, we have directed treatments to the effects of dystonia. Now we can attempt to direct treatment efforts to the causes of dystonia.

Non-genetic or secondary forms of dystonia may be caused by a brain injury at birth or later in life or by the use of certain drugs or tranquilizers.

## **The Diagnosis of Dystonia**

Currently, there is no specific laboratory test or x-ray that says whether a person has dystonia. Instead, the diagnosis of dystonia rests solely upon the neurological examination. Therefore, in order to correctly diagnose dystonia, doctors must be able to recognize the physical signs and be familiar with the symptoms, particularly because when dystonia begins, often, its symptoms may change significantly with different actions.

## **Current Forms of Treatment**

Unfortunately at this time, there is no cure for dystonia, but treatments are available. Treatment is designed to help the symptoms of spasms, pain, and disturbed postures and functions. Most therapies are symptomatic, attempting to cover up or release the dystonic spasms; therefore there is no single treatment program appropriate for every case.

The goal of any treatment is to achieve the greatest benefits while incurring the fewest risks. Establishing a satisfactory treatment scheme requires patience on the part of both the physician and the patient.

### DRUG THERAPY

Some of the medicines the doctor might consider include: Artane (trihexyphenidyl), Cogentin (benztropine), Valium (diazepam), **Klonopin** (clonazepam), Lioresal (baclofen), Tegretol (carbamazepine), Sinemet or Madopar (levodopa), Parlodel (bromocriptine), Symmetrel (amantadine). Additionally, newer drugs are currently being tested at different movement disorder centers.

## **BOTULINUM TOXIN INJECTIONS**

Botulinum Toxin A (**Botox®**) injections into selected muscles are helpful in treating many dystonic spasms, especially in focal dystonias.

Botulinum toxin, a complex protein produced by the bacterium *Clostridium Botulinum*, weakens a muscle sufficiently to reduce a spasm but not enough to cause paralysis. Botox® is a nerve "blocker," binding to nerve endings and preventing the release of chemicals (neurotransmitters) that activate muscles. Neurotransmitters carry the "message" from the brain that causes a muscle to contract. If the message is blocked, muscle spasms are reduced or eliminated.

Botox® injections are done with a small needle. There is usually only temporary discomfort at the site of injections. Effects are not usually noticed for 5 to 10 days and benefits last three to four months with minimal side effects until injections are repeated.

## **SURGERY**

Surgery may be considered when patients are no longer receptive to other treatments. It should be noted, surgery may lose its effect over the years, but it can possibly provide some relief.

Surgery is undertaken to interrupt, at various levels of the nervous system, the pathways responsible for the abnormal movements. Some operations intentionally damage small regions of the thalamus (thalamotomy), globus pallidus (pallidotomy), or other deep centers in the brain. Recently, chronic deep brain stimulation (DBS) has been tried with some success. Other surgical approaches include cutting nerves going 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).

There are a number of factors that may influence the success of the operation. Each patient is unique, and the muscles involved vary from one patient to another. It is for this reason that preoperative evaluation by a movement disorders expert is essential.

New modes of treatments are currently being researched including other forms of injectable medications and forms of surgery.

#### **COMPLEMENTARY HEALTHCARE**

Although stress clearly does not cause dystonia, many people have reported that symptoms worsen in stressful situations. Unfortunately, it is not possible to get rid of all stress, but a course of stress management or relaxation techniques are beneficial.

Sensory tricks, such as touching the chin, if one has cervical dystonia, can be effective in reducing the symptoms.

Some people with dystonia have also found benefit from complementary healthcare such as physical therapy, acupuncture, and massage therapy.

#### **Supportive Forms of Treatment**

By educating yourself with information, you have taken the first step in dealing with dystonia. Reassurance from family, friends, and others who have dystonia is beneficial. Support groups offer encouragement, camaraderie, and information about new treatments and medical advances. The Dystonia Medical Research Foundation maintains a network of support groups throughout North America.

#### **What is Being Done about Dystonia?**

Though the exact cause of dystonia is unknown, continued research offers hope that answers will be found. There is a great deal of current research, much of it being funded by the Dystonia Medical Research Foundation, with additional research funded by the National Institutes of Health.

Since its establishment in 1976, the Dystonia Medical Research Foundation has funded over \$15 million dollars in research, established support groups for people with dystonia **and** their families, and continues to battle misdiagnosis through expanding educational and awareness programs.

*This brochure on dystonia has been updated from the original material written by Charles H. Markham, MD., the Scientific Director of Foundation from 1985-1994 and from medical presentations at the Biennial Patient Symposiums. It was reviewed and updated by the current Scientific Director Mahlon R. DeLong, M.D.*