Dystonia is a disorder of muscle control; it can cause slow repetitive movements, abnormal postures and/or tremors of the musculature that are uncontrollable by the patient.

The symptoms of dystonia may include foot cramps, turning or dragging of the foot, worsening of handwriting, neck movements, rapid eye blinking or closing, various muscle tremors and sometimes difficulty with speaking; the symptoms may become more noticeable when the individual is tired or under stress.

The cause of dystonia is not known; researchers suspect that damage to the brain's basal ganglia or other regions that control movement and/or the brain's neurotransmitters; researchers divide dystonia into three groups - idiopathic, genetic and acquired.

Symptoms of dystonia can occur at any age but are often described as either childhood or early onset versus adult-onset with early onset beginning with symptoms in the limbs while adult onset usually begins in the neck and/or face; these symptoms often progress from intermittent to more frequent or fixed postures that can result in shortening of tendons.

Dystonia may be classified based on the regions of the body which are affected; in addition, they can be described as focal (localized to a specific area such as the neck) or as genetic forms that may start as focal then progress to other areas.

Currently, there are no medications to prevent or slow the progression of any dystonia; however, there are several drugs that can reduce the symptoms of dystonia (botulinum toxin, anticholinergic agents, benzodiazepines and dopaminergic agents) and other methods such as deep brain stimulation and/or physical therapy.

Researchers are still investigating what causes dystonia and are exploring ways to better treat dystonia with medications and/or surgery; genetic studies are ongoing to help understand the cause and provide ways to better treat genetically caused dystonia.

What is dystonia?
Dystonia is a disorder characterized by involuntary muscle contractions that cause slow repetitive movements or abnormal postures. The movements may be painful, and some individuals with dystonia may have a tremor or other neurologic features. There are several different forms of dystonia that may affect only one muscle, groups of muscles, or muscles throughout the body. Some forms of dystonia are genetic but the cause for the majority of cases is not known.

What are the symptoms of dystonia?

Dystonia can affect many different parts of the body, and the symptoms are different depending upon the form of dystonia. Early symptoms may include a foot cramp or a tendency for one foot to turn or drag -- either sporadically or after running or walking some distance -- or a worsening in handwriting after writing several lines. In other instances, the neck may turn or pull involuntarily, especially when the person is tired or under stress. Sometimes both eyes might blink rapidly and uncontrollably; other times, spasms will cause the eyes to close. Symptoms may also include tremor or difficulties speaking. In some cases, dystonia can affect only one specific action, while allowing others to occur unimpeded. For example, a musician may have dystonia when using her hand to play an instrument, but not when using the same hand to type. The initial symptoms can be very mild and may be noticeable only after prolonged exertion, stress, or fatigue. Over a period of time, the symptoms may become more noticeable or widespread; sometimes, however, there is little or no progression. Dystonia typically is not associated with problems thinking or understanding, but depression and anxiety may be present.

What do researchers know about dystonia?

The cause of dystonia is not known. Researchers believe that dystonia results from an abnormality in or damage to the basal ganglia or other brain regions that control movement. There may be abnormalities in the brain's ability to process a group of chemicals called neurotransmitters that help cells in the brain communicate with each other. There also may be abnormalities in the way the brain processes information and generates commands to move. In most cases, no abnormalities are visible using magnetic resonance imaging or other diagnostic imaging.

The dystonias can be divided into three groups: idiopathic, genetic, and acquired.

- **Idiopathic dystonia** refers to dystonia that does not have a clear cause. Many instances of dystonia are idiopathic.
- There are several genetic causes of dystonia. Some forms appear to be inherited in a dominant manner, which means only one parent who carries the defective gene is needed to pass the disorder to their child. Each child of a parent having the abnormal gene will have a 50 percent chance of carrying the defective gene. It is important to note the symptoms may vary. 
widely in type and severity even among members of the same family. In some instances, persons who inherit the defective gene may not develop dystonia. Having one mutated gene appears to be sufficient to cause the chemical imbalances that may lead to dystonia, but other genetic or even environmental factors may play a role. Knowing the pattern of inheritance can help families understand the risk of passing dystonia along to future generations.

- Acquired dystonia, also called secondary dystonia, results from environmental or other damage to the brain, or from exposure to certain types of medications. Some causes of acquired dystonia include birth injury (including hypoxia, a lack of oxygen to the brain, and neonatal brain hemorrhage), certain infections, reactions to certain drugs, heavy metal or carbon monoxide poisoning, trauma, or stroke. Dystonia can be a symptom of other diseases, some of which may be hereditary. Acquired dystonia often plateaus and does not spread to other parts of the body. Dystonia that occurs as a result of medications often ceases if the medications are stopped quickly.

**When do symptoms of dystonia occur?**

Dystonia can occur at any age, but is often described as either early, or childhood, onset versus adult onset.

Early-onset dystonia often begins with symptoms in the limbs and may progress to involve other regions. Some symptoms tend to occur after periods of exertion and/or fluctuate over the course of the day.

Adult-onset dystonia usually is located in one or adjacent parts of the body, most often involving the neck and/or facial muscles. Acquired dystonia can affect other regions of the body.

Dystonias often progress through various stages. Initially, dystonic movements may be intermittent and appear only during voluntary movements or stress. Later, individuals may show dystonic postures and movements while walking and ultimately even while they are relaxed. Dystonia can be associated with fixed postures and shortening of tendons.

**How are the dystonias classified?**

One way to classify the dystonias is based upon the regions of the body which they affect:

- Ralized dystonia affects most or all of the body.
- Focal dystonia is localized to a specific part of the body.
- Multifocal dystonia involves two or more unrelated body parts.
- Segmental dystonia affects two or more adjacent parts of the body.
- Hemidystonia involves the arm and leg on the same side of the body.

There are several different forms of dystonia. Some of the more common focal forms are:

Cervical dystonia, also called spasmodic torticollis or torticollis, is the most common of the focal dystonias. In cervical dystonia, the muscles in the neck that control the position of the head are affected, causing the head to turn to one side or be pulled forward or backward. Sometimes the shoulder is pulled up. Cervical dystonia can occur at any age, although most individuals first experience symptoms in middle age. It often begins slowly and usually reaches a plateau over a few months or years. About 10 percent of those with torticollis may experience a spontaneous remission, but unfortunately the remission may not be lasting.

Blepharospasm, the second most common focal dystonia, is the involuntary, forcible contraction of the muscles controlling eye blinks. The first symptoms may be increased blinking, and usually both eyes are affected. Spasms may cause the eyelids to close completely, causing "functional blindness" even though the eyes are healthy and vision is normal.

Cranio-facial dystonia is a term used to describe dystonia that affects the muscles of the head, face, and neck (such as blepharospasm). The term Meige syndrome is sometimes applied to cranio-facial dystonia accompanied by blepharospasm. Oromandibular dystonia affects the muscles of the jaw, lips, and tongue. This dystonia may cause difficulties with opening and closing the jaw, and speech and swallowing can be affected. Spasmodic dysphonia, also called laryngeal dystonia, involves the muscles that control the vocal cords, resulting in strained or breathy speech.

Task-specific dystonias are focal dystonias that tend to occur only when undertaking a particular repetitive activity. Examples include writer's cramp that affects the muscles of the hand and sometimes the forearm, and only occurs during handwriting. Similar focal dystonias have also been called typist's cramp, pianist's cramp, and musician's cramp. Musician's dystonia is a term used to classify focal dystonias affecting musicians, specifically their ability to play an instrument or to perform. It can involve the hand in keyboard or string players, the mouth and lips in wind players, or the voice in singers.

In addition, there are forms of dystonia that may have a genetic cause:

- DYT1 dystonia is a rare form of dominantly inherited generalized dystonia that can be caused by a mutation in the DYT1 gene. This form of dystonia typically begins in childhood, affects the limbs first, and progresses, often causing significant disability. Because the gene's effects are so variable, some people who carry a mutation in the DYT1 gene may not develop dystonia.
Dopa-responsive dystonia (DRD), also known as Segawa's disease, is another form of dystonia that can have a genetic cause. Individuals with DRD typically experience onset during childhood and have progressive difficulty with walking. Symptoms characteristically fluctuate and are worse late in the day and after exercise. Some forms of DRD are due to mutations in the DYT5 gene. Patients with this disorder have dramatic improvements in symptoms after treatment with levodopa, a medication commonly used to treat Parkinson's disease.

Recently, researchers have identified another genetic cause of dystonia which is due to mutations in the DYT6 gene. Dystonia caused by DYT6 mutations often presents as cranio-facial dystonia, cervical dystonia, or arm dystonia. Rarely, a leg is affected at the onset.

Many other genes that cause dystonic syndromes have been found, and numerous genetic variants are known to date. Some important genetic causes of dystonia include mutations in the following genes: DYT3, which causes dystonia associated with parkinsonism; DYT5 (GTP cyclohydrolase 1), which is associated with dopa-responsive dystonia (Segawa disease); DYT6 (THAP1), associated with several clinical presentations of dystonia; DYT11, which causes dystonia associated with myoclonus (brief contractions of muscles); and DYT12, which causes rapid onset dystonia associated with parkinsonism.

What treatments are available for dystonias?

Currently, there are no medications to prevent dystonia or slow its progression. There are, however, several treatment options that can ease some of the symptoms of dystonia, so physicians can select a therapeutic approach based on each individual's symptoms.

- Botulinum toxin. Botulinum injections often are the most effective treatment for the focal dystonias. Injections of small amounts of this chemical into affected muscles prevents muscle contractions and can provide temporary improvement in the abnormal postures and movements that characterize dystonia. First used to treat blepharospasm, such injections are now widely used for treating other focal dystonias. The toxin decreases muscle spasms by blocking release of the neurotransmitter acetylcholine, which normally causes muscles to contract. The effect typically is seen a few days after the injections and can last for several months before the injections must be repeated. The details of the treatment will vary among individuals.

- Medications. Several classes of drugs that affect different neurotransmitters may be effective for various forms of dystonia. These medications are used "off-label", meaning they are approved by the U.S. Food and Drug Administration to treat different disorders or conditions but have not been specifically approved to treat dystonia. The response to drugs varies among individuals and even in the same person over time. These drugs include:
Anticholinergic agents block the effects of the neurotransmitter acetylcholine. Drugs in this group include trihexyphenidyl and benztropine. Sometimes these medications can be sedating or cause difficulties with memory, especially at higher dosages and in older individuals. These side effects can limit their usefulness. Other side effects such as dry mouth and constipation can usually be managed with dietary changes or other medications.

GABAergic agents are drugs that regulate the neurotransmitter GABA. These medications include the benzodiazepines such as diazepam, lorazepam, clonazepam, and baclofen. Drowsiness is their common side effect.

Dopaminergic agents act on the dopamine system and the neurotransmitter dopamine, which helps control muscle movement. Some individuals may benefit from drugs that block the effects of dopamine, such as tetrabenazine. Side effects (such as weight gain and involuntary and repetitive muscle movements) can restrict the use of these medications. Dopa-responsive dystonia (DRD) is a specific form of dystonia that most commonly affects children, and often can be well managed with levodopa.

- Deep brain stimulation (DBS) may be recommended for some individuals with dystonia, especially when medications do not sufficiently alleviate symptoms or the side effects are too severe. DBS involves implanting small electrodes that are connected to a pulse generator into specific brain regions that control movement. Controlled amounts of electricity are sent into the exact region of the brain that generates the dystonic symptoms and interfere with and block the electrical signals that cause the symptoms. DBS should be conducted by an interdisciplinary team involving neurologists, neurosurgeons, psychiatrists, and neuropsychologists, as there is intensive follow-up and adjustments to optimize an individual's DBS settings.
- Physical and other therapies may be helpful for individuals with dystonia and may be an adjunct to other therapeutic approaches. Speech therapy and/or voice therapy can be quite helpful for some affected by spasmodic dysphonia. Physical therapy, the use of splints, stress management, and biofeedback also may help individuals with certain forms of dystonia.

What research is being done on dystonias?

The ultimate goals of research are to find the cause(s) of the dystonias so that they can be prevented, and to find ways to cure or more effectively treat people who are affected. The National Institute of Neurological Disorders and Stroke (NINDS), a part of the National Institutes of Health (NIH), is the Federal agency with primary responsibility for brain and neuromuscular research. NINDS sponsors research on dystonia both in its facilities at the NIH and through grants to medical centers and institutions throughout the country. Scientists at other NIH institutes also conduct research that may benefit individuals with dystonia.
Scientists at the National Institute on Deafness and Other Communication Disorders (NIDCD) are studying improved treatments for speech and voice disorders associated with dystonia. The National Eye Institute (NEI) supports work on the study of blepharospasm and related problems, and the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD) supports work on dystonia, including the rehabilitation for the disorder.

Scientists at NINDS laboratories have conducted detailed investigations of the patterns of muscle activity, imaging studies of brain activity, and physiological studies of the brain in persons with dystonia.

Treatment studies, using surgery or medication, are being conducted in many centers, including the NIH. To learn more about clinical studies on dystonia, please go to www.clinicaltrials.gov.

Recently, the Dystonia Coalition—a clinical research network for dystonia—has been established with support from the NINDS and the NIH Office of Rare Disease Research as part of the Rare Disease Clinical Research Network. For more information on the clinical studies and patient registry established by the Dystonia Coalition, see http://rarediseasesnetwork.epi.usf.edu/dystonia/.

The search for genes responsible for some forms of dystonia continues. In 1989 a team of researchers mapped a gene for early-onset torsion dystonia to chromosome 9; the gene was subsequently named DYT1. In 1997 the team sequenced the DYT1 gene and found that it codes for a previously unknown protein now called "torsin A." The discovery of the DYT1 gene and the torsin A protein provide the opportunity for prenatal testing, allow doctors to make a specific diagnosis in some cases of dystonia, and permit the investigation of molecular and cellular mechanisms that lead to disease.

The discovery of the mutation in "torsin A" has enabled scientists to study animal models into which the mutated gene has been introduced. Through research with patients informed by the latest discoveries from genetics and basic neuroscience, scientists and doctors hope to better understand dystonia and find more effective treatments.

Where can I get more information about dystonias?

For more information on neurological disorders or research programs funded by the National Institute of Neurological Disorders and Stroke, contact the Institute’s Brain Resources and Information Network (BRAIN) at:

BRAIN
P.O. Box 5801
Bethesda, MD 20824
(800) 352-9424
Information also is available from the following organizations:

Dystonia Medical Research Foundation
1 East Wacker Drive
Suite 2810
Chicago, IL 60601-1905
dystonia@dystonia-foundation.org
http://www.dystonia-foundation.org
Tel: 312-755-0198
Fax: 312-803-0138

National Spasmodic Torticollis Association
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http://www.torticollis.org
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American Speech-Language-Hearing Association (ASHA)
2200 Research Boulevard
Rockville, MD 20850
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Fred French Building 551 Fifth Avenue, at 45th Street
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Spasmodic Torticollis Dystonia/ST Dystonia
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