DYSTONIA

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Dystonia is a disorder of muscle tone. It causes involuntary muscle contractions, which force affected parts of the body into abnormal, repetitive movements or postures. Dystonia may affect any part of the body, including the limbs, trunk, neck, face, eyelids or vocal cords.

There are many different forms of dystonia. In general, dystonia is a chronic disorder but it is not life-threatening. Some forms of dystonia are painful. The prognosis, or long-term course, is difficult to predict. Individuals with some forms of dystonia may have muscle contractions that are severe and progressive. These individuals may be unable to perform normal motor function and become disabled. Others have milder forms of dystonia, which do not affect their ability to perform their usual activities of daily living. Their symptoms may only be noticeable after exertion, stress, or fatigue. Often, the symptoms stabilize and do not progress. Dystonia does not affect emotional health and intelligence.

Dystonia is the third most common movement disorder after Parkinson's disease and tremor. It affects approximately 30 people out of every 100,000 worldwide.

Researchers believe that dystonia is caused by an abnormality in a part of the brain called the basal ganglia, which processes messages that result in movement. Neurotransmitters are chemicals in the brain which transmit messages. Some neurotransmitters, including dopamine, are inhibitory and help the brain to control muscle movement. Other neurotransmitters are excitatory and produce movement. Researchers believe that an imbalance between these types of neurotransmitters in the basal ganglion leads to dystonia.

Dystonia has been classified according to its cause. In primary or idiopathic dystonia, which accounts for about half of all cases, there is no connection to any other disease or injury. Many of the primary dystonias are inherited by an autosomal dominant gene. Only one parent needs to carry the gene to potentially pass the disorder on to a child. However, only 30% to 40% of people with the mutated gene actually develop dystonia. This is referred to as variable penetrance. Other factors, such as the environment or interacting genes, influence whether or not the gene will express itself and cause symptoms. Individuals who have the gene but who do not develop the disorder are called carriers. Each child of a carrier has a 50% chance of inheriting the gene and becoming a carrier or having the disorder. Symptoms of the disorder may vary widely among family members with the disorder.

Secondary or acquired dystonia is caused by environmental factors or diseases which cause damage to the basal ganglia. These include birth injury, certain infections, or stroke. Reactions to certain drugs and heavy metal or carbon monoxide poisoning can also cause dystonia.

Another method used to classify dystonia is based on the part or parts of the body that are affected. In generalized dystonia, most or all of the body is involved. Focal dystonia is localized to a specific body part, such as an arm or the eyelids. Multifocal dystonia affects two or more unrelated body parts. This system further classifies the dystonias according to the specific muscles that are affected.

GENERALIZED DYSTONIAS

Early onset generalized dystonia is the most common hereditary form of dystonia. It usually begins between ages 7 and 10. Initially, the individual may have twisting movements of one limb. These movements may progress to involve other limbs or parts of the body. The progression varies depending on the age of onset and body part involved. Many children are disabled from this type of dystonia and are unable to perform simple motor tasks.

Dopa-responsive dystonia is a rare hereditary form of dystonia that usually begins in early childhood. The symptoms begin with difficulty walking and are usually worse later in the day and after exercise. It may be disabling. Dopa-responsive dystonia has been treated effectively with levodopa, a medication that is also used to treat Parkinson's disease.

FOCAL DYSTONIAS

Spasmodic torticollis is the most common of the focal dystonias. The neck muscles are affected and cause the head to twist and turn to one side. The head may also be pulled forward or backward. It usually begins around middle age. Some people may have a spontaneous remission. Unfortunately, it is usually short-lived.

Blepharospasm causes excessive, involuntary blinking. Usually both eyes are involved. At times, the eyelids may stay closed for several hours. During this time, the person is functionally blind, although vision is not affected. Individuals with blepharospasm may have increased blinking with bright lights, fatigue, and emotional stress.

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Oromandibular dystonia affects the jaw, lips and tongue. The jaw is pulled open or shut. Speech, chewing, and swallowing may be affected while the muscles are in spasm.

Spastic dysphonia only affects the muscles of speech. It results in a strained, breathy speech.

Writer's cramp affects the muscles of the hand and forearm. It only occurs during writing.

MULTIFOCAL DYSTONIAS

Meige's syndrome is a combination of blepharospasm and oromandibular dystonia.

DIAGNOSIS

The diagnosis of dystonia can be difficult because of the varying nature of the disorder. A complete medical history and neurologic examination are essential. The neurologist will ask detailed questions about the dystonia, including when it started, how it progressed, and conditions that make the symptoms better or worse. An important part of the history is a list of past and present medications. A family history will also be taken. Tests are ordered to determine wheteher there is an underlying injury or environmental factor that may be causing the dystonia. If the tests are normal, a diagnosis of idiopathic dystonia is made.

Genetic testing is helpful in the diagnosis of early onset dystonias. In 1997, a direct test for the genetic mutation was developed. It detects damage to the gene itself. It can also determine whether an individual is a carrier of the mutated gene. This information may be important for prospective parents. Genetic testing is performed on a blood sample. An important component of genetic testing is genetic counseling, which helps individuals understand the test results and their implications. Many testing sites will not perform genetic testing without genetic counseling.

TREATMENT

Treatment depends on the type of dystonia. Acquired dystonia may be helped by treating the underlying cause. Usually, a variety of treatments are used to help reduce or eliminate the muscle contractions, relieve pain and assist the individual to regain or maintain activities of daily living. Treatments are not yet available to cure dystonia or prevent its progression.

One approach to therapy is the use of medication. A combination of different types of medications may produce the best results with the least amount of side effects. The effectiveness of medications varies among individuals or in the same individual over time.

Medications that act on the neurotransmitters in the brain have been helpful. Some medications, including trihexyphenidyl, reduce the action of the excitatory neurotransmitters that produce movement. Others increase the action of the inhibitory neurotransmitters and help the brain to control movement. These include levodopa/carbidopa, bromocriptine, and the muscle relaxants diazepam and baclofen. Some medications that are typically used to control seizures can also help control the symptoms of dystonia.

Botulinum toxin has been used to treat many of the focal dystonias. Small amounts are injected into the affected muscle to provide temporary relief. The effect usually lasts for several months before the injections need to be repeated.

Physical therapy may help in treating the symptoms of dystonia. Stretching exercises may help the individual to maintain or regain range of motion in affected joints. Speech therapy often helps improve speech or swallowing. Some individuals have been helped with stress management, relaxation techniques, and biofeedback.

Some individual are able to obtain temporary relief with sensory "tricks," such as touching the body part involved or lifting a leg while standing. Different sensory tricks work for different people. The reason why sensory tricks work is not clear.

Surgery may be recommended for individuals whose symptoms have not been relieved by other means or who develop unacceptable side effects from the medications. In advanced generalized dystonia, surgical destruction of parts of the thalamus in the brain may help to control movement. This procedure can be risky, as important brain structures are located near the thalamus.

The focal dystonias may be helped by surgical cutting of the nerves to the muscles affected. This type of surgery can also be risky, as the results can be unpredictable and loss of muscle movement can lead to disfigurement.

Support groups can be very helpful for people with dystonia and their families. These groups provide information and group discussion and support. Just knowing that you are not alone can be invaluable in dealing with any chronic disorder.

RESEARCH

Researchers are studying dystonia to help identify factors that might contribute to the disorder. They are also studying the pathology of the disorder to understand exactly what is happening in the brain of an individual with dystonia. This research provides hope that new discoveries could help improve the diagnosis and treatment of all of the dystonias.

For additional information about dystonia, contact The National Institute of Neurologic Disorders and Stroke, PO Box 5801, Bethesda, MD 20824, 1–800-352–9424,

www.ninds.nih.gov.