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AMERICAN ACADEMY OF NEUROLOGY®

Dystonia: The Basics

WHAT IS DYSTONIA?

Dystonia is a movement disorder in which involuntary muscle contractions cause repetitive, twisting movements or abnormal postures. It may affect a single muscle, a group of muscles (such as the muscles in the limbs or neck), or the entire body and can cause varying degrees of disability and pain. Voluntary movements may trigger or worsen the condition.

WHAT ARE THE SYMPTOMS?

Dystonic movements and postures can either be confined to a single area—such as the hands, feet, or neck—or may spread to other parts of the body. Initial symptoms can be very mild and may appear only during periods of prolonged exertion or stress, or during a specific activity. If the disease progresses, patients may exhibit dystonic movements and postures while walking, or even while at rest. Sensory tricks, in which certain gestures and postures reduce dystonia symptoms, may be present in some forms of the condtion. Some forms of sustained dystonia may cause tendons to shorten, leading to permanent physical deformities in areas such as the neck, trunk, or limbs.

WHAT CAUSES IT?

Doctors don't know what causes most cases of dystonia. Researchers believe that abnormalities in the basal ganglia, an area of the brain that coordinates movement, may lead to the condition. It can be inherited from a parent who passes on one of several defective genes associated with the condition; or it can be acquired as a reaction to drugs, from a traumatic brain injury, or as a symptom of another disorder such as Parkinson's disease, Huntington's disease, Wilson's disease, encephalitis, or brain tumor.

HOW IS IT DIAGNOSED?

There is no single diagnostic test for dystonia. Instead, doctors rely on physical signs and symptoms, a thorough patient medical history, and a neurologic examination. They may order blood and urine tests, a magnetic resonance imaging (MRI) or computed tomography (CT) scan, and analysis of cerebrospinal fluid to rule out other conditions or disorders. They may also recommend genetic testing for specific forms of dystonia.

HOW IS IT TREATED?

For most cases of dystonia there is no cure, and no one treatment is universally effective. However, physicians may recommend a variety of therapies to manage symptoms. For example, muscle relaxants and anti-spasmodic, anti-epilepsy, anticholinergic, or anti-anxiety medication or antidepressants can help reduce or eliminate muscle spasms and pain. Doctors may also prescribe regular injections of botulinum toxin (for example, Botox, Dysport, Myobloc, or Xeomin), which temporarily weakens the affected muscles, reducing their activity.

If none of these treatments is effective, a procedure called deep brain stimulation, in which surgeons implant electrodes into the brain to block the signals that cause the condition, may be an option. Physical therapy, occupational therapy, and speech therapy may offer some relief for patients.

WHAT RESEARCH IS BEING DONE?

Organizations such as the National Institute of Neurological Disorders and Stroke (**ninds.nih.gov**), the Dystonia Medical Research Foundation, and the Dystonia Coalition (**rarediseasesnetwork.org/dystonia**) support research to uncover the mechanisms in the nervous system that lead to symptoms, create models of dystonia to use in experiments, and discover targeted therapies.

For more *Brain & Life* articles on dystonia, go to **BrainLifeMag.org/Dystonia**.

For more resources and support, contact:

- American Speech-Language-Hearing Association (ASHA): **asha.org**; **800-638-8255**
- The Bachmann-Strauss Dystonia & Parkinson Foundation: **dystonia-parkinsons.org**; **212-509-0995, ext 204**
- Benign Essential Blepharospasm Research Foundation: **blepharospasm.org**; **409-832-0788**
- Dystonia Medical Research Foundation: **dystonia-foundation.org**; **800-377-DYST (3978)**
- National Spasmodic Torticollis Association: **torticollis.org**; **800-487-8385**
- ST/Dystonia, Inc: **spasmodictorticollis.org**; **888-445-4588**

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