

## **Life in Motion Dystonia Fact Sheet**

### **Overview and Symptoms**

Dystonia is a movement disorder that causes your muscles to contract without your control. This can cause twisting or jerking repetitive movements or abnormal and uncomfortable positions or postures. Dystonia may affect any body part, any area of the body, half of the body, or the entire body. In most people with dystonia, the cause is not known. Several forms of dystonia, including one called *DYT1*, are due to a change in a gene. These forms can be passed from parent to child. Some cases of dystonia are caused by exposure to toxins or poisons or to certain prescription medicines used to treat psychiatric disorders.

There are many forms of dystonia, including the following:

- Cervical dystonia (CD) or spasmodic torticollis is the most common form of focal dystonia. CD affects the muscles that control the movement of the head and neck, causing the person's head to turn to the side, or up or down. Pain may accompany the movements.
- Limb dystonia affects the arm, hand, leg, or foot and includes writer's cramp, an involuntary posturing of the hand that occurs only when writing.
- Spasmodic dysphonia or laryngeal dystonia affects the vocal cords and causes the voice to be either breathy or harsh and strangled.
- Oromandibular dystonia affects the mouth and tongue, and causes difficulty with speaking or swallowing.
- Blepharospasm affects muscles around the eyes causing eye closure and difficulty with vision.
- Childhood onset generalized dystonia is the most severe form. Unlike the focal dystonias listed above, this form of dystonia usually begins in the leg at a young age and often spreads to affect the whole body. Many cases are due to changes (mutations) in the *DYT1* gene.

Because of the involuntary movements, dystonia can interfere with work and activities of daily living. Sometimes, the abnormal postures cause social embarrassment. Cervical dystonia is often painful.

### **Diagnosis**

Dystonia is not a common disorder and may be mistaken for other more common diseases. In determining if you have dystonia, your physician will want to interview and examine you for:

- Your complete medical history, especially any exposure to legal or illegal drugs, or poisons.
- Your family history.
- Your general neurological condition, with particular attention to an assessment of dystonia, and the actions that make it better or worse.
- Your results from certain tests performed to rule out other conditions.

The doctor also may recommend a genetic test to determine if you have an inherited form of dystonia.

### **Treatment**

Not all dystonia needs to be treated. If you and your doctor agree that treatment is needed,

your choices may include the following:

- Injection therapy with a botulinum neurotoxin (BoNT) type A as onabotulinumtoxinA (Botox<sup>®</sup>) or abobotulinumtoxinA (Dysport<sup>®</sup>) or incobotulinumtoxinA (Xeomin<sup>®</sup>) or botulinum neurotoxin type B as rimabotulinumtoxinB (Myobloc<sup>™</sup>) is the most effective treatment for focal dystonia; injections are typically limited to a specific region of the body. It typically requires repeated treatments at about three-month intervals and is usually associated with minimal side effects.

- Physical therapy, bracing, relaxation and learning techniques may assist in accommodating the dystonia. Stretching is important to prevent muscle shortening. Bracing may be effective if it reduces the dystonia without being uncomfortable. Techniques such as learning to write with the non-dystonic hand may also be beneficial.
- Oral medications may be very helpful in children with generalized dystonia, but are usually not completely effective in adults. In addition, oral medication may cause reversible side effects that limit the doses. These oral medications may include: benzodiazepines such as diazepam (Valium<sup>®</sup>) and clonazepam (Klonopin<sup>®</sup>); the anti-spasmodic baclofen (Lioresal<sup>®</sup>); anticholinergics such as trihexyphenidyl (Artane<sup>®</sup>) or diphenhydramine (Benadryl<sup>®</sup>); or neuroleptic drugs such as clozapine (Clozaril<sup>®</sup>) or olanzapine (Zyprexa<sup>®</sup>). A small group of patients with a specific type of childhood-onset dystonia (dopa-responsive dystonia or DRD) improve dramatically with levodopa.
- Intrathecal baclofen therapy (ITB Therapy<sup>™</sup>) may be most effective in those with dystonia and spasticity involving the lower body. Surgery is required to implant a pump, which delivers anti-spasm medication directly into the space around the spinal cord. Complications may occur and may be serious in some patients.
- Two different types of brain surgery have been tried—lesioning and deep brain stimulation (DBS). During a lesioning surgery, a small selected area of the brain called the globus pallidum is destroyed. This disrupts abnormal brain activity in this region, partially restoring normal movement. Deep brain stimulation (DBS) uses implanted electrodes to stimulate these same areas of the brain. The electrical stimulation interferes with the abnormal activity, creating the same effect as a lesion. The effect lasts as long as the stimulation continues. Complications are possible.
- Support groups and other forms of psychological counseling are very important for psychological adjustment and maintaining or improving self-image.

### **Questions to Ask Your Doctor**

- Are my symptoms likely to get worse or better over time?
- Will my symptoms spread to other parts of my body?
- Will my children get dystonia?
- How do I know if I am a good candidate for treatments like BoNT injections, ITB or DBS?
- What are the treatments that could help me and are there side effects?
- How can I contribute to research studies on dystonia?

If you would like more detailed information on the symptoms, diagnosis, and treatment options or additional support (including the Dystonia Discussion Forum), please visit [www.wemove.org](http://www.wemove.org).