What is the Cause?

There are multiple causes for generalized dystonia. Dystonia may result from changes in certain genes, birth injury, exposure to certain drugs, head injury, infection, and other secondary causes. Dystonia symptoms that occur only on one side of the body strongly suggest a secondary cause. Dystonia resulting from secondary causes often occurs with additional movement and neurological symptoms, depending on the nature and severity of injury to the nervous system.

For many people who develop generalized dystonia, there is no identifiable cause.

Common Misdiagnoses

Generalized dystonia symptoms should not be confused with:

- Parkinson's disease
- Tremor
- Cerebral palsy
- Orthopedic and structural conditions
- Behavioral disorders or mental illness

What is Generalized Dystonia?

Dystonia is a neurological disorder characterized by uncontrollable muscle contractions resulting in involuntary postures. These postures can be accompanied by movements that are patterned and twisting, and may resemble tremor.

Generalized dystonia refers to dystonia that is not limited to a single part of the body but affects multiple muscle groups throughout the body. Generalized dystonia typically affects muscles in the torso and limbs, and sometimes the neck and face. Patients have difficulty moving their bodies freely and controlling their body movements. It is important to note that dystonia does not target vital organs such as the heart.

Generalized dystonia typically begins during childhood or adolescence, often without additional neurological symptoms. Initial symptoms may be a turned or twisted foot and/or leg. However, dystonia in children can be associated with more complex neurological or metabolic diagnoses. Generalized dystonia can occur with or without a family history. The symptoms may always be present or only occur in episodes.

What Support is Available?

Having access to credible information and connection to others in the dystonia community can be vital to living well with generalized dystonia. Dystonia Medical Research Foundation DMRF Canada can provide educational resources, self-help opportunities, contact with others, volunteer opportunities, and connection to the greater dystonia community.

What is DMRF Canada?

Dystonia Medical Research Foundation (DMRF) Canada is a charitable organization that funds medical research toward a cure, promotes awareness and education, and supports the well being of affected individuals and families.

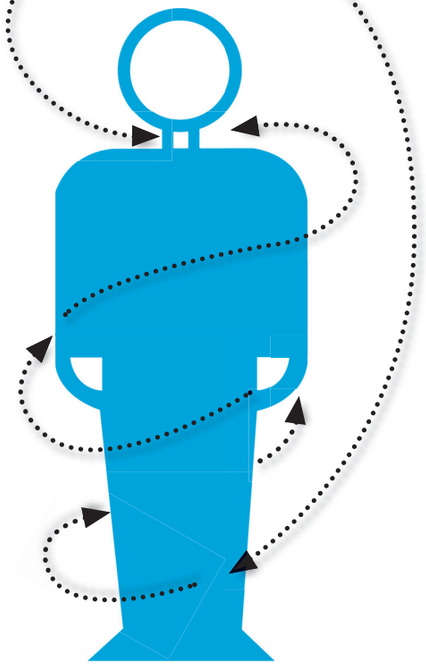
To learn more about dystonia and DMRF Canada, contact:

DMRF Canada
Po Box 1009 Stn Toronto Dom
Toronto On M5K 1P2
Phone: 416.488.6974
Toll free: 800.361.8061
www.dystoniacanada.org
Email: info@dystoniacanada.org

For more in-depth information, visit dystoniacanada.org/generalizeddystonia

Many thanks to past DMRF Clinical Fellow Kelly Andrzejewski, DO, PhD for reviewing the content of this brochure

Generalized Dystonia



Dystonia Medical Research Foundation Canada
Fondation de Recherche Médicale sur la Dystonie Canada

serving all dystonia-affected persons
désservant toutes personnes atteintes de dystonie

Are there Different Types?

There are several genetic subtypes of generalized dystonia. For example:

Early Onset Generalized Dystonia (DYT1/TOR1): DYT1 dystonia typically begins around age 10 years with the twisting of a foot or arm. Symptoms tend to progress to involve additional limbs and the torso, but usually not the face or neck. In rare cases, the vocal cord muscles are affected. The symptoms tend to be less severe the later in life they start and if they start in a hand or arm. About 30% of individuals who have the DYT1 genetic mutation will develop dystonia. If a person does not manifest symptoms before the age of 28 years, they will usually remain symptom free for life—even if they have the DYT1 mutation. The DYT1 mutation is responsible for about 90% of early onset generalized dystonia in individuals of Ashkenazi Jewish ancestry and up to about 50% of early onset generalized dystonia in other ethnicities.

Adolescent Onset Generalized Dystonia (DYT6/THAP): DYT6 may occur as generalized dystonia or remain focal to a specific part of the body. Although the symptoms may resemble DYT1 dystonia, onset of DYT6 is usually in the late teens and symptoms are more likely to occur above the neck. Symptoms typically affect the muscles of the tongue, vocal cords, and face. Patients often have difficulty speaking. About 40% of individuals who have the mutation will develop dystonia.

Testing for DYT1, DYT6, and other known dystonia genes is available. A genetic counselor can help families understand what genetic testing may be appropriate.

What Kind of Doctor Treats Dystonia?

Individuals with dystonia are encouraged to seek treatment from a neurologist (or child neurologist) with special training in movement disorders. A multidisciplinary team of medical professionals may be appropriate to tailor treatment to the needs of the patient.

What Treatments are Available?

A movement disorder specialist will develop a treatment plan that is customized to each patient. Most individuals require a combination of therapies. Treatment to lessen dystonia symptoms may include oral medications such as anticholinergics, baclofen, and benzodiazepines combined with botulinum neurotoxin injections, and/or surgical procedures such as deep brain stimulation (DBS).

Specific treatment may be needed to prevent loss of motion in the joints and/or curvature of the spine due to the dystonic postures. Complementary therapies to support overall functioning and wellness may include occupational therapy, physical therapy, speech/voice therapy, and other interventions depending on a person's symptoms. Individuals with childhood onset generalized dystonia may have increased risk for depression so monitoring and addressing emotional and mental health is often an important part of the treatment strategy.

Is Dystonia Fatal?

No, dystonia is almost never fatal. In extreme cases, the development of increasingly frequent or continuous episodes of severe generalized dystonia causes a medical emergency called status dystonicus. If untreated, this rare condition can cause life-threatening complications. Status dystonicus is a treatable condition and, with prompt medical attention, symptoms typically can be brought under control.

Living with Dystonia

Living well with generalized dystonia is possible. The early stages of symptom onset, diagnosis, and seeking effective treatment are often the most challenging. In some cases, treatment can dramatically reduce or suppress symptoms.

Individuals and families living with dystonia are strongly encouraged to:

- Seek treatment from a neurologist who specializes in movement disorders.
- Learn about dystonia and treatment options.
- Develop a multi-layered support system of support groups, online resources, friends, family, and mental health professionals, if needed.
- Investigate complementary therapies that support overall functioning and wellness.
- Get active within the dystonia community.

Dystonia can occur with other movement symptoms:

Dopa-responsive dystonia includes symptoms of dystonia and parkinsonism (slow movement, muscle rigidity, tremor, balance instability). Symptoms typically respond to a drug called levodopa.

Myoclonus-dystonia is characterized by rapid jerking movements (myoclonus) alone or with dystonia. Additional features may include obsessive compulsive disorder, depression, anxiety disorders, and alcohol abuse.

Paroxysmal dyskinesias are episodic movement disorders. The involuntary movements may include dystonia, chorea (uncoordinated jerking), ballism (rapid, flinging movements), and/or athetosis (writhing).

Rapid-onset dystonia-parkinsonism is characterized by abrupt onset of dystonia with parkinsonism. Additional features may include anxiety, depression, and seizures.

X-linked dystonia-parkinsonism is characterized by parkinsonism, often followed by dystonia later in life. Affects individuals of Filipino descent almost exclusively.