

# What Is Dystonia?

## So You've Been Diagnosed with Dystonia.... Now What?

Dystonia is a disorder that affects the nervous system. Abnormal signaling from the brain causes muscles to tighten and twist involuntarily. These muscle spasms force areas of the body into awkward movements and positions. It may be painful and interfere with daily activities. It is considered a neurological movement disorder.

Dystonia affects over 50,000 in Canada, of all ages and backgrounds. Most common forms affect adults but it can also occur in childhood especially if genetic. It is almost never fatal but rarely may occur as a symptom of neurodegenerative diseases, and these conditions may affect life span.

There are several forms of dystonia, and dystonia may be a symptom of many diseases and conditions. The words used to describe your specific diagnosis may be confusing.

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***To accurately describe a dystonia diagnosis, several specific pieces of information must be accounted for:***

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### 1. What are the Clinical Features?

Clinical features are the signs and symptoms of dystonia, which can vary a great deal from patient to patient. These include age at which symptoms began, body distribution, certain qualities about the symptoms, and the presence of other movement disorder symptoms or other neurological features. Doctors use these factors to guide diagnosis and treatment.

#### **Age**

Dystonia can occur at any age. At this time, there is no way to predict the rollout of a prognosis, but the age at which symptoms begin often affects if, and how, dystonia might spread to other parts of the body. When symptoms begin during childhood, they are often more likely to spread and affect multiple muscle groups. When symptoms begin in adults, the symptoms are more likely to remain localized to a particular body area such as the neck or an arm.

# What Is Dystonia?

## Body Distribution

Dystonia symptoms can affect the body in several anatomical patterns:

- **Focal dystonia:** Only one body region is affected, for example the neck or the hand.
- **Segmental dystonia:** Two or more connected body regions are affected. For example, the neck and the face.
- **Multifocal dystonia:** Two or more non-connected body areas are affected. For example, the face and an arm.
- **Generalized dystonia:** The trunk and at least two other body areas affected, and usually at least one leg.
- **Hemidystonia:** Symptoms are restricted to muscles on only one side of the body.

## Duration or Quality of Symptoms

Certain special qualities about dystonia symptoms also guide diagnosis and treatment.

The symptoms typically fall into one of several patterns:

- **Chronic:** Dystonia that is present throughout the day.
- **Task-specific:** Dystonia that occurs only during a particular activity or task.
- **Diurnal fluctuations:** Dystonia that fluctuates during the day, often in patterns related to sleep and waking.
- **Paroxysmal:** Sudden episodes of dystonia usually induced by a trigger.

Dystonia can occur in isolation or in combination with other movement disorders. When dystonia occurs in isolation (sometimes referred to as 'primary' dystonia), it is the only present neurological disorder (with the exception of tremor).

Dystonia can also occur in combination with other movement symptoms such as myoclonus or parkinsonism. Treating the additional movement symptoms must then be part of the treatment plan.

## 2. What is Known About the Cause?

In many cases, individuals develop dystonia without any obvious cause. However, if a cause can be identified, this may guide treatment. When describing dystonia by the cause, it may be characterized as primary, secondary, or dystonia-plus:

Dystonia may be inherited or acquired. Inherited dystonias are those with a proven genetic origin. Several genes known to cause dystonia have been identified.

# What Is Dystonia?

Acquired dystonias, or secondary dystonias are due to a specific life event or series of events, for example birth injury, drug exposure, brain injury, infection, and other factors. Psychogenic or functional dystonias may be considered acquired dystonias, though this in an area that remains in debate even among experts.

Dystonia can also occur as a symptom of numerous diseases and conditions. These include specific vascular conditions, infections, brain tumors, metabolic conditions, neurodegenerative disorders, demyelinating disorders, and structural conditions.

However, most cases of dystonia are idiopathic (or primary dystonia), meaning that there is no obvious or identifiable cause. This includes cases with or without a family history. There are families with multiple members who are affected by dystonia—suggesting an obvious genetic component—but no specific gene is known to be the culprit. Many of the focal dystonias that occur in adulthood fall under this category. As more genes are identified, certain idiopathic forms may be reclassified as inherited. Some primary dystonias include genetic forms (such as DYT<sub>1</sub> dystonia for which a gene has been identified).

## 3. Forms of Dystonia

Doctors use these classifications to guide diagnosis and treatment. Forms of dystonia include:

**Focal dystonias** affect a specific group of muscles or body parts, including:

- **Blepharospasm** affects the muscles of the eyelids and brow.
- **Cervical dystonia** affects the neck and sometimes the shoulders.
- **Oromandibular dystonia** (*cranial dystonia*) includes forceful contractions of the face, jaw, and/or tongue.
- **Spasmodic dysphonia** (*laryngeal dystonia*) affects the vocal cords.
- **Hand dystonia** (*writer's cramp*) affects the fingers, hand, and/or forearm.
- **Lower limb dystonia** affects the leg, foot, and/or toes.

**Musicians' dystonias** are task-specific dystonias sometimes given this name because constant use of various muscles and movements can contribute to dystonia among musicians related to their performance.

**Early-onset generalized dystonia (DYT<sub>1</sub> and non-DYT<sub>1</sub>)** is characterized by twisting of the limbs and torso.

# What Is Dystonia?

**Dopa-responsive dystonia** refers to a group of dystonias that respond to a medication called levodopa.

**Myoclonus dystonia** is a hereditary form that includes prominent jerk-like movements called myoclonus

**Paroxysmal dystonias and dyskinesias** involve episodic abnormal movements which occur only during attacks.

**X-linked dystonia-parkinsonism** is a hereditary form of dystonia (also called Lubag disease) that includes symptoms of parkinsonism.

**Rapid-onset dystonia-parkinsonism** is a hereditary form of dystonia that includes symptoms of parkinsonism.

**Secondary dystonias** are forms triggered by factors such as trauma, medication exposure and toxins.

- **Trauma:** Dystonia may follow trauma to the head and/or to a specific area of the body.
- **Drug-induced** (*Tardive dystonia & dyskinesias*): Specific drugs can cause dystonia.
- **Neurological and metabolic disorders:** Dystonia can occur as a symptom of multiple disorders such as stroke and Parkinson's disease

**Psychogenic dystonia** is secondary to psychological/psychiatric causes or an underlying pain syndrome.

For more details and information visit [www.dystoniacanada.org/whatisdystonia](http://www.dystoniacanada.org/whatisdystonia) or contact us at **1.800.361.8061** or [info@dystoniacanada.org](mailto:info@dystoniacanada.org).