Dystonia is a neurological movement disorder. It causes involuntary muscle contractions, which lead the affected parts of the body to develop abnormal postures, which may or may not be associated with tremor. Dystonia may affect just one part of the body or several different parts. When it develops in adults (late-onset) it usually confines itself to one part of the body but in the case of early-onset (in late childhood and early teens), it can generalise ie spread to two or more contiguous body parts.

Generalised dystonia / Early-onset dystonia
Generalised dystonia is a rare form of dystonia, which usually begins in late childhood / early teens. It is sometimes known as primary torsion dystonia or Oppenheim's dystonia. Typically, the condition starts in a limb and can then spread to adjoining areas. Involuntary spasms can occur in a foot or leg and then progress to include other limbs and the trunk. The most common age of onset is around nine years old. Cases of generalised dystonia starting after the mid-twenties are unusual.

Symptoms may include:
- Muscle spasm with or without pain
- Twisted postures of the limbs or trunk
- Abnormal fixed postures of the limbs or trunk
- Turning in of the foot and/or leg and/or arm
- Rapid jerking movements
- Unusual walking with bending and twisting of the torso

Initial symptoms will often spread over a number of months, usually up to two years, to lead to a ‘generalised’ form of dystonia. However, some patients may only develop dystonia in one or two body areas (focal/segmental dystonia respectively). Symptoms will then typically plateau, although they may wax and wane.

There are two distinct forms of generalised dystonia:

1. Primary Where there is no identifiable cause other than genetic factors
2. Secondary Where there is an underlying cause such as another neurological condition, drugs or a stroke

In cases of early-onset primary dystonia there is a 50–60% likelihood that the person affected will have a genetic mutation known as DYT1 (the first gene identified for dystonia). DYT1 dystonia is due to a fault in functioning of the DYT1 gene. The DYT1 gene codes for torsin A, a protein that is involved with key functions of the cell, especially in areas of the brain involved in the processing of dopamine (involved in movement).

The DYT1 mutation occurs in all ethnic groups but is 3–5 times more common in Ashkenazi Jews compared to other populations. Testing for DYT1 is technically easy and widely commercially available.

DYT1 dystonia is inherited in an autosomal dominant fashion but has a markedly reduced penetrance. This means that the gene mutation can be passed to the child by one parent who is a carrier (i.e. there is a 50% chance of their child having the gene mutation). However, the reduced penetrance of the mutation means that only 30% of carriers of the mutation are likely to develop any symptoms of dystonia during their lifetime.
Furthermore, the penetrance is age dependent: if a carrier of the mutation reaches 25 without developing symptoms of generalised dystonia, they are unlikely ever to develop dystonia. Even if symptoms of dystonia develop, there is great variability in the range and severity of the symptoms that result. For instance, while some patients develop severe generalised dystonia, others may just have mild focal dystonia in the limbs. Once the condition has plateaued, further progression is highly unusual.

While genetic counselling is generally available for those undergoing a test for the DYT1 gene, the psychological and social implications of an autosomal dominant disorder with markedly reduced penetrance and very variable expression, are complicated. Patients and their families will need time to review their situation and to consider their options.

**Non-DYT1 early-onset primary dystonia**

Some patients with generalised dystonia/early-onset primary dystonia may not be carriers of the DYT1 gene yet may still have dystonia caused by a genetic mutation. For instance, there is a rare form of dystonia called dopa-responsive dystonia that is caused by a mutation in gene DYT5. This form of dystonia usually starts in childhood and affects the limbs. There is often a strong diurnal (daily) variation (ie. the child can seem unaffected in the morning and then their movements become progressively worse throughout the day). This condition can respond dramatically to daily treatment by the drug called levodopa. Because of the positive effect of the drug, most physicians will now suggest that a child presenting with dystonic symptoms should be tried on a course of levodopa to see if there is any marked improvement. There are also several other even rarer genetic mutations that have been linked with particular cases of generalised dystonia (ie. DYT4 and DYT13) in individual families. Currently, gene tests are not available for these genetic defects.

In cases where a young patient is not a DYT1 gene carrier and has not responded to a simple course of levodopa, the physician will want to explore other avenues to establish whether the dystonia is ‘secondary’ ie. is the result of other progressive or degenerative conditions. These further examinations may involve a simple MRI scan of the brain as well as testing for other rare conditions such as Wilson’s disease (a rare but treatable genetic condition in which the body does not process copper properly) or neuronal brain iron accumulation syndrome (NBIA – a genetic degenerative condition in which iron is deposited in the basal ganglia).

### What causes dystonia?

Dystonia is believed to be due to a problem in an area of the brain called the basal ganglia, which are structures deep within the brain that control voluntary movements and postures. Although the precise way in which these structures malfunction to cause dystonia is not completely understood, much research is ongoing and is progressing towards a greater understanding of the condition.

### Getting a Diagnosis

Although descriptions of dystonia have appeared in medical literature since the early part of this century, getting an accurate and prompt diagnosis has been more unusual than typical for too many people. The usual clinical tests come up negative, so doctors can sometimes be as mystified as the patient. Getting referred to a neurologist who specialises in movement disorders is often the only way an accurate diagnosis can be reliably found.
How may generalised dystonia be treated?

Although at the present time there is no known cure for dystonia, several treatment options are available.

As every person with generalised dystonia is unique, treatment needs to be tailored to the needs of the individual as no single course of treatment will be appropriate for all cases. The primary intention of all treatment is to help reduce the symptoms, principally the effects of muscle spasm, pain and awkward postures. The ultimate goal of treatment being to enhance and maintain the quality of life of everyone affected.

**Oral medications** will generally be the first type of treatment tried. A trial of levodopa is appropriate in all young patients with early-onset primary dystonia to exclude the possibility of dopa-responsive dystonia. Generally a course of the drug will be tried for at least two months before the effectiveness is judged. If, as is unfortunately usually the case, levodopa is ineffective, an anti-cholinergic drug such as trihexyphenidyl will often be tried which can often be helpful in controlling muscle spasms and tremor. In some cases, second line treatments will be tried which may include clonazepam (a strong muscle relaxant) or tetrabenazine (helps to control tremor and involuntary spasm) and baclofen (an effective muscle relaxant). A combination of medications is often advised by a neurologist. These drugs can have side effects which you should discuss with your doctor.

**Botulinum toxin injections** are the most common and effective treatment for the thousands of adults with focal dystonia. However, they have a limited role for people with generalised dystonia. Isolated areas that are affected by dystonia may be treated, such as the jaw, hands, feet or leg, with some functional benefit.

“Many people do find significant relief from treatments to manage painful symptoms...”

A surgical technique known as deep brain stimulation has recently been shown to provide remarkable and sustained benefit for selected individuals. It works by inserting fine electrodes in the brain that will stimulate areas of the globus pallidus to damp down the spurious signals causing the muscle spasms. It is rapidly emerging as a significant early treatment for dystonia, particularly for patients with severe primary generalised dystonia. Selection of patients for surgery is done carefully after an extensive series of tests at a movement disorder clinic in order to ensure the patient’s condition is likely to benefit from the surgery.

**Dystonia and Pain**

The degree of pain associated with dystonia tends to vary greatly from person to person, from no pain to severe pain. In many cases, treatments that address the dystonic contractions and spasms will also help to relieve the pain. Botulinum toxin injections and muscle relaxant medications may be quite effective at reducing pain.

The emotional toil of chronic pain may make pain worse. Anxiety, stress, depression, anger and fatigue interact in complex ways with pain and may decrease the body’s production of natural painkillers; moreover, negative feelings may increase the level of substances that amplify sensations of pain, causing a vicious cycle of pain.

Management of the pain can provide physical and emotional benefits. It is important that anyone in pain due to their dystonia tries to get help from one of the various specialists in pain management. Many people do find significant relief from treatments to manage painful symptoms.
What the Society may offer

The Dystonia Society is dedicated to providing information and support to everyone affected by dystonia in the UK and to raising awareness of the condition and the needs of everyone affected. The Society is also committed to ensuring that everyone with dystonia has access to the treatment they need.

Our Helpline is open Mondays to Fridays between 10am and 4pm and offers an opportunity to discuss concerns in confidence, and to obtain information on dystonia and its various treatments, including ways of making living with dystonia easier.

Call our helpline on:
0845 458 6322

Important note
The contents of these pages are provided only as information and are in no way intended to replace the advice of a qualified medical practitioner. The Society strongly advises anyone viewing this material to seek qualified medical advice on all matters relating to the treatment and management of any form of medical condition mentioned. Furthermore, rapid advances in medicine may cause information contained here to become outdated after some months.