Generalised dystonia and pain
Some people with generalised dystonia report experiencing pain. There are a number of medications that can be helpful in managing pain. Many people also find attending pain management sessions helpful. To learn more about managing pain, you can get our Dystonia and pain leaflet through the helpline or from the website.

Dystonia and mental / emotional health
Mental (and emotional) health is a sensitive topic for many people with dystonia as many cases of dystonia are initially mistaken for a mental health (or psychological) condition. In the vast majority of cases, dystonia is a neurological illness and does not have a mental health cause.

However, it is also increasingly understood, that although mental health conditions do not normally cause dystonia, there can be an important relationship in some cases between dystonia and mental health conditions such as stress, depression and anxiety. This relationship can be two way - the symptoms of dystonia can cause anxiety or depression but also anxiety and stress can make the physical symptoms of dystonia worse.

If you are affected by a mental health problem, it is important to get treatment. To learn more, you can get our Dystonia and mental health leaflet through the helpline or from the website.

Other support
Physiotherapy can be helpful in managing generalised dystonia. It needs to be provided by a specialist physiotherapist who understands dystonia as inappropriate exercises may make symptoms worse. Occupational therapists can advise on specialist equipment which assists posture, movement and coping with daily living.

The Dystonia Society
The Dystonia Society is dedicated to providing information and support to everyone affected by dystonia in the UK. Our services include a helpline, advocacy, regional support groups and events about dystonia across the UK.

You can find out more and sign up for our free e-newsletter on our website. Alternatively you can email or call the Society.

Helpline
0845 458 6322
Website
www.dystonia.org.uk

March 2015
Dystonia is a neurological movement disorder that results in abnormal postures or movements, with or without tremor. It affects at least 70,000 in the UK.

**Generalised dystonia**

Dystonia can affect just one part of the body or several different areas. When it develops in adults it usually confines itself to one part of the body. However, dystonia which develops in children or young adults (early-onset) can often spread to two or more areas of the body.

**Generalised dystonia** is a rare form of dystonia that is usually early-onset (most often appearing in late childhood / early teens). Typically, the condition starts in a limb and then ‘generalises’ to other areas. Involuntary spasms can occur in a foot or leg and then progress to include other limbs and the trunk. In most cases, symptoms progress and stabilise within a 5-year period. Cases of generalised dystonia starting after the mid-twenties are unusual. Symptoms may include:

- Muscle spasm with or without pain
- Twisted or abnormal fixed postures of the limbs or trunk
- Twisted or abnormal neck position
- Turning in of the foot, leg and/or arm
- Rapid jerking movements
- Unusual walking with bending and twisting of the torso

**Types of generalised dystonia**

Generalised dystonia can be **inherited** (caused by genetic factors), **idiopathic** (where there is no identifiable cause) or **acquired** (there is a cause such as another medical condition, drugs or a stroke).

Generalised dystonia is sometimes caused by a mutation in the DYT1 gene. If someone inherits this mutation there is 30% chance they will develop DYT1 dystonia. If a carrier reaches the age of 30 without developing dystonia there is a good chance the dystonia never will develop. Testing for DYT1 is technically easy.

There are a number of other genes which cause primary early-onset generalised dystonia including the DYT5 gene which causes a rare type of generalised dystonia highly responsive to treatment by dopamine called **dopa-responsive dystonia**.

If no genetic cause can be identified, the physician will explore if the dystonia is “acquired”. This may involve a MRI scan of the brain and testing for other rare conditions such as Wilson’s disease (a 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).

**Treatment of generalised dystonia**

There is currently no known cure for dystonia but treatment options are available. Treatment differs between individuals but the goal of all treatments is to reduce the symptoms.

**Oral medications** will generally be tried first. A trial of levodopa (usually for at least 2 months) is appropriate in all early-onset idiopathic /inherited dystonia to identify dopa-responsive dystonia. If this is ineffective, an anticholinergic drug such as trihexyphenidyl can often be helpful in controlling muscle spasms and tremor. Second line treatments may include clonazepam (a strong muscle relaxant) or tetrabenazine (helps to control tremor and involuntary spasm) and baclofen (another muscle relaxant). A combination of medications is often used. These drugs can have side effects that you need to discuss with your doctor.

**Botulinum toxin injections** can sometimes be helpful in reducing symptoms in some isolated areas that are affected by dystonia such as the jaw, hands, feet or leg.

A surgical technique known as **deep brain stimulation** can provide sustained benefit in some cases. It works by inserting fine electrodes in the brain to damp down the signals causing the muscle spasms. Selection of patients is done carefully with extensive testing to ensure the patient’s condition is likely to benefit from the surgery.