

# Generalised dystonia

which you should 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 spurious 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.

## Dystonia and Pain

The degree of pain varies greatly between people with dystonia from no pain to severe pain. Treatment can often relieve pain. However, the emotional impact of dystonia may make pain worse. Anxiety, depression and other emotions interact in complex ways with pain, for example by decreasing the body's production of natural painkillers. Management of the pain can provide physical and emotional benefits. Many people find significant relief from treatments to manage painful symptoms provided by specialists in pain management so referral to these specialists is important.

Our services include the following:

- **Our helpline** offers an opportunity to discuss concerns in confidence and to obtain information on dystonia.

**Helpline: 0845 458 6322**

- **Our regional support groups** run by volunteers provide an opportunity to share experiences and meet others.
- **Our website** has information on dystonia and a lively forum. The address is [www.dystonia.org.uk](http://www.dystonia.org.uk). You can also sign up on the website for our free **e-newsletter**.
- **The organisation** supports research into potential treatments and ways of coping.
- **Join us** – become a member and receive our quarterly newsletter. Call: 0845 458 6211 or email [info@dystonia.org.uk](mailto:info@dystonia.org.uk).

Dystonia affecting the limbs, trunk and other body areas

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# What is dystonia?

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 and is believed to be the result of abnormal functioning of the basal ganglia, an area of the brain involved in controlling movement.

## 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 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

## Types of generalised dystonia

Generalised dystonia can either be **primary** where there is no identifiable cause other than genetic factors or **secondary** where there is an underlying cause such as another medical condition, drugs or a stroke.

Primary generalised dystonia is often 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 'secondary' ie. is there an identifiable cause? 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 primary dystonia to identify dopa-responsive dystonia. If this is ineffective, an anti-cholinergic 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 (an effective muscle relaxant). A combination of medications is often advised by the neurologist. These drugs can have side effects