



Dystonia Fact Sheet

Key Facts

- Dystonia is a movement disorder in which muscle contractions cause twisting and repetitive movements or abnormal postures.
- More than 500,000 people are affected by dystonia across Europe.
- It can develop at any time from childhood through to adulthood, and can occur spontaneously or as a result of another condition.
- Although not terminal symptoms can severely disrupting a person's life.

What is dystonia?

Dystonia is a neurological movement disorder involving involuntary and abnormal movements and postures which can affect just one part of the body or several different areas.¹ It affects males and females of all ages; in adults, dystonia tends to remain located in a specific part of the body (focal form) such as the neck, hands, face or eyelids. However, if it starts in childhood, it often spreads to other parts of the body (generalized) and can be particularly disabling.²

The scale of the problem

Dystonia is the third most common movement disorder after Parkinson's disease and essential tremor.³ Epidemiological studies indicate that there are more than 500,000 people across Europe with some form of the disorder, many of whom are unaware that they have dystonia. The disorder can be difficult to diagnose, which means that many patients remain untreated, their symptoms unrecognised.¹

How severe is dystonia?

Dystonia is not a life-threatening disorder but, in its more serious forms, it can have a devastating effect on the life of patients and their families.¹ Symptoms are often specific to each individual, and there is no way to predict exactly how the disease will progress, although most cases of dystonia will usually stabilize within five years of onset. No medication or therapy can prevent the disease from progressing, and there is no cure,

although prompt diagnosis and treatment can often minimize the impact of symptoms and improve or maintain a person's ability to function in everyday activities.³ Remission from symptoms, though unusual, can occur, although only in around 5% of cases.²

Causes of dystonia

In the majority of cases the underlying cause of the condition is unknown. Where no cause can be identified, the dystonia is said to be **primary** or **idiopathic**.¹ Dystonia which develops in childhood is often inherited through one or more affected genes, although not everyone who inherits the gene develops dystonia. Dystonia which develops in adults may also be inherited. Some people with dystonia may have suffered from an illness or injury which has damaged a part of the brain known as the basal ganglia (**secondary dystonia**), such as a stroke, a brain injury, encephalitis or Parkinson's disease.¹

Diagnosis

There are no specific tests which determine whether a person has dystonia. Many family doctors are unfamiliar with it, and therefore clinical examination by a specialist doctor is required.¹

Primary or secondary dystonia is usually confirmed through a series of general tests.

- ▲ **Recent medical history and family history** e.g. whether the patient has recently suffered a head injury, or whether they have a relative who has developed dystonia.
- ▲ **Blood and urine tests** check the function of organs, such as the liver to check for infection or high levels of toxins in the body.
- ▲ **Genetic testing** can identify abnormal genes associated with some types of dystonia and can also confirm whether or not the person's dystonia is secondary to a genetic condition, such as Huntington's disease.
- ▲ **A magnetic resonance (MRI) scan** checks whether there is any damage to the brain, or whether there is another condition affecting the brain, such as a tumour.⁴

Main features of the disease

Symptoms can vary depending on the form of dystonia - some of which are described below.⁴

- ▲ **Cervical dystonia** (also known sometimes as **torticollis**) is the most common form of dystonia and affects the muscles in the neck, leading to the head and neck being twisted or pulled in one particular direction resulting in neck pain and stiffness.
- ▲ **Blepharospasm** affects the muscles around the eyes. Symptoms include eye irritation, sensitivity to light (photophobia), uncontrollable blinking and closing of the eyes effectively causing short periods of temporary blindness.
- ▲ **Generalised dystonia** usually begins around the time that a child reaches puberty, starting in one of the limbs before spreading to other parts of the body. Symptoms

include muscle spasms; abnormal or twisted posture of the limbs or torso; a foot, leg or arm turning inwards; body parts jerking rapidly.

- ⤴ **Dopa-responsive dystonia** often begins during childhood, (between ages six and sixteen), and usually involves an abnormal, stiff way of walking with the foot bent or turned, although it may also affect the arms and torso.
- ⤴ **Oromandibular dystonia** affects the muscles in the mouth and jaw causing the mouth to pull outwards and upwards.
- ⤴ **Writer's cramp** causes involuntary cramps and movements in the muscles of the arm and wrist, and commonly affects people who do a lot of writing. Other forms of task-specific dystonia affect musicians, golfers and typists.
- ⤴ **Laryngeal dystonia** causes the muscles of the larynx (voice box) to spasm. Depending on whether the muscles spasm outwards or inwards, the spasm causes the voice to sound either strangled or very quiet.

Treatments and therapies

Several different types of treatment are available.

Botulinum toxin prevents the release of the chemical messenger, acetylcholine from the nerve endings, which helps to reduce muscle contractions, and causes muscle weakness. However, once the nerve endings grow back, the dystonic symptoms return so the treatment has to be repeated every two or three months.⁵

Levodopa is used to treat dopa-responsive dystonia. It works by increasing the levels of the neurotransmitter, dopamine.⁴

Anticholinergics are used to treat some types of focal dystonia, although they are not effective in all cases. They work by blocking the release of the neurotransmitter, acetylcholine, which is known to cause muscle spasms in some cases of dystonia.⁴

Muscle relaxants work by increasing the levels of the neurotransmitter gamma-aminobutyric acid (GABA), which helps to relax affected muscles. **Diazepam** and **clonazepam** are two examples, and they are often used to treat cases of dystonia that fail to respond to other types of medication.⁴

Surgery is usually only considered for patients in whom other forms of treatment have not been successful.⁵ Surgical approaches include:

- ⤴ **Selective peripheral denervation** - used for cervical dystonia, the surgeon makes an incision in the neck, then severs the nerves connected to muscles that are prone to spasms.⁴
- ⤴ **Thalamotomy and pallidotomy** for generalised forms of dystonia.¹
- ⤴ **Deep brain stimulation** involves the insertion of tiny electrodes into a part of the basal ganglia through holes drilled in the skull. The electrodes are connected to a small pulse generator implanted under the skin, usually on the chest or lower

abdomen. The pulse generator sends signals to the basal ganglia to block the abnormal nerve impulses which lead to the symptoms for dystonia.⁴

- ▲ **Non-pharmaceutical** treatments of many kinds like acupuncture, relaxation techniques, homeopathy and hypnosis have been tried with varying degrees of success, but with little evidence of long-term benefit. Physiotherapy and speech therapy have proved to be invaluable in many cases.¹

Impact on the lives of those affected and carers

The symptoms of dystonia can be both painful and inconvenient. The severity of symptoms varies enormously, but for some they can totally disrupt normal life – preventing patients from working, driving, socialising and many other day to day functions. Continual spasms and contortions can be exhausting and severely limit an individual's life choices.

Additionally, a lack of awareness and understanding of the condition can make sufferers feel embarrassed by their symptoms, which may appear bizarre and disfiguring. The resulting avoidance of social interaction can be very isolating. Even family, friends and work colleagues may find it difficult to accept the problems faced by patients.¹

Unmet needs

The aim of most research is to support the discovery of improved therapies and ultimately a cure. The Dystonia Medical Research Foundation supports research in furthering the fundamental understanding of dystonia, uncovering the mechanisms in the nervous system that lead to symptoms, creating models of dystonia to use in experiments, and discovering targets for new and improved therapeutics designed specifically to treat dystonia.³

In addition, there is still a significant need to promote awareness and understanding of dystonia amongst the public and the medical profession.¹

Further information

- European Dystonia Federation <http://www.dystonia-europe.org/europe/index.htm>
- National Institute of Neurological Disorders and Stroke <http://www.ninds.nih.gov/disorders/dystonias>
- Dystonia Medical Research Foundation <http://www.dystonia-foundation.org/>
- Brain Research Trust <http://www.brt.org.uk/dystonia>



References

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