

Dystonia – Neurology Services in the UK

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Dystonia?

Dystonia is a movement disorder characterised by, patterned, directional and sustained muscle contractions that produce abnormal, often twisting postures or repetitive movements. (Howard L Geyer and Susan S Bressman 2007)

There is currently no known cure

It is a neurological disorder affecting movement but not intellect.

Dystonia can affect just one part of the body or several different areas



It is an umbrella term covering a number of complex conditions affecting the eyes; face; neck, speech, limbs and trunk. It can be primary or secondary

The word [primary](#) describes a case in which the dystonia is the only neurological disorder that the person has. Primary dystonias include some genetic forms (such as [DYT1 dystonia](#) in which a gene has been identified) and forms for which a cause is not usually found (such as most [focal dystonias](#)).

Genetic dystonias may be described using certain terms: Before any of the dystonia genes were discovered, the word [idiopathic](#) was often used to describe the forms that were presumed to be genetic. By definition, idiopathic means "of unknown cause." Today, if a person's dystonia is described as *idiopathic*, that simply means that the cause is not known—it cannot be attributed to trauma, drug exposure, a gene mutation, or another disease or condition.

Secondary dystonias are often accompanied by other neurological problems. They begin suddenly at rest and are associated with different hereditary and environmental causes. Environmental causes include head trauma, stroke, a tumour, multiple sclerosis, infections in the brain, injury to the spinal cord, or after chemotherapy, drugs or toxins that affect the basal ganglia, thalamus or brain stem.

They may be associated with other hereditary neurological syndromes. Dystonia may be the first sign in a patient with [Huntington's disease](#), and is secondary to many other neurological diseases. These include [Parkinson's disease](#), [Wilson's disease](#) and Ataxia telangiectasia. Examples of metabolic disorders causing secondary dystonia are Lesch-Nehlan syndrome, Niemann-Pick disease and Leigh's disease. All of these causes are rare.

'Dystonia-plus' syndromes Dopamine, (often called 'dopa' which is in fact an intermediate chemical in dopamine's production) is a chemical messenger widely used in the nervous system in passing nerve impulses between nerve cells (neurotransmission). Dopa-responsive dystonia is an important form that can be successfully treated with drugs such as levodopa (eg [Madopar](#), [Sinemet](#)). Typically it begins in childhood or adolescence and leads to progressive difficulty in walking and in some cases spasticity (limb stiffness). The symptoms may fluctuate during the day from relative mobility in the morning to increasingly worse disability in the afternoon, evening and after exercise.

Generalised dystonia also known as primary torsion dystonia or dystonia musculorum deformans. The usual age of onset is between 5 and 16 years. Parents or teachers may notice an abnormal turning in of the foot, an awkward gait or contractions of many different muscle groups. The involuntary dystonic movements may progress quickly to involve all the limbs and torso, but the rate of progression usually slows after adolescence. A genetic basis for generalised dystonia has now been confirmed.

Focal dystonias - Spasmodic torticollis; Blepharospasm; Hemifacial spasm; Oromandibular dystonia; Orofacial-buccal dystonia (Meiges or Brueghels syndrome); **Spasmodic dysphonia; Writer's cramp; Adult-onset primary dystonia** This is a rare subtype of focal dystonia localised to the trunk of the body, but may spread to involve the neck muscles. The dystonia does not spread to the leg. Unlike other forms of focal dystonia it is more common in men than women. The twisting trunk movements have been likened to the Leaning Tower of Pisa,

Incidence and Prevalence

- ✿ It is thought that there are more than 40,000 with Dystonia in the UK that is 1:1500 but some neurologists and epidemiologist suggest it could be higher
- ✿ It is estimated to be about ten times more common than Motor Neurone Disease



The approximate number of those in the UK with dystonia is probably at least **40,000**. This figure is likely to be much higher than this possibly 80 – 120,000

These can be broken down to specific types of dystonia these are approximate figures as not all are known to Doctors as they may not be diagnosed or receiving treatment:

Cervical Dystonias **15000**

Blepharospasm **10000**

Writer's cramp and other limb dystonia **10000 (75% with writers cramp)**

Generalised **1000**

Laryngeal/Oromandibular/others **5000**

Treatment Options

- ♣ **Drug Therapy**
- ♣ **Botulinum Toxin Injections**
- ♣ **Surgical Intervention**
- ♣ **Physiotherapy**
- ♣ **Coping strategies**
- ♣ **Cognitive Behavioural Therapy**
- ♣ **Complementary Therapies**

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Benzhexol (Trihexyphenidyl), Benztropine, Diazepam, Baclofen
Drug therapy works on interfering with neurotransmitters actions on the muscles. They tend to work better in children than adults. Most of the drugs that can help have very unpleasant side effects and need to be given in very low doses and slowly increased. Some patients find the side effects worse than the condition itself.

Botulinum toxin injected into the muscles blocks neurotransmission and stops muscle action

Surgical intervention – Deep brain stimulation implanted into Globus Pallidus to interfere with nerve pathways; selective denervation strips out the specific nerves which are acting on the muscles in a permanent Botox effect.

Physiotherapy – This needs to be provided by specialist Neuro-Physiotherapist, 2 studies going on in Glasgow and Aberdeen looking at the methods developed by Jean Pierre Bleton in Paris

Geste antagoniste

Cognitive Behavioural Therapy

Relaxation, Shiatsu, Massage

Treatment Centres across the UK

- ♣ 128 clinics treating blepharospasm
- ♣ 108 clinics treating cervical dystonia or spasmodic torticollis
- ♣ 28 clinics treating laryngeal dystonia
- ♣ 15 centres implanting DBS for dystonia for adults
- ♣ 9 centres implanting DBS for dystonia in children
- ♣ 1 paediatric Complex Motor Disorders team
- ♣ 1 centre undertaking Selective Peripheral Denervation for cervical dystonia
- ♣ 65 members of Dystonia Nurses Network

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We have tried to map the services available and we hope to expand on this further once we have had an opportunity to analyse the data from the responses to a questionnaire which we sent out to our members. We had 60% response rate. Some centres only treat specific types of dystonia whereas others treat all.

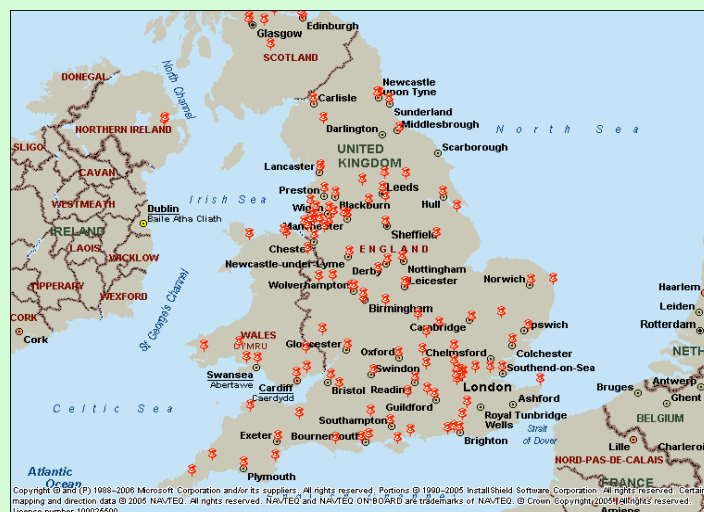
The services on offer from these centres vary some centres within DGH's have General Neurology clinics where those with dystonia are diagnosed and treated in the same clinics as those with other neurological conditions; some attend movement disorder clinics where they are diagnosed and treated with other movement disorders; some may attend specific dystonia clinics where they receive services from a Movement Disorder neurologist or Specialist ENT Consultant and / or a nurse or therapist, where they can meet others with the same type of dystonia and they benefit from a more holistic approach to care.

Some of our members value being treated at a specialist centre as these are seen as centre however these are often teaching hospitals where they don't see the same person more than once, consultations take place with a large entourage of junior doctors and medical students in the consulting room at the same time and this can be quite intimidating and people do not feel able to ask questions to clarify information given.

An example of good care is where people are regularly treated at a local hospital or in their own home by a clinician skilled and competent to deliver that care we currently know of only one service like this and this is from the Walkergate Park Centre in Newcastle.

Many movement disorder neurologist we have spoken to have suggested it is helpful to have a diagnosis at a specialist centre but once the treatment regime has been shown to be effective it would be ideal if this could be regularly provided more locally by a nurse or therapist. Some centres provide a service whereby the consultant undertakes the initial diagnosis and treatment and then the nurse or therapist continues with the treatment regime referring the patient back to the consultant for review. We know of several centres where this happens including: Hope Hospital, Salford; Musgrove Park Hospital, Taunton; Walkergate Park Centre in Newcastle; Oxford Eye Hospital; and Hurstwood Park Neurological Centre, Hayward's Heath. Although this list is not exhaustive.

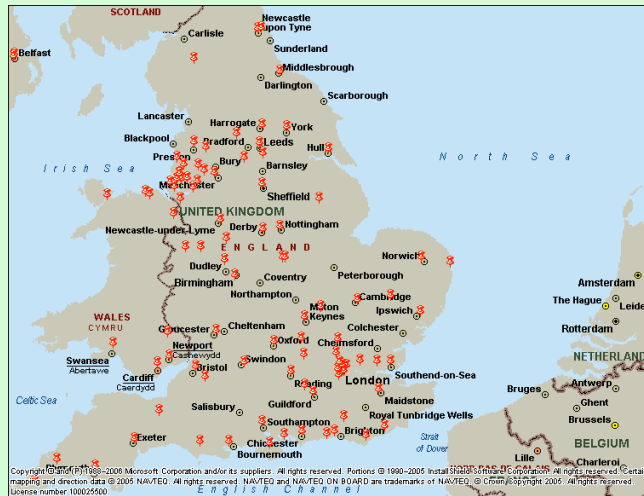
Blepharospasm Clinics



We also know that in some centres this is not the case. Treatments are given in very busy clinics where there is no time to ask questions on either side, people then experience long time intervals between treatments which leads to an increase in symptoms. The recommended interval is about 12 weeks but often because of the pressure on clinics this can be much longer

As far as DBS for dystonia for adults is concerned access and provision varies across the UK as funding is increasingly becoming a problem. In some areas there appears to be no problem however in other areas there appears to be a postcode lottery. Funding decisions in the form of refusals are being made by people in PCT's who do not understand dystonia and how beneficial DBS can be for these people and this statement is based on discussions I have had with them. Because this treatment options has only been available for about ten years there is not a huge amount of evidence available which can make it difficult to challenge these decisions. However the dystonia society are in the process of commissioning a service evaluation of DBS for dystonia and we hope that all 15 centres will participate. It is hoped that this will provide up to ten years worth of evidence of the outcomes of this procedure.

Cervical Dystonia Clinics in England & Wales



As far as DBS for children is concerned there are 9 centres providing this service however the Evelina Children's hospital have developed a multi-disciplinary team specialising in Complex Motor Disorders in Children and young people. This team led by Dr Jean-Pierre Lin has a dedicated physio; OT; SALT; specialist nurse; and psychologist. This team provide a holistic service from initial assessment and to discharge from the service which is likely to be for several years. This service focuses not just at the medical model of care but more holistically at all other aspects of the needs of this group. Education, socialisation and family life. This service is for are children and young people who have either primary or secondary dystonia including metabolic conditions.

Laryngeal Dystonia Clinics



The one centre providing selective peripheral denervation surgery for cervical dystonia is in Derriford Hospital Plymouth and takes referrals from all over the UK. It is a NICE approved procedure so there is no difficulty with funding. Mr James Palmer who carries out this procedure suggests that there is a 40% chance of eradicating the effects of dystonia. As well as the surgery there is a neurophysiologist and physiotherapists to provide follow-up care including exercises to build up the strength of their neck muscles prior to discharge. Mr Palmer believes there are approximately 400 patients who would benefit from this procedure.

The dystonia nurses network is made up of nurses working with patients with dystonia some are movement disorder nurse specialists, ophthalmology nurses, and some are dedicated dystonia nurses. Some inject Botulinum Toxin and provide support etc others just provide support and do not inject. The surgical nurses are mostly Movement Disorder or PD nurses who are involved in the monitoring and programming of the stimulator.

We know of at least one Physiotherapist who injects botulinum toxin. We also know of Speech and Language Therapists who have particular expertise in providing speech therapy for spasmodic dysphonia.

The vision

- ♣ **Short term – early diagnosis; effective management and access to support and services to maximise independence and an improved quality of life.**
- ♣ **Development of a Pathway of Care**
- ♣ **Long term – A CURE**



We need to encourage awareness across health and social care and also other statutory and voluntary services to ensure greater understanding of this complex condition and improved service provision. This includes funding and commissioning of services.

People affected by dystonia want to be treated with dignity and respect and to be accepted in society and not to be treated as a freak. They want to be able to work or go to school and college and to contribute to the family budget and family life.

We plan to develop a pathway of care from onset of symptoms to effective management of the condition and its impact on their life including education, work and finance and socialising.

Most of all our members want a cure for dystonia and the Dystonia Society support research into new treatments and to finding a cure.