

Dystonia: A Guide to Good Practice for Health and Social Care Professionals

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Foreword

Dystonia causes involuntary and sometimes painful muscle spasms as a result of incorrect signals from the brain. These muscle spasms can force affected parts of the body into abnormal movements or postures. Dystonia can affect speech, sight and mobility.

There are thought to be over 70,000 adults and children in the UK who have some form of dystonia. At present there is no cure, although there are treatments which can help to alleviate symptoms. As a health or social care professional, it is likely that you encounter dystonia: it is estimated that there are at least 7 people with dystonia in each GP practice.

Expert care is vital at every stage in helping those with dystonia to achieve the best possible quality of life. This document provides:

- An authoritative guide to best practice for health/social care professionals who are involved in the treatment and support of people with dystonia and their families.
- Guidance for managers responsible for configuring health services on the key issues relating to dystonia and best practice for delivery of services.
- Information about the impact of dystonia on people's lives – to give an insight into common problems and to enable the professionals involved to take positive steps towards resolving them.

Although an overview of diagnostic and treatment processes for dystonia is included, this is intended as general information only and not suitable to be used as an algorithm to support the work of specialists such as neurologists and others responsible for treating and diagnosing dystonia.

Why is this guide important?

The Dystonia Society's principal aim is to ensure that everyone affected by dystonia has access to the most appropriate treatment, support and information. It also works to increase awareness of this condition amongst health professionals and the public.

We know from our members and from callers to our helpline that the condition is often poorly understood. It can sometimes take many frustrating years to get an accurate diagnosis. Once a diagnosis has been achieved, there may be difficulties in accessing proper treatment. This guide has been compiled with the help of many medical, social care and educational experts who, like us, wish to ensure that these problems become a thing of the past, and that everyone who has dystonia gets the care and support they need.

Note: This document was published in 2011 and will be reprinted as new information or recommendations become available. The most up to date version is available on the Dystonia Society's website: www.dystonia.org.uk

To find out more about dystonia, contact the Dystonia Society www.dystonia.org.uk or go to the online learning module devised by the Dystonia Society and British Medical Journal. Enter learning.bmj.com in any browser to go to the BMJ website. Once registered, you will find the dystonia module under Neurological Problems. The module will help you to understand some of the common forms of dystonia and treatments.

SECTION 1 About dystonia

1.1 Dystonia in brief

Description

A neurological condition characterised by involuntary and sustained muscle spasms which can force affected parts of the body into abnormal movements or postures. Dystonia is an umbrella term covering a number of conditions. In most cases, it affects the motor pathways in the brain that control recruitment and movement of muscles causing them to perform an action that leads to unwanted spasms.

Impact

Dystonia leads to sustained abnormal postures or repetitive movements of the affected body part. Typically, the abnormal postures are not fixed, and slow writhing movements can occur (athetosis) where the dominant muscle activity switches from agonist to antagonist and back again. Dystonia can affect movement, posture, speech, sight and mobility but not intellect. Living with dystonia can be painful and debilitating, as well as embarrassing and stigmatising. Work, social activities and quality of life may be significantly impacted.

Pain

The experience of pain in dystonia varies between patients. Some experience extremely intense levels of pain, others very little. However, serious levels of pain are reported by large numbers of patients, most commonly in cervical dystonia, and pain management is an important aspect of treatment.

Parts of body affected

The most common dystonias affect the neck or eyes. Others parts of the body affected include trunk, limbs, hand, voice, mouth and tongue. Adult-onset dystonia is usually focal, affecting one or two parts of the body, while early-onset dystonia often generalises to affect multiple parts of the body.

Cause

Causes vary and include gene mutations, brain lesions, premature birth, disorders of body biochemistry known as inborn errors of metabolism, and exposure to drugs or chemicals. Some focal dystonias may arise from repetitive activity leading to over-excitability of the regions of the brain associated with the muscles involved. However, a high proportion of cases have no identifiable cause.

Pathology

Most of the clinical evidence points to the basal ganglia as the site of pathology in dystonia. The basal ganglia are situated at the base of the forebrain and are associated with a variety of functions, including motor control (Warner & Bressman 2007).

Diagnosis

Cases of dystonia often require specialists to establish the specific diagnosis, but the hallmarks of dystonia, irrespective of cause, are identifiable by all health professionals. Depending on the part of the body affected, diagnosis is usually by either a neurologist specialising in movement disorders or another specialist such as an ear, nose and throat surgeon. Dystonia often goes unrecognised by healthcare professionals and frequently patients are not diagnosed for many years after symptom onset. GPs and other medical professionals therefore need to be alert to the possible symptoms of dystonia so they can make prompt referrals to the relevant specialist.

Sleep

Dystonia is usually abolished by sleep and returns immediately on waking, except a rare form called dopa-responsive dystonia in which there may be marked fluctuations in function throughout the day: very good in the morning but worsening during the course of the day, but improved by sleep.

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Prognosis

Rarely fatal but there is currently no cure. However, in many cases it can be effectively managed with botulinum toxin, medication, surgery and other therapies.

Incidence

Not precisely known, but the Dystonia Society believes that there are at least 70,000 people in the UK affected by dystonia. This equates to a prevalence of 1 in 900. At least 8,000 of these are children and young people. Defazio (2010) suggests that, based on minimum prevalence estimates, primary dystonia should be considered the third most frequent movement disorder after essential tremor and Parkinson’s disease.

History

Dystonia has been known about for over 100 years. For some time it was thought to be of psychiatric origin and patients were given various psychiatric treatments. Only in recent decades has it been recognised as an organic neurological disorder (Warner & Bressman 2007).

1.2 Classification

Classification of dystonia is based on 3 axes (Albanese et al 2010)

1. By cause

PRIMARY DYSTONIAS

Primary pure dystonias: torsion dystonia is the only clinical sign (apart from tremor), and there is no identifiable exogenous cause or other inherited or degenerative disease.

Primary plus dystonias: torsion dystonia is a prominent sign but is associated with another movement disorder, for example myoclonus or parkinsonism. There is no evidence of neurodegeneration.

Primary paroxysmal dystonia: torsion dystonia occurs in brief episodes with normality in between. These disorders are classified as idiopathic (often familial although sporadic cases also occur) and symptomatic because of a variety of causes.

Heredodegenerative dystonias: dystonia is a feature, amongst other neurological signs, of a heredodegenerative disorder. Example: Wilson’s disease.

SECONDARY DYSTONIAS

Dystonia is a symptom of an identified neurological condition, such as a focal brain lesion, exposure to drugs or chemicals. Examples: dystonia due to brain tumour, off-period dystonia in Parkinson’s disease, tardive dystonia which is drug-induced, some forms of cerebral palsy including premature delivery and birth injuries, metabolic disorders.

2. By age at onset

EARLY-ONSET (variably defined as 2–30 years):

Usually starts in a leg or arm and frequently progresses to involve other limbs and the trunk.

LATE-ONSET

Usually starts in the neck (including the larynx), the cranial muscles or one arm. Tends to remain localised with restricted progression to adjacent muscles.

SECTION 1 About dystonia

3. By distribution

FOCAL: single body region

SEGMENTAL: contiguous body regions

Example: cranial and cervical, cervical and upper limb

MULTIFOCAL: non-contiguous body regions

Example: upper and lower limb, cranial and upper limb

GENERALISED: both legs and at least one other body region (usually one or both arms)

HEMIDYSTONIA: half of the body

(usually secondary to structural lesion in contra-lateral basal ganglia)

Patients describe experiencing dystonia

“ It felt like my head was trying to twist down my back.

“ The pain is like having a red hot poker in my neck.

The psychological and emotional pain was so engulfing – it seemed unending.

I feel I have been evicted from my own life.

SECTION 1 About dystonia

1.3 Types of dystonia by classification and symptoms

TYPE OF DYSTONIA	USUAL CAUSE	DISTRIBUTION	USUAL AGE OF ONSET	MUSCLES AFFECTED	SYMPTOMS
Primary generalised dystonia	Primary Pure	Generalised	Early onset	Throughout the body, particularly the trunk	▪ Turning in or dragging of foot or leg ▪ Clumsy or unsteady walking ▪ Painful twisting postures ▪ Can lead to permanent deformity ▪ Spread to involve other parts of body
Cervical dystonia	Primary Pure	Focal (sometimes part of multifocal or segmental)	Late onset	Neck	▪ Causes head to twist ▪ Can be extremely painful ▪ Often associated with tremor
Blepharospasm	Primary Pure	Focal (sometimes part of multifocal or segmental)	Late onset	Around the eyes	▪ Excessive blinking ▪ In more severe cases, eyes can spontaneously clamp shut
Oromandibular dystonia	Primary Pure	Focal (sometimes part of multifocal or segmental)	Late onset	Jaw, tongue and/or mouth	▪ Strange movements of face and mouth ▪ In some cases, eating / swallowing difficult
Laryngeal dystonia or spasmodic dysphonia	Primary Pure	Focal (sometimes part of multifocal or segmental)	Late onset	Vocal cords	▪ Affects speech – voice either strangled or breathy
Focal hand dystonia	Primary Pure	Focal	Late onset	Forearm and/or hand (also called writer’s cramp)	▪ Hand and/or fingers contort, twist or go into spasm when used ▪ Often specific to tasks
Myoclonus dystonia	Primary Plus	Multifocal	Early onset	Neck, trunk and arms	▪ Jerking movements combined with other symptoms of dystonia
Dopa-responsive dystonia	Primary Plus	Generalised	Early onset	Throughout the body, particularly trunk and legs	▪ Turning in or dragging of foot or leg ▪ Clumsy or unsteady walking /affects mobility ▪ Painful twisting postures ▪ Symptoms worsen as day goes on ▪ Good response to drug levo-dopa
Paroxysmal dystonia	Primary Paroxysmal	Focal or generalised	Early onset	All or part of the body	▪ Episodes during which dystonia affects the body – often hemidystonia or generalised ▪ Episodes last from minutes to hours ▪ Between episodes no sign of a problem
Tardive dystonia	Secondary (caused by drugs)	Usually multifocal	Late onset	One or more of face, neck, tongue, trunk, arm, leg	▪ Face and/or tongue movements ▪ Spasms of trunk, neck, arm and/or leg
Secondary dystonia	Many causes	Any site singly or in combination	Any age	From focal to total body involvement with difficulty speaking and feeding	▪ Spasms of face, trunk and/or limbs ▪ Difficulty feeding, sitting, lying, sleeping ▪ Difficulty with speech or unable to speak

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1.4 Early-onset dystonia

Age of onset is sub-classified as follows:

- Infantile dystonia: first symptoms start before age 2 years
- Childhood dystonia: first symptoms start between ages 2–12 years
- Juvenile dystonia: first symptoms start between ages 13–25 years

Early-onset primary (or idiopathic) dystonias

Early onset primary dystonias tend to be generalised and are most often of genetic origin. There are currently at least 12 known types of dystonia caused by gene mutations, of which 7 or 8 affect children, including:

- Most commonly, a mutation in the DYT1 gene which causes primary generalised dystonia
- A mutation in the DYT5 gene which causes Dopa-responsive dystonia, treatable with L-dopa

It is hoped that scientists will identify more dystonia genes as this may lead to greater understanding and treatment of what are predominantly non-degenerative conditions (Carr 2009).

These genetic forms of dystonia can occur in a sporadic, autosomal dominant, autosomal recessive or x linked manner. Heritable childhood onset dystonia is common amongst some ethnic groups (for instance DYT1 dystonia is more common in Ashkenazi Jewish people).

If a parent is found to be positive for a DYT1 gene mutation, this does not mean that their children will automatically get dystonia. The child may not inherit the gene and, even if the child does, they will not necessarily develop dystonia as penetrance is low – often only 30% in most genetic types of dystonia (Warner & Bressman 2007). It is important for the parents to speak to a geneticist before any decisions are made about future children, in order to be able to make an informed decision.

Secondary dystonia

Early-onset secondary dystonia can result from a wide variety of neurological conditions or inherited metabolic defects. Symptoms begin suddenly at rest and are associated with different hereditary and environmental causes.

Environmental causes include head trauma, stroke, brain tumour, infections in the brain, injury to the spinal cord and a variety of drugs or toxins that affect the basal ganglia, thalamus or brain stem. 15% of children and young people who have cerebral palsy may have persistent dystonic symptoms (Carr 2009). Dystonic symptoms may be overlooked and underdiagnosed (Lin, 2011; McClelland 2011).

Dystonia symptoms may be associated with other hereditary neurological syndromes such as Huntington's disease, Wilson's disease and Ataxia telangiectasia. Metabolic disorders causing secondary dystonia are Lesch-Nehan syndrome, Niemann-Pick disease, Leigh's disease and Hallervorden-Spatz Disease.

All of these conditions are very rare (Edwards, Quinn & Bhatia 2008) in adult practice but collectively more common in children's hospitals with neurological, neurodisability and metabolic services.

For more information about how dystonia affects children and young people, see section 7.6.

SECTION 1 About dystonia

1.5 Treatments in brief For more information see sections 3.3 and 3.4.

While there is as yet no cure for dystonia, there are a number of different treatments that can be offered. These vary according to the type of dystonia, and the individual requirements and responses of the patient.

Botulinum Toxin injections

In most cases of focal dystonia in adults, the usual treatment is regular injections of botulinum toxin into the affected muscles. The frequency of injections is usually around every 12 weeks. Botulinum toxin affects the nerves at their junction with the muscle. It prevents the release of acetylcholine from the nerve endings and thereby prevents the involuntary muscle contractions.

Drug treatments

Oral medications are often the mainstay of treatment for treating dystonia in children and young people. They can tolerate higher doses than adults, but efficacy is variable, and several medications may be required in combination or in series to achieve symptom control at the expense of reduced alertness and increased somnolence. There is no one drug that is the definitive treatment and other approaches may enable effective management (see section 3.4 for more information). Drugs are sometimes also used to treat dystonia in adults. However documentation of benefit in well-designed studies is lacking (see section 3.3 for more information).

Surgery

Where all other treatments have failed to provide adequate improvement, Deep Brain Stimulation (DBS) is considered a good option. In this procedure, two fine electrodes are inserted into the brain powered by a battery implanted in the chest. The electrodes send a pulse that blocks the signals from the brain that cause the involuntary muscle spasms. Some other surgical treatments are also occasionally used (see section 3.3 for more information).

Therapeutic interventions

A number of studies have reported motor improvement in patients with writer's cramp and other forms of focal dystonia following physical treatment and sensory and motor retraining.

Cognitive Behavioural Therapy (CBT)

There is currently little research evidence about the use of CBT in dystonia, but the principles on which CBT is based suggest that it may be helpful in the management of some cases of dystonia. It may also help associated symptoms such as depression, anxiety, anger, sleep problems and chronic pain. Currently, CBT should be classified as experimental treatment.

Young people and parents describe experiences of early-onset dystonia



My daughter's tongue was going into spasm which caused problems with eating and speaking.

The spasms in my arm stopped me writing – I had to teach myself to write left handed. Eventually, they were so bad I stopped going to school and stayed in my room.

The symptoms started at 12 and it severely dented my self-esteem.

SECTION 2 Identifying and referring cases

2.1 The role of non-specialist medical professionals in identifying dystonia

Getting an accurate diagnosis of dystonia can be difficult and can take many years because of a lack of awareness amongst the public and medical professionals. People affected by dystonia are often referred to, and receive inappropriate treatment from, a variety of specialists such as physiotherapists and psychiatrists before getting a correct diagnosis. Non-specialist medical professionals have a role in addressing this:

General Practitioner

The GP is usually the first person to be approached by someone who develops symptoms of dystonia. The GP's key role is to recognise that the patient in their surgery may have a movement disorder and to make a prompt referral to a specialist. Dystonia is something which all GPs need to be aware of. The Dystonia Society estimates that each GP practice has an average of 7 patients affected by dystonia at any time of whom at least one-third will be undiagnosed.

Dystonia can be difficult to diagnose because of its variability in presentation, the wide spectrum of causes and the coexistence of other movement disorders. It is often mistaken for a number of other more common conditions or is assumed to be psychological. It is therefore vital that GPs are alert to the symptoms and willing to consider movement disorders if treatment for other conditions does not work or if symptoms persist. If in doubt, it can be helpful to ask the patient to prepare a symptom diary including drug, lifestyle and dietary changes to aid diagnosis and eliminate other conditions. Section 2.3 shows the symptoms for the different types of dystonia and indicators that may prevent misdiagnoses.

Physiotherapist

Physiotherapists may frequently see patients with dystonia before they are diagnosed. Cervical dystonia, in particular, is often misdiagnosed as a neck injury and referred for physiotherapy. Some physiotherapy treatments such as deep massage and traction can aggravate dystonia and increase pain and spasms in the affected muscles. Physiotherapists need to be able to spot the signs of dystonia to avoid treating inappropriately.

Psychiatrist/psychologist/counsellor

Dystonias are frequently misdiagnosed as psychological problems. In these cases, the client may be sent for psychiatric assessment or for therapeutic support. Therapists and psychiatrists therefore need to be aware that the presumed diagnosis may be incorrect and should be alert to signs of dystonia. Psychiatrists also need to be aware that dystonia can be triggered by drugs used to manage psychiatric symptoms, including dopamine receptor blockers. It is therefore vital that baseline assessments of movement are made before the patient starts a course of these drugs. Regular reassessment is required throughout the duration of the medication to ensure that any symptoms are identified quickly and appropriate changes in medication are made to eliminate or reduce the symptoms. The longer these drugs are causing symptoms of dystonia or dyskinesia, the less likely it is that the symptoms will be reversed. Where signs of dystonia appear, prompt referral needs to be made to a neurologist specialising in these disorders.

Other medical professionals

Other medical professionals may also encounter someone with undiagnosed dystonia when caring for them because of another health problem. If they notice strange muscle spasms, tremors or body movements, it is important that the patient is encouraged to seek a referral through their GP to a neurologist for diagnosis and management.

SECTION 2 Identifying and referring cases

2.2 Referral

If a GP or other medical professional suspects a neurological movement disorder, they should enquire about general health, approximate timing of onset of the problem and note which body parts are affected, exclude serious co-existing medical disorders and offer support. A referral should be made to the following specialists who will be responsible for making the diagnosis and treating dystonia:

Type of Dystonia	Referral to:
Laryngeal dystonia	Laryngologist or ENT (Ear, Nose and Throat) Surgeon
Blepharospasm	Neurologist specialising in movement disorders or an ophthalmologist
Oromandibular dystonia	Neurologist specialising in movement disorders or an ENT (Ear, Nose and Throat) Surgeon
Any dystonia affecting children	Paediatric movement disorder neurologist or paediatric neurologist
All other types of dystonia	Neurologist specialising in movement disorders

People with dystonia describe the experience of seeking a diagnosis



My foot started twisting in when I was 14. I spent the next 7 years being told my symptoms were psychosomatic during which time the symptoms spread to my legs, arms and neck.

My doctor told me it was all in my head. He made me feel that I was mentally ill.

When I got the diagnosis after 5 years, I was full of joy knowing it wasn't my imagination. I really did have an illness.

SECTION 2 Identifying and referring cases

2.3 Common GP misdiagnoses / indicators to avoid misdiagnoses

TYPE OF DYSTONIA	MUSCLES AFFECTED	SYMPTOMS
Primary generalised dystonia	Throughout the body, particularly the trunk and limbs	▪ Turning in or dragging of foot or leg ▪ Clumsy or unsteady walking ▪ Painful twisting postures
Cervical dystonia	Neck	▪ Causes head to twist ▪ Can be extremely painful ▪ Often associated with tremor
Blepharospasm	Around the eyes	▪ Excessive blinking ▪ Usually present in both eyes ▪ In more severe cases, eyes can spontaneously clamp shut
Oromandibular dystonia	Jaw, tongue and mouth	▪ Strange movements of face and mouth ▪ In some cases, eating / swallowing difficult ▪ Can be jaw open / jaw closed mix ▪ Tongue protrusion or twisting
Laryngeal dystonia or spasmodic dysphonia	Vocal cords	▪ Affects speech – voice either strangled or breathy
Focal hand dystonia	Forearm / hand (also called writer’s cramp)	▪ Hand / fingers contort, twist or go into spasm when used ▪ Often specific to tasks
Myoclonus dystonia	Neck, trunk and arms	▪ Jerking movements combined with other symptoms of dystonia
Dopa-responsive dystonia	Throughout the body, particularly the trunk and legs	▪ Turning in or dragging of foot or leg ▪ Clumsy or unsteady walking / affects mobility ▪ Painful twisting postures ▪ Symptoms worsen as day progresses
Paroxysmal dystonia	All or part of the body	▪ Episodes of dystonia or dyskinesia lasting minutes to hours ▪ Between episodes no sign of a problem
Tardive dystonia	One or more of face, tongue, trunk, neck, arm, leg	▪ Face and/or tongue movements ▪ Spasms of trunk, neck, arm and/or leg

SECTION 2 Identifying and referring cases

SYMPTOMS COMMONLY MISDIAGNOSED AS:	INDICATORS FOR A GP THAT DIAGNOSIS MAY BE DYSTONIA
Orthopaedic problem Psychological problem	▪ Movement worse at certain times of day or when stressed and tired ▪ Movements often not present when sleeping ▪ Does not respond well to splinting or other corrective therapeutic techniques ▪ Movement still present when patient unaware of being observed
Neck damage, Head trauma Pulled muscle/muscle strain Slept the wrong way Psychological problem	▪ Does not respond to physiotherapy or pain killers ▪ Does not clear up over time ▪ Symptoms sometimes ease with sensory tricks (such as putting a finger on the chin) ▪ Movement still present when patient unaware of being observed
Dry eye, Eye strain / tired eyes / tic (eg. from overuse of computer or reading) Myasthenia Gravis Psychological problem	▪ Does not respond to dry eye treatment ▪ Does not clear up over time ▪ Dark glasses may ease symptoms ▪ Symptoms sometimes respond to sensory tricks such as touching the corner of the eye
Damage to the jaw, Tic Bruxism (teeth grinding) Dental problem, Orthopaedic or Psychological problem	▪ Does not respond to orthopaedic procedures ▪ No damage shows up on x-rays ▪ Does not clear up over time ▪ Movement still present when patient unaware of being observed
Throat cancer, Acid reflux, Nerve damage Psychological problem	▪ Tests or scans of throat negative ▪ Does not clear up over time
Tendonitis, Orthopaedic or Psychological problem	▪ Not usually present all the time ▪ Tends to be task specific such as writing or playing a musical instrument
Psychological problem	▪ Family history of similar problems ▪ Writers cramp quite common ▪ May be associated with compulsive disorders
Orthopaedic problem Psychological problem	▪ Movement better in the morning and worse at end of the day or when stressed and tired ▪ Movements often not present when sleeping ▪ Other family members affected ▪ Movement still present when patient unaware of being observed
Psychological problem Epilepsy, Stroke	▪ Family history of similar problems ▪ Patient usually does not lose consciousness during an episode
Psychological problem Epilepsy, Chorea	▪ History of having been prescribed antipsychotic or antiemetic medication ▪ History of illegal drug taking

SECTION 3 Diagnosing and treating dystonia

3.1 The role of the consultant neurologist or other specialist

Diagnosis and treatment of dystonia is usually the responsibility of a consultant neurologist specialising in movement disorders. Most consultant neurologists have a good knowledge and understanding of the common neurological conditions, including movement disorders such as dystonia. However not all provide treatment for all types of dystonia and patients are sometimes referred to tertiary centres, particularly for the rarer forms of dystonia. Some examples are listed below:

Type of Dystonia	Diagnosis and treatment by:
Laryngeal dystonia	Laryngologist or ENT (Ear, Nose and Throat) Surgeon
Blepharospasm	Neurologist specialising in movement disorders or an ophthalmologist
Oromandibular dystonia	Neurologist specialising in movement disorders or an ENT (Ear, Nose and Throat) Surgeon
Any dystonia affecting children	Paediatric movement disorder neurologist or paediatric neurologist

Consultant neurologists work in collaborative networks with specialist nurses, therapists and other physicians and surgeons, with the neurologist providing clinical leadership in these teams. This ensures a holistic approach to care. Once the diagnosis has been made, treatment can often more effectively be provided by other members of the team, leaving the consultant neurologist free to manage the more complex cases.

3.2 Diagnosis

Dystonia can be difficult to diagnose because of its variability in presentation, wide spectrum of causes and coexistence with other movement disorders. Diagnosis is based on clinical presentation. The core manifestation is abnormal postures and involuntary muscle spasms (with or without tremor) and the recognition of specific features eg. ‘geste antagoniste’, overflow and mirror movements. It is also a case of eliminating other conditions. The classification of dystonia is important for providing appropriate management, prognostic information, genetic counselling and treatment. It is important to be aware that some psychogenic disorders have symptoms which can appear similar to dystonia and these need to be eliminated.

The neurologist will take a detailed history about the onset of symptoms and their impact and carefully observe the patient to see how the dystonia is affecting them. The observation will be done whilst the neurologist is chatting to the patient perhaps gathering other information. For instance:

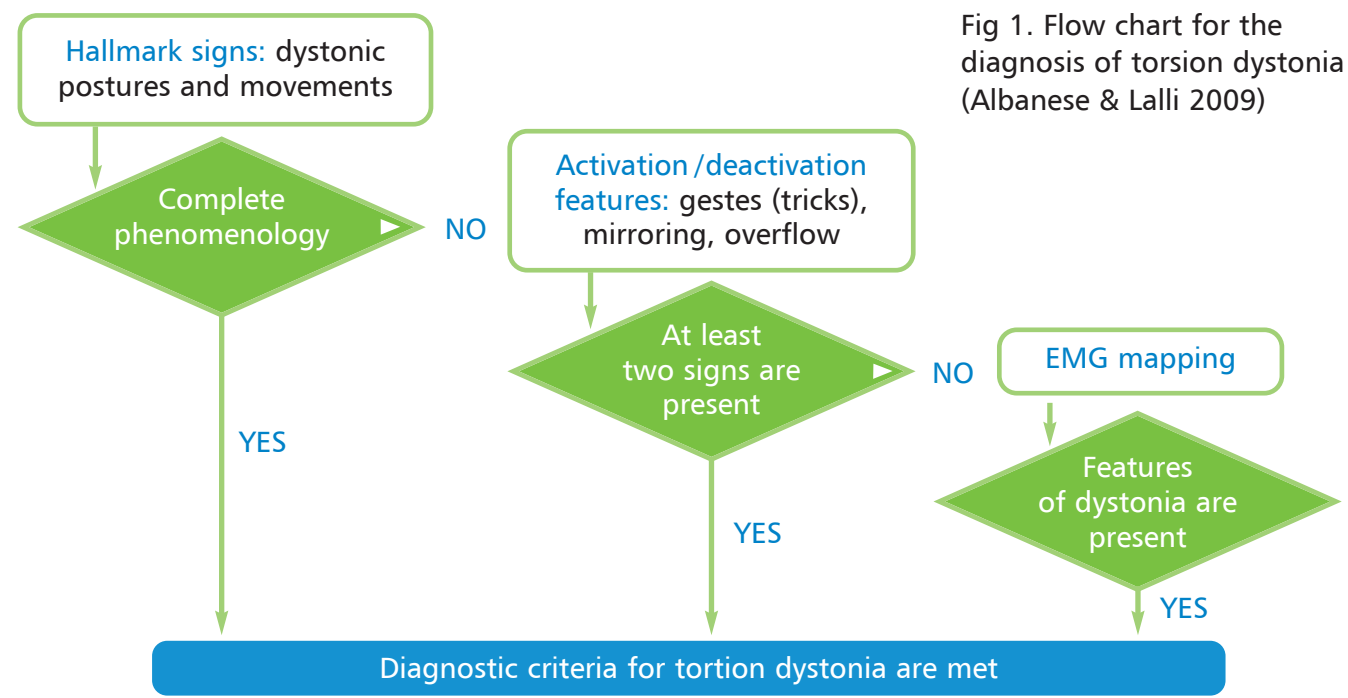
- With a possible case of cervical dystonia, the neurologist will be noting how the head is held and moved and what muscles are obviously pulling and twisting the neck.
- If checking for blepharospasm, observation will be how and when the eyelids are closing and what is happening with the muscles of the face and eyelids.

Structural brain imaging (MRI) is required in generalised or hemidystonia and if there are any features to suggest a secondary form of dystonia. However, MRI is not routinely required when there is a confident diagnosis of primary focal dystonia in adult patients, as this is almost always a normal study.

Neurophysiological tests are not routinely recommended for the diagnosis or classification of dystonia. However, multiple simultaneous electromyography (EMG) recordings from various muscles may contribute to the clinical assessment by showing characteristic features of dystonia.

SECTION 3 Diagnosing and treating dystonia

Assessment of dystonia may be performed using a validated rating scale (eg. Burke-Fahn-Marsden Dystonia Rating Scale, TWSTRS, CDIP-58). They are most useful for measuring the effectiveness of certain treatments such as deep brain stimulation. The use of a structured flow chart such as the example shown in (Fig.1) below may increase diagnostic accuracy.



Other tests can be carried out but may not be routinely used unless diagnosis is proving difficult or in the case of rarer conditions, these may also be used where other members of the family may also be affected. These can include those recommended by Albanese et al 2006 & 2010, for instance:

- Pre-synaptic dopaminergic scan can be useful to differentiate between dopa-responsive dystonia (DRD) and juvenile Parkinson’s disease. This can also be useful to distinguish dystonic tremor from parkinsonian tremor.
- Appropriate investigations are required if the initial presentation or the course of the symptoms suggest hereditary or secondary symptomatic dystonia
- Testing of DYT1 gene in individuals with young onset dystonia can prevent the need for other more invasive tests. Testing of family members of an index case with DYT1 dystonia should only be done with appropriate genetic counselling.
- DYT6 testing can be helpful in early-onset dystonia or familial dystonia with cranio-cervical predominance.
- Individuals with early-onset myoclonus dystonia affecting the arms, neck or trunk, particularly if positive for autosomal-dominant inheritance, may lead to testing for the DYT11 gene.
- Testing for the PNKD gene (DYT8) can be helpful in symptomatic individuals with PNKD.

Diagnosis of children

The diagnosis of dystonia in children can be difficult and, as with adults, it is most often a case of eliminating other conditions. For this reason a comprehensive holistic history and assessment of the child and the family must take place so that an accurate diagnosis can be made. This should usually be done in a specialist paediatric movement disorder clinic so that the subsequent treatment plan can be put into effect as soon as possible. Parents should be kept well-informed and supported throughout this process.

SECTION 3 Diagnosing and treating dystonia

Giving a diagnosis

Many patients receiving a diagnosis will know little if anything about dystonia. Sufficient time should be allowed during the consultation for a careful explanation. The patient's key concerns are likely to be:

- What is the condition and what is the likely cause?
- What is the prognosis? Will it get better or worse?
- Is there any immediate medical management available to help relieve my symptoms?
- What will the longer term management involve?
- Where will I be treated? Will it be locally or at a larger centre some distance away?
- Where can I get more information about dystonia?
- Are there sources of support if I need it?
- When the diagnosis has sunk in, who can answer any other questions I might have?

Before giving the diagnosis, the clinician and his team should therefore prepare for the consultation to ensure that the relevant information and support are available should they be needed. The Dystonia Society can supply relevant leaflets to clinics.

3.3 Treatment for adults

Botulinum Toxin injections

In most cases of focal dystonia, the usual treatment is regular botulinum toxin injections into the affected muscles, usually around every 12 weeks. Botulinum toxin affects the nerves at their junction with the muscle. It prevents the release of acetylcholine from the nerve endings and thereby prevents the involuntary muscle contractions.

The frequency can vary for some forms of dystonia at the consultant's clinical discretion. It should not be used more often than every 8 weeks as there is an increased risk of antibody development.

- Botulinum Toxin (BoNT) A (or type B if there is resistance to type A) can be regarded as first-line treatment for primary cervical dystonia and blepharospasm.
- BoNT-A can be effective for writer's cramp and is probably effective in other types of upper limb dystonia, but often EMG guided injections are required to pinpoint the overactive muscles.
- BoNT-A is usually effective for adductor-type and abductor-type laryngeal dystonia. However in mixed or atypical abductor laryngeal dystonia it does not work well or consistently.

Notes:

- BoNT injections are relatively safe and efficacious when repeated treatments of recommended doses are performed over many years but excessive doses result in increased risk of side effects at each session. Cumulative doses can result in antibody formation. Doctors should refer to Summaries of Product Characteristics for information on indications and dosing etc.
- BoNT injections are usually performed by direct clinical assessment; EMG or ultrasound -assisted targeting may improve clinical outcome.
- BoNT should not be used in patients affected by neuromuscular junction abnormalities or if there is local infection at the injection site. The recommended dosage should not be exceeded.
- BoNT is not licensed for treatment of dystonia in children (see section 3.4).

SECTION 3 Diagnosing and treating dystonia

Drug treatments

Levodopa: A diagnostic levodopa trial is warranted in every patient with early-onset dystonia in case they have dopa-responsive dystonia. Following a positive diagnostic trial with levodopa, chronic treatment with levodopa should be initiated and adjusted according to the clinical response.

Anticholinergic agents: The absolute and comparative efficacy and tolerability of anticholinergic agents in dystonia is poorly documented in children and there is no proof of efficacy in adults. Therefore no recommendation can be made to guide prescribing.

Tetrabenazine / benzodiazepines: Frequently used in the treatment of dystonia but documentation of benefit in well-designed studies is lacking.

There is a lack of evidence to give recommendations for the use of antiepileptics.

Surgery

Pallidal Deep Brain Stimulation (DBS)

Considered a good option, particularly for primary generalised or segmental dystonia after medication or botulinum toxin injections have failed to provide adequate improvement. In this procedure, two fine electrodes are inserted into the brain powered by a battery implanted in the chest. The electrodes send a pulse that blocks the signals from the brain that cause the involuntary muscle spasms.

In general, DBS is less effective in secondary dystonia with the exception of tardive dystonia. DBS can have side effects and involves a life-long commitment by the patient (and family) as ongoing follow-up is required and may necessitate travel to a DBS centre. DBS can be also considered for cervical dystonia if other treatments have not worked.

Assessment requires a specialised multi-disciplinary team. Some patients may see DBS as a potential 'cure' and can be very disappointed if after full assessment they are not thought to be a suitable candidate. Care must be therefore taken to manage patient expectations, and the conveying of such a decision must be handled sensitively.

Selective peripheral denervation

An alternative approach to treat medically refractory cervical dystonia. There is a significant risk of recurrence of symptoms. Insufficient evidence exists to use this treatment in primary dystonia but the procedure can be indicated in patients where secondary dystonia is combined with spasticity.

Therapeutic interventions

A number of studies have reported motor improvement in patients with writer's cramp and other forms of focal dystonia following physical treatment and sensory and motor retraining. New randomised controlled studies on these potentially useful interventions for patients with upper limb dystonia may provide more evidence.

Coping strategies

There are a number of techniques called 'geste antagoniste' that can be adopted to help manage symptoms. These can include touching the chin to stop the head turning or tilting in cervical dystonia; and touching the temple in blepharospasm to stop the eyelids closing. For some people, symptoms can also be mitigated through focusing on another activity such as talking or playing a musical instrument. It should be noted however that these coping strategies have not been scrutinised in any formal studies. In addition, some people do find the symptoms come back more aggressively when these techniques have been used.

SECTION 3 Diagnosing and treating dystonia

A note on the use of Cognitive Behavioural Therapy (CBT) in a dystonia treatment plan

People with dystonia who are referred for CBT may be concerned because they believe it to imply that the doctor thinks their dystonia is ‘all in the mind’. Good communication with the patient is therefore essential so that the patient understands why CBT is being recommended. There is currently little research evidence about the use of CBT in dystonia, but the principles on which CBT is based appear to support the theory that it could be helpful in the management of some cases of dystonia. It may also help associated symptoms such as depression, anxiety, anger, sleep problems and chronic pain. CBT should be classified as an experimental treatment.

People with dystonia describe the experience of successful treatment

“ Having botulinum toxin injections has allowed me to work and get on with my life.

Botulinum toxin reduces the rapid movement/ closure of the eyelids and tremor in the hands – giving me more control.

Once the deep brain stimulation was correctly tuned, my spasms stopped and no longer appear to exist.

Getting effective treatment felt like someone had put my brain back in my head.

3.4 Treatment for children

Oral medications are often the mainstay for treating dystonia in children and young people. They may respond well and can tolerate higher doses than adults, but effects may be short-lived or the medication may cause side effects such as somnolence, drooling, poor trunk and neck control and difficulties concentrating in class. Mood and behavioural disturbances may further limit the use of drugs. There is no one drug that is the definitive treatment but often a combination of several drugs and other treatments can enable effective management. When the cause of dystonia is unknown and the brain MRI scan is normal, a trial of levodopa is required to diagnose the rare genetic disorder known as dopa-responsive dystonia which can be managed for years on small doses of levodopa once or twice a day.

All dystonia management should be tailored to individual needs of children and goal-directed under the guidance of experienced doctors and therapists. The Multidisciplinary Team (MDT) is vital to supporting families of children with dystonia and can help create strategies for coping with many essential functions. Some of the management options that may be offered include:

Medication:		
Levodopa	Trihexyphenidyl	Baclofen* (oral and/or intrathecal)
Diazepam*	Lorazepam	Clonazepam
Nitrazepan	Carbamazepine	Tetrabenazine
Chloral hydrate	Clonidine	Dantrolene Sodium

* Limited license for use to treat muscle spasms in children

SECTION 3 Diagnosing and treating dystonia

- **Botulinum toxin A injections**
These are not licensed for children. However, they are often used for management directed at specific muscle groups that interfere with function of the neck, jaw, hands, elbows, hips, knees, ankles or feet. Great care must be exercised to avoid botulism through overdosage or excessively frequent injections. Very disabled children with little or any voluntary movements are particularly vulnerable to respiratory difficulties after excessive botulinum toxin A injections.
- **Intrathecal baclofen (ITB)**
This approach offers regional and total body dystonia control without some of the risks of somnolence attendant on oral medication. This will sometimes be used to treat secondary dystonia not amenable to Deep Brain Stimulation.
- **Deep Brain Stimulation (DBS)**
This is very effective in primary dystonias. It should be recognised that the longer the duration of dystonia, the greater the risk of skeletal deformities, dependency (through lack of opportunity) and potential lowered efficacy of DBS. DBS should be considered when dystonia is rapidly progressive and disabling and when two or more drugs have failed to bring adequate relief of dystonia or the drugs are poorly tolerated. Loss of a major skill such as walking, manual ability, speech or feeding are signs that functional neurosurgery should be considered. Unfortunately, many forms of secondary dystonia that are associated with focal or generalised brain injury will preclude the use of DBS.

Dystonia onset before puberty is particularly disruptive to a child’s growth and development. The stress of dystonia may prevent the onset of puberty. The effect of puberty in a dystonic child may accelerate the appearance of contracture and deformity which further diminishes function and limits opportunity.

Children with primary dystonia may need to be referred to other specialist departments for treatment for conditions that result from dystonia (e.g. musculoskeletal and orthopaedic problems). Those with secondary dystonia are also very likely to be under the care of different teams at different stages. It is essential that full communication is established and maintained between the neurology/movement disorder team and these other specialist teams so that treatment is optimal and appropriate.

3.5 Tertiary-based neurology teams

These are teams who work in specialist centres, often based in teaching hospitals. These centres offer diagnosis, treatment and monitoring of most types of dystonia. For dystonia, the team has one or more neurologists specialising in movement disorders, usually supported by a multidisciplinary team including dystonia nurses.

There are also 17 neurosurgical centres in the UK, of which 8 are designated centres offering Deep Brain Stimulation in both adults and children with dystonia. There is currently only one centre offering Selective Peripheral Denervation for cervical dystonia.

3.6 The role of a dystonia nurse

Dystonia nurses are general nurses who have a detailed knowledge and understanding of dystonia and its management. They provide vital continuity of care as they often see the same patient at each visit and their clinic slots are frequently longer than those of a neurologist. Whitaker et al (2001) states that nurse practitioners provide a more flexible, much appreciated, safe and cost-effective service for this client group. Wider use of outreach nurse practitioners should therefore be encouraged.

SECTION 3 Diagnosing and treating dystonia

Some dystonia nurses are trained to give botulinum toxin injections either in the clinic or as an outreach service. Through continuity, they can learn the specifics of each patient’s treatment, including how best to site the injections for maximal effect with lowest dose. Dystonia nurses are sometimes also able to adjust the dose of botulinum toxin within agreed parameters, if necessary seeking advice from the consultant through agreed protocols (Whitaker et al (2001)). Other dystonia nurses provide support to the consultant in the clinic. They can offer patients counselling and advice either before or after treatment, particularly for those who are newly diagnosed or are experiencing side effects from treatments, or when there is an exacerbation of symptoms.

In centres where DBS (Deep Brain Stimulation) is carried out, patients may be seen by specialist DBS nurses who provide advice, liaise with neurosurgical staff and sometimes make adjustments to the settings of the implanted neuro-stimulator with guidance from the neurologist. The specialist nurse is also very important in managing an Intrathecal Baclofen (ITB) service in children with secondary dystonias not suitable for DBS. The diagram below highlights the roles and responsibilities of the dystonia nurse. Dystonia nurses may do all or some of these roles depending on the requirements of the service.

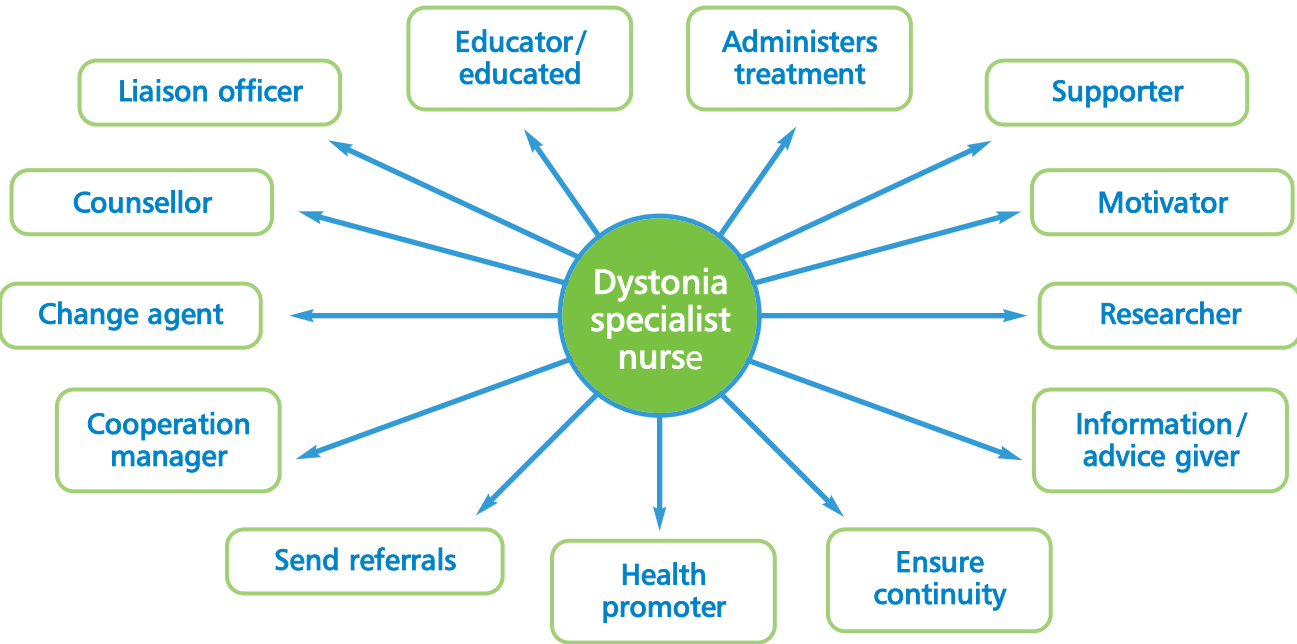


Fig 2. The role and responsibilities of a dystonia specialist nurse (adapted from King (2007))

3.7 Outreach/home visit models

Patients living a significant distance from their treatment centre can experience pain and posture problems during the journey to receive treatment. Outreach services can address this through providing services closer to or in people’s homes. They benefit patients as it means only the specialist clinician has to travel a significant distance.

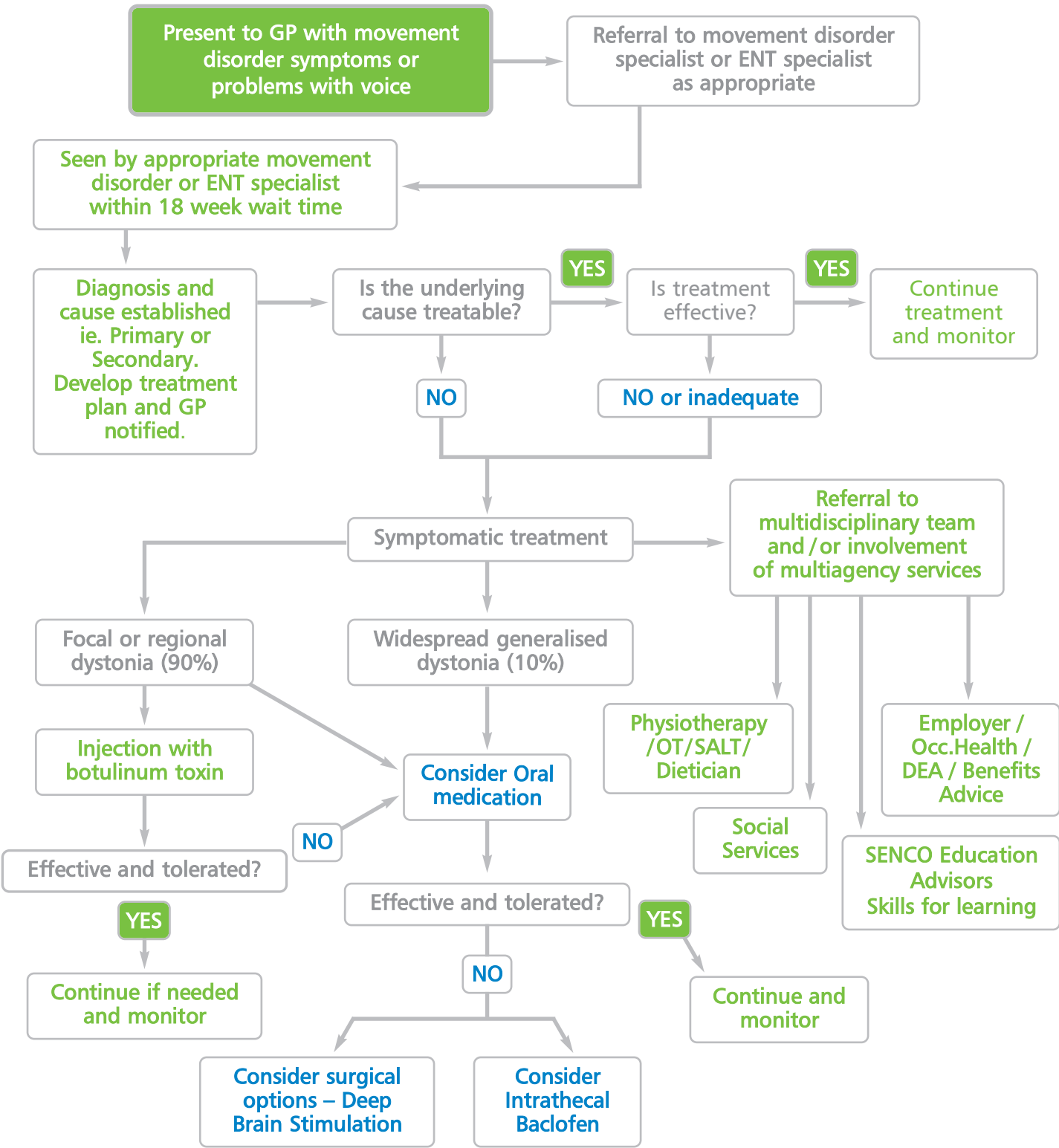
Outreach services for dystonia are usually provided by a specialist nurse or sometimes a neurologist. They are based in local community hospitals and health centres, and occasionally services are delivered in the patients’ own homes. Referral to these services is usually by one of the neurologists at the main treatment centre who do the initial assessment and treatment and then refer for treatment locally.

Local services can also often offer more time for advice and support. In addition, they provide continuity of care as usually the same clinician treats the patient each time. Over time, this enables the clinician to adjust the specifics of their treatment to the patient’s needs, including how best to site the injections for maximal effect with lowest dose.

SECTION 4 Patient pathway flowcharts

4.1 Recommended patient pathway for adults with dystonia

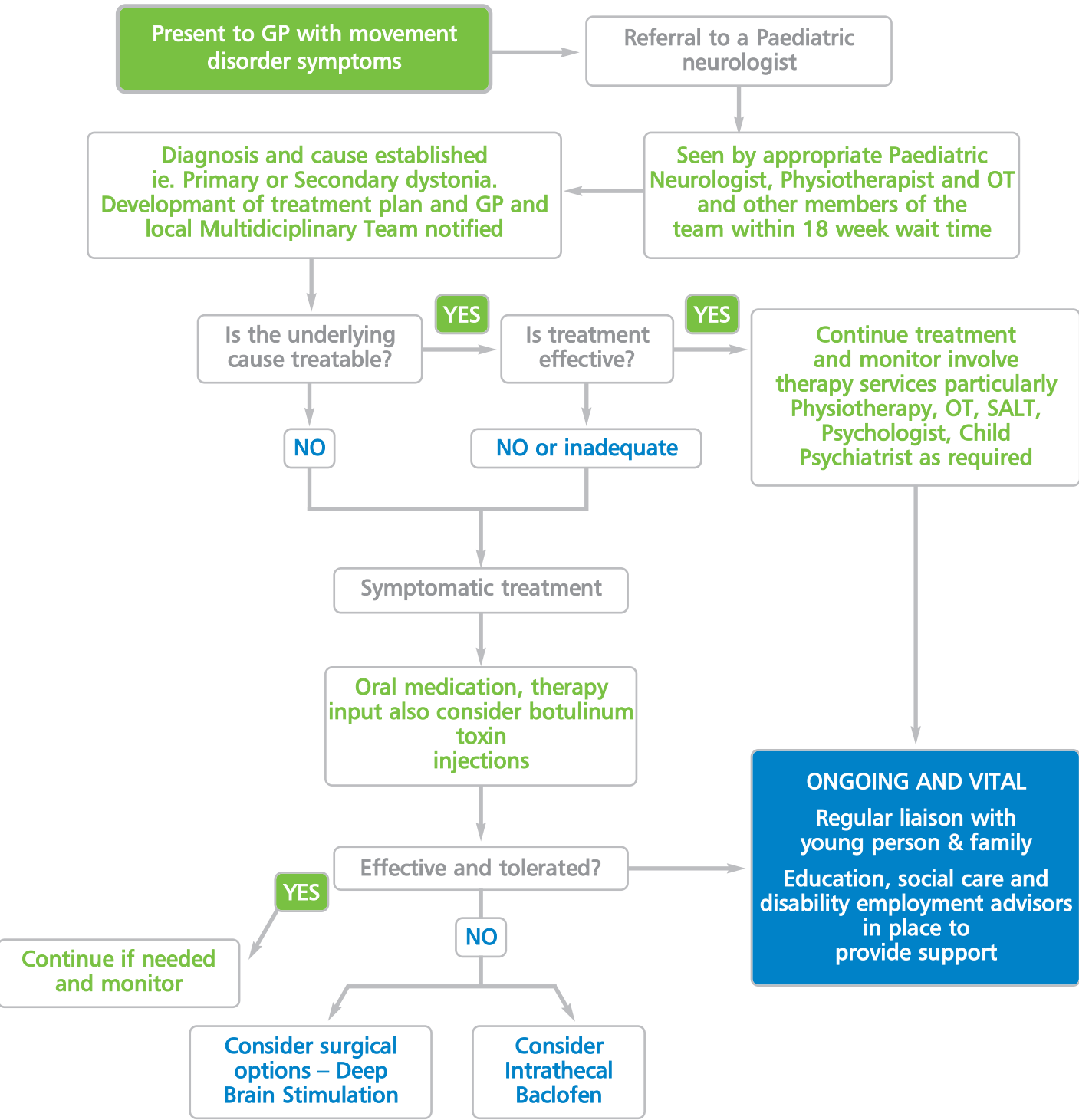
(adapted from NW Neurosciences Partnership 2006)



SECTION 4 Patient pathway flowcharts

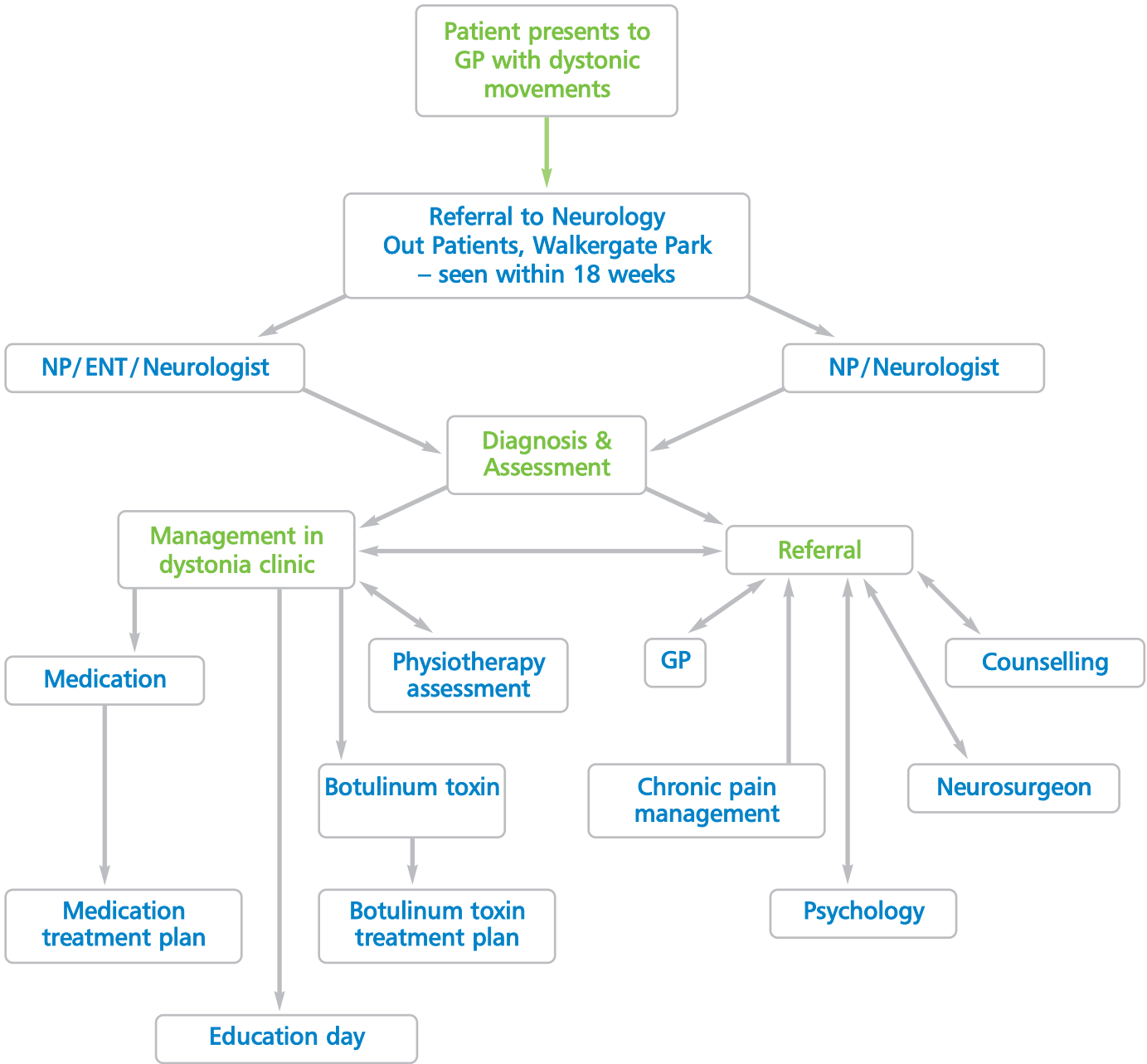
4.2 Recommended patient pathway children and young people with dystonia

(adapted from NW Neurosciences Partnership 2006)



SECTION 4 Patient pathway flowcharts

4.3 Example of specific pathway: Walkergate Park Adult Dystonia Patient Pathway



SECTION 5 Additional support for patients with dystonia

As part of a patient's treatment programme for dystonia, the neurological team will also consider a number of other supplementary treatment options.

5.1 Physiotherapy

For focal dystonias, the use of rehabilitative physiotherapy in treating dystonia is well developed and structured. It aims to give patients as much independence as possible, in a palliative way. The objective of physiotherapy is to correct the affected function through specific interventions. This type of therapy is demanding for both the patient and the therapist (Bleton (2007)).

In some areas Specialist Neurological Physiotherapists have also been trained to offer regular treatment and support for those with dystonia who do not need to be seen by the consultant neurologist. This treatment can include Botulinum toxin injections, physical therapy and counselling, support and advice.

For generalised dystonias, while therapeutic handling strategies can be useful, they tend not to have carryover (ie. when the therapist releases their handling, the dystonic posture /movement tends to return). Physiotherapists usually have a wider, more supportive role including:

- Supporting patient, parents and carers in other settings (eg. school) about aspects including handling, positioning, and adapting activities to promote active participation
- Advising on general and specific exercise to maintain general health and wellbeing or to address specific goals such as reducing pain /discomfort, strengthening and range of movement.
- Promoting functional mobility and advising on equipment such as seating, wheelchairs, mobility aids, leisure equipment
- Identifying issues which may warrant referral to other services such as Orthopaedics for management of contracture and deformity.

5.2 Pain management

Pain resulting from dystonia can be in the muscles affected by spasms, or in joints where bone surfaces rub together due to twisting of posture and limbs. Sometimes, the resulting intractable pain can dominate a patient's life and may be unresponsive to medication including that used to manage dystonia. These patients need to be referred to a pain specialist urgently to reduce the impact on health and wellbeing. Although the British Pain Society guidelines (2007) suggest that patients should only be referred to pain specialists when all other treatments have failed, it is now recognised that a referral should be offered when indicated by persistent pain causing distress, disability, and a negative impact on quality of life.

5.3 Dietary support

Adequate nutrition is essential to the wellbeing of children and adults with severe dystonia. A number of types of dystonia can affect nutrition:

- Dysphagia (difficulty swallowing) can arise from oromandibular and lingual dystonia when it can be difficult to chew or move food around the mouth to prepare it for swallowing.
- Dysphagia can also sometimes be a side effect of botulinum toxin injections for cervical dystonia. It is usually a short lived problem but can be frightening when it happens.

SECTION 5 Additional support for patients with dystonia

- Some people, particularly children with generalised dystonia, may have difficulty with swallowing food safely. They may also use more calories as a result of the muscle spasms.

Dystonia can also lead to an inadequate diet:

- Those who have excessive movements may also find it almost impossible to keep still whilst eating making hand to mouth feeding very difficult. As a result, finger foods and feeding cups are often used but these may not provide enough nutrition.
- Texture modified diets (e.g. puree diets) may be recommended due to dysphagia and can have negative impact on nutritional status due to their poor nutritional content.

Nutrition can be monitored using a screening tool (e.g. MUST) during home/clinic visits or inpatient admissions. A referral to the dietitian should be made if weight loss has occurred or if malnutrition is a concern. A dietetic referral is also required when dysphagia is present. The dietitian can suggest appropriate use of texture modified diets, food fortification and nutritional supplement drinks (eg. Build Up™, Complian™, Ensure Plus™ and Fortisip™) to supplement diet.

5.4 Speech and language therapy

A number of dystonias can affect speech. Where speech difficulties occur referral should be made to a speech and language therapist (SALT):

- Patients with spasmodic dysphonia will be given techniques to help them speak. These can include breathing exercises and ways to make best use of the voice and sound they have.
- Those with oromandibular, lingual and generalised dystonias with articulation difficulties can be given mouth and swallowing exercises to help them reduce the risk of choking by chewing and swallowing safely.

Within the NHS there are very few SALTs who have experience of dystonia so not everyone who may need to see one is referred. However, if a health professional is concerned about a patient's speech and swallowing issues they should always seek the advice of a SALT.

5.5 Occupational therapy

Occupational therapy can help people with dystonia with practical everyday tasks, enabling them to live as independently as possible – at home, in employment or in education. Support can include:

- Identifying ways problematic everyday tasks can be done differently, including recommending alterations or adaptations in the home, school or workplace environment.
- Advice on disability equipment and other aids.
- Assessing the needs of patients who may qualify for a Disabled Facilities Grant towards the cost of adapting their home.
- Referral to other services – for example, speech and language therapy, or employment -related advice centres.

Specialist Neurological OTs actively support and promote best practice in occupational therapy for people with neurological conditions.

SECTION 5 Additional support for patients with dystonia

Specialist equipment

The occupational therapist may make recommendations for specialist equipment. In each area, the local NHS and local authority have a duty to provide certain daily living and mobility equipment including wheelchairs. Provision is based on assessed need and can be for short or long term use. Patients should be discouraged from choosing equipment without being properly assessed as it can result in the equipment not helping or causing more problems than it solves. This is particularly the case with mobility aids and wheelchairs if the user has postural problems. The equipment may seem comfortable at first but it may encourage fixed postures and muscle tightening, leading to more body deformity.

Statutory provision may not cover top-of-the-range bespoke equipment but it should fit the need. Where appropriate, children in particular should be given the option of a motorised chair that they can control themselves to help maintain fine motor skills and increase their sense of independence. In certain situations a voucher may be issued (the NHS contribution) towards the equipment which can be topped up with other funds to pay for higher specification equipment. It may also be possible to apply for financial support towards the cost from charitable funds.

5.6 Podiatry

Patients with dystonia may experience gait problems and struggle to look after their own feet due to mobility and dexterity issues or to problems caused by uncontrollable muscle spasms. Podiatrists help them address these problems using foot orthotics to assist with gait problems.

5.7 Psychological support

Dystonia is not a mental health condition but it can cause severe depression and anxiety due to pain, stigma, employment difficulties and social isolation. Psychological therapies and counselling can therefore play an important role. This can include helping newly-diagnosed patients to deal with the diagnosis, or assisting those already diagnosed to cope with the more stressful aspects of their condition.

Where appropriate, a referral can also be to a clinical neuropsychologist who specialises in treating neurocognitive problems caused by problems with the brain. They bring a psychological viewpoint to treatment, to understand how brain pathology may affect and be affected by psychological factors. They also can offer an opinion as to whether a person is demonstrating difficulties due to brain pathology or as a consequence of emotional or other (potentially) reversible cause. Neuropsychologists can have an impact on the management of dystonia by ensuring that appropriate treatment is being used to manage each problem that may have both a physical and psychological basis.

5.8 Social support

Dystonia can cause difficulties with all or some of the activities of daily living. Where this occurs, patients should be referred to social services for support eg. domiciliary care, equipment or home adaptations. Social care professionals may not have much knowledge of dystonia or how complex and painful the condition can be. The multi-disciplinary team therefore needs to ensure the responsible social workers are aware the kind of help and support that is necessary. A number of other specific social interventions may also be required:

SECTION 5 Additional support for patients with dystonia

- **Visual impairment due to blepharospasm:** A consultant ophthalmologist should complete an assessment to determine if the patient is sight impaired (SI) or severe sight impaired (SSI) and a certificate issued and sent to Social Services. Advice about visual impairment can be obtained from the Royal National Institute for the Blind or Action for Blind People.
- **Problems working:** Referral to an Occupational Health and/or Disability Employment Advisor.
- **Patient unable to work:** Referral to welfare rights officer or Citizens Advice Bureau to complete applications for benefits and allowances.
- **Patient rejected for one or more benefits:** Appeal decision by sending more information about how the condition affects them (patients are often turned down for benefits due to ignorance of the condition).
- **Access further or higher education:** Support from the Disability Advisor or Learning Support Coordinator at the college or university to ensure reasonable adjustments are made.
- **Patient wishes to continue driving:** Neurologist or dystonia nurse to advise if condition stable enough. If so, confirmation to be sent to DVLA.
- **Carer struggling or experiencing mental health problems:** Referral should be made to carer's support organisation.

5.9 Genetic Counselling

Adults who have genetic forms of dystonia and are considering having children may have concerns about their children developing dystonia. They may decide to seek genetic counselling to help inform their decision making. Also where parents have an infant or young child who has dystonia which may have a genetic cause, they may want to seek genetic counselling with regard to future siblings. If they choose to have genetic counselling they should seek the advice of their neurologist/paediatric neurologist and ask for a referral. There may be a wait and the counselling may not be available through the NHS except in exceptional circumstances.

5.10 Complementary therapy

People with dystonia often seek out complementary therapies and report varying degrees of benefit (Lim 2007). These therapies include exercise, acupuncture, chiropractic, massage, homeopathy, yoga, Chinese medicine, osteopathy, reflexology, shiatsu, aromatherapy and meditation. The Alexander Technique has also been found by some to be helpful. Formal studies into the effects of these therapies have been limited, so no recommendations can be given. However, therapies which help to increase relaxation, relieve stress and calm symptoms are felt by some patients to be useful in managing their condition (Lim 2007).

5.11 Additional support for young people with dystonia

Young people may benefit from some or all of the referrals listed above. In addition, they may have the following additional needs.

SECTION 5 Additional support for patients with dystonia

Special Educational Needs (SENCO) support:

SENCOs in nursery, primary and secondary schools can play a crucial role in the early years by:

- Ensuring appropriate educational assessments are carried out
- Putting an individual education plan in place
- Liaising with other specialists (e.g. paediatric occupational therapists) to ensure that suitable equipment and other adjustments are provided
- Training for school staff

Pupils with dystonia can be bullied or ostracised. It is essential that this is addressed proactively, through information-sharing, discussions and briefings. The Dystonia Society can provide guidance if necessary.

Transition from children’s services to adult services

This is an area of particular concern which requires good planning and handling. Moving a young person with dystonia to adult services in health/social care and education can be a fraught process. Outcomes can be mixed if best practice guidelines are not followed. The process should start with a referral to education and social care services when the child turns 13 so that the process can begin in a timely way with regular discussion in planning. Transition is a process spanning the period from 16-25 years of age. Consideration of the young person’s circumstances will determine the precise transition needs, including time-critical educational needs.

5.12 Checklists of additional support

The following support may be appropriate for specific types of dystonia:

TYPE OF DYSTONIA	SUPPORT NEEDED	ACTION	OUTCOME
Generalised Dopa-responsive Cervical Focal hand Tardive Paroxysmal	Exercises to increase or maintain range of movement and mobility	Referral to physiotherapist with expertise in movement disorders	Improved range of movement and mobility
Oromandibular Cervical Generalised	Dietary advice	Referral to dietitian with expertise in dystonia	Improved nutritional status
Laryngeal Oromandibular	Exercises to help with speech	Referral to Speech and Language Therapist (SALT) with expertise in dystonia	Improved voice function/projection
Blepharospasm	Advice about visual impairment	Referral to: www.rnib.org.uk and www.actionforblindpeople.org.uk	
All genetic dystonias	Advice about risks of passing dystonia to children	Referral to a Genetic Counsellor	Informed decision about having children

SECTION 5 Additional support for patients with dystonia

The following support may also be appropriate for all types of dystonia:

PROBLEM	ACTION	OUTCOME
Significant level of pain not helped by current medication	Referral to pain specialist	Pain more effectively managed
Difficulties with gait or other foot problems	Referral to a podiatrist	Improved gait through foot orthotics
Psychological issues	Referral to a neuropsychologist, psychologist or counsellor	Programme such as CBT or counselling improves psychological wellbeing
Difficulty with all or some of the activities of daily living	Referral to social services	Support to enable coping with the activities of daily living
Difficulty working	Referral to Occupational Health and/or Disability Employment Advisor www.dystonia.org.uk	Work arrangements adapted
Unable to work	Assistance from welfare rights officer or Citizens Advice Bureau to complete applications for benefits and allowances www.citizensadvice.org.uk www.dialuk.info	In receipt of all appropriate statutory benefits and allowances
Patient turned down for benefits	Advised to appeal decision by sending more information about how the condition affects them www.citizensadvice.org.uk www.dialuk.info	In receipt of all appropriate statutory benefits and allowances
Patient wishes to continue driving	Neurologist or dystonia nurse to advise if condition stable enough and if so letter/report written confirming this for DVLA www.dft.gov.uk/dvla/	Able to continue to drive once DVLA and motor insurance company have agreed
Carer struggling or experiencing mental health problems	Referral to carer’s organisation www.carersuk.org	Carer receiving necessary support
Young person having difficulties with accessing education	Contact the SENCO at school if under 16 or the Disability Advisor or Learning Support Coordinator if 16+ at the college or university	Able to access education with appropriate support and assistance and provision of equipment and other resources to aid study
Young person is of the age to move towards adult health, social and education services	Referral to education and social care services when child turns 13 so that process can begin in a timely way	Smooth transition to adult services

SECTION 6 Impact of dystonia on other medical support

Dystonia can affect the treatment provided by other medical professionals.

6.1 The role of the GP

Working alongside multidisciplinary neurological team

Ideally treatment for dystonia will be provided by a multi-disciplinary neurological team. In this case, the GP needs to track the progression of the patient's dystonia treatment, usually through clinic letters from the patient's consultant, to ensure that any proposed treatment for other conditions does not conflict with this.

It is important GPs recognise that not every symptom a dystonia patient presents with is necessarily related to the dystonia. There is a danger that delays may occur in diagnosing another perhaps more serious condition if there is a wait while a referral is being made to the neurologist, or if the patient is advised to bring the new symptom up at their next visit to the neurologist.

The GP may also be involved in issuing repeat prescriptions for dystonia-related medication, endorsing patients' applications for welfare benefits, DVLA and blue badge applications etc, in making referrals to other specialist clinicians or service providers, and in assisting with the transition of children to adult services.

Areas where there is no multidisciplinary neurological team

In areas where there is no multi-disciplinary team, the GP may be involved directly in supporting the specialist managing the dystonia. In this case, the GP will need to be aware of the additional referrals appropriate to best practice in managing dystonia (see section 5).

6.2 Radiography

Radiographers may encounter patients with dystonia when they come in for radiological tests such as x-rays and scans in connection with the dystonia or for some other health issue.

These patients may have great difficulty getting into the correct position or keeping still during the procedure. It is important that the radiographer is sympathetic and talks to the patient and to others involved in their care, ideally in advance of the appointment.

They should find out what helps or aggravates their movements and spasms, whether they need supports to help posture or to alleviate spasms, and whether there is a better time of day to carry out the procedure. Some patients may require sedation to be able to keep still for the procedure.

6.3 Dentistry

Dentists need to understand the nature of the condition and how it affects the patient's ability to sit still and open their mouth. They may need to assist by helping them to find a comfortable position to reduce muscle spasms during examination and treatment.

Some forms of dystonia, such as oromandibular and lingual dystonia, can cause the lining of the mouth and the tongue to become damaged by uncontrolled chewing and biting. This leads to ulceration and infection which can be very painful. These forms of dystonia can also make opening the mouth in a controlled way very difficult and as a result having a dental examination and treatment is almost impossible. It may require treatment to be provided under sedation or general anaesthetic, with the attendant risks discussed if necessary in advance with relevant medical professionals.

SECTION 6 Impact of dystonia on other medical support

Dentists need also to be aware of particular issues relating to patients who have had DBS (Deep Brain Stimulation). Diathermy is contra-indicated, and some patients may require prophylactic antibiotics to prevent infection. Specialist advice should be sought from the patient's DBS nurse if necessary.

6.4 Psychiatry

Tardive dystonias can be caused by some anti-psychotic medication. It is therefore likely that many people with tardive dystonia will also be under the care of a psychiatrist. For all types of dystonia, care needs to be taken when psychiatric treatment is being planned as some medication can have an impact on existing dystonia.

To find out more about dystonia, contact the Dystonia Society www.dystonia.org.uk or go to the online learning module devised by the Dystonia Society and British Medical Journal. Enter learning.bmj.com in any browser to go to the BMJ website. Once registered, you will find the dystonia module under Neurological problems. The module will help you to understand some of the common forms of dystonia and treatments.

SECTION 7 The social impact of dystonia

Dystonia has an impact beyond the symptoms and pain observed in medical consultations. A study undertaken in 2006 showed that dystonia had a significant impact on health-related quality of life including deterioration in physical functioning, general health, vitality and social functioning. Also reported by some clients were mental health issues including depression and anxiety (Lim 2007).

7.1 Isolation and feeling ostracised

In 2008, the Dystonia Society conducted a questionnaire to which 1800 members responded. Findings included:

- 75% felt that dystonia had an impact on their social life.
- 60% were worried about its impact on their family and friends.
- 80% felt very self-conscious about how dystonia makes them look.

Description of the effects of dystonia included:

- Feeling that they 'just want to hide away from others' because they have been bullied and teased by people who do not understand why they move oddly or have strange facial expressions.
- Unable to go out alone because either they are too unsteady or they are simply afraid to leave the house. Some struggle because of the pain and fatigue caused by dystonia which makes them feel worthless.
- Finding that going into supermarkets and department stores can be quite intimidating as managers and staff treat them as if they are drunk or up to no-good. They may be watched, followed round or asked to leave because they are upsetting other shoppers.

7.2 Problems with driving

For people with blepharospasm, being able to continue to drive legally can be a problem. They will need to satisfy both their insurance company and the DVLA that they are safe to drive. This can be straightforward, but sometimes there are problems because the correct paperwork is not available to prove that they can keep their eyes open safely while driving. The same situation can apply to those with severe cervical dystonia if they are unable to maintain a safe driving position or are unable to turn their head when manoeuvring.

7.3 Difficulties with welfare benefits

People with dystonia are often turned down for benefits due to lack of understanding about the conditions by the claims officer making the assessment. It is a significant challenge to explain the impact of dystonia on a complex and rigid benefits application form. It is therefore essential that as much information as possible about how dystonia affects that individual is included in the application. Most rejections are reversed on appeal but the process often generates significant anxiety.

7.4 Difficulties with personal care

People with dystonia can sometimes have difficulty with simple daily tasks, such as washing, dressing, eating, drinking and going to the toilet. In addition, inability to keep the mouth, head or tongue still or the whole body from writhing can create difficulties going to the dentist, optician, podiatrist /chiropodist, hairdresser or beautician. This can on occasion result in neglected dental care, poor foot care, limited vision and poor body image.

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7.5 Impact on employment

In the Dystonia Society's 2008 survey, 39% of respondents reported that they were not in work. Of those in work, 38% said they could only work part of a day while others had to have one or two rests during the day. Some lost their job because it was simply too dangerous for them to continue.

7.6 The social impact of dystonia on children and young people

In addition to the difficulties already mentioned, particular challenges faced by children and young people include:

Education

- The physical challenge of coping with education while experiencing spasms, pain and fatigue or side effects from medication. Most are able to attend mainstream education, but coping with a full day or carrying heavy bags around can be challenging.
- Getting adequate support especially in primary and secondary schools. It is often difficult to get an agreed statement of Special Educational Needs (SEN). Even where a SEN is in place, the provision of extra classroom assistance and other services varies widely between local authorities.
- Isolation and bullying. Some respondents to our questionnaire report difficulties because other children do not understand their disability or because they are seen as different.

Social activity

- Restrictions on leisure activity: Young people report being less active than their friends because they lack the physical ability or are simply too tired or are in pain. Some find they have to plan outings carefully so they do not get into awkward situations or get too tired.
- Access to special equipment: Many children and young people report difficulty accessing equipment such as wheelchairs in a timely way. With growing bodies and changing postures, equipment quickly becomes unsuitable and the assessment process has to be repeated.
- Lack of independence: Young people often have to rely on parents to travel/go out. This issue increases as they get older.
- Stigma: Young people with dystonia report sometimes being refused service in shops, pubs and restaurants because people do not understand the odd movements and think they are drunk.

People with dystonia describe their experience of social isolation



I found difficulty going out and about. It required determination as I experienced lots of insulting behaviour from members of the public.

I finally made myself go out but then a man started laughing at me about my jerky movements. I went home immediately and didn't go out again for days.

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7.7 The impact of dystonia on parents and carers

In 2009, the Dystonia Society conducted a questionnaire of parents of those with dystonia. This showed clearly that the impact on carers of a child or young person with dystonia is often enormous. Common issues include:

- Giving up work or being made redundant because they took too much time off to care for their child.
- Difficulty juggling full-time work with taking their child to frequent hospital and clinic visits, especially when these are far from home.
- Impossible to have a full family life. Many have had to completely re-adjust their family and working life to ensure that their child is properly cared for.
- For many parents or carers, these problems were at their worst while trying to get an adequate diagnosis and effective treatment.

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SECTION 9 Further sources of information

www.dystonia.org.uk

The Dystonia Society's website.

learning.bmj.com

Access to the British Medical Journal's online dystonia learning module produced in association with The Dystonia Society.

www.dystonia-foundation.org.com

Dystonia Medical Research Foundation website.

www.emedicine.medscape.com

'Dystonias' E A Moberg-Wolff, A R Thiyagarajah and S A Barna
13th January 2010.

www.direct.gov.uk

Government website for access to information about welfare benefits and disability rights including access to education.

www.citizensadvice.org.uk

The website of the Citizens Advice Bureau who can help with getting benefits and advice.

www.dialuk.info

The website of the Disability Information Advice Lines which can help you to access information in your area about benefits and other support.

www.rnib.org.uk

The website of the Royal National Institute of Blind which can help with registering as sight impaired.

www.actionforblindpeople.org.uk

A useful website for those with a visual impairment.

www.dft.gov.uk/dvla/

The website of the Driver Vehicle and Licensing Agency for information about driving with a medical condition.

www.medicalert.org.uk

The website of the MedicAlert Foundation which provides necklets and bracelets for people with medical conditions.

www.carersuk.org

A useful website for carers of those with a disability.

Dystonia Society Helpline: 0845 458 6322

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*This report was researched and compiled by **Val Wells**, Service Development Manager for The Dystonia Society.*

SECTION 11 About The Dystonia Society

Your notes:

The Dystonia Society exists to promote the welfare of people who are affected by any form of the neurological movement disorder known as dystonia. The Society aims to do this by promoting awareness of the disorder, by supporting research and by undertaking welfare initiatives. It does this on a national level and through its network of

The Society is a charity and was established in 1983 by a small group of people affected by dystonia, with the support of the late Professor David Marsden. It has since grown significantly in size and ambition. Those on its Board of Trustees are elected by the membership of the charity and the Society has a dedicated staff team to carry out the day to day work of the organisation. The Society also has three patrons and a Royal patron who provide invaluable support. It receives little government funding and relies almost entirely on voluntary donations to fund its services.

The organisation currently has four principal areas of focus:

1. Improving access to treatment services
2. Providing improved support and information for individuals affected by dystonia
3. Progressing research activities
4. Raising awareness of the condition

Among its many activities, the Society runs a busy helpline and website, organises 'Living with Dystonia' days around the UK, and works proactively with government departments and health commissioners to improve services for those with dystonia. It funds research into dystonia, and its conferences attract leading clinical speakers and members from all over the country.

The Society has forged links with many senior neurologists and movement disorder specialists in the UK, and with dystonia organisations in Europe and the United States. It is a leading member of the European Dystonia Federation.

The Dystonia Society

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Your notes:

