

# Dystonia: A Guide to Best Practice

For health and social  
care professionals

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# Foreword

Dystonia is a neurological condition causing involuntary and sometimes painful muscle spasms resulting in abnormal movements or postures. It can affect movement, posture, speech, visual performance and mobility. There are thought to be over 70,000 adults and children in the UK who have some form of dystonia.

## 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 enable the professionals involved to take positive steps towards resolving them.

## Some experiences of dystonia

“My dystonia made me wonder whether I could carry on. I lost my job as my neck was resting on my right shoulder and the pain was excruciating.

*I could deal with the condition and people staring if I was not so tired and had no pain. I feel like dystonia is a person that has taken over my life.*

*I would love to have my life back – I have two small children, and I don't want them to say when they're older, we couldn't do this or that because my mum was always unwell.*

# SECTION 1

## Introduction to dystonia

### 1.1 Dystonia: a short overview

#### 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, visual performance and mobility. 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.

#### Parts of body affected

In adults, the most common dystonias affect the neck or eyes. Other 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.

#### Pain

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

#### Psychological impact

In the vast majority of cases, dystonia is an organic condition. However, prevalences of depression, anxiety and obsessive-compulsive disorder amongst those with dystonia are often higher than that expected in the general population. The cause of these psychiatric symptoms is thought to be both secondary to the dystonic symptoms and also caused by disruption to normal brain circuitry. Stress and anxiety can also exacerbate the physical symptoms. Neuropsychiatric features are also now recognised in children and young people with some 'combined' dystonias such as some types of dystonia-myoclonus.

#### 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

A majority of clinical evidence points to the basal ganglia as the site of pathology in dystonia. However, studies have also identified the cerebellum as playing a causal role.

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# Introduction to dystonia

### Diagnosis

People with dystonia often require specialists to establish the specific diagnosis, but the hallmarks of dystonia, irrespective of cause, should be 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 or ophthalmologist.

### Sleep

Dystonia is usually abolished by sleep and returns immediately on waking except sleep can sometimes be interrupted by the pain resulting from dystonia. An exception to this pattern is 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.

### Prognosis

Currently there is no cure but dystonia is very rarely fatal. The condition can be effectively managed with medication, botulinum toxin injections, surgery and other therapies. Spontaneous remissions have been reported in a small minority of cases. There is no cure, but most types of dystonia do not affect lifespan.

### 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. Defazio (2010) suggests that, based on minimum prevalence estimates, idiopathic and inherited dystonias combined should be considered the third most frequent movement disorder after essential tremor and Parkinson's disease.

### History

The term dystonia was first used to describe childhood torsion disorders by Oppenheim in 1911. Over the years, the condition has been variously understood as a psychological disorder or as a pathology of the basal ganglia. Since the mid 1970s and primarily as a result of the work of Dr David Marsden and Dr Stanley Fahn, there is now a unanimous view that focal and generalised dystonia are organic brain disorders.

## 1.2 How dystonia is classified

The classification of dystonia was revised in 2013 (Albanese 2013). The new classification is based on 2 axes: clinical and aetiological.

### Clinical axis

#### Time of onset

**EARLY ONSET DYSTONIA** appearing up to the age of 20 (often generalises to affect multiple body areas). Broken into sub-categories as follows:

INFANCY (0–2 years)

CHILDHOOD (3–12 years)

ADOLESCENCE (13–20 years)

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**ADULT ONSET DYSTONIA** appearing after the age of 20 (usually remains localised with restricted progression to adjacent muscles). Broken into sub-categories as follows:

EARLY ADULTHOOD (21–40 years)

LATE ADULTHOOD (>40 years)

### Body 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:** the trunk and at least two other sites  
*(often includes arms and/or legs).*

**HEMIDYSTONIA:** half of the body  
*(usually secondary to structural lesion in contra-lateral basal ganglia).*

### Temporal pattern

**PERSISTENT:** symptoms persist to approximately same extent throughout the day.

**ACTION-SPECIFIC:** symptoms only present when performing a particular task  
*(such as writing or playing music).*

**DIURNAL:** symptoms fluctuate through the day in a regular circadian pattern.

**PAROXYSMAL:** symptoms occur in brief episodes with normality in between.

### Isolated / Combined

**ISOLATED:** torsion dystonia does not appear in combination with any other movement disorder (with the exception of tremor).

**COMBINED:** torsion dystonia combined with other movement disorders  
*(such as myoclonus or parkinsonism).* There is no evidence of neurodegeneration.

## Aetiological axis

### Inheritance

**INHERITED:** linked to a known gene locus or identified genetic defect.

**ACQUIRED:** dystonia has an identified environmental cause such as a brain lesion, another neurological disorder or 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, neurodegenerative and metabolic disorders.*

**IDIOPATHIC:** dystonia has no identified cause.

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### 1.3 Types of dystonia by classification and symptoms

TYPE OF DYSTONIA	CLASSIFICATION	DISTRIBUTION	USUAL AGE OF ONSET	MUSCLES AFFECTED	SYMPTOMS
<b>Generalised dystonia</b>	Isolated, Persistent Inherited or idiopathic	Generalised	Early onset	Throughout the body, particularly the trunk	<ul style="list-style-type: none"> <li>● Turning in or dragging of foot or leg</li> <li>● Clumsy or unsteady walking</li> <li>● Painful twisting postures</li> <li>● Can lead to permanent deformity</li> <li>● Spread to involve other parts of body</li> </ul>
<b>Cervical dystonia</b>	Isolated, Persistent Mainly idiopathic	Focal (sometimes part of multifocal or segmental)	Adult onset	Neck	<ul style="list-style-type: none"> <li>● Causes head to twist ● Can be extremely painful</li> <li>● Often associated with tremor</li> </ul>
<b>Blepharospasm</b>	Isolated, Persistent Mainly idiopathic	Focal (sometimes part of multifocal or segmental)	Late adulthood	Around the eyes	<ul style="list-style-type: none"> <li>● Excessive blinking ● In more severe cases, eyes can spontaneously clamp shut</li> </ul>
<b>Oromandibular dystonia</b>	Isolated, Persistent Mainly idiopathic	Focal (sometimes part of multifocal or segmental)	Adult onset	Jaw, tongue and/or mouth	<ul style="list-style-type: none"> <li>● Strange movements of face and mouth</li> <li>● In some cases, eating / swallowing difficult</li> </ul>
<b>Laryngeal dystonia or spasmodic dysphonia</b>	Isolated, Persistent Mainly idiopathic	Focal (sometimes part of multifocal or segmental)	Adult onset	Vocal cords	<ul style="list-style-type: none"> <li>● Affects speech – voice either strangled or breathy</li> </ul>
<b>Focal hand dystonia</b>	Isolated, Task-specific Mainly idiopathic	Focal	Adult onset	Forearm and/or hand (also called writer's cramp)	<ul style="list-style-type: none"> <li>● Hand and/or fingers contort, twist or go into spasm when used ● Often specific to tasks</li> </ul>
<b>Myoclonus dystonia</b>	Combined, Persistent Inherited or idiopathic	Multifocal	Early onset	Neck, trunk and arms	<ul style="list-style-type: none"> <li>● Jerking movements combined with other symptoms of dystonia</li> </ul>
<b>Dopa-responsive dystonia</b>	Isolated, Diurnal Mainly inherited	Generalised	Early onset	Throughout the body, particularly trunk and legs	<ul style="list-style-type: none"> <li>● Turning in or dragging of foot or leg</li> <li>● Clumsy or unsteady walking / affects mobility</li> <li>● Painful twisting postures ● Symptoms worsen as day goes on ● Good response to drug levo-dopa</li> </ul>
<b>Paroxysmal dystonia</b>	Isolated, Paroxysmal Inherited or idiopathic	Focal or generalised	Early onset	All or part of the body	<ul style="list-style-type: none"> <li>● Episodes during which dystonia affects the body – often hemidystonia or generalised</li> <li>● Episodes last from minutes to hours</li> <li>● Between episodes no sign of a problem</li> </ul>
<b>Tardive dystonia</b>	Isolated, Persistent Acquired (caused by drugs)	Usually multifocal	Usually adult onset	One or more of face, neck, tongue, trunk, arm, leg	<ul style="list-style-type: none"> <li>● Face and/or tongue movements</li> <li>● Spasms of trunk, neck, arm and/or leg</li> </ul>
<b>Acquired dystonia</b>	Isolated or combined Persistent, Acquired	Any site singly or in combination	Any age	From focal to total body involvement with difficulty speaking and feeding	<ul style="list-style-type: none"> <li>● Spasms of face, trunk and/or limbs</li> <li>● Difficulty feeding, sitting, lying, sleeping</li> <li>● Difficulty with speech or unable to speak</li> </ul>

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USUAL AGE OF ONSET	MUSCLES AFFECTED	SYMPTOMS
Early onset	Throughout the body, particularly the trunk	<ul style="list-style-type: none"> <li>● Turning in or dragging of foot or leg</li> <li>● Clumsy or unsteady walking</li> <li>● Painful twisting postures</li> <li>● Can lead to permanent deformity</li> <li>● Spread to involve other parts of body</li> </ul>
Adult onset	Neck	<ul style="list-style-type: none"> <li>● Causes head to twist ● Can be extremely painful</li> <li>● Often associated with tremor</li> </ul>
Late adulthood	Around the eyes	<ul style="list-style-type: none"> <li>● Excessive blinking ● In more severe cases, eyes can spontaneously clamp shut</li> </ul>
Adult onset	Jaw, tongue and/or mouth	<ul style="list-style-type: none"> <li>● Strange movements of face and mouth</li> <li>● In some cases, eating / swallowing difficult</li> </ul>
Adult onset	Vocal cords	<ul style="list-style-type: none"> <li>● Affects speech – voice either strangled or breathy</li> </ul>
Adult onset	Forearm and/or hand (also called writer's cramp)	<ul style="list-style-type: none"> <li>● Hand and/or fingers contort, twist or go into spasm when used ● Often specific to tasks</li> </ul>
Early onset	Neck, trunk and arms	<ul style="list-style-type: none"> <li>● Jerking movements combined with other symptoms of dystonia</li> </ul>
Early onset	Throughout the body, particularly trunk and legs	<ul style="list-style-type: none"> <li>● Turning in or dragging of foot or leg</li> <li>● Clumsy or unsteady walking / affects mobility</li> <li>● Painful twisting postures ● Symptoms worsen as day goes on ● Good response to drug levo-dopa</li> </ul>
Early onset	All or part of the body	<ul style="list-style-type: none"> <li>● Episodes during which dystonia affects the body – often hemidystonia or generalised</li> <li>● Episodes last from minutes to hours</li> <li>● Between episodes no sign of a problem</li> </ul>
Usually adult onset	One or more of face, neck, tongue, trunk, arm, leg	<ul style="list-style-type: none"> <li>● Face and/or tongue movements</li> <li>● Spasms of trunk, neck, arm and/or leg</li> </ul>
Any age	From focal to total body involvement with difficulty speaking and feeding	<ul style="list-style-type: none"> <li>● Spasms of face, trunk and/or limbs</li> <li>● Difficulty feeding, sitting, lying, sleeping</li> <li>● Difficulty with speech or unable to speak</li> </ul>

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# Introduction to dystonia

## 1.4 Early onset dystonia

### Early onset inherited or idiopathic dystonias

Early onset dystonias tend to be generalised. Many are of known genetic origin. There are currently more than 20 known types of dystonia caused by gene mutations. Some of these affect children including:

- A mutation in the DYT1 gene which causes generalised dystonia
- A mutation in the DYT5 gene which causes dopa-responsive dystonia which is treatable with L-dopa

The majority of genetic forms of dystonia are autosomal dominant although some types of genetic dystonia occur in an autosomal recessive or x linked manner. Heritable childhood onset dystonia is more 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 inherit the gene, the penetrance is only around 30%. 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.

Children and young people with early-onset motor symptoms of genetic disorders such as dystonia-myoclonus (DYT11) and benign hereditary chorea (TITF1) may inadvertently be suspected of exhibiting features of attention-deficit hyperactivity disorder (ADHD) because of their apparent 'fidgetiness'.

Inherited dystonias are relatively rare and their prevalence is dwarfed by that of children with acquired dystonia manifesting in infancy.

### Early onset acquired dystonias

Early onset acquired dystonia can result from a wide variety of neurological conditions, inherited metabolic defects or environmental causes.

Environmental causes include disorders associated with reproductive casualty such as premature birth, perinatal brain damage (including birth asphyxia, also known as hypoxic ischaemic encephalopathy, and perinatal jaundice, also known as kernicterus), head trauma, hypoxia and haemorrhage and a variety of drugs or toxins that affect the basal ganglia, thalamus or brain stem. At least 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).

Making an appropriate diagnosis can be difficult in children with normal brain imaging. Such normal imaging may be found in up to 20% of children diagnosed with cerebral palsy (Korzeniewski 2008) but normal scans may be found in up to 50% of children with dystonia of prematurity (Towsley 2011). However 'normal imaging' may prompt the need for a search for a neurometabolic or genetic cause of dystonia or may reflect the lack of power for current imaging techniques to detect small changes.

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### Early onset acquired dystonias (continued)

Dystonia symptoms may be associated with other hereditary neurological syndromes such as Huntington's disease, Wilson's disease and Ataxia telangiectasia. Metabolic disorders causing acquired dystonia include Lesch-Nyhan syndrome, Niemann-Pick disease, Leigh's disease and neurodegeneration with brain iron accumulation (NBIA): and newly described forms of NBIA such as BPAN (see Hayflick 2013).

A relatively new class of causes of dystonias in childhood are the 'interferonopathies' producing infantile, late infantile and childhood nigrostriatal necrosis such as Aicardi-Gouttieres Syndrome Type 6 (Rice 2012) and other interferon-related biomarkers (Rice 2013) opening up a whole new spectrum of progressive dystonias with possible new therapeutic approaches.

Autoimmune mechanisms of dystonia are increasingly recognised in children and young people under 20 years of age, including other acute movement disorders such as chorea and myoclonus with and without neuroimaging changes (Hacohen 2012, 2013). Untreated in the acute phase of the illness, these autoimmune encephalopathies can result in long-standing and often very disabling movement disorders, for instance after herpes simplex type 1 encephalitis (Hacohen 2013).

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

### Age at onset, co-morbidities and progression in early onset dystonia

The following information is based on a cohort of 279 children referred to a UK specialist paediatric movement disorder service including 18% children with inherited / idiopathic dystonia, 72% non-progressive acquired dystonia and 10% hederodegenerative dystonia.

Median onset ages in this cohort were 3 years for inherited / idiopathic dystonia and 0.25 years for acquired dystonias. Only 33% of the children experienced a period of normal motor development. In this sample, co-morbidities were identified in 63% of all cases and spasticity co-morbidity was present in 34% of children with acquired dystonia.

At referral (which was median 4.8 years after onset for inherited / idiopathic dystonia and 7.8 years after onset for acquired dystonia), carers perceived dystonia deterioration in 60% of cases, stabilisation in 32% and improvement in 8% (Lin 2014).

### 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 1

# Introduction to dystonia

### 1.5 The impact of dystonia

#### The effect of adult onset dystonia

Dystonia has a significantly adverse impact on quality of life (QoL) measures as well as on mental health. Among those with focal dystonia, QoL reported at baseline on day of treatment included (Statistics from Gudex 1998):

<b>MOBILITY</b>	53% of people report mobility problems.
<b>DAILY ACTIVITIES</b>	64% report problems with daily activities and 23% report problems with self-care.
<b>PAIN</b>	83% report severe or moderate pain.
<b>MENTAL HEALTH</b>	65% report moderate or severe anxiety/depression. It is increasingly accepted that both stress and anxiety may also exacerbate symptoms of dystonia and that psychological problems are caused not only by the symptoms but also possibly by the condition itself.

For people with focal dystonia, QoL is significantly worse for all these dimensions than the general population. Untreated cervical dystonia also has a significant effect on employment status. In one study of those in employment at the onset of cervical dystonia, 69% reported reduced productivity, 31% reported reduced hours or responsibilities and 19% reported unemployment as a result of their condition (Molho 2009).

In addition, 75% of people with dystonia mentioned an impact on their social life and 80% feel self-conscious about how their condition makes them look (Dystonia Society 2011).

#### The effect of early onset dystonia

The effect of early onset dystonia is widely variable. It can moderately or severely affect one or more of mobility, posture, verbal and written communication, daily living activities and self-care. Pain is often a significant feature of severe cases.

The impact on development, educational and social opportunities cannot be underestimated. Early-onset dystonias reduce the confidence and self-reliance of children and young people through reduced independence and increasing demands of almost all activities. Fatigue, arising from this increased effort as well as difficulty getting to sleep and a fragmented night's sleep further interferes with activity and participation.

## SECTION 2 Dystonic storms, "brittle" dystonia and status dystonicus

Sometimes people with dystonic symptoms develop a rare condition called status dystonicus which is intense and frequent episodes of severe generalised dystonia. An individual episode of status dystonicus is called a 'dystonic storm' or 'dystonic attack'. They usually occur in individuals who already have generalised dystonia.

Although the exact numbers are not known, it is thought that mild to moderate dystonic storms are a fairly regular experience for some individuals with generalised dystonia, especially if it is an acquired dystonia. Most severe cases of status dystonicus occur when generalised dystonia appears in combination with other neurological condition or metabolic conditions or if it is has been caused by a traumatic or infective event or trigger.

If a person's swallowing or breathing is affected by a dystonic storm, they may require emergency medical attention. In very severe cases, there may be renal involvement and individuals may be admitted to an intensive care unit setting where they may be sedated with medication or need temporary mechanical ventilation to support breathing and allow relaxation of dystonic muscles.

#### Childhood dystonic storms, brittle dystonia and status dystonicus

In this situation, the child is in an unstable motor condition in which the dystonia suddenly flares up in severity, interrupting all normal activities and often requiring additional medication or initiation of an emergency care plan.

A simple scheme for dystonia severity assessment and planning (DSAP) has recently been published (Lumsden *et al* 2013) in which SAP also stands for 'soon as possible' to denote the urgency of getting the dystonia under control.

Five DSAP levels exist:

#### LEVEL 1: Child sits comfortably and can participate as usual in their normal activities of the day

#### LEVEL 2: The child cannot sit comfortably or tolerate sitting at all

If the child is constantly slipping from level 1 to level 2 we consider this to be unstable dystonia and the background management needs to be adjusted at home following telephone and outpatient consultations in the local hospital with usual paediatric team. If this happens several times a month, the dystonia may be described as 'brittle' in the same way as asthma may be referred to as 'brittle', necessitating emergency plans and frequent hospital reviews for advice to adjust medication.

#### LEVEL 3: The child has difficulty lying and sleeping

This is a potentially very dangerous condition and needs to be managed urgently in a children's ward where underlying precipitating factors such as respiratory or urinary infections are excluded or if present, are managed with antibiotics. Dystonia-relieving medication, fluids and nutrition are also adjusted and the goal is to achieve good periods of sleep with medication where possible without depressing respiration.

## SECTION 2 Dystonic storms, “brittle” dystonia and status dystonicus

**LEVEL 4: Excessive dystonic muscle contractions lead to sweating, fever, muscle damage, dehydration and poor urinary output and hypotension (low blood pressure).**

The child is managed in a High Dependency Unit (HDU). Nutrition, fluid and medication may need to be delivered by intravenous infusion or via nasogastric tube if the child is too sick to cooperate. In this condition, oxygen is often required by mask or nasal prongs.

**LEVEL 5: Multi-organ failure requiring support.**

There is marked muscle breakdown (known as rhabdomyolysis). Multi-organ support is required such as oxygen, mechanical ventilation, renal dialysis, management of clotting disorders and liver support, usually in the setting of a Paediatric Intensive Care Unit (PICU). Deep sedation is essential and support of breathing as a consequence of this is almost invariable. Severe muscle breakdown can lead to renal and hepatic failure as well as disorders of blood clotting referred to as disseminated intravascular coagulation. There may be a need to completely paralyse muscles temporarily and artificially ventilate the child to prevent further muscle breakdown.

A practical guide to managing the child with Status Dystonicus or severe dystonia has recently been reported using the DSAP grades (described above) for guidance (Allen 2014). This guide helps management as the dystonia severity dictates but more work is required determine the best way of terminating status dystonicus, a condition which may last days, weeks or even months and may be fatal in up to 10% of cases (Fasano 2012).

### 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 3 Diagnosing dystonia

Getting an accurate diagnosis of dystonia can take many years because of a lack of awareness among the public and medical professionals. People affected by dystonia are often referred to, and receive inappropriate management from a variety of specialists. Physiotherapists and psychiatrists should consider the possibility of a diagnosis of dystonia in unusually persistent disorders of posture and movement. Patients thought to have dystonia should be referred to the following professionals for diagnosis and management:

Type of Dystonia	Diagnosis and treatment by:
All types of dystonia	Consultant neurologist specialising in movement disorders
Laryngeal dystonia	Laryngologist or ENT (Ear, Nose and Throat) Surgeon
Blepharospasm	Neurologist specialising in movement disorders or an ophthalmologist
Any dystonia affecting children	Paediatric neurologist or neuro-developmental paediatrician. Some cases may be referred to a paediatric movement disorder neurologist.

To ensure a holistic approach to care, consultant neurologists need to work in collaborative networks with specialist nurses, therapists and other physicians and surgeons, with the neurologist providing clinical leadership in these teams.

### 3.1 The diagnostic process

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 e.g. ‘geste antagoniste’, overflow and mirror movements. It is also a case of eliminating other conditions.

The use of a structured flow chart such as shown in Figure 1 may help diagnostic accuracy.

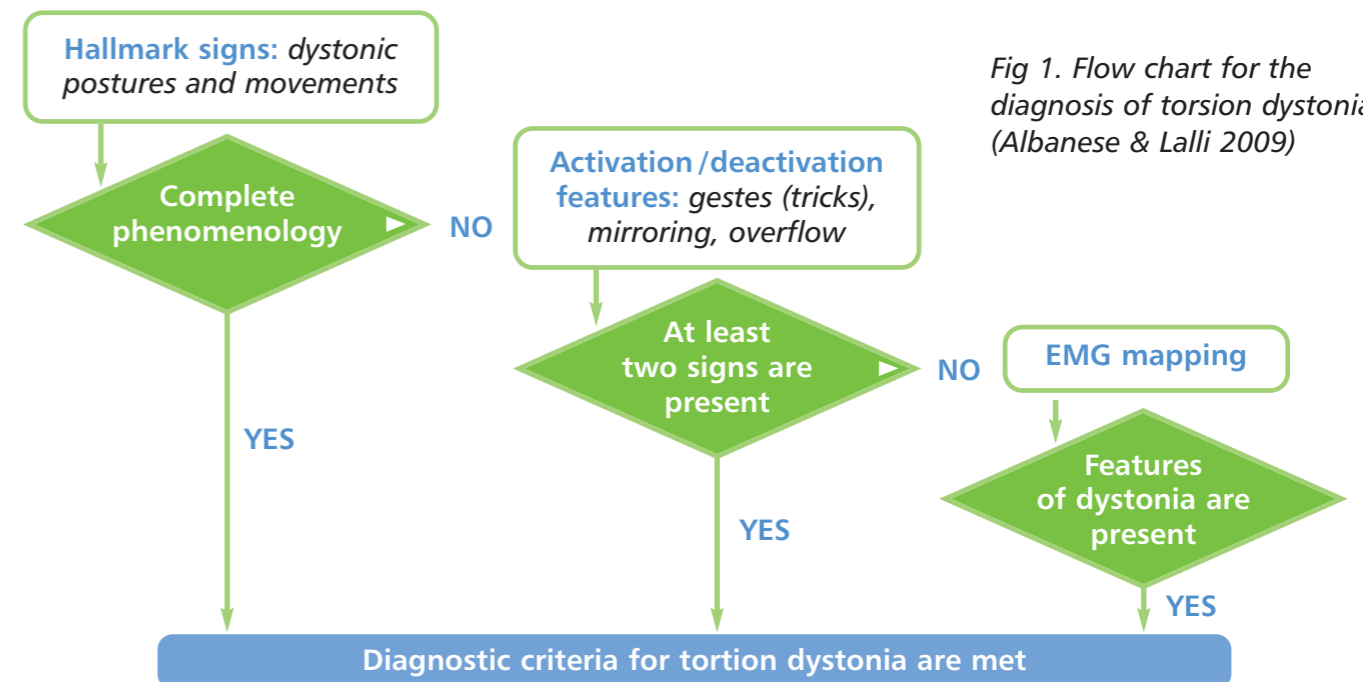


Fig 1. Flow chart for the diagnosis of torsion dystonia (Albanese & Lalli 2009)

## SECTION 3

# Diagnosing dystonia

It is important to be aware that some psychogenic disorders have symptoms which can appear similar to dystonia and these need to be eliminated.

Dystonic symptoms can sometimes temporarily disappear when the patient is concentrating and, for this reason, dystonia is sometimes missed even by experienced neurologists. Helpful techniques to aid diagnosis can include:

- Asking the patient to sit in silence for several minutes (for blepharospasm).
- Where cervical dystonia is suspected, asking them to close their eyes.
- Giving the patients a biscuit to check for oromandibular dystonia.

Because the symptoms of dystonia are diverse, patients may not realise that symptoms in different areas of the body are part of the same underlying condition and so fail to report some relevant symptoms. Where dystonia is suspected, it is therefore important to ask about symptoms in all the main body areas affected by 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.

### Additional tests

Structural brain imaging (MRI) is required in generalised or hemidystonia and if there are any features to suggest an acquired form of dystonia. However, MRI is not routinely required when there is a confident diagnosis of idiopathic (or inherited) 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.

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 presence of degenerative or structural pathology or acquired 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.

## SECTION 3

# Diagnosing dystonia

### 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. Parents should be kept well-informed and supported throughout this process.

Nevertheless, even in specialist hands, establishing a genetic diagnosis may represent 'work in progress' as large referral centres discover new gene mutations. This process of exact diagnostic characterisation often requires collaboration of several centres and can take many years. New technologies are simplifying and reducing the costs of these searches. Screening multiple dystonic conditions with a single blood test will certainly shorten the length of time required to obtain definitive diagnoses and also identify cases that require fresh searches for as yet uncharacterised genetic causes.

A lack of genetic diagnosis may prevent appropriate counselling regarding future affected children but also affects decisions about management since the 'genetic dystonias' tend to respond more favourably to deep brain stimulation (see below). Cheaper and faster diagnostic pathways are therefore needed for children's dystonia services.

### 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 include:

- An explanation of the condition and its prognosis.
- Management options – including timing and location.
- Additional sources of information – including details of the Dystonia Society helpline and website.

### Additional sources of support:

- The Dystonia Society can supply relevant leaflets to clinics to aid this process.
- An important new source of information is online peer support groups using forums or Facebook pages. These can link people with dystonia, parents and carers together – but users need to be advised that they are usually moderated by unpaid volunteers and non-professionals. The Dystonia Society can provide information as to what online peer support options are available.

## SECTION 4

# Management of dystonia in adults

## 4.1 Botulinum toxin injections

In most cases of focal dystonia, the usual management 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. The patients who benefit from botulinum toxin injections are treated at intervals, adjusted to the duration of the benefit of the management (usually 2 to 4 months, sometimes longer).

Botulinum Toxin (BoNT) can be regarded as first-line management for **idiopathic (or inherited) 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 management using recommended doses is performed over many years but excessive doses result in increased risk of side effects at each session. Prolonged use of botulinum toxin has been associated with resistance resulting from antibody in some cases.

- 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.
- Transient dysphagia can follow BoNT injections for cervical, laryngeal, tongue and jaw dystonia. Patient should be able to contact directly a member of the medical team providing the injections and be seen to assess the severity of the dysphagia. Severe dysphagia can expose the patient to aspiration.
- BoNT is not licensed for management of dystonia in children (see section 5.2).

### Nurse and physiotherapist injectors

The majority of BoNT injections are currently provided by consultants and senior doctors – but injections in many cases can be effectively administered by nurses or physiotherapists provided appropriate training, supervision and patient review is provided by the consultant responsible for patient care (although some complex injections such as those to treat laryngeal dystonia are done only by consultants). Dystonia nurses are sometimes also able to adjust the dose of botulinum toxin within agreed parameters, if necessary seeking advice from the consultant according to agreed protocols (*Whitaker et al (2001)*).

In 2012, nurses and physiotherapists provided around a quarter of all injections and in some clinics the proportion was substantially above 50%.

## SECTION 4

# Management of dystonia in adults

### Where BoNT is not working

BoNT injections provide significant relief for the majority of patients in the categories identified above. However, a substantial minority of patients report receiving limited relief. In these cases the following steps should be considered:

- Review BoNT dosage and selection of muscles being injected by clinical examination.
- Where injected muscles are deep below the skin, consider using electromyographic (EMG) or ultrasound machine.
- Review patient one month after injection.
- If biological resistance to BoNT is suspected, perform a frontalis test. Patient should not be considered to have antibodies until this is done and proven conclusive.
- If patient is resistant to toxin, consider BoNT A holiday, reviewing the patient regularly, with possible physiotherapy and management using oral medication. Try a new injection at 12 months. If still resistant, discuss surgery where appropriate.
- If patient not immune but not responding after trying the above, consider second opinion from another BoNT injector.

### Outreach services

Botulinum toxin injections can be effectively provided through outreach services based in local community hospitals and health centres. In rural areas, outreach services address an important patient need. 35% of patients currently have a round trip travel time of more than 2 hours to their injection centre and, among this group, 55% report travel as a problem. The problem of travel is exacerbated by the fact that many patients are elderly – the average age of someone receiving botulinum injections is 60 years with 23% of patients over 70 years.

## 4.2 Oral medication

### 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 management with levodopa should be initiated and adjusted according to the clinical response.

### Anticholinergic agents

Anticholinergic drugs are the only group that have been consistently considered efficacious (Cullis 1989; Lang 1989) although only small and methodologically poor Randomised Controlled Trials (RCTs) are available. For many patients the side effects are not manageable. It is important to increase very slowly the dosage over 2 or 3 months to minimise side effects. The drug should be avoided in elderly patients (after the age of 70). Alcohol is contra-indicated with this drug.

### Tetrabenazine / benzodiazepines

Sometimes 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.

It is essential to monitor the effects of management using oral medications carefully and, if more than one healthcare professional is involved, to be clear who is responsible for this.

## SECTION 4

# Management of dystonia in adults

### 4.3 Surgery

Pallidal Deep Brain Stimulation (DBS). Considered a good option, particularly for idiopathic (or inherited) generalised or cervical dystonia and dystonic tremor, 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 procedure is carried out while the patient is asleep. The electrodes send a pulse that blocks the signals from the brain that cause the involuntary muscle spasms. DBS is funded by the NHS (and in England is approved as a specialised treatment by the National Commissioning Board).

In general, DBS is less effective in acquired dystonia with the exception of tardive dystonia. DBS can have side effects and involves a life-long commitment by the patient (and family) as on-going follow-up is required and may necessitate travel to a DBS centre. More information on DBS can be obtained from the nearest DBS implanting centre.

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.

After the DBS operation, the patient will need regular follow-up as it can take several months to reach full effectiveness. In addition, the patient may require a range of allied healthcare support (see section 4.4 below).

A number of other surgical approaches are also used in some cases. Selective peripheral denervation is an alternative approach to treat medically refractory cervical dystonia. There is a significant risk of recurrence of symptoms. Insufficient evidence exists to use this intervention in idiopathic or inherited dystonia but the procedure can be indicated in patients where acquired dystonia is combined with spasticity.

### 4.4 Allied healthcare

BoNT, oral medication and surgery are effective treatments and can substantially mitigate symptoms for patients with dystonia but they are not a complete solution. In a Dystonia Society survey, patients reported that on average they only mitigate around half their symptoms. A number of allied healthcare interventions should therefore also be considered.

#### Physiotherapy

For focal dystonias, the use of rehabilitative physiotherapy is well developed and structured. It aims to give patients as much independence as possible. 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)). Studies have shown that physiotherapy also enhances the effectiveness of BoNT in treating cervical dystonia but, at present, they have not identified which techniques are the most beneficial. In a Dystonia Society survey, among those referred to specialist neurophysiotherapy, 74% of patients with cervical dystonia and 62% of those with focal hand dystonia reported it as being helpful.

Due to the specialised nature of dystonia, this treatment needs to be provided by a neurophysio-therapist familiar with the condition.

## SECTION 4

# Management of dystonia in adults

#### Psychological therapy

Dystonia is not a mental health condition but it can cause depression and anxiety due to the pain, stigma and social isolation of the condition. The overall rate of psychiatric symptoms amongst individuals with dystonia is often higher than that seen in the general population, with onset of the psychiatric symptoms prior to the physical symptoms of dystonia in some cases.

Psychological therapies such as cognitive behavioural therapy (CBT) and counselling can therefore play an important role. In addition, as stress and anxiety can exacerbate the physical symptoms of dystonia, effective management of these can also improve the outcome of managing the physical symptoms.

Mindfulness (autogenic training or meditation) can also be helpful. In a Dystonia Society questionnaire, mindfulness was reported helpful by 65% of those who tried it.

#### 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. Sometimes, the resulting intractable pain can dominate a patient's life and may be unresponsive to medication including that used to manage dystonia. Among those with cervical dystonia, a high prevalence of substantial pain is reported. Pain is also reported for other types of adult onset dystonia in some cases. Referral to pain management programmes has been shown to be effective for management of chronic pain caused by conditions such as dystonia.

#### 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 (**laryngeal dystonia**) 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** and **lingual dystonias** with articulation difficulties can be given mouth and swallowing exercises to help them reduce the risk of choking by chewing and swallowing safely.

## SECTION 5

# Management of dystonia in children

The management of dystonia in children is undergoing intense review. These guidelines will be regularly amended to provide the most comprehensive overview to carers and health providers as new evidence regarding diagnosis, assessment and management of dystonia in childhood emerges.

### 5.1 Oral medication

Oral medications are often the mainstay for managing dystonia in children and young people. They may respond well and can tolerate higher doses of anticholinergics than adults, but the effect 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 method of managing dystonia but often a combination of several drugs and other therapies can enable effective management.

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 a vital resource to support families of children with dystonia and can help devise strategies for coping with many essential functions. Easily identified stages in dystonia severity have recently been described (Lumsden 2013) and this scheme has further been incorporated into a practice guideline for the management of status dystonicus, dystonic storms and brittle dystonia (Allen 2014) – see section 2.

Some of the management options that may be offered include:

#### Types of Medication

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.

If a trial of levodopa is unsuccessful, then **trihexyphenidyl** is usually the first line of management. This is often well tolerated in children although anticholinergic side effects need to be monitored. However, beneficial reductions in dystonia can vary from child to child and may not be tolerated in older children who may experience feeling muddled-headed or blurred vision at higher doses.

**Oral Baclofen\*** and **diazepam\*** are often useful but side effects of drowsiness, drooling and droopiness of trunk (and neck control) are common at higher doses. The undesirable advent of “the three d’s” should always be discussed when these medications are prescribed. A problem with the sedative medicines such as the benzodiazepines (diazepam, nitrazepam, midazolam, lorazepam) is the liability to cause respiratory depression, copious secretions and chest infections when used regularly and especially in higher doses: side effects generally not encountered with clonidine or gabapentin. (\*Limited license for use to treat muscle spasms in children).

**Clonidine** is useful but can cause somnolence, but this property is very useful in very severe cases of dystonia which often manifests as ‘brittle/unstable dystonia’ or status dystonicus (see section 2).

**Gabapentin** is sometimes used and is currently being evaluated as a promising mainstream medication with many positive benefits in controlling uncomfortable dystonic postures, mood, behaviour and most importantly in helping to improve sleep quality and duration (Lin unpublished).

**Tetrabenazine** is sometimes used but the results are disappointing. Nevertheless the use of tetrabenazine in status dystonicus has been reported.

Licensing of the safest medicines in children with dystonia is urgently required.

## SECTION 5

# Management of dystonia in children

### 5.2 Other treatment

#### Botulinum toxin A injections

These are licensed for treating lower limb spasticity in children with cerebral palsy but are not licensed for the treatment of children’s dystonia. However, they are often used off licence to treat dystonia in children in order to manage pain and spasm in specific muscle groups that interfere with function of the neck, hands, elbows, hips, knees, ankles or feet. Good practice guidelines for use of botulinum toxin A in children are available which advise on maximum dosage and administration frequency and technique so that systemic botulism is not seen. They highlight caution in giving botulinum toxin A to the most functionally impaired children.

#### 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 acquired dystonia not amenable to Deep Brain Stimulation.

#### Deep Brain Stimulation (DBS)

This is very effective in inherited and idiopathic dystonias. Response to DBS in the treatment of dystonia is thought to decline with the proportion of life lived with dystonia; so for inherited and idiopathic dystonias, DBS should be offered early, preferably within 5 years of onset of dystonia. In addition, it should be recognised that the longer the duration of dystonia, the greater the risk of skeletal deformities and dependency (through lack of opportunity). 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 is a sign that functional neurosurgery should be considered.

Unfortunately, many forms of acquired dystonia that are associated with focal or generalised brain injury will preclude the use of DBS. Careful evaluation of potential DBS candidates in childhood should be performed by experienced teams with access to multi-disciplinary assessment and follow-up by psychology, occupational therapy, physiotherapy and speech and language specialists, particularly with regard to setting realistic pre-operative goals and measuring outcomes from a variety of functional assessment scales (Gimeno 2012, 2013, 2014). Detailed neuroimaging and neurophysiology evaluations are also mandatory prior to proceeding to DBS (McClelland 2011). However recent reports indicate that children with acquired dystonia can experience improved quality of life and achieve important goals after DBS (Gimeno 2012, 2013) as well as improved upper limb function (Gimeno 2013). An important area for all healthcare delivery is an appropriate understanding of functional priorities using a tool known as the Canadian Occupational Performance Measure (COPM) in children with dystonia, irrespective of cause (Gimeno 2013) and these can then be quantified in terms of performance and satisfaction before and after an intervention such as DBS (Gimeno 2014). Such approaches follow the WHO International Classification of Function (ICF) which focuses on ‘activity’ and ‘participation’ rather than simply focussing on the impairment measure i.e. just measures of dystonia.

## SECTION 5

# Management of dystonia in children

### Physiotherapy

For generalised dystonias, while therapeutic handling strategies can be useful, they tend not to have carryover (i.e. 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 (e.g. school) about aspects including handling, positioning, and adapting activities to promote active participation.
- Advising on general and specific exercise to maintain general health and well being 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.

Due to the specialised nature of dystonia, this treatment needs to be provided by a neurophysio-therapist familiar with the condition.

### Psychological therapy

Dystonia is not a mental health condition but it can cause severe depression and anxiety both due to the pain, stigma and social isolation of the condition and prevalence of mental health problems among people with dystonia is high. There is also evidence that dystonia can affect mental health even before physical symptoms appear. Psychological therapies such as cognitive behavioural therapy (CBT) and counselling can therefore play an important role. In addition, as stress and anxiety can exacerbate the physical symptoms of dystonia, effective management of these can also improve the outcome of physical treatment.

### 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 or limbs. Sometimes, the resulting intractable pain can dominate a patient's life and may be unresponsive to medication including that used to manage dystonia. Among those with generalised dystonia, a high prevalence of substantial pain is reported. Referral to pain management programmes has been shown to be effective for treatment of chronic pain caused by conditions such as dystonia.

### Speech and language therapy

If the early onset dystonia affects eating or speaking, then speech and language therapy can be helpful – see section 4.4.

## SECTION 5

# Management of dystonia in children

### 5.3 Other considerations

Dystonia onset before puberty is particularly disruptive to a child's growth and development. Any growth spurt such as occurs at puberty may accelerate the appearance of contracture and deformity which further diminishes function and limits opportunity.

Children with idiopathic or inherited dystonia may need to be referred to other specialist departments for treatment for conditions that result from dystonia (e.g. musculoskeletal and orthopaedic problems). 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.

Close links must be forged with education services and a Statement of Special Educational Needs may be implemented to provide the child with the necessary support in school.

### *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.*

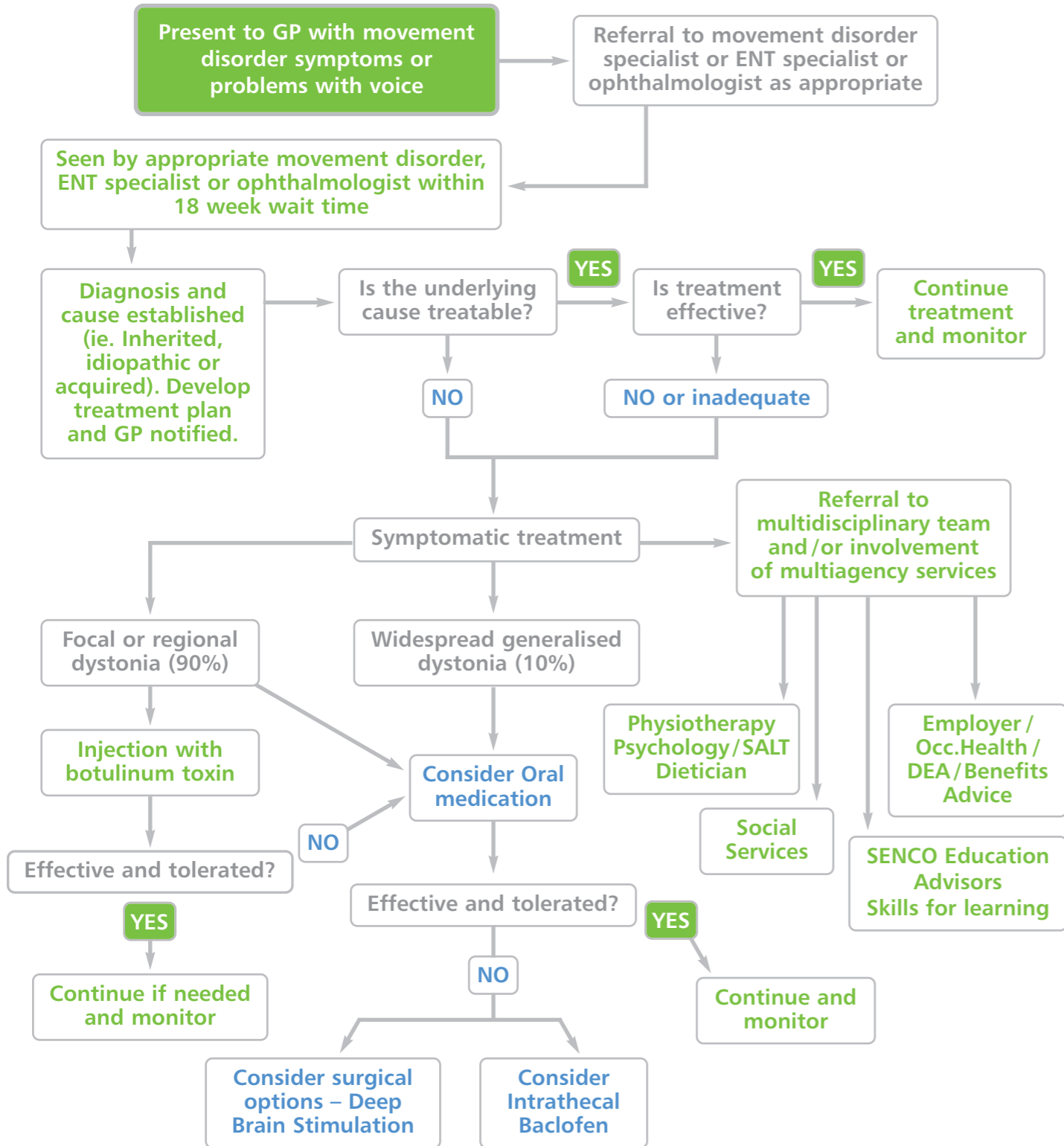
*Getting effective treatment felt like someone had put my brain back in my head.*

## SECTION 6

# Patient pathway flowcharts

### 6.1 Recommended patient pathway for adults with dystonia

(adapted from NW Neurosciences Partnership 2006)

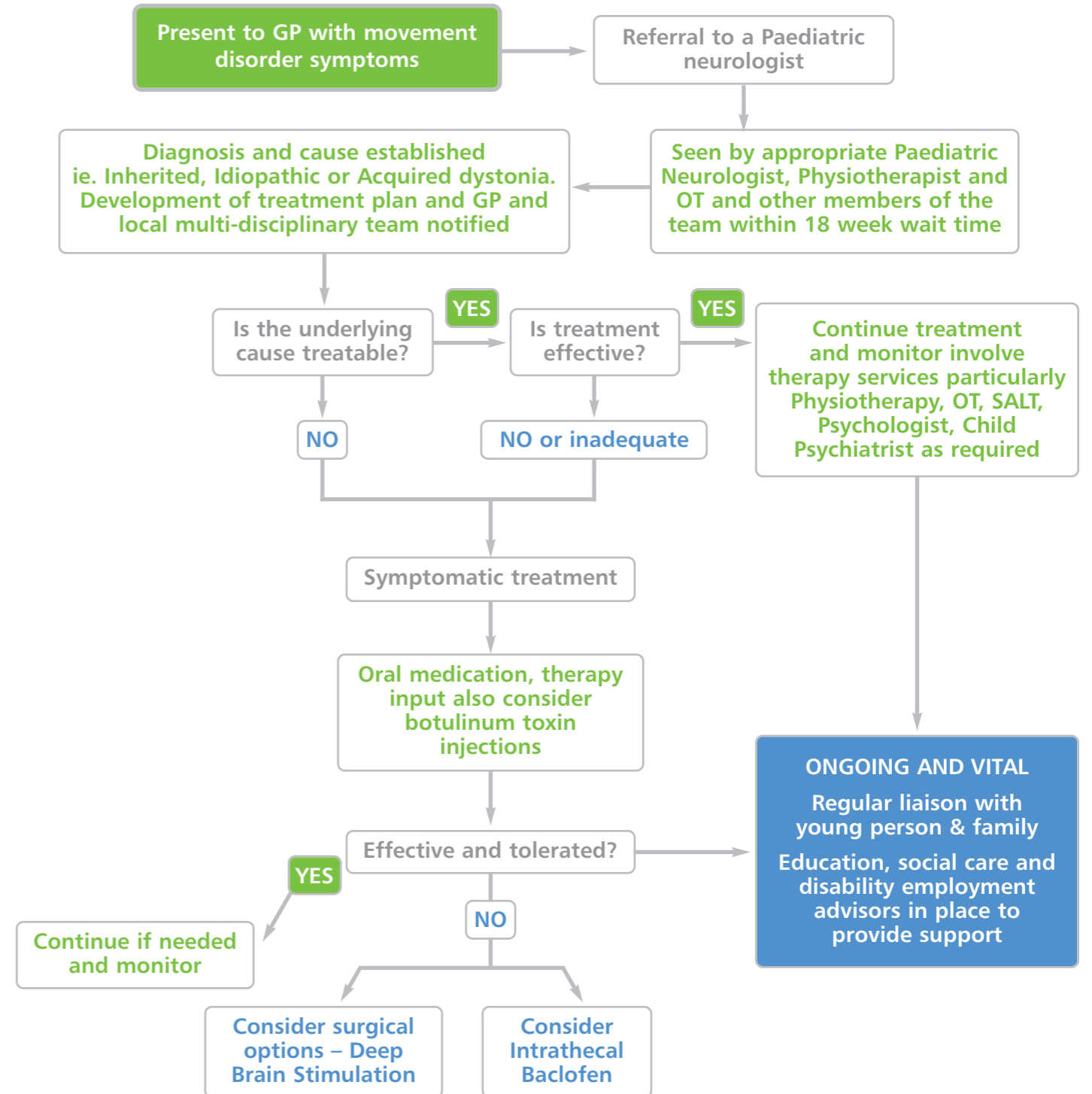


## SECTION 6

# Patient pathway flowcharts

### 6.2 Recommended patient pathway for children and young people with dystonia

(adapted from NW Neurosciences Partnership 2006)



## SECTION 7

# Other support

### 7.1 Dietary support

A number of types of dystonia can affect nutrition:

- Dysphagia (difficulty swallowing) can arise from cervical, 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, laryngeal, tongue and jaw dystonia. (N.B. see additional comments on dysphagia following botulinum toxin injections in section 4.1).
- 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 a 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.

### 7.2 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 provided can include identifying ways problematic everyday tasks can be done differently, advice on equipment and referral to other services. Examples of equipment include body suits to assist with posture and transportable back/neck supports.

#### Specialist equipment

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.

### 7.3 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.

## SECTION 7

# Other support

### 7.4 Complementary therapy

People with dystonia often seek out complementary therapies and report varying degrees of benefit (Lim 2007). 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).

In a Dystonia Society questionnaire, mindfulness (autogenic training) was reported helpful by 65% of those who tried it. Other therapies tried include acupuncture, chiropractic, homeopathy, hypnotherapy, osteopathy, reflexology and the Alexander Technique. All were reported helpful in the range of 15–30% of those who tried them.

### 7.5 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 to their local medical genetics service. Referral of children with idiopathic dystonia can be helpful as geneticists can sometimes aid diagnosis.

### 7.6 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 e.g. domiciliary care, equipment or home adaptations.

A number of other specific social interventions may also be required:

- Visual impairment due to **blepharospasm**. A consultant ophthalmologist should complete an assessment to determine 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.
- Patient wishes to continue driving. Neurologist or dystonia nurse to advise if condition stable enough. If so, confirmation to DVLA.
- As appropriate, young people with dystonia should be referred to Special Educational Needs (SENCO) support.

## SECTION 8 References

Albanese A, Barnes M P, Bhatia K P, Fernandez-Alvarez E, Filippini G, Gasser T, Krauss J K, Newton A, Rektor I, Savoirdo M and Valls-Sole (2006) 'A systematic review of the diagnosis and treatment of primary (idiopathic) dystonia and dystonia plus syndromes: report of an EFNS/MDS-ES Task Force'. *European Journal of Neurology* Vol.13 pp. 433–444

Albanese A, Asmus F, Bhatia KP, Elia AE, Elilob B, Filippini G, Gasser T, Krauss JK, Nardocci N, Newton A, Valls-Solè J, (2011). *EFNS guidelines on diagnosis and treatment of primary dystonias*. *Eur J Neurol* 18:5 – 18

Albanese A, Bhatia K, Bressman S, DeLong M, Fahn S, Fung V, Hallett M, Jankovic J, Jinnah H, Klein C, Lang A, Mink J, Teller J (2013). *Phenomenology and classification of dystonia: A consensus update*. *Movement disorders*, Vol 28 No 7. 863–873

Allen N, Lin JP, Lynch T, King M (2014). *Status dystonicus: a practice guide*. *Dev Med Child Neurol.*; 56(2):105–12

Bleton J-P (2007) *Role of the Physiotherapist* in Warner T T and Bressman S B (ed.) *Clinical Diagnosis and Management of Dystonia*. London. Informa Healthcare 2007.

Dauer W, Burke R, Greene P and Fahn S (1998). *Current concepts on the clinical features, aetiology and management of idiopathic cervical dystonia*. *Brain* 121, 547–560

Carr L (2009) Consultant Paediatric Neurologist Robert Surtees Memorial Lecture. The Annual Conference of the Dystonia Society, London, 7 November 2009

Cullis P, Walker P (1989). *The treatment of Spasmodic torticollis*. In: Quinn NP, Jenner PG, eds. *Disorders of movement*. San Diego: Academic Press, 1989:295–301

Defazio G (2010) *The epidemiology of primary dystonia: current evidence and perspectives*, *European Journal of Neurology* Vol 17 (suppl. 1) pp 9–14

Delnooz C, Horstink M, Tijssen M, van de Warrenburg B (2009). *Paramedical treatment in primary dystonia: A systematic review*. *Movement disorders*, Vol 24. No 15, pp 2187–2198.

Dias F, Kummer A, Doyle F, Harsanyi E, Cardoso F, Fontenelle L, and Teixeira A (2011). *Psychiatric disorders in primary focal dystonia and in Parkinson's disease*. *Neuropsychiatric disease treatment*, 7: 111–116.

Edwards M, Quinn N and Bhatia K (2008) *Parkinson's Disease and Other Movement Disorders*. Oxford Specialist Handbooks in Neurology, Oxford University Press 2008.

Gimeno H, Tustin K, Selway R, Lin JP (2012). *Beyond the Burke-Fahn-Marsden Dystonia Rating Scale: Deep brain stimulation in childhood secondary dystonia*. *Eur J Paediatr Neurol*. 2012;16(5):501-8.

Gimeno H, Lumsden D, Gordon A, et al (2013). *Improvement in upper limb function in children with dystonia following deep brain stimulation*. *Eur J Paediatr Neurol*. 17(4):353-60.

Gimeno H, Gordon A, Tustin K, Lin JP (2013). *Functional priorities in daily life for children and young people with*

*dystonic movement disorders and their families*. *Eur J Paediatr Neurol*. Mar;17(2):161–8

Gimeno H, Tustin K; Lumsden D, Ashkan K, Selway R, Lin JP (2014). *Evaluation Of Functional Goal Outcomes Using The Canadian Occupational Performance Measure (COPM) Following Deep Brain Stimulation (DBS) In Childhood Dystonia*. *Eur J Paediatr Neurol*. pii: S1090-3798(14)00005–1

Great Britain, Department of Health, National Institute for Health and Clinical Excellence (2004) *Selective peripheral denervation for cervical dystonia (IPG 80)*

Gudex C, Hawthorne M, Butler A, Duffey P (1998). *Effect of dystonia and botulinum toxin treatment on health-related quality of life*. *Movement disorders*, Vol 13, No 6

Fasano A, Ricciardi L, Bentivoglio A, Canavese C, Zorzi G, Petrovic I, Kresojevic N, Kostić V, Svetel M, Kovacs N, Balas I, Roubertie A, Mishra D, Mariotti P, Temudo T, Nardocci M (2012). *Status dystonicus: Predictors of outcome and progression patterns of underlying disease*. *Movement Disorders*, Volume 27, Issue 6, pages 783–788, May.

Hacohen Y, Wright S, Siddiqui A, Pandya N, Lin JP, Vincent A, Lim M (2012). *A clinico-radiological phenotype of voltage-gated potassium channel complex antibody-mediated disorder presenting with seizures and basal ganglia changes*. *Dev Med Child Neurol*. Dec; 54(12):1157–9

Hacohen Y, Wright S, Waters P, Agrawal S, Carr L, Cross H, De Sousa C, Devile C, Fallon P, Gupta R, Hedderly T, Hughes E, Kerr T, Lascelles K, Lin JP, Philip S, Pohl K, Prabahkar P, Smith M, Williams R, Clarke A, Hemingway C, Wassmer E, Vincent A, Lim MJ (2013). *Paediatric autoimmune encephalopathies: clinical features, laboratory investigations and outcomes in patients with or without antibodies to known central nervous system autoantigens*. *J Neurol Neurosurg Psychiatry*. Jul; 84(7):748–55

Hacohen Y, Deiva K, Pettingill P, Waters P, Siddiqui A, Chretien P, Menson E, Lin JP, Tardieu M, Vincent A, Lim MJ (2013). *N-methyl-D-aspartate receptor antibodies in post-herpes simplex virus encephalitis neurological relapse*. *Mov Disord*. Sep 6

Hayflick S, Kruer M, Gregory A, Haack T, Kurian M, Houlden H, Anderson J, Boddaert N, Sanford L, Harik S, Dandu V, Nardocci N, Zorzi G, Dunaway T, Tarnopolsky M, Skinner S, Holden KR, Frucht S, Hanspal E, Schrandt-Stumpel C, Mignot C, Héron D, Saunders DE, Kaminska M, Lin JP, Lascelles K, Cuno SM, Meyer E, Garavaglia B, Bhatia K, de Silva R, Crisp S, Lunt P, Carey M, Hardy J, Meitinger T, Prokisch H, Hogarth P, (2013). *β-Propeller protein-associated neurodegeneration: a new X-linked dominant disorder with brain iron accumulation*. *Brain*. Jun;136(Pt 6):1708–17. doi: 10.1093/brain/awt095. Epub 2013 May 17. PMID: 23687123

Korzeniewski S, Birbeck G, DeLano M, Potchen M, Paneth N. (2008) *A systematic review of neuroimaging for cerebral palsy*. *J Child Neurol* 23: 216-27

Lang (1989). *Drug treatment of dystonia In: Quinn NP, Jenner PG, eds. Disorders of movement*. San Diego: Academic Press, 313–21.

## SECTION 8 References

Lim V K (2007) *Health Related Quality of Life in Patients with Dystonia and their Caregivers in New Zealand and Australia*. *Movement Disorders* Vol. 22, No. 7, pp. 998–100

Lin JP (2011). *The contribution of spasticity to the movement disorder of cerebral palsy using pathway analysis: does spasticity matter?* *Dev Med Child Neurol*. 2011 Jan; 53(1): 7–9. doi: 10.1111/j.1469–8749.2010.03843

Lin JP, Lumsden D, Gimeno H, Kaminska M (2014). *The impact and prognosis for dystonia in childhood including dystonic cerebral palsy: a clinical and demographic tertiary cohort study*. *J Neurol Neurosurg*

Lumsden D, Kaminska M, Gimeno H, Tustin K, Baker L, Perides S, Ashkan K, Selway R, Lin J–P (2013). *Proportion of life lived with dystonia inversely correlates with response to pallidal deep brain stimulation in both primary and secondary childhood dystonia*. *Developmental Medicine and Child Neurology*

Lumsden D, Lundy C, Fairhurst C, Lin JP (2013). *Dystonia Severity Action Plan: a simple grading system for medical severity of status dystonicus and life-threatening dystonia*. *Dev Med Child Neurol*. 55(7):671–2

Malone M, Strube M, Scogin F (1998). *Meta-analysis of non-medical treatments for chronic pain*. *Pain*, 34 (3): 231–244

McClelland V, Mills K, Siddiqui A, Selway R, Lin JP (2011). *Central motor conduction studies and diagnostic magnetic resonance imaging in children with severe primary and secondary dystonia*. *Dev Med Child Neurol*. Aug; 53(8):757–763. doi: 10.1111/j.1469–8749.2011.03981

Molho E, Agarwal N, Regan K, Higgins D, Factor S (2009). *Effect of cervical dystonia on employment: A retrospective analysis of the ability of treatment to restore pre morbid employment status*. *Movement disorders*, Vol 24. No 9, pp 1384–97

NHS Commissioning Board (2013). *Clinical Commissioning Policy*. Deep brain stimulation in movement disorders.

North West Neurosciences Partnership (Position statement 2006) *Recommendations regarding the use of Botulinum Toxin in Adult Patients with Spasticity and Dystonia – A Report of the North West Botulinum Toxin Subgroup*

Peall K, Smith D, Kurian M, Wardle M, Waite A, Hedderly T, Lin JP, Smith M, Whone A, Pall H, White C, Lux A, Jardine P, Bajaj N, Lynch B, Kirov G, O'Riordan S, Samuel M, Lynch T, King M, Chinnery P, Warner T, Blake D, Owen M, Morris H (2013). *SGCE mutations cause psychiatric disorders: clinical and genetic characterization*. *Brain*. Jan;136(Pt 1):294–303

Rice G, Kasher P, Forte G, Mannion N, Greenwood S, Szykiewicz M, Dickerson J, Bhaskar S, Zampini M, Briggs T, Jenkinson E, Bacino C, Battini R, Bertini E, Brogan P, Brueton L, Carpanelli M, De Laet C, de Lonlay P, del Toro M, Desguerre I, Fazzi E, Garcia-Cazorla A, Heiberg A, Kawaguchi M, Kumar R, Lin JP, Lourenco C, Male A, Marques W Jr, Mignot C, Olivieri I, Orcesi S, Prabhakar P, Rasmussen M, Robinson R, Rozenberg F, Schmidt JL, Steindl K, Tan TY, van der Merwe WG, Vanderver A,

Vassallo G, Wakeling EL, Wassmer E, Whittaker E, Livingston J, Lebon P, Suzuki T, McLaughlin PJ, Keegan L, O'Connell M, Lovell S, Crow Y. (2012). *Mutations in ADAR1 cause Aicardi-Goutières syndrome associated with a type I interferon signature*. *Nat Genet*. Nov;44(11):1243–8

Rice G, Forte G, Szykiewicz M, Chase D, Aeby A, Abdel-Hamid M, Ackroyd S, Allcock R, Bailey K, Balottin U, Barnerias C, Bernard G, Bodemer C, Botella M, Cereda C, Chandler K, Dabydeen L, Dale R, De Laet C, De Goede C, Del Toro M, Effat L, Enamorado N, Fazzi E, Gener B, Haldre M, Lin JP, Livingston J, Lourenco C, Marques W Jr, Oades P, Peterson P, Rasmussen M, Roubertie A, Schmidt J, Shalev S, Simon R, Spiegel R, Swoboda K, Temtamy S, Vassallo G, Vilain C, Vogt J, Wermenbol V, Whitehouse W, Soler D, Olivieri I, Orcesi S, Aglan M, Zaki M, Abdel-Salam G, Vanderver A, Kisand K, Rozenberg F, Lebon P, Crow YJ (2013). *Assessment of interferon-related biomarkers in Aicardi-Goutières syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study*. *Lancet Neurol*. Dec;12(12):1159–69

Ron M (2009). *Primary focal dystonia – a disease of brain and mind: motor and psychiatric manifestations have a common neurobiological basis*. *Journal Neurosurgery Psychiatry* 80:1059

Slawek J, Friedman A, Potulska A, Krystkowiak P, Gervais C, Banach M, Ochudlo S, Budrewicz S, Reclawowicz D, Rynkowski M, Opala G (2007). *Factors affecting the health related quality of life of patients with cervical dystonia and the impact of botulinum toxin type A injections*. *Funct Neurol*, 22(2): 95–100

Tassorelli C, Mancini F, Balloni L, Pacchetti C, Sandrini G, Nappi G and Martignoni E (2006). *Botulinum toxin and neuromotor rehabilitation: An integrated approach to idiopathic cervical dystonia*. *Movement disorders*, Vol 21. No 12, pp 2240–2243

The British Neurotoxin Network (2012). *Responses to questionnaire*. [www.neurotoxinnetwork.org](http://www.neurotoxinnetwork.org)

The Dystonia Society (2011). *Dystonia: A guide to good practice for health and social care professionals*

The Dystonia Society (2012). *Responses to questionnaire*

Towsley K, Shevell M, Dagenais L (2011). *Population-based study of neuroimaging findings in children with cerebral palsy*. *Eur J Paediatr Neurol* 15: 29–35

Walkergate Park Centre for Neurorehabilitation and Neuropsychiatry (2010). *Pathways for diagnosis and management of dystonia*. Lesley Kidd, Nurse Manager, Neurology Rehabilitation Outpatients

Warner T T and Bressman S B (ed.) (2007) *Clinical Diagnosis and Management of Dystonia*. London. Informa Healthcare

Whittaker J, Butler A, Semlven J and Barnes M P (2001) *Botulinum toxin for people with dystonia treated by an outreach nurse practitioner: A comparative study between a home and a clinic treatment service*. *Archives of Physical Medicine and Rehabilitation* Vol. 82 Iss.4 pp 480–484

## SECTION 9

# Acknowledgments

*The Dystonia Society is grateful to the following medical professionals who kindly contributed to this report.*

*This guide is an update of the Dystonia Society's 2011 Guide to Good Practice. The following reviewed and contributed material to the updated document:*

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*The following contributed material to the Dystonia Society's 2011 Guide to Good Practice which has been incorporated in this updated document:*

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*Publication was funded with  
an unrestricted grant from:*

**Medtronic UK Ltd**  
Building 9, Croxley Green Business Park,  
Hatters Lane, Watford, Herts WD18 8WW

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and compiled by:*

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## SECTION 10

# Further information

The Dystonia Society's website  
[www.dystonia.org.uk](http://www.dystonia.org.uk)

The British Neurotoxin Network  
[www.neurotoxinnetwork.org](http://www.neurotoxinnetwork.org)  
*A coalition of botulinum toxin injectors from across the UK*

British Medical Journal's online dystonia learning module  
Access at: [www.learning.bmj.com](http://www.learning.bmj.com)  
*Produced in association with The Dystonia Society*

Dystonia Medical Research Foundation  
[www.dystonia-foundation.org](http://www.dystonia-foundation.org)  
*A non-profit organisation in the United States focusing on research*

The Dystonia Coalition  
[www://rarediseasesnetwork.epi.usf.edu/Dystonia/](http://www://rarediseasesnetwork.epi.usf.edu/Dystonia/)  
*An international collaboration of medical researchers and patient advocacy groups*

Dystonia Europe  
[www.dystonia-europe.org](http://www.dystonia-europe.org)  
*A collaboration of European patient advocacy groups*

**Dystonia Society Helpline: 0845 458 6322**

## SECTION 11

# About the Dystonia Society

The Dystonia Society exists to promote the welfare of people who are affected by dystonia through providing support and information, promoting awareness, advocating for best practice, supporting research and linking the dystonia community together. It does this on a national level and through its network of local groups.

Activities include a helpline, an advocacy service, events across the UK, a self-management project and provision of information online and in hard copy. The Society works closely with government departments, health commissioners and clinicians to improve services.

The Society has close links with the British Neurotoxin Network, the Neurological Alliance, the international Dystonia Coalition and Dystonia Europe.

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