# Childhood Dystonia

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***Conflict of interest statement:***
The author declares that there are no financial or commercial conflicts of interest.

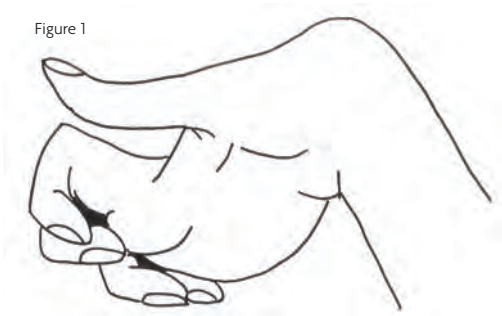
**Summary**
- Dystonia means involuntary muscle contractions causing repetitive movements and twisted postures
- The commonest clinical picture in children is dystonic cerebral palsy following hypoxic brain injury
- A trial of levodopa is warranted in cases without a clear secondary cause
- Management is often challenging, and must be holistic

Dystonia is a movement disorder in which involuntary muscle contractions cause repetitive movements and twisted postures. Dystonia causes significant morbidity in sufferers, and may even be fatal in severe cases. It may be a primary, genetic disorder, or secondary to a large number of other disorders. In children, these are mainly neurometabolic and degenerative. A thorough history, examination, and targeted use of investigations can provide the diagnosis in a subset of children, and help identify those in whom esoteric tests are warranted. Management is usually challenging, with a lack of robust evidence for treatment strategies in children. This article summarises an approach to the child with dystonia, and provides a framework for management.

## Defining dystonia

Dystonia is defined as "a movement disorder in which involuntary sustained or intermittent muscle contractions cause twisting and repetitive movements, abnormal postures, or both".[1] The postures produced by co-contraction of agonist and antagonist muscle groups include hyperextension of the back and neck, torticollis, foot inversion, upward extension of the great toe, and 'spooning' of the hands (Figure 1).[2] Dystonia is often more prominent when voluntary movement is attempted, or in certain postures. Muscle tone may be normal at rest, enabling the clinician to differentiate dystonia from hypertonia. Dystonia may be generalised (or multi-focal), or localised to specific regions of the body, such as in torticollis.[1] In childhood, the commonest clinical picture is one of cerebral palsy with elements of spasticity and dystonia together.[3] However primary dystonia and dystonia secondary to other causes also occur.[1]

Figure 1



## Aetiology

Primary dystonia occurs as an isolated presentation and has a genetic (or presumed genetic) aetiology (Table 1). Inheritance is often autosomal dominant; a careful family history may reveal previously undiagnosed relatives with milder phenotypes. Dystonia occurring secondary to another disease process affecting the basal ganglia is the more common finding in children (Table 2). Psychogenic pseudo-dystonia is an important differential diagnosis.

## Clinical approach

The aims of the clinical assessment will be to confirm the presence of dystonia, and assess associated co-morbidities, functional impact, aetiology, perpetuating factors and complications.

### *History*

A summary of key elements of the history is provided in Table 3.

### *Examination*

The key aims of the examination are to characterise the dystonia and the degree of functional impairment, document associated motor disorders, review growth parameters and home video footage.

Firstly, inspect from a distance: note the use of orthoses, plot the height, weight and head circumference on a growth chart, looking specifically for malnutrition or microcephaly. Next observe more closely: assess if the dystonia is isolated, or if there is additional chorea, athetosis, or spasticity. Ask the child to walk if they can, preferably with shoes and clothes on at first, and then off. Video is very useful as gait can be very difficult to evaluate as children move swiftly around. Use functional techniques to bring out movement disorders: holding their fingers "as near to the nose as possible without touching it" (tremor), heel- toe walking and turning (ataxia), walking on the heels looking for inserted movements of hands and feet (Fogg sign). If you can see dystonia, note whether it is generalised, focal or segmental, and postural or fixed.

Next move them to the couch (even if wheelchair bound): assess the character of the dystonia and any additional movement disorders. Examine the cranial nerves with emphasis on fundi, eye movements, dysarthria, dysphagia (offer water if they drink orally), and tongue thrusting. Examine the limbs for evidence of other movement problems, e.g. dysmetria, intention tremor, spasticity, or neuropathy. Assess function through handwriting, drawing spirals, and performing tasks such as pouring water into a cup. It is also useful to video this, looking for posture and movement during a simple activity. Home videos can provide excellent insights, and should be reviewed.

### *Grading severity*

Severity of the current episode of dystonia should be determined. Features of increasing severity of dystonia include being unable to sleep, sit or lie

**Table 1: Childhood-onset primary dystonia[4]**

| Gene | Disease | Inheritance | Gene product & | location |
|---|---|---|---|---|
| DYT1 | Idiopathic torsion dystonia | AD | Torsin A | 9q34 |
| DYT3 | X-linked dystonia-parkinsonism | XL | TAF 1 | Xq13-1 |
| DYT4 | Whispering dysphonia | AD | TUBB4a | 19p13.12-13 |
| DYT5a | AD Segawa syndrome (Dopa responsive dystonia) | AD | GCH1 | 14q22.1-q22.2 |
| DYT5b | AR Segawa syndrome (TH deficiency) | AR | TH | 11p15.5 |
| DYT6 | Adolescent/adult-onset Idiopathic torsion dystonia (mixed) | AD | THAP1 | 8p21-q22 |
| DYT11 | Myoclonus-dystonia syndrome | AD | SGCE | 7q21.3 |
| DYT12 | Rapid onset dystonia-parkinsonism | AD | ATP1A3 | 19q12-q13.2 |

**Table 2: Causes of secondary dystonia[5]**

| Cerebral Palsy following hypoxic brain injury (commonest cause) | |
|---|---|
| **Metabolic** | |
| Biotinidase deficiency | Mitochondrial diseases |
| Creatine deficiency | Mucopolysaccharidoses |
| Galactosaemia | Neuronal ceroid lipofuscinoses |
| Glutaric acidura type 1 | Neurotransmitter disorders |
| GM1 and GM2 gangliosidosis | Niemann-Pick C |
| Hartnup disease | Proprionic acidaemia |
| Homocystinuria | Sulphite oxidase deficiency |
| Hypoparathyroidism | Tyrosinosis |
| Krabbe disease | Vitamin E deficiency |
| Lesch-Nyhan | Wilson disease |
| Metachromatic leukodystrophy | |
| Methyl-malonic acidaemia | |
| **Metabolic** | |
| Ataxia telangiectasia | Neuroaxonal dystrophy |
| Ataxia with oculomotor apraxia type 1, 2 | Panthothenate kinase 2-associated |
| Infantile bilateral striatal necrosis | neurodegeneration (PKAN2) |
| Juvenile Huntington's | Pelizaeus-Merzbacher disease |
| Neuroacanthocystosis | Spinocerebellar ataxias |
| **Drugs/Toxins** | |
| Phenothiazines | |
| Haloperidol | |
| Metoclopramide | |
| **Other** | |
| Alternating hemiplegia of childhood | Porencephaly |
| Basal ganglia infarction | Sandifer syndrome |
| Basal ganglia neoplasm | Striatal necrosis |
| HIV infection | Vascular malformations |
| Kernicterus | |

**Table 3: History**

**Birth history**

Pregnancy complications, Gestation, Mode of delivery, Cord gas results, Neonatal resuscitation, Encephalopathic features

**Early life**

Feeding, Seizures, Hospital admissions, Medical diagnoses

**Development**

Milestones achieved, Delay, Regression, School

**Family history**

Family tree, Consanguinity, Movement disorders, Neurological disorders, Stillbirths or early deaths

**Dystonia**

Age of onset, Progression, Focality, Diurnal variation, Functional impact, activities of daily living

**Dystonia exacerbating factors**

Gastro-oesophageal reflux, Constipation, Dental caries, Orthopaedic problems, including dislocated hips, fractures, Other causes of pain, Infection, Drug addition or withdrawal, Boredom, Emotional abuse/frustration/fear

**Dystonia complications**

Swallowing problems, Failure to thrive, Anxiety, depression, Aspiration pneumonia, Status dystonicus (potentially fatal exacerbation with multisystem dysfunction)

**Co-morbidity**

Spasticity, Oculogyric crises. Chorea. Other neurological problems

comfortably, and being systemically unwell. Children who show signs of systemic illness require urgent assessment and treatment for status dystonicus. Several formal grading scores are available.[6]

### *Investigation*

Investigation and treatment are interlinked, as a therapeutic trial of levodopa is often used as a diagnostic tool. This should be considered in any child with dystonia without an obvious secondary cause. Those with Segawa disease (dopa-responsive dystonia) typically show a dramatic improvement within a few days.[3,4,7]

Other investigations will be guided by the clinical findings and response to levodopa (when used), and should be directed at the possible underlying causes (Tables 1 and 2).

### Management strategies

There is a lack of robust evidence to inform pharmacotherapy for dystonia, therefore strict recommendations of first, second and third line medications are not practical.[4] Therapeutic strategies tend to vary with individual clinician preference and experience. As well as dystonia-specific therapy, identifying and treating precipitating factors is paramount (Table 3). Spasticity is a common co-morbidity, and it can be difficult to differentiate between spasticity and dystonia in some children. In these cases a pragmatic approach to symptom control should be taken.[4] Medications should be reviewed periodically, addressing whether the drug has had a positive effect on quality of life and the side effects. If there is no improvement with second line medication, consider discussion with colleagues at a complex case review or referring to a quaternary movement disorders clinic. As well as medication, supportive management in a multidisciplinary team including physiotherapy, occupational therapy, speech therapy and psychosocial support is essential.[3] Management is summarised in the algorithm (Figure 2, adapted from[3]).

### Status dystonicus

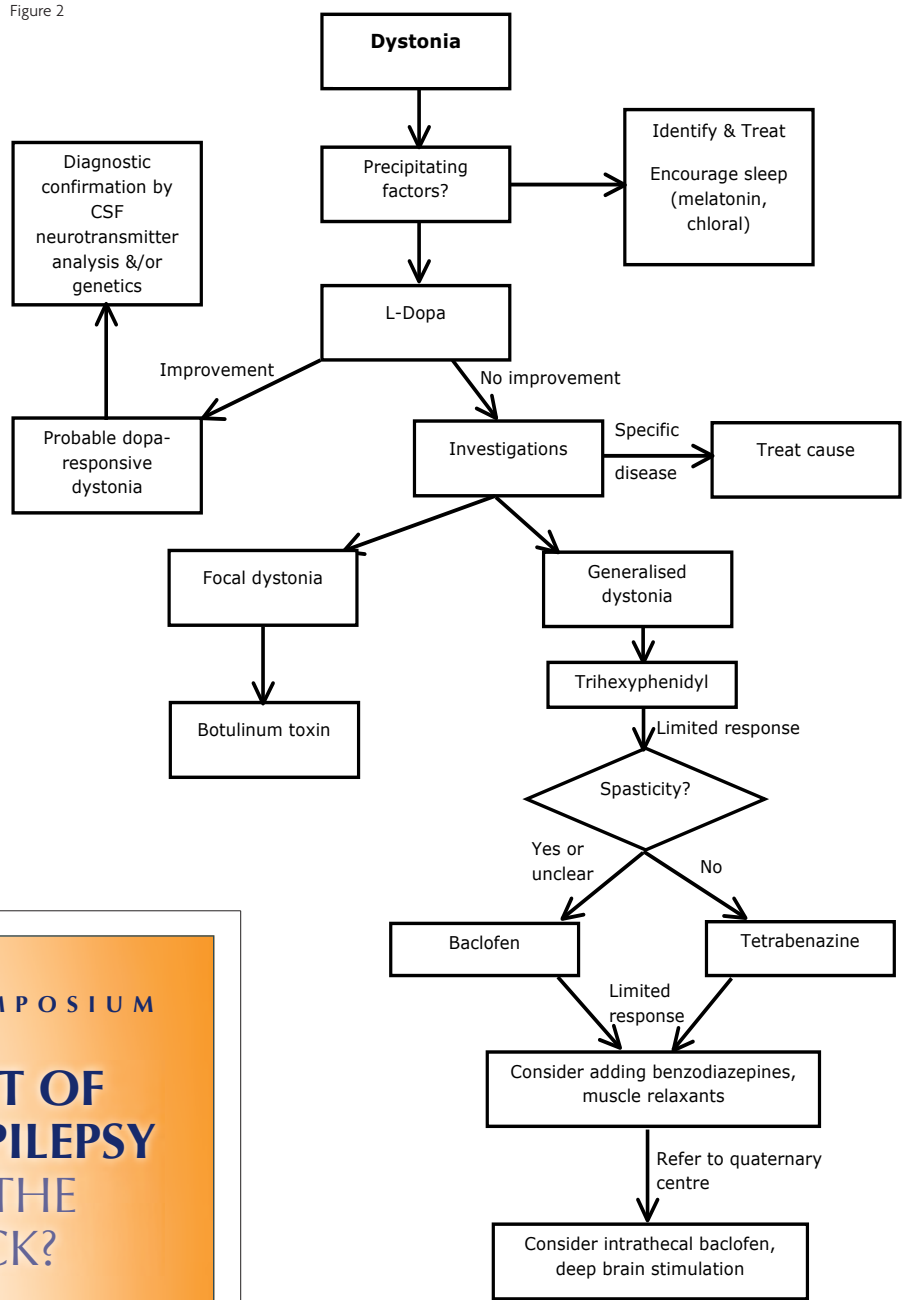
Status dystonicus is a potentially fatal episode of severe generalised dystonia. Complications include bulbar and respiratory compromise, and metabolic disorders such as rhabdomyolysis leading to acute renal

failure.[8] It usually occurs in children with known chronic dystonic disorders, but may occur in previously well children with acute illness affecting the basal ganglia or central nervous system. Children with status dystonicus should be managed in a hospital setting, and will often need intensive care. It is important to address precipitating factors (Table 3) and treat complications.[3] Supportive care such as invasive ventilation and haemofiltration for rhabdomyolysis may be needed. Therapy should be aggressive, with a slow weaning process. Treatment options include benzodiazepines, clonidine, propofol, and deep sedation with barbiturates. Surgical management, such as deep brain stimulation, will be required in up to one third of cases.[9] Once the dystonia severity has lessened, a slow wean of therapy can begin.

## Conclusion

Childhood dystonia is a challenging condition. A multitude of external and internal factors often play a part in influencing dystonia, no matter what the underlying cause. A pragmatic, multidisciplinary approach is vital. ◆

Figure 2



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