Adult-onset dystonia:
The importance of effective and timely treatment
Introduction
This brief guide has been developed to support the appropriate commissioning of treatment for dystonia.
Dystonia is a very complex condition which has far-reaching consequences for patients, primary and secondary healthcare services, society and the wider system.
The guide aims to raise the profile of dystonia as a long term condition, and seeks to demonstrate the whole system impact when patients are either not treated at all, or outside of effective treatment time parameters – in particular, it examines the negative impact on the quality of life.
This guide does not attempt to introduce a ‘one size fits all’ approach to the management and treatment of dystonia, but seeks to provide an opportunity for commissioners and providers to work together, to ensure that key service and treatment standards are commissioned and managed at a local level.
It does not promise to be a panacea, but rather a discussion document that can be used by secondary care clinicians with their local commissioners, to ensure that dystonia services and treatment are both better understood and commissioned appropriately within the new NHS.
What is adult-onset dystonia?
Dystonia is 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. It is estimated to affect 70,000 people in the UK.
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 muscles involved in movement, posture, speech, sight and mobility but does not affect 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.
Dystonia is a chronic disorder, but the vast majority of dystonias do not impact cognition, intelligence, or shorten a person’s life span. The main exception to this is dystonia that occurs as a symptom of another disease or condition that can cause such complications.
Adult-onset dystonia represents around 90% of all primary dystonias. It appears in mid-to-late life and usually only affects a single body area. In a minority of cases (around 25% of cases) it spreads to a second area and in around 10% of cases to three or more areas. Symptoms can include twisting or pulling of the neck (around 65% of cases), forced eye closure and/or functional blindness (20%), difficulties with eating and/or speaking (20%) and hard/arm problems including writing (20%) (based on information from Dystonia Society members n.b. numbers add to more than 100% as for some people more than one area is affected).
Dystonia causes varying degrees of disability and pain, from mild to severe. Whilst there is currently no cure, there are treatments available.

So why does it matter?
Dystonia affects quality of life, employment, daily activity and social interaction as well as causing pain and mental health problems.
Unmanaged dystonia places a range of unnecessary burdens on the NHS.
The effect of adult-onset dystonia on the individual
Dystonia has a significantly adverse impact on all quality of life (QoL) measures as well as on mental health. Among those with focal dystonia, QoL reports were as follows (at baseline on day of treatment) (Statistics from Guox 1998):
- **Mobility**: 53% of people report mobility problems
- **Daily activities**: 64% report problems with daily activities and 23% report problems with self-care
- **Pain**: 83% report severe or moderate pain
- **Mental Health**: 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.

On all these dimensions, QoL for people with focal dystonia is significantly worse than the general population. Untreated neck dystonia also has a significant effect on employment status. In one study, 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).

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.

Experience of dystonia

The effect of untreated adult-onset dystonia on the health and social care services
There is consensus that failure to manage adult-onset dystonia results in substantial burdens for the NHS. These can include:
- **Avoidable visits to GPs**
- **Avoidable steps and delays in a patient pathway**
How is adult-onset dystonia treated?

Botulinum toxin (BoNT) injections
In most cases of focal dystonia, the usual first line treatment is regular BoNT injections into the affected muscles, usually around every 12 weeks. BoNT 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. It also has a significant beneficial effect on pain. BoNT is a very effective treatment when administered at the appropriate time and frequency.

Medications
In a minority of cases, BoNT injections are not effective due to the patient developing immunity, unmanageable side effects or because the dystonia is too complex or widespread to identify injection sites. In these cases, medications such as anticholinergics, GABA agonists and anti-convulsants individually or in combination are often tried although evidence in well defined trials is lacking. However, for many patients, these medications either do not work or the side effects are unmanageable but a minority find them of significant benefit.

Surgery
As a last resort for patients with severe dystonia, Deep Brain Stimulation (DBS) is considered to be a good option. In addition, other surgical interventions such as peripheral denervation or myotomy/myomectomy are sometimes tried.

Other allied health treatments:

Physiotherapy
Studies have shown that physiotherapy enhances the effectiveness of BoNT in treating neck dystonia and high percentages of patients report it as being helpful. Because of the specialised nature of dystonia, this treatment needs to be provided by a neuro-physiotherapist familiar with the condition.

Psychological support
Because of the high prevalence of mental health conditions among patients with dystonia, psychological therapies such as cognitive behavioural therapy (CBT) are often desirable.

Pain management
Referral to pain management has been shown to be effective for treatment of chronic pain caused by conditions such as dystonia.
If the NHS is to succeed in its QIPP challenge, dystonia services must be robustly and effectively commissioned on the same principles as other chronic diseases. Services need to be clinically effective, patient centred, responsive, integrated, timely and multidisciplinary.

- The commissioning of high quality dystonia services based on these principles will improve outcomes for patients
- The commissioning of well coordinated services for dystonia including timely access to BoNT and allied healthcare professional input will reduce the number of steps a patient takes within an individual pathway; it will also result in a reduction in the number of hand-offs between professionals
- The commissioning of timely treatment for dystonia through BoNT will result in fewer GP appointments
- The commissioning of an effective treatment pathway for dystonia patients with an appropriate integrated treatment plan with access to a full multidisciplinary team, will improve the patient experience and outcome – enabling them to actively manage their condition. Patients will also benefit from reduced pain and reduced risk of abusing pain relief medication, and improved mental health.

The appropriate commissioning of timely dystonia BoNT treatment will have enormous impacts on the whole system, allowing patients to be more active and independent and to remain in employment.

Conclusion

Currently, the best treatment option for dystonia is the injection of BoNT into the affected muscles. Whilst the access to BoNT is improving, and treatment regimes are timely in most instances, there remains inconsistency in:

- Access of initial and subsequent effective treatment
- Providing a holistic approach to patient care
- The coding and charging of services
- Adverse impact on quality of life

If CCGs are to commission services effectively and appropriately, we need to ensure that dystonia treatment is available to all, that clinical variation is reduced or avoided completely, and that patient outcomes are improved.

This guide has been developed to illustrate the importance of effective treatment for dystonia, and is aimed at encouraging positive and open communication between providers and commissioners to ensure that dystonia patients get access to the treatment and services that they need and deserve.

References


The British Neurotoxin Network (2012). Responses to questionnaire.


The Dystonia Society (2012). Responses to questionnaire.