



Cervical dystonia

The big picture

Europe's largest centre for the treatment of cervical dystonia (dystonia of the neck muscles) is the National Hospital for Neurology in Queen Square, London.

Professor Kailash Bhatia is responsible for this clinic which currently treats over 450 patients with cervical dystonia.

Here Professor Bhatia highlights some of the important issues.

The figure of 40,000 is usually quoted as the total incidence of dystonia in the UK but Professor Bhatia believes may be an understatement. "This figure is too low. I believe the true figure is closer to three times this value," he says. Cervical dystonia (often referred to as spasmodic torticollis) is the most common form of dystonia. It accounts for around two thirds of the total number of cases of dystonia in the UK – in other words 25,000 people are affected by cervical dystonia.

To the Professor's dismay, it is still a very under-recognised condition. Patients are still reporting that it can take up to three or four years before a diagnosis is made and they are finally seen by a neurologist. "Unfortunately there is still misdiagnosis at the primary care level," he says. "People are often sent to see a rheumatologist, osteopath or physiotherapist or even psychiatrist. It is only after doing the rounds, there will eventually be a diagnosis of dystonia and people come to the clinic. We need to raise the awareness of dystonia amongst all these specialists, particularly as there is an effective treatment available through botulinum toxin injections," he says.

"It is important to identify people affected by cervical dystonia early on. The pain and the strong spasms resulting from the dystonia are not necessary. These symptoms can be effectively dealt with by regular botulinum toxin injections. As these symptoms can cause anxiety and depression, cervical dystonia can have a big effect on the quality of a person's life," he adds.

Botulinum toxin as a therapy for dystonia has been around in the UK for over twenty years. "We were the first centre to start injections back in 1987. These were done under Professor Marsden. I still treat patients who attended the very first clinics and who are still finding the treatment effective," the Professor comments.

He acknowledges that in medical terms this is a relatively short time interval. However, the treatment has by now been used by thousands of people throughout the world and is safe. "There has not been any suggestion in the medical literature that people who have been having this treatment for focal dystonia over eight or ten years have had any long-term negative effects. It is a safe treatment though there is still a need to research any long-term effects." As regards the recent reports of generalised weakness resulting from botulinum toxin injections, he is keen to point out that these may be related to its use in children with spasticity with high doses – quite a different usage to that in adults with dystonia.

Some Society members have raised questions about the way that botulinum toxin appears to 'thin' muscles. Professor Bhatia reassures: "As the injections are stopped, so the muscles will start to fill out. It's very unlikely that after you stop the injections they will remain thin forever."

As with all types of dystonia, people with torticollis can find their dystonia goes into remission.

"The ball park figure in our experience is about one in ten cases go into remission. It's more common for this to happen in the cases of younger onset – people under 40 years old. However, we do know of this happening to people in their 50's and 60's," comments Professor Bhatia. "Unfortunately, for most people the dystonia will reoccur after five years. Permanent remission rates are much lower – well under 5%."

In most cases cervical dystonia settles down after two or three years to being a stable condition without much change in the muscles affected. It is only in the first few years that it is likely to spread. "If you're anxious, tired, depressed, it can temporarily get worse but it will normally settle down again after the stress is removed," he says.

"Cervical dystonia accounts for around two thirds of the total number of cases of dystonia in the UK..."

Cervical dystonia

The big picture continued

There has been a belief that if your head turns to one side, you should try to develop the muscles on the other side. "I personally don't think there is good scientific evidence to show that is beneficial but graded physiotherapy exercises along with botulinum toxin injections could be useful. There is no rule that works for everyone. There is a need for much more research into this area," comments Professor Bhatia.

A number of puzzles remain concerning cervical dystonia. For instance, as with most other types of dystonia, it is more common in women than men (by a ratio of 3:1 in the case of cervical dystonia). Torticollis is more common pulling to the left than to the right. The likeliest time for people to report cervical dystonia is in their 50's. Why does it affect people in the 50-70's age range frequently? Why is it that when you are young you get generalised dystonia but, when you're older, dystonia tends to be restricted to certain areas such as the neck? These questions amongst others await some answers in the future.

What causes cervical dystonia? This is still a mystery. The scientific community still has no clear idea of the mechanisms underlying the condition. However, Professor Bhatia and his colleagues have a hunch: they believe that there are actually a number of 'cervical dystonias'. Various pieces of their research already point to the fact that people with cervical dystonia are not a homogenous group so that although people with cervical dystonia all have similar symptoms, the underlying causes are likely to be varied due to a mix of environmental and genetic factors.

Professor Bhatia and colleagues are currently formulating an exciting project that could provide some very important answers to this question by potentially finding genes associated with each type of cervical dystonia. Then the way will be open to producing individualised treatments that could cure the problem at a cellular level, rather than just masking the symptoms as is currently the case.

Research update

Mechanisms of childhood dystonia



Dystonia that starts in childhood can be one of the most severe and disabling forms. Recently the Society made an award of £25,000 towards continuing the important work of Dr Tom Warner and his team. This work is shedding light on the mechanisms that cause malfunctioning in the cells controlling the movement of affected children.

Childhood dystonia is often caused by a genetic change (mutation) in the DYT1 gene. This gene encodes a protein called torsinA, which is found in most cells, especially those in our brains. By understanding how this gene and the associated protein work in healthy children and how they differ in children with dystonia, Dr Warner's team are gaining important insights into why the dystonic movements arise. It is hoped that in the future this research may identify novel treatment strategies for this incurable condition. In addition, the study of the DYT1 gene is also relevant to the more common forms of focal dystonia appearing in adulthood (such as cervical dystonia and blepharospasm), as work has shown that this gene can act to increase the susceptibility of some people to develop these forms of dystonia.

The team has investigated the way in which mutant torsinA causes dystonia in nerve cell models and has identified a novel protein that is directly associated with it. This protein, called snapin, is involved in the release cycle of the packets of chemical messengers (called synaptic vesicles) which allow our nerve cells to communicate to each other and ultimately make the brain work. The team has also found that the normal torsinA protein is involved in regulating this release process, a function which is lost by the mutant form. These findings are of great significance as, for the first time, they provide a link between the mutated DYT1 gene and abnormal nerve cell signalling which leads to dystonic movements.

Cervical dystonia

DBS for Cervical dystonia?

Deep Brain Stimulation (DBS) is widely known as a potentially successful treatment in cases of generalised dystonia, but does it have a role in cervical dystonia?

*Mr Jeremy Rowe, consultant neurosurgeon at the Royal Hallamshire Hospital in Sheffield talks to **DystoniaMatters!** about the potential uses of DBS for this form of dystonia.*



We take our neck movements for granted and do not even think about them when they are normal. To manage even basic functions such as swallowing or eating requires neck control. Therefore it is important when considering treatments for cervical dystonia to look at the impact of the condition on all aspects of the lives of those affected..

The appropriateness of DBS for cervical dystonia will depend on how effective the existing treatments are for the individual patient, the extent of the muscle groups being affected by the dystonia and, of course, the impact that this has in terms of the activities of daily living.

Patients with spasmodic torticollis would most likely be treated in the early stages with botulinum toxin to manage their symptoms. It can work very well particularly if the affected the muscles are in a limited area. However the more extensive the dystonia is, then the harder it can be to get adequate symptom control with botulinum toxin. In addition with time there can be a loss of efficacy or resistance, or more rarely adverse reactions to the botulinum toxin.

Mr Rowe said: "Patients would only come to me to consider neurosurgery when they have failed in the conventional medical management of drugs and botulinum toxin."

When talking to patients about surgery, Mr Rowe always talks about the risks. "I would divide those risks up in different ways", he says. "The most significant risk is the risk of causing a bleed or a stroke when

"Should a patient decide to go ahead, there is then a very rigorous assessment to ensure that they and their symptoms are suitable for DBS."

inserting the electrodes in the initial DBS operation. This risk is small and is usually cited at around 1%, but it is real," he says. There are also potentially less important risks related to equipment malfunction etc which may require further surgery.

Should a patient decide to go ahead there is a very rigorous assessment to ensure that they and their symptoms are suitable for DBS. The tests are extensive and involve MRI scans of the head and neck both for planning surgery and to ensure the dystonia has not caused too much degeneration in the spine. There is also a battery of neuro-psychology tests that are undertaken. As part of the assessment a patient will also see a specialist movement disorder nurse and will be video recorded and assessed using the standard disease rating scales to provide a baseline.

One of the rewards of Mr Rowe's work is the interaction and relationship between his multi-disciplinary colleagues. He says "It is not a neurosurgeon operating in isolation. DBS needs strong backup from the movement disorder nursing team, neurology services team, neuro-psychology and the radiological imaging group, amongst others.

How does DBS contrast with selective denervation?

Mr Rowe points out some of the differences: DBS is different from selective denervation in that it does not involve destructive surgery. Electrodes are inserted into the brain without destroying the neural connections and these electrodes can be taken out without having caused permanent changes. With selective denervation the aim is to cut nerves permanently. Cervical denervation can be attractive because it offers a one-off permanent solution. However sometimes further operations are required as muscles that were initially denervated grow back at a later stage. "We must also recognise that following the initial operation for DBS further operations may be required when the battery needs replacing, though rechargeable batteries are in development," he concluded.

(See interview with Mr James Palmer about this treatment.)

Selective Denervation for torticollis



Mr James Palmer, Consultant Neurosurgeon and Medical Director at Derriford Hospital in Plymouth, is the principal surgeon in the UK carrying out this procedure.

Dystoniamatters! asks the questions.

● What is Selective Denervation (SD)?

This is a surgical operation to cut the nerves controlling the overactive muscles that are causing the symptoms of dystonia. The aim of the operation is to introduce a permanent paralysis to the muscles causing the problems.

● When should people consider surgery?

People normally consider SD when botulinum toxin treatment has been tried and has failed or is starting to fail. It is important to see patients early after the failure of previous treatments, not years later. If patients are referred early then results can be really good and offer a chance of providing a permanent fix for their cervical dystonia. Unfortunately, patients who are referred to me after many years often don't do well: the constant twisting of torticollis will have caused destruction of the joints in the neck over the years. However, even in these cases the procedure can often reduce pain and improve head position though it is difficult to return the neck to having a good range of movement once the bones have been damaged.

● When is surgery not appropriate?

If the cervical dystonia involves too many muscle groups, SD may not be feasible because it would involve trying to cut too many nerves. Some nerve pathways always need to be maintained to preserve neck movement. Some types of torticollis are easier to treat than others. The best results are usually obtained in cases of a rotation or twist of the neck. The hardest examples to treat are cases where the neck is pulled up and backwards

because there are so many muscles involved. This wouldn't necessarily preclude the operation taking place, but I would have to advise the patient that the chances of success were less.

● What are the benefits of this surgery?

For the right patient, there is a very reasonable chance (maybe 40%) of eradicating the effects of dystonia. Even in other cases there should be marked – and permanent – improvement in position and body image which for me is one of the most rewarding reasons for carrying out this procedure. Following surgery, a neurophysiologist or a physiotherapist are on hand to teach the patient exercises to build up the strength of their neck muscles. This is important.

● What are the disadvantages?

In a few cases there is a risk of incurring temporary swallowing problems. In extremely rare cases this can become permanent. As in the case of botulinum toxin, swallowing problems may follow injections but the symptoms do ease. Surgery can certainly leave you with several numb patches at the back of the head because some of the sensation nerves have to be removed. However it is an operation involving muscles which usually heals very well and there aren't many risks involved in this type of surgery.

● Is it difficult to get funding for this type of surgery?

Because it's approved by the National Institute for Clinical Excellence (NICE) and has a tariff, our hospital is quite happy to operate and provide treatment. It's free for every member of the NHS. I've not had a problem getting funding so far.

● Why are there so few neurosurgeons performing this type of operation?

More are not needed! When I first started, I worked out there were about 400 patients in the UK who might benefit from this operation. One surgeon can probably treat all 400 in their career. The technique is very different from other types of neurosurgery. I'm sure as I get older I will need to make sure another surgeon is trained to take over. You only need one or two surgeons in the UK to do this procedure as it needs particular expertise.

My story

Trip to the spa changed my life...

By Lisa Todd

I was only 42, a busy working mum, always planning getaways and trips to the theatre with friends and family. Then dystonia took away my spontaneity, crushed my social life and left me frightened of even shaking hands with someone in case it set off the spasms. Worse of all, I now live in terror of being hugged, even by my own family.

It happened one Wednesday in October, four and a half years ago. A girlfriend and I had decided to treat ourselves to a day at a luxury spa, warding off the winter blues with a float in the Dead Sea pool. After lunch we retired to the poolside for a pot of tea in our dressing gowns. I laid my head back for a snooze, only for the chair I was sitting on to collapse, throwing me backwards so that my head hit the ground and I was left winded and crying. I had sat down on a broken chair.

Invisible dog collar

Within 24 hours my head had started pulling to the left as if my neck was in an invisible dog collar and someone was yanking on the lead every other second. I couldn't keep still for a moment. But I assumed I'd just pulled a muscle and it would heal soon enough.



● Lisa Todd with her daughter Olivia at their home in Windsor

A few days later, I took the train to Bath with my daughter Olivia to visit her grandmother. As soon as she saw me, my mum said 'what on earth's happened to you?' My head was still twitching uncontrollably and the spasms contorted my face with the searing pain. I was terrified.

The next day I went to see a physiotherapist at the Bath Clinic. Puzzled, she sent me to see a rheumatologist and then the tour of specialists began. They all drew a blank.

At first I had no idea of the seriousness or permanence of the situation and did not want to pursue compensation because of the stress; all I wanted to do was get better! As the treatment bills mounted and the suffering continued, I did want to sue the pants off them and at least have some recompense for the devastation!

Meanwhile for nine weeks I struggled from rheumatologist to osteopath to physiotherapist in a desperate attempt to find out what had happened to me. None of the health professionals knew what the problem was although there are at least 40,000 known sufferers of dystonia in the UK. Dystonia is almost an unheard of condition, with both diagnosis and the on-going day to day challenges, remaining hurdles to be overcome.

During this time my neck pulled ferociously and I could not stand still, sit still or walk in a straight line. My neck muscles pulled in searing, painful spasms, my head contorted onto my left

shoulder. In bed I had to lie on my side and hold the pillow to stop my head from pulling me over. I lay on the couch when friends visited and couldn't hold my head up. I couldn't drive my daughter to school, cook a meal or keep my head still in the bath. It was how you imagine Chinese water torture to be and I felt suicidal for the first time in my life.

'Dystonia is an almost unheard of condition, with both diagnosis and the on-going day to day challenges, remaining hurdles to be overcome.'

My story *continued*

by Lisa Todd

I was given the number of a neurologist and was told I would need botulinum toxin. Two days later I was in a neurologist's waiting room unable to sit still. As he approached he took one glance and said I had spasmodic torticollis and need botulinum toxin injections.

After two sets of injections and with Christmas approaching, I was unable to lift my head off the couch. I was also being spoon fed M&S cottage pie by my husband and was unable to write a card or wrap a present for my little girl. In between (and many tears later) I co-ordinated my own programme of rehabilitation determined not to be beaten by this monster that had inhabited my body.



● *Walking in our garden*

Alternative therapies

I had cranial osteopathy, massage, acupuncture, remedial pilates and lots of gentle work in the swimming pool. (None of these treatments were available on the NHS and our financial resources soon became strained.) Cranial osteopathy rebalances the cranial fluids and helps improve the head position with the objective being to feedback to the brain that a straight head position is normal and the muscles do not need to fight. Acupuncture works by releasing the endorphins and therefore lifting the mood making it easier to cope and also provides relief from pain and stiffness in the muscles.

My injections followed at 3 monthly intervals. Botulinum toxin injections receive lots of media coverage mainly to do with cosmetic enhancement. The drug provides relief by temporarily paralysing the nerve endings which helps to calm the spasms. It is not administered in pin pricks, as for wrinkles, but in larger doses depending on the type of dystonia.

I was terrified each time I attended the clinic for further injections and suffered from the side effects. I would certainly rather have tried other 'non drug' therapies but could not have survived without it.

With more osteopathy, acupuncture and healing, together with huge support from my family, it is now nine months since I had the last injections and I am apparently in mild remission. In reality I can't get in or out of a bath, am unable to drive, go to the dentist, hairdresser, do the school run, go to the pictures, get on a plane, go to Waitrose unaccompanied, or wear high heels!

On the positive side I have started a painting course at my local college and it is fantastic to be out with other people doing something creative. I try to swim three times a week. After three years I can now turn my head to breathe when swimming, having progressed from doggie paddle to using one arm whilst needing to stop to breathe – a huge feeling of achievement. I love water and it provides great therapy. I have had some Shaw technique lessons which concentrate on taking the strain off the neck whilst swimming. Dystonia does nothing for your confidence, self esteem or sex appeal (you so feel like the world is looking at you with your crooked head) and there are many activities that are on my wish list – driving is number one!

The chance of remission is small but even mild remission is infinitely better than nothing! The injections have helped to manage my dystonia and I believe the complementary therapies and exercise have significantly improved my outlook for a better future.

Cervical dystonia

dystonia affecting the muscles of the neck (includes torticollis, antecollis, retrocollis or laterocollis)

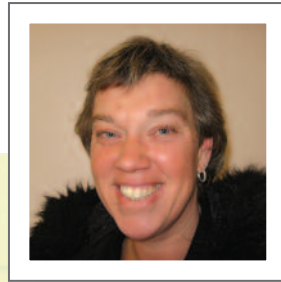
IPSEN Sponsored by Ipsen Limited

The dystonia society

To obtain the above leaflet about cervical dystonia, including information about spasmodic torticollis, or if you have any other questions about dystonia, please visit our website: www.dystonia.org.uk or call our helpline: **0845 458 6322**

Case notes

Mrs Caroline Minger



Condition: Cervical dystonia **Age:** 44

Address: Lives in Rainham, Essex with husband Pierre and has three children, Natasha (22) Francesca (18) and Mark (12)

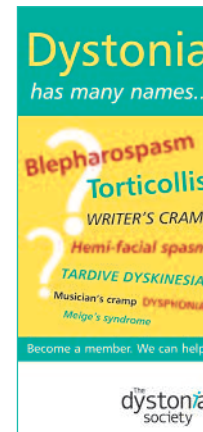
Patient notes:

"I started with tremors in my neck and head. This made simple tasks become more difficult, like putting on makeup. It was hard to keep my head still and my family couldn't understand. 'Don't be so nervous,' they said. But I couldn't help it. My head just wouldn't keep still. This went on for a few years and I was prescribed with anti-depressants. I knew there was something else wrong and eventually I was referred to the Queen's Hospital in Romford under Professor Findley. I'd never heard of the condition before when I was told I may have dystonia. I started on botulinum toxin injections about three years ago, these are repeated every 2 to 3 months. The injections do help tremendously. I started seeing Liz about the same time as commencing the injections. First it was home visits and then to the nurse-led clinic. I really appreciate the help and support I receive from Liz on a regular basis."

Liz Edmonds RGN works for Havering Primary Care Trust as a motor neurone disease and dystonia nurse specialist and operates her nurse-led clinic from the Disablement Services Centre at Harold Wood hospital.



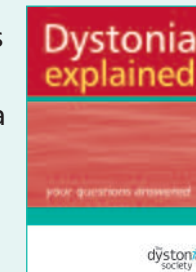
Diagnosis: The great thing about Caroline is that she is very receptive to education about her condition and she is aware of when support is necessary and where it should come from. She has had to cope with problems other than cervical dystonia which have been very hard because she is a sensitive person. She is coping because she knows the support is on-going which I think is highly important for her well-being.



Dystonia Society

Information leaflets available:

- Dystonia explained
- Driving & dystonia
- Blepharospasm
- How to find a therapist
- Hemifacial spasm
- Self-help techniques
- Writer's cramp
- Sensory tricks & dystonia
- Myoclonus dystonia
- What special educational equipment is available
- Paroxysmal dystonia
- Getting the best from neurological services
- Dystonia & genetics
- Graded neck exercises
- Laryngeal dystonia (*Spasmodic or spastic dysphonia*)
- Oromandibular dystonia
- Cranial dystonia (*Meige's syndrome*)
- Cervical dystonia (*Spasmodic torticollis*)
- Tardive dystonia / dyskinesia
- Online resources (*other websites*)
- Dopa responsive dystonia
- What you need to know about my condition (*care information*)
- Generalised dystonia
- Your starter for ten (*for telling people about dystonia*)
- Facial relaxation exercises for blepharospasm
- Access to Disability Living Allowance
- Deep brain stimulation (DBS)
- Ptoisis eyelid props / lundie loops
- Botulinum toxin treatment
- Access to the Blue Badge Scheme
- Statutory benefits
- Blepharospasm Sight Impairment services available
- Dystonia Guidebook for Kids (8-12 yrs)
- Blepharospasm or facial dystonia exercises
- A Guidebook for Young Adults who have dystonia (18-30 yrs)
- Coping strategies for oromandibular dystonia
- Coping strategies for blepharospasm
- Coping strategies for paroxysmal dystonia
- Coping strategies for myoclonus



We also have a selection of videos, DVD's and books that can be bought or borrowed. Please call our office number on 0845 458 6211 or our Helpline 0845 458 6322

for more information. Or you can visit our website www.dystonia.org.uk

Notice board

Coping with cervical dystonia



Tips from members:

- I go swimming and take part in Pilates classes. These are gentle exercises, they help to relieve stress and I feel good afterwards.
“Pilates develops the body uniformly, corrects wrong postures, restores physical vitality, invigorates the mind and elevates the spirit.” Joseph Pilates
- I avoid anything that jolts the neck and shoulders such as lifting heavy weights above the head as I find it causes more spasms and pain afterwards.
- I enjoy listening to music and watching TV as it helps me to relax. When I am relaxed the pain and spasms are easier to cope with.
- I put a newspaper down the back of my jumper to help with driving! It seems to support the neck and shoulders.
- When I am in a lot of pain or discomfort I use heated wheat packs to relieve it.
- I find that gentle massage is really helpful as it seems to relax me and makes the pain and stiffness easier.
- Having physiotherapy really helps but it is important to have a therapist that knows about dystonia and how it affects you.
- I use relaxation and meditation techniques, they seem to make the dystonia less active.
- I know that avoiding getting stressed and anxious makes my dystonia better.
- I have found that if I avoid lots of tea and coffee and other caffeine drinks, my dystonia seems to be less troublesome.
- I find that I need to eat a healthy diet regularly, otherwise I get really tired which makes the dystonia worse.

- I use a variety of high backed chairs or seats to provide head support, which reduces the discomfort I get from my dystonia.
- I touch the right hand side of my head or face which seems to realign the head and neck and stops my head turning.
- I have trained myself to use my PC mouse with the left hand so that if I need to, I can support my head with the right hand.
- I now drive an automatic car so that I can support my head with my right hand if I need to.
- I try to keep people on my left hand side as it is much easier to see and/or speak to them without straining.
- I have an excellent pillow on my bed which provides good support. Mine is a Tempura foam one but there are others you can buy.
- A neuro-physiotherapist has given me graded neck exercises which I do every morning. *(A video providing graded neck exercises is available from UK Office.)*
- A trainer at my gym has developed a fitness programme incorporating several exercises for my neck, shoulders and upper back which I do regularly.



The above is feedback from others with dystonia and may be called sensory tricks. We have no clinical evidence that they will work universally, and therefore it is up to yourself if you want to try them out. Please do feedback anything you try and find works for you.