National and international news and views
Networking Dystonia patients

New Zealand Dystonia Patient Network Inc.

News+Views

Newsletter 71, November 2014

2014 Seminar and AGM

Our 2014 Seminar was held at the Novotel Christchurch on Saturday 17 May 2014 and guest speakers were Dr Jon Simcock , Bryon Cope, Anne Recordon and David Barton.

Our first speaker Dr Jon Simcock spent 20 years as Medical Advisor to the Neurological Foundation and recently retired as a Neurologist.

Jon told us a little of the Neurological Foundation which has no government funding. Their mission is to alleviate suffering through research and education. The Foundation gives scholarships for Neuroscientists and to doctors for overseas study.

Jon spoke briefly about the Brain Bank Research Centre in Auckland where they currently hold 150 brains. He was optimistic about the rate of change and the increase of knowledge he has seen over the last 20 years.

One of Jon's earliest patients diagnosed with Dystonia is dopa-responsive. Jon traced her family history over generations and found other family members also with Dystonia.

Developing a diagnostic test for the medical profession would help research, and some of his findings were that mumps, rubella, dust and tremor history either affect or in some cases worsen an existing Dystonic condition.

Bryon Cope – another of our excellent speakers – is a Depression Support Facilitator. His role includes community education and delivering individual and group peer support for those people who have been affected by depression.

Bryon has a Diploma in Mental Health and Addiction studies, and has worked in this role for over five years. He is a father of two and lives in North Canterbury.

Having experienced depression Bryon was able to give us valuable insights, as well as being a very entertaining speaker.

David Barton (Past President, NSDA) shared some updates from the 3-day NSDA meeting in Chicago he attended in May this year. The National Spasmodic Dysphonia Association was founded 25 years ago (1989) so was celebrating the Silver Anniversary of the first meeting.

Spasmodic Dysphonia (SD) is the voice that results from dystonia in the vocal cords, or laryngeal dystonia. SD is a focal dystonia - just like blepharospasm or spasmodic torticollis (cervical dystonia) or writer's cramp. Several of these focal dystonias also have their own organizations.

Highlights from the NSDA Symposium in May 2014

The NSDA has recently been approached by Scott Adams, the famous cartoonist, who created the Dilbert character, and he is happy to help with SD awareness. Scott Adams has SD himself, and is very eloquent about the challenges it has thrown up in his career and journey through life. As with any famous person who has dystonia, a big plus is their familiarity, so ordinary patients can say "This person also has dystonia, just like me". It helps deal with a situation where

friends and family think you are unique, and are perhaps less sympathetic because they have never seen examples of other people like yourself.

Other highlights, in no particular order, from the presentations, were as follows:

A procedure called 'Pre-implantation genetic diagnosis' is now available for some forms of dystonia. Apart from the ethical issues, where people may differ on its desirability, this testing allows couple to try for a 'test-tube' baby – often there are several embryos to choose from, and the clinician can choose an embryo which does *not* carry a gene for dystonia. Understanding of the DYT1 gene means an embryo can be tested at the in-vitro fertilisation stage. If the gene is present, the prospective parents can made the decision whether to proceed.

Dr Christy Ludlow said the disorder is plastic, and fluid; meaning it can evolve over time. Focal dystonia is a functional problem rather than something concrete, such as a lesion in a particular place in the brain. Researchers have found differences in post-mortem brains in many different places. The disorder is thought to have more to do with faulty brain circuitry rather than a problem in a command centre.

The role of Genetics was discussed. In just about every case, no other members of the family have it (the audience were asked to put up their hands – no-one did). This makes a focal dystonia much harder for geneticists to study, so scientists are very excited when they find family combinations – e.g. mother/daughter.

Current thinking is that the genetic risk factor is for *any* focal dystonia - not a specific one like BEB, WC or SD. Penetrance is only thought to be around 25%. That is, 3 out of 4 may carry a gene for focal dystonia but won't go on to develop symptoms. Research has discovered genes like DYT1, DYT4, DYT6, but does not, currently, appear to be leading to focal dystonias. Perhaps focal dystonia is *one* single disorder, and this could explain why you can see a range of dystonias in some large families, so say if one person has CD, others could have BEB or WC.

Doctors now believe there is no one single definite cause for focal dystonia; instead there is likely to be an interaction of factors – partly genetic, and then these can be triggered by injury/trauma. Some mild risk factors have been noted: dust exposure, frequent voluntary voice use, history of tremor and other dystonias.

Doctors have noticed that symptoms change over a period of time. They are not sure why – a response to prolonged Botox treatment, or maybe the disorder itself is evolving. They have noticed changes in dopamine levels in dystonia patients – going up with conscious activity and down when resting; but they do not recommended sinemet (only for DRD = dopa-responsive dystonia).

A major thrust of current research is to develop a diagnostic test for focal dystonia – to make sure research conclusions apply to the right population etc. Currently in a study there are 177 patients, and an 'SD Attributes' inventory has been developed. The doctor will check off items, like voice breaks, normal voice when whispering etc. to lead to a major and perhaps secondary diagnosis – to establish a percentage for each type.

Interesting information from Japan has come in. Doctors had thought that the Japanese population was free from focal dystonia, or the condition was very rare. They now think this view was wrong, the reason being that the condition was not being diagnosed.

There are two types of SD – ABductor, where the vocal cords flip open, giving a breathy voice; and ADductor, where the larynx suddenly slams shut, breaking up the voice. Doctors find it quite difficult to reliably distinguish between the two types, and they can be mixed. Here are some diagnostic sentences to elicit symptoms:

"We eat eels every day" or counting numbers from 60 to 69. A patient who struggles with this vocal task probably has ABSD.

Difficulty with saying "the puppy bit the tape", or counting numbers from 80 to 89 can illustrate a case of ADSD.

There was an interesting talk about Botox. Originally there was 'just Botox', but now there are seven different serotypes: A, B, C, D, E, F and G. Botox is actually just a brand-name, and technically we should use the full name, 'botulinum neurotoxin'. Other brands include Dysport, Xeomin, and Myobloc. The Chinese have recently started producing botulinum toxin – some is counterfeit, some is correctly labelled. One doctor speculated there could be a risk of BSE (mad cow disease), because the Botox is manufactured in a gelatine base rather than albumin.

A new development with the neurotoxin is subtypes A1-A6. Why is this important? Each subtype produces distinct antibodies so if a patient develops immunity to one, a different subtype can be tried. The doctors stressed that these are not yet commercially available.

A talk about surgery followed, and Dr Blumin stressed that this treats the symptoms, not the underlying cause. One technique is thyroplasty - inserting a shim to keep the vocal cords apart. The long-term results are not good. One doctor mentioned it is really a sensory trick or *geste antagoniste*, where the brain is temporarily fooled into not producing the twists and spasm typical of dystonia.

Surgeons have seen patients where vocal cords respond to a shim by changing their shape from straight to curved, so that the voice can once again be distorted; their conclusion is that dystonia is really a brain disorder, and the brain wants to produce the dystonic voice.

The main surgical technique at present is SLAD-R , or selective laryngeal adductor deinnervation-reinnervation. Reasons for: the treatment is permanent, patients can come off the Botox roller-coaster, and convenience (no returning for frequent treatment). There is not a lot of long-term data on this SLAD-R surgery though. It is not effective for tremor, and is not recommended for people over 65, due to slow nerve regeneration. Recovery from the surgery takes at least six months.

Dr Blitzer (the first ever doctor to inject Botox for SD) is very anti-surgery; he says it is like a sensory trick and the dystonia will come back.

The medical panel discussed whether vocal rest after a Botox shot is advisable. They recommend speaking after the injection - it helps spread the toxin into the target muscle.

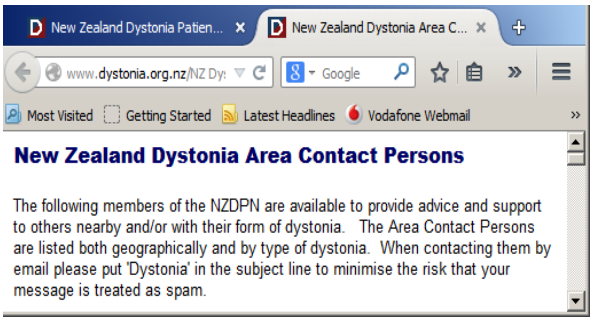
Area Contact Persons

We would welcome expressions of interest from any member of the Network who would like to serve as an Area Contact Person.

The role involves 'being available' for enquiries from the newly diagnosed or others who want to talk about dystonia with another patient who has 'been there, done that'. Dystonia can be a socially isolating disorder, and it is very helpful to be able to reach out to others who understand. From the Area Contact Person's perspective it is helpful to 'pay forward' the support you yourself have received, and it helps with your own acceptance of the disorder when you help others.

Although we have Support Groups in several areas of New Zealand, they are not always easily accessible to people in the less populated areas.

Our Area Contact Persons are listed on our website with their contact details - usually an email address.



If you would like to volunteer, please email info@dystonia.org.nz. You can change your mind at any time if you wish to discontinue.

Changes to the Executive Committee

We would like to thank all members of the Committee who have recently retired for their service to the Network.

A group of members requested a special meeting in June, and subsequently the Chair and Treasurer resigned. The Committee are happy to answer any questions about these events at the next Annual General Meeting. This is set down for Auckland on May 9th next year. The Committee are working to move forward from this situation, and have commissioned a review to the

incomplete accounts presented to the AGM, so that we can meet our obligations to the Charities Commission.

Further details about the 2015 Seminar will be published in the next newsletter and at the Network's website www.dystonia.org.nz

2015 MAY						
SUNDAY	MONDAY	TUESDAY	WEDNESDAY	THURSDAY	FRIDAY	SATURDAY
					1	2
3	4	5	6	7	8	9
10	11	12	13	14	15	16
17	18	19	20	21	22	23
24	25	26	27	28	29	30
31						

A special General Meeting was held at the Domain Lodge, Auckland on 23 September and those in attendance unanimously elected Philippa Hooper and David Barton to the Executive Committee. The Minutes of this Special General Meeting are available at the Network's website: www.dystonia.org.nz

The NZDPN is governed by an Executive Committee, elected each year at the Annual General Meeting.

Your NZDPN Executive Committee is:

Chairperson (Acting) Alison Fitzpatrick
alisonfitzpatrick@yahoo.com.au
Secretary Desiree Sargon
desiree.sargon@roche.com
Treasurer (Acting) David Barton
dsbarton@ihug.co.nz
Network Manager Philippa Hooper
phooper@paradise.net.nz
E-mail Contact and Information Officer
Alison Fitzpatrick alisonfitzpatrick@yahoo.com.au
Webmaster
David Barton - dsbarton@ihug.co.nz

To contact by mail please write to:
Executive Committee
New Zealand Dystonia Patient Network
PO Box 305 375,
Triton Plaza
AUCKLAND 0757

Our 3-fold mission is:

* To support dystonia patients with information, advice and networking opportunities
* To increase awareness about dystonia – among the medical community and the general public
- To encourage and facilitate research, with the aim of seeking better treatments, prevention, a cure.

Donations and membership

The NZDPN is a Health Promotion Association registered with the New Zealand Charities Commission. As well as encouraging research into dystonia and promoting awareness of our condition, our mission is to provide information and support to all those affected by dystonia. We are a 'grass-roots' organisation. Most of our leaders have dystonia themselves, and we are entirely reliant on donations, membership contributions and other charitable grants.

The Network invoices members once each year, in February, for the Annual Subscription. Membership is $25 per annum and applies for the calendar year in which the payment is made. Donations are also welcome and are tax deductible. Internet Banking details are:

NZ DYSTONIA NETWORK
NATIONAL BANK, WAIKANAE
06-0577-0110415-00
Please complete the 'Code', 'Reference' or 'Particulars' fields to let us know whether a deposit is a donation or membership. To send a cheque our address is:
The Treasurer,
NZDPN,
PO Box 305 375,
Triton Plaza,
Auckland 0757

PLEASE NOTE - Receipts will be automatically issued for amounts of **$100** and over. Receipts for lesser amounts will only be issued upon request.

Support Group Leaders can be contacted by Emailing info@dystonia.org.nz or contact details are available on our Web-site www.dystonia.org.nz

(Charities Registration: CC10565).

Disclaimer: Nothing in this newsletter is intended to serve as medical advice on dystonia. The NZDPN recommends that you consult your own doctor(s) and other health professional(s) regarding your diagnosis and treatment.