National and international news and views
Networking Dystonia patients

New Zealand Dystonia Patient Network Inc.

News+Views

Newsletter 74 – November 2015

2016 Seminar

The Network is pleased to announce that our annual Seminar and AGM will be held at the Sudima Hotel in Rotorua on Saturday 7 May next year.

All members are invited to attend for a fun day of learning, networking and companionship, with friends, partners, family and friends.

The Committee are devising a programme and choosing speakers. The Seminar will be followed by the Annual General Meeting.

More details will follow in the first newsletter of 2016, including the program, notice of the AGM and a registration form. Please also check the Network's website at www.dystonia.org.nz for updated information.

The contact details for booking direct with the Sudima Hotel if you wish to stay at the same venue as the meetings are:

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Phone: +64 7 348 1174
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PHYSICAL ADDRESS
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Rotorua 3010

Highlights from an address by Dr Mark Simpson to the Annual Seminar this May

Definition of Dystonia

Dystonia is a hyperkinetic movement disorder characterised by sustained or intermittent muscle contractions causing abnormal (often repetitive) movements, postures, or both. Dystonic movements are typically patterned and twisting and may be tremulous. Dystonia is often initiated or worsened by voluntary action and associated with overflow muscle activation.

Dystonia has been recorded in the medical literature for the last 100 years. Looking back through accounts of disability, historians speculate that the Flemish painter Brueghel was portraying dystonia cases back in the 16th century.

Historical representations of Meige's syndrome (left) and Writer's cramp (right)

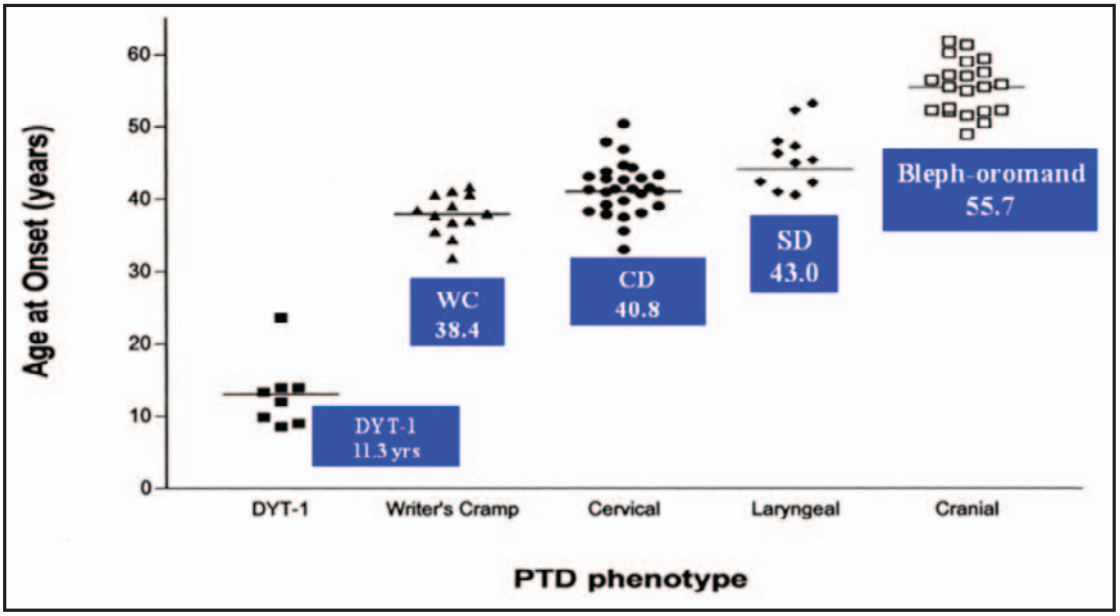
Age of Onset

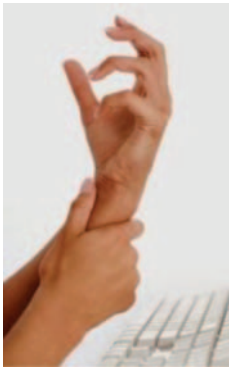
Members will know that dystonia can impact one or more areas of the body. The term 'Focal' is used to describe dystonia that affects a particular part of the body. Often the dystonia is task-specific - for example embouchure dystonia affects the lips, but not when eating; only when needing the fine motor control required to play a wind instrument, such as the trumpet or French horn. And spasmodic dysphonia usually only affects conversational voicing while other functions of the larynx are normal.

Examples of focal dystonia are

- Writer's cramp (WC) or focal hand dystonia - affects writers, golfers
- Cervical dystonia (CD) or spasmodic torticollis - posture of the head and neck is affected
- Spasmodic dysphonia (SD) or laryngeal dystonia - the symptom is a broken, effortful voice
- Blepharospasm - the muscles around the eyes squeeze shut inappropriately

Researchers have explored the connection between the type of dystonia and its age of onset. The graph below shows five different types of dystonia. In each case the average age is given, and the dots represent individual cases and how they are spread out.





Dr Simpson's interpretation of the diagram :

- Most dystonia begins as focal dystonia.
- Most cases of genetic dystonia begin in childhood, and rarely begin after age 26.
- The range of age of onset of focal dystonia is small
- It is unusual for a patient to present with writer's cramp at age 70, or blepharospasm at age 20
- When dystonia occurs early in life, syndromic and structural causes should be carefully excluded.

Why is this information useful? Firstly, scientists may be able to make connections between aging in the brain and how the dystonia manifests, thus shedding light on the cause. Secondly, the information helps with diagnosis - if a patient in their twenties presented with symptoms like those of blepharospasm, a movement disorder specialist would suspect the problem was unlikely to be a simple (primary) case of blepharospasm, and would look for an underlying disorder; whereas if the patient was in their fifties, the doctor may proceed with treating the symptoms as a more uncomplicated case of blepharospasm.

Secondary dystonia - the needle in the haystack

Metal and Mineral Metabolism
Wilson Disease
Neurodegeneration with brain iron accumulation Type I (formerly Hallervorden-Spatz disease
Neuroferritinopathy
Idiopathic basal ganglia calcification (Fahr disease)

Lysosomal Storage Disorders
Niemann-Pick disease type C
GM1 Gangliosidosis
GM2 Gangliosidosis
Metachromatic Leukodystrophy
Krabbe Disease
Pelizaeus-Merzbacher Disease
Neuronal ceroid lipofuscinosis (Batten disease)
Fucosidosis

Inborn errors of Metabolism
Lesch-Nyhan Syndrome
Aromatic amino acid decarboxylase deficiency
Triosephosphate Isomerase Deficiency
Guanidinoacetate Methyltranferase Deficiency
Molybdenum cofactor deficiency
Glucose Transport Defects

Amino and Organic Acidurias
Glutaric Acidemia type I
Homocystinuria
Priopionic acidemia
Methlymalonic Aciduria
4-hydroxybutyric aciduria
3-methylglutaconic aciduria
2-oxoglutaric aciduria
Hartnup's Disease

Mitochondrial disorders
Leigh Disease
Leber's Hereditary Optic Neuropathy
Mohr-Tranenberg syndrome-dystonia, deafness

Trinucleotide repeat disorders
Huntington's disease
Spinocerebellar ataxia-3 (Machado-Joseph disease) and other SCAs

Parkinsonian syndromes
Parkinson's disease
Progressive supranuclear palsy
Multiple system atrophy
Corticobasal ganglionic degeneration
Juvenile-onset parkinsonism
X-linked dystonia-parkinsonism (Lubag)
Rapid-onset dystonia-parkinsonism

Other degenerative processes
Ataxia-telangiectasia
Chorea-acanthocytosis
Rett syndrome
Infantile Bilateral Striatal Necrosis
Neuronal intranuclear inclusion disease
Ataxia with vitamin E deficiency
Progressive pallidal degeneration
Sjogren-Larsson Syndrome
Ataxia-Amyotrophy-Mental Retardation-Dystonia Syndrome

Secondary dystonia

Secondary dystonia is a consequence of some other underlying disorder. Clues that suggest a case of dystonia is secondary:

- Previous injury to the brain - e.g. stroke as a child
- Atypical site for age of onset
- Leg onset in an adult, cranial onset in a child
- Hemidystonia
- Presence of signs other than dystonia
- Abnormality on brain imaging

This table presented by Dr Simpson illustrates the difficulty of investigating a diagnosis of secondary dystonia. There are a huge number of possibilities to work through!

Report from NSDA Symposium, San Francisco, May 2015

Note: the report is specific to spasmodic dysphonia (dystonia that affects the larynx) but has features of interest to all types of dystonia)

Overview of Spasmodic Dysphonia:

Dr. Mark Courey covered the 'ABCs of Spasmodic Dysphonia', including the history of medical understanding of the condition, its causes, diagnosis, current treatments and research. This localised dystonia of the larynx (sometimes called Laryngeal Dystonia) is characterised by involuntary movements of muscle groups controlling the vocal cords. It results in reduction in voice-related quality of life, and therefore often in reduced social functioning and mental health. Patients' perceptions of their own vocal quality are often different from clinical analysis. There are compensatory mechanisms. And for many patients, treatment helps.

First identified in 1871, our voice disorder was initially described as "talking while being choked" and known at points in time as "nervous hoarseness", "hysterical voice" and "spastic dysphonia." By 1968, two primary types of SD were identified (AD and AB), though both kinds appear to result from a defect in the same part of the brain. It is identified as a "spectrum disorder" with varying types of muscle group involvement and varying degrees of severity.

The etiology, or cause, of SD is unknown, but clearly the condition is neurogenic in origin (originating in the brain and neural connections). There appears to be some genetic component. It is not degenerative (i.e. patients don't generally get worse over time). It sometimes co-occurs with other neurological conditions.

Diagnosis of SD is based on identification of symptoms rather than definitive testing. It is an interdisciplinary medical condition and teams often include otolaryngologists (ENT doctors), neurologists and speech and language pathologists ("SLPs"), previously known as speech therapists.

Currently available treatments include Botox injections (a temporary solution to hyperactivity of a muscle group that controls opening or closing of the larynx by preventing the connection between a nerve ending and the spasming muscle group) and surgery (a more permanent solution to the problem that is effective in some cases). Voice therapy can also be useful, especially when coupled with one of the treatments above, to better control airflow and vocalisation.

Dr. Courey also described some areas of research that have helped or are helping to understand this condition. The motor sensory function has been located in a particular part of the central nervous system, where this condition appears to originate. But the research continues to identify other parts of the brain that may play a part in SD, including the location where the intention to create speech occurs, the location where the onset of breath needed to phonate occurs and the areas which respond to feedback from the larynx and adjust the larynx muscles as feedback is received. This results in a complex system, making it difficult to identify when and where the abnormal activity originates.

SD Research: Drs. Gerald Berke, John Houde, Carlie Tanner

Dr. Berke described the goal of the committee put together by Dr. Christie Ludlow to produce a clear definition of Spasmodic Dysphonia, and its various manifestations, to differentiate it from other vocal disorders and to provide a scientifically sound basis for diagnosis of SD, which is essential when undertaking further scientific research. This has proved more difficult than expected at the outset because of the many individualistic forms of the disorder. Her paper on the agreed-upon definitions will come out soon. Dr. Berke also described work being done on drugs that may improve alcohol-sensitive forms of SD, studies continuing to look at areas of the brain involved in speech and the use of high-speed magnetic imaging.

Dr. Tanner described research she has done in the broad category of movement disorders at Kaiser in Northern California, helping us better understand the frequency of the various dystonias and sub-types in the population. Her comments focused primarily on SD, but included some comparative information about other dystonias. Females constitute about 80% of those affected. Average age of onset 45 – 55. Among the dystonias, Blepharospasm (spasms in the eyelids) was most frequent, Torticollis (spasms in the neck and head, formerly called "Wry Neck" and now sometimes called "Cervical Dystonia") second most frequent, Spasmodic Dysphonia third. Occurrence seems to go up with age, but at some point approaching old age goes back down. Risk factors appear to include: frequent upper respiratory infections (especially strep throat), surgeries, family neurologic diseases, mumps, measles, stress, voice overuse. (Oddly cigarette smoking was inversely correlated, but not a reason to take up smoking!) Genes are a minor factor, with SYT1 and THAP1 having been identified. Median time from onset to diagnosis: 5 years and 4.5 doctor visits on average.

Dr. Houde described current research being done to better understand how the central nervous system controls speech, particularly the role of the feedback loop during phonation, its timing and how sensory feedback affects SD. He referred to a "runaway feedback process" in people affected by this condition. At UCSF, this area is being studied with magnetic signals (both MEG and EMG are being used) to understand the timing of onset of the feedback perturbation that may be a major factor in SD.

Botulinum Toxin Injections: Drs. Dinish Chhetri, Mark Courey, Edward Damrose, Elizabeth DiRenzo

This panel described the various methods used to inject botulinum toxin for temporary relief of SD, including the particular muscle group injected for various forms of the condition, bilateral versus unilateral injections, points of injection, frequency, dose size and the trade-off between the initial period of breathiness and the length of decreased symptoms. Methods continue to vary significantly among practitioners. These can include 'point-touch', EMG-assisted etc. although fortunately we did not hear about any injections being given under general anaesthetic (which still happens in some situations). Benefits to patients with abductor SD, mixed AD/AB and tremor are more limited.

Surgical Options: Drs. Herbert Dedo, Shinya Hiroshiba, Gerald Berke; moderator Sarah Schneider

This panel described a number of surgical techniques being used to provide a more permanent solution for SD. Dr. Dedo provided history of his pioneering work in SD surgery. Dr. Hiroshiba reviewed work being done in Kyoto, Japan involving thyroplasty. Dr. Berke described SLAD/R (Selective Laryngeal Denervation and Reinnervation) surgeries involving denervation of the spasming laryngeal muscle group and reinnervation using a nerve from another location. Video clips demonstrated various surgical techniques. These surgical procedures are generally not available to patients where AB SD or tremor is dominant.

In memoriam - Dr Michael Taylor

We were deeply saddened to learn that one of our members, Dr Michael Taylor, died recently. Our Chairperson, Alison Fitzpatrick, shares her memories of this remarkable man.

Associate Professor Michael Taylor was one of my Chemistry lecturers at the University of Auckland in the 1980s. He was a kind and patient teacher and supervisor who always had time for his students.

Even though he had a distinguished research background at Oxford University, he never came across as superior or arrogant in any way.

Dystonia put an end to my own dreams of a career in Chemistry, and I went on to do other things. By a series of happy coincidences, I met up with Michael again in 2013. Imagine my surprise when I found out that a movement disorder had caused him to take early retirement from the Chemistry department! Undeterred by setbacks, Michael lived a full life outside the laboratory. He was hugely involved in the Native Forest Restoration Trust, the Royal Forest and Bird Protection Society and the Ornithological Society of New Zealand. He loved model aircraft, genealogy and poetry. We used to recommend books to each other and he would dutifully read mine and then report back with his verdict.

Michael came to our seminar in May, and found the day fascinating. He said it gave him a lot to think about and he intended to do some further reading on the topics discussed. Unfortunately, he became very unwell over the next few months, and died in October. The phrase "scholar and gentleman" was surely invented to describe Michael. It was such a privilege to know him and have him as friend and mentor. He will be greatly missed.

Generous donation

The Network wishes to acknowledge an extremely generous donation of $1000 made recently by a member who wishes to remain anonymous. This gift will help meet the costs of the current newsletter. Unlike the annual membership subscription, any donations are tax deductible because of the Network's status as a registered charity. The Treasurer will issue donors with an official receipt which can be submitted to the IRD at the end of the Financial Year.

A big THANK YOU to all those who are able to support the Network's Mission. All contributions are welcome.

Update from the Chair

From Alison Fitzpatrick:

Season's Greetings to everyone and we hope you enjoy our third newsletter for 2015. A warm welcome to new members who have joined the Network over the last few months. We look forward to seeing as many of you as possible at our May seminar in Rotorua next year.

We really appreciate your support because it makes a big difference to our modest financial situation. We thank those who are able to make a contribution.

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.

Governance of the NZDPN

The Executive Committee is elected each year at the Annual General Meeting.

Chairperson: Alison Fitzpatrick
Deputy Chairperson: Barbara Murrell
Secretary: Desiree Sargon
Treasurer: David Barton
Committee Members: Roger Terry, Alex Weir
Network Manager: Philippa Hooper

Left to right: Roger Terry, Alison Fitzpatrick, Barbara Murrell, Philippa Hooper, Des Sargon

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Donations and membership

The NZDPN is a Health Promotion Association registered with the New Zealand Charities Commission (Registration: CC10565). 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 to the calendar year in which the payment is made. Receipts are issued for amounts over $100, and otherwise on request. Donations are also welcome and are tax deductible if $5 or over.

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NZ DYSTONIA NETWORK
ANZ BANK, WAIKANAE
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Please complete the 'Code', 'Reference' and/or 'Particulars' fields to let us know your name, and whether a deposit is a donation or membership.

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