



Dystonia Medical Research Foundation

Calgary Chapter

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CHAIRPERSON'S CORNER

Hello everyone. Hope your winter has been a happy healthy one. Weather was not as extreme as in some years, so that was good news. We were away again for most of the winter, but watched TV to see what was happening back here: our thoughts are with you even when we aren't. Our winter, of course, was an easy one in Southern California.

Normally this time of year, we are all thinking of our Walk & Wheel, but that tradition has passed. Last year's physical participation was so low that interest has obviously waned. There are still, however, members of our group who will continue to do their own walks and fundraising this time of year: kudos to them, and their faithful supporters. Any of you who wish to participate this way are thanked and commended, and if you cannot do your own walk and wish to support one who does, that too would be appreciated. Just contact myself or Heather, and we can add your contributions. This was and still can be an important fundraiser for us.

I was interested to see the information about the 5th International Dystonia Symposium, held in Barcelona, Spain last October. It was a collaborative effort of the European Dystonia Federation, the Dystonia Medical Research Foundation, and the Dystonia Coalition. A record 560 attendees from 38 countries attended, including established experts, newer researchers, clinicians and some young investigators who had their travel sponsored by various patient organizations. Following is the latest "About Dystonia" to come out of that gathering.

"As researchers learn more about dystonia, the definition and classification of dystonia is changing. A Dystonia Classification Committee met just before the symposium to revisit the currently used descriptions and classification schemes. Because dystonia occurs in

diverse forms and with a spectrum of clinical presentations and symptoms, creating an accurate system to classify dystonia is essential for guiding proper diagnosis and treatment.

It has become clearer than ever that dystonia is more than a "movement disorder." Research is demonstrating, for example, that individuals with dystonia display differences in how their brains process sensory information compared to unaffected individuals. There may be additional non-motor components that may be overlooked.

The explanation for non-motor findings most likely lies in the fact that the brain structures implicated in dystonia are associated with not only movement but also behavior, cognition, and emotions. Depression is one of the clinical features that is associated with some forms of dystonia. Non-motor clinical features are sometimes overlooked in the treatment process. As a result, quality of life may be compromised. These insights lead to more effective treatment of the "whole person."

Rating scales used to assess dystonia in patients are being expanded and revised to more accurately measure motor symptoms as well as additional factors such as impact on daily living, pain, emotional health, and overall quality of life.

Dystonia is not just a disorder of the basal ganglia, as once thought. Symptoms in some forms of dystonia appear to arise as well from the complicated interaction between the basal ganglia and the cerebellum. Other brain areas including the motor cortex, thalamus, cerebellum, and corpus callosum are implicated in dystonia.

The basal ganglia remain an important point of focus in dystonia research. The concepts of "reduced inhibition" in controlling movement and abnormal neural plasticity as significant contributors to dystonia are linked to basal ganglia dysfunction.

Researchers continue to uncover new genes associated with specific forms of dystonia since the discovery of the DYT1 gene in patients with the most common form of generalized primary dystonia. 21 genes and gene markers are known to date. Various relationships and associations among different dystonias previously thought to be separate forms are being uncovered.

Molecular genetic studies point to a growing number of proteins believed to contribute to the disorder at the cellular level. A new understanding of how these proteins interact with each other is taking shape.

The role of the protein torsinA in neurons of individuals with DYT1 dystonia is becoming clearer. TorsinA is now known to interact with many proteins including other dystonia-related proteins such as THAP1 and SGCE.

Researchers continue to study the role of a protective factor, an additional mutation occurring in the DYT1 gene that appears to reduce the risk of a person with the dystonia-causing mutation from developing symptoms.

Although deep brain stimulation (DBS) for dystonia has proven to be highly effective, it is not the only viable surgical option. Additional therapeutic stimulation methods, such as cerebellar and premotor cortical stimulation, are under investigation. There is also a renewed consideration of ablative brain surgeries such as the pallidotomy to treat dystonia.

Physical therapy may be an underrated therapeutic tool. Larger, more complete studies are needed.

Increased brain plasticity appears to play a major role in some forms of dystonia. This might point toward possible biomarkers to assist in diagnosis and identify individuals at risk of developing dystonia. Increased plasticity appears to be a quality that distinguishes dystonia from other movement disorders.

Contrary to previous ideas about cell loss in dystonia, preliminary studies suggest that there may be a subtle loss of cerebellar Purkinje cells in adult onset primary focal dystonia.

Several speakers stressed the role of training and experience in treatment success with both botulinum neurotoxin and deep brain stimulation (DBS) and highlighted the importance of patients consulting qualified physicians and medical teams.”

It must have been exciting to be part of such a knowledgeable group and it is by these gatherings that all the wonderful research being done on our behalf is discussed and enlarged upon. The next International Symposium is expected to be held in 2015.

Time to think of our next meeting. We will gather at our usual spot, on May 12, Saturday morning from 10 to noon. We tried working on a speaker, but nobody from Foothills Neurology was available, but please plan to attend. I look forward to the support and information we gain from each other each time we assemble. Now that we meet only in the Spring and Fall, it is important that we have this personal support.

I look forward to seeing many of you, but if you cannot join us, please let us know how you are doing. As always, you are in our thoughts.

Marg Roy

ITEM OF INTEREST

ITEM 1

Researchers Identify Gene Associated with Primary Cervical Dystonia

As many of you will already have heard, a team of researchers led by Mark LeDoux MD, PhD, and including current DDMRF grant recipient Zbigniew Wszolek MD and Medical & Scientific Advisory Council member Joel Perlmutter, MD have discovered that a mutation in the C1Z1 gene appears to cause adult onset primary cervical dystonia.

The C1Z1 gene makes a protein expressed in certain neurons and which seems to be involved in cell cycle activities.

Gene discovery is a critical step toward identifying new therapeutic targets that may eventually lead to revolutionary treatments that interrupt or reverse the dystonia mechanism.

Gene discovery is also an important step toward improved diagnosis

Many thanks to the team of researchers who made these findings possible

ITEM 2

There is some new work going on with ultrasound that looks very interesting. A friend of Syeeda's first mentioned it to her, I mentioned it to Rae and she has done a lot of research on the internet and by phone into what is happening around the world. So far, there is no dystonia connection but I know that we and our friends and families fight other medical problems too.

What first attracted our attention was a headline **"Patients' Tremors Stopped After New Non-Invasive, Pain-Free Brain Surgery."** This article was about patients with Essential Tremor who had been given a single treatment of ultrasound by Dr Jeff Elias, associate professor of neurology at the University of Virginia. Dr Elias used ultrasound waves, focused to a specific point located by using an MRI machine, to treat the part of the brain that was causing the shaking. "This MRI-guided focused ultrasound allows us to actually visualize the entire treatment, while it's being delivered," said Elias. "And we still have the ability to interact with a patient and refine and polish the procedure." The procedure is unique in that it can safely and immediately deliver results, is reportedly pain-free, and there is virtually no recovery time.

One patient's hands shook so badly that she stopped eating at the table with her family. She said, "I pretty much ate over the sink. I could not use a knife and fork and just didn't want to sit at the table and be embarrassed." After the treatment, results were immediately clear, she could feed herself and write her name. Her handwriting had been illegible before. Another patient said that after the procedure she could take up some old passions like cooking and quilting, and was even thinking of playing the piano again.

The more we looked into new uses for ultrasound, the more we found. Work is being carried out in the USA, at Sunnybrook in Toronto, in Switzerland, in Israel – and quite probably other places too.

Rae found the following article on the Focused Ultrasound Surgery Foundation website. They are in Virginia.

Michael Milken joins Focused Ultrasound Foundation Council

CHARLOTTESVILLE, VA, March 7, 2012 -- The Focused Ultrasound Foundation announced today that medical research activist, philanthropist and financier Michael Milken has joined its Council, a select group of advisors and advocates who are helping to advance the organization's mission. A champion of medical innovations since the 1970's, Milken will support the Foundation in accelerating the development and adoption of focused ultrasound therapies for many of today's most devastating illnesses.



About Focused Ultrasound

An early-stage medical technology with the potential to revolutionize the treatment of many life threatening and disabling conditions, focused ultrasound is a non invasive therapeutic technology that uses concentrated ultrasound energy to treat tissue deep in the body while leaving surrounding healthy areas intact. Focused ultrasound therapy is being studied as a potential breakthrough in non invasive surgery and as an alternative or complement to radiation therapy. Its use is also being investigated in new treatments that deliver chemotherapeutics and other drugs to precise targets within the body at higher concentrations with less toxicity than currently possible. Researchers around the world are investigating the technology's use in treating many forms of cancer—including bone, brain, breast, liver, pancreas, prostate and thyroid—and neurological conditions such as essential tremor, Parkinson's disease, epilepsy, obsessive compulsive disorder and stroke. In the United States, a focused ultrasound treatment for uterine fibroids has been approved by the FDA. *[In Israel, they have been treating uterine tumours this way, without general anaesthetic or*

hospitalization, and the patients can normally return to work the next day. Treatments for additional indications are available abroad.

About the Focused Ultrasound Foundation

Based in Charlottesville, Virginia, the Focused Ultrasound Foundation is dedicated to improving the lives of millions of people worldwide by accelerating the development and adoption of focused ultrasound therapy. Our programs are designed to provide an “adrenaline rush” in the field and achieve progress faster. We work to clear the path to global adoption of focused ultrasound by coordinating research activities, creating effective partnerships, fostering collaboration and building awareness for “medicine’s best kept secret.” The Foundation is dedicated to ensuring that focused ultrasound finds its place as a mainstream therapy for cancer, brain tumours, Parkinson’s disease, epilepsy, stroke and other life-threatening conditions within years, not decades. Since its establishment in 2006, the Foundation has become the largest non-governmental source of funding for focused ultrasound research.

The Foundation is supporting clinical studies in essential tremor and brain tumours, as well as pending clinical trials for Parkinson’s disease, breast cancer and epilepsy. Complete information about the Foundation and its work can be found online at www.fusfoundation.org.

Rae spoke to Dr Elias’s coordinator in Virginia. She asked him if they would be looking at using ultrasound for cases of dystonia. He told her that if their success continues they will be trying their techniques on other conditions.

I look forward to following the progress of these procedures and trials. *HM*

INTRODUCING: Estela, who is a member of our Calgary support Group.

My name is Estela Strasser, I am 63 years old; I was born and raised in La Paz, Bolivia South America. I have three brothers, Peter, Benjamin and Alejandro; they live in Bolivia with their families. My mother was from Spain and my father from Poland (both deceased). I have three children, Jacqueline, Audrey and David; and three grandchildren Lukas, Jakob and Yael.

I am retired from the Bank of Montreal, I moved to Canada when I was 20 years old. Some of my activities involve reading, exercising, going to the movies, shopping, visiting with friends, taking different courses, and doing volunteer work.

I have Cervical Dystonia/Spasmodic Torticollis; I had Deep Brain Stimulation (DBS) surgery in June 2005, when I was 57 years old. This is how it started:

When I was in my early teens (13-14), I started with tremors in my hands. Since the tremors increased when I was nervous, the obvious conclusion was that it was nerve related. We (my mother and I) never sought any kind of medical advice for it.

After being diagnosed with Dystonia I realized the hand tremors were the first symptoms of my Dystonia. The hand tremors gradually became worse (it was hard to hold a cup of tea or bring a spoon of hot soup to my mouth); I often spilt things much to my embarrassment.

To control my tremors I started using "tricks". I would tighten the muscles of my upper body especially my upper back to control the tremors. In my late teens the tremors started to decrease but never completely disappeared.

At the age of 20 after completing part of my studies in Bolivia, I moved to Los Angeles, California to learn English, complete my studies and return home. Instead I met Frank (my ex-husband), fell in love and married him. Shortly thereafter we moved to Canada to start our new life. It was not long before Frank noticed tremors in my head; he was the first one to notice them, or maybe the first one to say something.

At first and for a long time, still thinking it was related to nerves I never sought medical attention over it, I just ignored it.

I have also suffered from migraine headaches since I was 12 years old until the present. In my late twenties-early thirties I sought help for the headaches. It was during this time I was referred a couple of times to neurologists. While consulting with them regarding the migraines I took the opportunity to enquire about my tremors; but they had no answers for me.

Later in my mid thirties my maternal grandmother died three or four years after developing Parkinson's. At that point I decided to seriously seek medical help. The medical findings were negative for Parkinson's but I was diagnosed with Familiar Tremor which they told me was incurable and would worsen in severity with age.

Over the next while I tried various prescribed medications that were used to treat Epilepsy. Not only did these medications fail to control the tremors but the side effects were unbearable.

Meanwhile my shoulders and neck became increasingly tight and painful. I tried massage therapy, physiotherapy, chiropractic treatments and acupuncture to bring relief but nothing really helped. Later in one of my visits to a Calgary Sports Clinic a young Physiotherapist asked the clinic doctor to see me after she unsuccessfully tried to treat me with acupuncture. After briefly checking me, he knew instantly what my problem was and referred me to a Neurologist from Toronto who was temporarily working at The Department of Clinical Neurosciences at the Foothills Medical Centre. He was the first one to diagnose me with Cervical Dystonia/Spasmodic Torticollis in 2000.

I received my first Botox treatments from him; however I had a bad reaction to the injections. After three months when the injections wore off I did not want to continue with the Botox treatment.

During this time they were doing a study at the clinic with Myobloc and my new and permanent Neurologist Dr. Oksana Suchowersky (Principal Investigator) asked me if I wanted to be part of it.

For four years I received Myobloc injections every three months with fairly good pain relief. But three years into the study I was experiencing increased and continuous head tremors. My head was progressively twisting to the right and back resulting in much greater pain than ever.

Not only was I having to live with excruciating pain but my daily functioning was also becoming more difficult due to the pain and my awareness of people noticing my symptoms. Driving my car had become more difficult and dangerous. I would secure my head with pillows but shoulder checks were getting harder to perform. My bank job of twenty five years was being affected by my condition, physically and emotionally.

I became very self conscious, being in public and performing my daily duties as a Branch Assistant Manager.

During my next appointment with my Neurologist in mid 2004 for another Myobloc injection the doctor noticed my marked change in appearance and explained to me that the injections were no longer having an effect.

When she told me this I felt as though my whole world was crumbling. The injections had been my only hope to live a fairly normal life. I was devastated and started to cry. I asked the doctor what was I to do now to which she replied "have you given any thought to the surgery?" to which I replied "what surgery" since I had never heard of this new treatment called Deep Brain Stimulation Surgery (DBS) for Dystonia. The neurologist asked her nurse to give me literature to read and the website to become familiar with the procedure. With my children's help I did a considerable amount of research. One of the things I learned during my research was that Dystonia has a higher rate among ethnic groups one being Eastern European Jews which is where my roots are from.

I also learned that the condition Spasmodic Dysphonia is a form of Dystonia; I have a first cousin from my dad's side of the family who suffers with this condition.

I decided that regardless of the risks involved without the surgery I would very soon be unable to drive or work.

At the end of 2004 I firmly decided to seek the surgery; in early 2005 upon meeting with the neurosurgeon I was told that I was a good candidate for the surgery. After having the surgery briefly explained to me and what to expect during and after the surgery, I was on the surgical wait list.

On June 27, 2007, the day of my 57th birthday I underwent DBS surgery performed by Dr. Z. Kiss at the Foothills Hospital. The surgery was 100% successful and I have had my life back since. The tremors are gone, my head is straight once more and I am 90% pain free.

We are very blessed to have the best Neurologists and Neurosurgeons right here in Calgary. I am for ever grateful to them and to the entire team of nurses at The Foothills Neurosciences Medical Centre and The University of Calgary Movement Disorders Clinic.



ON THE LIGHTER SIDE

I had this in an email from a friend:

Subject: Well, I Never Knew That

Q: Why do men's clothes have buttons on the right while women's clothes have buttons on the left?

A: When buttons were invented, they were very expensive and worn primarily by the rich. Since most people are right-handed, it is easier to push buttons on the right through holes on the left. Because wealthy women were dressed by maids, dressmakers put the buttons on the maid's right! And that's where women's buttons have remained since.

Q: Why are zero scores in tennis called 'love'?

A: In France, where tennis became popular, round zero on the scoreboard looked like an egg and was called 'l'oeuf,' which is French for 'egg.' When tennis was introduced in the US, Americans (mis)pronounced it 'love.'

Q: Why do people clink their glasses before drinking a toast?

A: It used to be common for someone to try to kill an enemy by offering him a poisoned drink. To prove to a guest that a drink was safe, it became customary for a guest to pour a small amount of his drink into the glass of the host. Both men would drink it simultaneously. When a guest trusted his host, he would only touch or clink the host's glass with his own.

Q: Why are people in the public eye said to be 'in the limelight'?

A: Invented in 1825, limelight was used in lighthouses and theatres by burning a cylinder of lime which produced a brilliant light.. In the theatre, a performer 'in the limelight' was the centre of attention.

Q: In golf, where did the term 'Caddie' come from?

A. When Mary Queen of Scots went to France as a young girl, Louis, King

of France, learned that she loved the Scots game 'golf.' So he had the first course outside of Scotland built for her enjoyment. To make sure she was properly chaperoned (and guarded) while she played, Louis hired cadets from a military school to accompany her. Mary liked this a lot and when returned to Scotland (not a very good idea in the long run), she took the practice with her. In French, the word cadet is pronounced 'ca-day' and the Scots changed it into 'caddie.

I had this one from Teresa recently:

My inconclusive travel plans 2012

I have been in many places, but I've never been in Cahoots. Apparently, you can't go alone. You have to be in Cahoots with someone.

I've also never been in Cognito. I hear no one recognizes you there.

I have, however, been in Sane. They don't have an airport; you have to be driven there. I have made several trips there, thanks to my friends, family and work.

I would like to go to Conclusions, but you have to jump, and I'm not too much on physical activity anymore.

I have also been in Doubt. That is a sad place to go, and I try not to visit there too often.

I've been in Flexible, but only when it was very important to stand firm.

Sometimes I'm in Capable, and I go there more often as I'm getting older.

One of my favourite places to be is in Suspense! It really gets the adrenalin flowing and pumps up the old heart! At my age I need all the stimuli I can get!

I may have been in Continent; but I don't remember what country I was in. It's an age thing.

And from another friend:

Subject: What Confucius DIDN'T say

Man who wants pretty nurse, must be patient.

Lady who goes camping must beware of evil intent.

Man who leaps off cliff jumps to conclusion.

Man who runs in front of car gets tired,

Man who runs behind car gets exhausted.

War does not determine who is right, it determines who is left.

Man who drives like hell is bound to get there.

Man who live in glass house should change clothes in basement.

Finally CONFUCIUS DID SAY. . .

"A lion will not cheat on his wife, but a Tiger Wood!"

Remind yourself that it's OK not to be perfect

Subscriptions are now due for 2012. Please mail to Brian, contact info below. Thank you.

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NEXT MEETING

DATE Saturday, May 12, 2012 at 10.00 a.m.

LOCATION DEVELOPMENTAL DISABILITIES RESOURCE CENTRE
4631, RICHARDSON WAY, SW, CALGARY

SPEAKER None

MEMBERSHIP FORM 2012

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