NATIONAL SPASMODIC DYSPHONIA ASSOCIATION
300 Park Boulevard, Suite 415 • Itasca, IL 60143 • Phone 800-795-6732 • Fax 630-250-4505 • E-mail NSDA@dysphonia.org • Website www.dysphonia.org

NSDA Western Regional Symposium

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Compiled by Pat Hill, *NSDA Area Contact Person Coordinator*

Disclaimer: Notes compiled based on a patient interpretation of the medical presentations. Any advice here is general in nature and is not supposed to apply to individual cases – please consult your healthcare professional with any questions.

Morning Session

Charlie Reavis, NSDA president, started off the meeting by telling us about the dedication of the NSDA toward research, awareness, and patient support. To date, we now have 80 support groups and approximately 300 Area Contact Persons. The NSDA provides grant money for research, and publishes newsletters, brochures, books, and DVDs, Symposiums are held to promote awareness and offer support to SD people, and the NSDA is a proud member of the Dystonia Advocacy Coalition. NSDA volunteers also attend medical conventions to raise the profile of SD with healthcare professionals. NSDA's on-line services include the Bulletin Board (at www.dysphonia-bb.org) and this site has just passed the milestone of 40,000 posts since it was set up 12 years ago.

Gerald Berke, M.D., Chairman of UCLA Head and Neck Surgery, was the first speaker and he described the function of the larynx – voicing generated by air from our lungs causing the vocal cords to vibrate. The air pressure required is about the same as that required to support 5 cc of water (a teaspoon), which is not a lot. Dr Berke gave the analogy of the process being like the noise made when we fill a balloon with air and then let it go.

Dr. Berke gave an overview of the different therapies used for SD. In the 300+ surgeries that he has performed, he has noted significant differences in patients. The neural anatomy in the larynx can vary considerably. He has seen nerves that are thin, or fat, or swollen, which leads him to believe that SD may actually consist of several disorders: Adductor SD, Abductor SD, Adductor SD with compensation, functional disorders, Abductor SD with compensation, SD with essential tremor, and SD with Muscle Tension Dysphonia. Psychogenic causes have been ruled out. In his time at UCLA Dr Berke has noted that SD cuts across all walks of life and social cultures.

Therapies

Botulinum toxin injections - botulinum toxin is injected into the neuromuscular conjuncture, NOT the muscle, with placement under the thyroid cartilage.

The injection takes effect pre-synaptically (before the muscle) and stops the muscle from emitting the neuro-transmitter. Botulinum toxin was initially weaker in the early years when this was first done, but has been produced in a stronger formulation over the years.

In 1992, the 'Point-Touch' technique was developed where the injection is done through the front of the throat and is very effective without using a scope. Dr. Berke mixes 100 units of botulinum toxin (normal amount in a vial) with 4 cc of saline dilution. He advises patients not to use botulinum toxin for cosmetic reasons as this could create antibodies against botulinum toxin. He has also learned that some patients have a significant sensitivity to botulinum toxin and he

has had to administer fractional doses. Botulinum toxin is generally very safe.
Speech Therapy is a form of therapy used to help with compensatory habits and articulation. This helps somewhat.

Myomectomy involves the cutting of muscles and has been used but is not as effective.

Selective Laryngeal Adductor Denervation-Rennervation surgery involves severing the particular nerve that delivers the message to spasm and grafting on one of the eight strap muscles nearby to replace it. Unilateral surgery has been done but bilateral has proven to be more effective. Dr. Berke cautioned that this surgery is not for everyone and a patient must be able to tolerate three hours of anaesthesia.

Laurie Ozelius, Ph.D., Associate Professor in Genetics and Genomic Sciences, and Associate Professor in Neurology at Mt. Sinai School of Medicine in NY, spoke about the DYT1 genome that is the primary genetic marker for the inheritance of dystonia. This can be inherited maternally and paternally. With DYT6, Primary Torsion Dystonia, speech is involved but it is not SD. Onset is from 5 to 62 years of age. It is possible to be a host of dystonia and not show any symptoms. In other words, a parent could carry a gene and not exhibit symptoms but could pass the gene onto their offspring.

A genome consists of 3 billion pieces and it currently costs $10,000 to run DNA sequencing and testing. Mutations, however, are most likely within the 2% of the protein coding, so checking only this part of the DNA for SD may make it more simple to detect in the future. Genome sequencing is used to identify different genes compared to the rest of the patient's family.

Tania Fuchs, Ph.D. is currently working on a grant to identify SD by endophenotype. It has been discovered in other genetic forms of dystonia that if a subject has two stimuli applied to their fingers, people who carry the gene but do not exhibit symptoms, take longer to identify the stimuli.
Kristina Simonyan, Ph.D. at Mount Sinai is doing research on imaging in SD that can then be compared and correlated to genome patterns.

Christy Ludlow, Ph.D., Professor of Communication Services and Disorders and Director of the Laboratory of Neural Bases of Communication and Swallowing at James Madison University, VA, stated that there are now 13 grants for SD research funded through the National Institutes of Health. She lauded the efforts of the NSDA and its advocates in raising the profile of SD in the research community. Dr Ludlow strongly encouraged all to keep up the work of advocacy and support for SD as it is making a difference.

Dr. Ludlow reviewed a study coming out of a study at the University of Utah:

- 63% of patients are female
- 76% had a gradual onset
- 27% had a voice tremor
- 96% sought treatment
- 89% received botulinum toxin injections
- 59% tried voice therapy
- 7% underwent surgery

Using various treatments, patients reported:

- 21% were better
- 34% were worse
- 45% remained the same

People who had SD with tremor responded better to medications.

Quality of Life in the SD population was also addressed through this study. Depression was an issue, as a secondary condition in addition to the disorder. Depression prevalence was noted in:

- 28% with SD
- 36% with MTD
- 25% with anxiety and SD
- 29% with MTD and SD

It was advised that these people would benefit most from a support group.

Dr. Christine Tanner in a study has compared SD to other head and neck disorders to determine possible co-existing risk factors.

- SD with tremor included the risk of head and neck tremors, immune disorder, thyroid problems, and panic disorder
- SD alone included the risk of hand tremor, blepharospasms, and anxiety.
- SD with tremor within the family members had the risk of tics, ocular disease, meningitis, cancer and compulsive disorder
- SD only with immediate family (parents and sibling) had a risk of tremors, tics, asthma, and cancer
- SD with/without tremor in extended family had a higher risk of cancer

Environmental triggers for SD could include:

- thyroid problems
- mumps
- URI frequency
- accidents

SD appears to have a genetic predisposition with increased risk of tremors, blepharospasm, meningitis and mumps. There is also a possibility that individuals have been exposed to a virus that a neuron is susceptible to. People who are vaccinated have less of a chance of being impacted by SD.

Additional studies being conducted through funding through the NIH include: MRIs are being used by Dr. Guenther to study SD and structural changes in the brain. Tomography is being used to study dopamine transmitters. SD is being measured to determine interruptions in communication. Drs. Houde and Niziolek are using speech auditory feedback to study how to help SDers improve speech. The use of lidocaine is being studied by Dr. Watson to determine how it can change symptoms of SD versus MTD. Dr. Carlie Tanner at Kaiser is studying the frequency of SDers misdiagnosed. Mark Hallett is studying genetic markers of focal dystonias. There is also the Dystonia Coalition Project that is trying to develop a standard test for diagnosing SD, measuring treatment results and the change in diagnosis, and the excitability of the cortex.

Overall there are studies ongoing in abilities to diagnose, differences in individuals physiologically, genetic risk factors, brain dysfunction, and treatment options.

The forum was then opened up to questions. These are the answers to a variety of them.

- People who are NOT good candidates for SLAD-R surgery are: AbSD, no response to botulinum toxin, tremor (this varies), and general anesthesia risk. It is advised to be examined and have a consultation if you are considering this surgery. Go to a doctor who is an expert in this area and has done multiple surgeries.
- People who have late onset SD: it is probably genetic but has been triggered by an environmental episode.
- Focal dystonia, like SD, is at one site, but segmental dystonia are two adjacent dystonias.
- Medications have been used to treat SD but are more individual and something that should be worked out with a person's physician. Medications usually only address conditions like anxiety or tremors. Stress can worsen SD as it can with any neurological disorder. It just makes it more apparent.
- SD is a complicated disorder, so if you try some treatment like acupuncture and it works for you, that's fine; but the treatment cannot be generalized to the entire SD population.
- There is no literature that shows that DBS (Deep Brain Stimulation) is effective for SD. In France it was shown to help people with

Meige's Syndrome but was less effective with the voice.

- Laryngeal Dystonia is a breathing disorder separate from SD.
- Blepharospasm is a condition where the eyes close involuntarily. You cannot drive with this condition.
- SD is believed to be genetic but may be triggered by other things, like bronchitis, emotional trauma, surgery, an accident, etc.
- Unilateral botulinum toxin injections may travel to the other vocal cord and is effective for that reason, but there are too many variables to accurately compare the benefits of unilateral vs bilateral injections.
- There may actually be sensors in the vocal cord muscles that send the message back to the brain not to spasm. So there is not only message input to the muscle but also output messages to the brain to correct the condition.
- AdSD has vowel breaks, whereas with AbSD the voice breaks before the vowel with voiceless sounds.
- Studies being done on SD are listed on NIHreporter.gov and can also be found on the NSDA website. Search for 'spasmodic' + 'dysphonia'.
- Medicines for dealing with tremors vary. Inderol is used but it may have cognitive side effects.
- There may not be a cure for SD, but in the future it is hoped that they may be able to prevent it; for instance, there may be a vaccine for a virus that might set off the genome.
- Botulinum toxin injections may vary due to abnormal neural anatomy or different genotypes.
- SDers can often be without spasm during period of crying or laughing. Researchers found out from a study on monkeys that emotional expression is a different system than the speaking system. The neurological pathway is not the same.
- Long term data shows that people benefit from botulinum toxin over many years but others may choose surgery.
- Fine wire EMGs is typically used to study muscles for research.
- Hormone therapy or PMS has not been correlated to SD. It is thought that retention of body fluids during a woman's period might cause edema which may make a voice disorder worse.
- If a person has thyroid surgery, the surgeon must be careful not to injure nerves in that area. Nerves can heal/regrow but you should always have an experienced surgeon so damage is not done to the voice.
- People who use their voices for their occupation like radio announcers, ministers, and singers may put their voices at risk from overuse.
- Botulinum toxin A is more effective and less expensive than Botulinum toxin B.
- There are not many treatments for AbSD – at this time only thyroplasty and botulinum toxin.

Brain donations were discussed. To date only two brains from SD people have been studied, and abnormalities have been found. In the first brain there was a cortical bulbar pathway abnormality and in the second one there were abnormalities in the brain stem. Harvard collects the brain donations and does exhaustive studies on them for a couple of years. Brain donations are strongly recommended as the most conclusive source of study for SD. There are currently 35 people who will be donating their brains, but more are needed.

Afternoon Session

Dr. Tom Hofmann, Chair of the Psychology Department at Hodges University in Florida, talked about the Quality of Life with SD. There are three stages of grieving:

- Grieving the huge loss of your voice by getting in touch with your emotions.
- Acceptance of your condition
- Growth and thriving

Depression can cause a great expenditure of energy, resources and money, so it's important that you work through it.

Treatment options are:

- Get in touch with your emotions and talk to people with similar conditions - a support group can be understanding and empathetic
- Seek professional help from a chronic illness therapist

The longer a person lives with SD, the better the quality of life, because the person has learned how to cope skills, adjusted emotionally and learned social skills. Negativity can cause a chain reaction as it will feed upon itself and become a self-fulfilling prophecy. Positive thought can make you less fearful. If positive thinking does not come automatically, "Fake it until you make it." Pretend that you are positive and it will become a habit. Do not let negative thoughts enter your thinking nor comments. Tasks that you can attack are ordering, phone calls, voice mails, addressing groups, interviews, dates, and relating to your spouse.

There can be individual challenges such as:

- Personality
- Phase of your life
- Previous success at a task
- Effectiveness of medical treatment
- Social network
- Sense of humor
- Importance of a task
- How competent you feel

It's important to always be moving forward. You should have a Plan A and Plan B, in case something doesn't work.

- Have a plan to address changes in your life like retraining through Vocational Rehab., retirement, stop working, or telecommute.
- Look at your thoughts and bring about an attitude adjustment and judge public reactions to you.
- Get help via amplifiers, email, texting, telephone amplifiers, help from others or by educating others, gesturing, body language, or carrying a card that explains that you have a voice disorder.

Dr. Frank Prochaska was the final speaker of the day. He is Chair of Management at Colorado Technical University. His message was to be caring and positive. He said that most people have "flat" spots in their lives and we need to help each other. His favorite quotes were:

- "If you don't have something positive to say, don't say it."
- "Attitude is the only thing you have control of."
- "Stay on life's path. Don't let SD be a monster in your life. Get back to yourself and who you are."

The NSDA thanks all of the speakers and attendees for making the meeting a success.