Movement Disorder Experts Go Beyond Call of Duty on Behalf of Dystonia Patients

2015 Research Projects

Congressional Hearing on Dystonia Educates Legislators
The Baron family of Rhode Island organized the first-ever Providence Dystance4Dystonia Zoo Walk at Roger Williams Park on June 27. Over 400 people attended to support dystonia research and bring visibility to the disorder.

Rosemary Young and her supporters turned out for the Detroit Dystance4Dystonia Zoo Walk on June 30. The event attracted 500 attendees and kicked off a number of awareness efforts in Michigan.

June Tritley, Perry Patten, and the St Louis MO/IL Metro Dystonia Support Group held the first-ever St. Louis Dystance4Dystonia Zoo Walk on August 30. More than 200 people attended to show their support.

On September 12, over 700 people representing 40 teams gathered for the 3rd Annual Dystance4Dystonia Cincinnati Zoo Walk organized by Melissa Phelps. The Zoo Walk was preceded by “Go Blue for Dystonia Day” in Gallatin, KY and surrounding counties.

Jennifer Kassis celebrated her birthday on September 19 by hosting the very first Binghamton Dystance4Dystonia Zoo Walk. The walk attracted robust attention from the local press, resulting in much-needed awareness in the NY/PA region.

The Flanagan Family, who held the original Dystance4Dystonia Zoo Walk, continued their support of the DMRF with the 4th Annual Cleveland Zoo Walk on September 19. Many thanks to: Karen Flanagan, Jane Ann Flanagan, and Gale Flanagan.

The 2nd Annual Dystance4Dystonia Pittsburgh Zoo Walk was a great success thanks to MaryRae Nee, Ed Cwalinski, Shayla Anthony, and the Western Pennsylvania Dystonia Support Group.

Congratulations and thanks to the Metherell family for organizing the 3rd Annual Toss4Dystonia Cornhole Tournament in Buffalo to raise awareness and research funds. Local media covered the Toss, expanding dystonia awareness further in Western NY.

Going the Distance for Dystonia
Thousands Attend Zoo Walks and Local Events in Honor of Local Families

DMRF supporters across the country are increasing dystonia awareness and funding medical research by hosting local events. These powerful grassroots efforts are on track to raise $200,000 to support the DMRF in 2015. Many of this year’s Dystance4Dystonia Zoo Walks and events were held in September during Dystonia Awareness Month. See page 16 for more in "People on the Move."
The DMRF thanks Allergan, Ipsen, Merz, and US WorldMeds for their lead sponsorship of the 2015 Dystance4Dystonia Zoo Walks.

Pamela Sloate organized the very first Dystance4Dystonia Bronx Zoo Walk on October 4, attracting over 500 people. The teams represented families impacted by dystonia, movement disorder clinicians, and researchers creating a truly unifying atmosphere.

Many thanks to Kohl’s Cares for partnering with the DMRF to generously support a number of Dystance4Dystonia events including the Stomp Out Dystonia 5K, and Zoo Walks in Providence, St Louis, and Cincinnati.
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What is Dystonia?
Dystonia is a disorder that affects the nervous system. Improper signaling from the brain causes muscles to contract and twist involuntarily. Dystonia can affect a single body area or multiple muscle groups. There are several forms of dystonia, and dozens of diseases and conditions include dystonia as a significant symptom. For more information visit: www.dystonia-foundation.org

On the Cover:
Dystonia Awareness Month took place in September. DMRF supporters hosted events, took part in the Dystonia Moves Me campaign, and used their creativity to bring visibility to dystonia. Read more on pages 2 and 16.
Dear Friends,

The highlighted quote is evocative and in many ways captures the drive behind the DMRF’s mission, but we prefer to replace the word “chasing” with “achieving.” Our dream is to close the Foundation’s doors because there is no longer a need for the organization. Our dream is a day when no individual or family is ever again burdened by dystonia. We have made progress toward our dream, and that progress gives us confidence that the ultimate goal will be achieved.

When the founding leaders of the DMRF set out to find a cure for dystonia, there was no way to imagine how long or winding the journey would be. This kind of undertaking is indeed a marathon, not a sprint. And yet, as supporters of the DMRF, we hope each of you is encouraged by the achievements made so far. Fueling dystonia research by raising awareness at the National Institutes of Health, supporting innovative research projects, funding brilliant investigators, attracting bright young researchers to the field, and exploring inventive new ways to fund research like the Department of Defense Congressionally Directed Medical Research Program—these are things the DMRF has done for decades because we know these efforts produce results.

The DMRF is the pace-setter in this marathon toward a cure. We feel a responsibility to lead and blaze the trail. We take this role seriously. Our track record provides the confidence—and evidence—that we are on the correct path.

In the meantime until we have a cure, people impacted by dystonia have daily, immediate needs. The DMRF will continue to make sure the dystonia community has access to the latest information to inform decisions about treatment and care, to promote greater public awareness of dystonia, and to provide coping support for those who are struggling. We will do this every day until we can greet the researchers at the finish line and celebrate the cure together.

Art Kessler
President

Janet L. Hieshetter
Executive Director

Join us in Washington, DC for Dystonia Advocacy Day • April 12–13, 2016
Movement Disorder Experts Go Beyond Call of Duty on Behalf of Dystonia Patients and Support of Science

Doctors & Researchers Support DMRF Events

A number of movement disorder specialists are going above and beyond their professional duties by supporting the dystonia community at DMRF events. Prominent movement disorder experts have spoken and attended Zoo Walks throughout the country. Two neurologists ran the TCS New York City Marathon as part of Team DMRF.

On October 4, over 500 people gathered at the Bronx Zoo for the first ever New York City Dystance4Dystonia Zoo Walk. Most attendees represented teams of 10 or more assembled in honor of a person or family impacted by dystonia, or by a prominent healthcare institution. One of the largest teams—including more than 30 people—was a group from Albert Einstein College of Medicine led by Dr. Kamran Khodakhah. Dr. Khodakhah is Professor and Harold and Muriel Block Chair in Neuroscience at Albert Einstein. His research explores the role of the cerebellum and basal ganglia in motor function and in dystonia and other movement disorders.

Dr. Khodakhah explains, “For my colleagues and me, the walk was particularly meaningful because it reminded us of the ultimate reward that our hard work might secure: a cure for dystonia. It was also crystal clear to us that every dollar raised by such activities in support of dystonia research is mixed with an incalculable amount of love.”

Dr. Joel Perlmutter, Head of the Movement Disorders Section and Elliott Stein Family Professor of Neurology at Washington University spoke at the first ever St Louis Dystonia Zoo Walk on August 30. “The Zoo Walk is a terrific idea. The enthusiasm was palpable,” he remarked. “The DMRF has an incredibly important role for helping us not only to provide education and support for people and families with dystonia but also to support research.”

For individuals with dystonia, it can feel like a marathon just to get through the day due to the challenge of living with a chronic, debilitating disorder. On November 1, Drs. Pedro Gonzalez-Alegre and Patrick Hogan represented the DMRF at the TCS New York City Marathon. The doctors belong to a team of runners from across the country who volunteered their time, energy, and legs to raise dystonia awareness and research funds.

Before embarking on the marathon, Drs. Hogan and Gonzalez-Alegre had already devoted their careers to caring for dystonia patients and research into improved treatments.

Dr. Gonzalez-Alegre is Co-Director of the Parkinson’s Disease and Movement Disorders Center and Associate Professor of Neurology at Pennsylvania Hospital in Philadelphia. His research on dystonia is dedicated to understanding the disease mechanism of specific dystonia-causing genes, which frequently cause dystonia.
“The Bronx Zoo Walk was a great event for the DMRF. The large turnout demonstrates the compassion participants feel for each other and their interest in supporting dystonia research. The participants consisted not only of people with dystonia and their families, but also neuroscientists and medical professionals; all were there to defeat dystonia.”

– Stanley Fahn, MD, Founding & Emeritus Director, Movement Disorders Division, Columbia University Medical Center

How Can I Participate in Dystonia Research?

You can support dystonia research discoveries in more ways than one. Consider these opportunities to have an impact:

Ask Your Doctor
Your movement disorder specialist may be participating in a clinical trial that needs volunteers.

Search Online for NIH Studies
Search for dystonia clinical studies supported by the National Institutes of Health at www.nih.gov/health/clinicaltrials/

Register as a Brain Donor
Individuals with all types of dystonia as well are urgently needed. For info visit www.dystonia-foundation.org/brain

Join the Global Dystonia Registry
Patient registries help researchers better understand dystonia by collecting information directly from patient volunteers. Join the Global Dystonia Registry at https://globaldystoniaregistry.org/

Donate
Financial contributions to the DMRF support cutting-edge research by the world’s foremost thought leaders and up-and-coming experts. Use the envelope enclosed in this newsletter to make a gift or donate online at www.dystonia-foundation.org/donate

See page 8 for more information about Team DMRF at the TCS NYC Marathon.

Drs. Pedro Gonzalez-Alegre and Patrick Hogan ran the TCS NYC Marathon to support the DMRF.

in children. “My research is devoted to better understanding why people develop dystonia and finding better ways to treat it,” he explains, “but I wanted to join Team DMRF because of all the families I have met who share the burden of living with dystonia every day. They have been truly an inspiration for me.”

Dr. Hogan is Director of the Puget Sound Regional Movement and Motility Disorder Center in Tacoma, Washington. He has cared for patients with dystonia for 35 years and was the first in Washington to use therapeutic botulinum toxin injections for dystonia in the early 1990s. “Despite the best treatment, I continue to witness the devastating effect that dystonia can have on a person’s life and those around them,” he explains. “I was happy to be able to run the New York Marathon and at the same time help achieve the goal of a cure for dystonia.”
Big Discoveries from Tiny Worms

Roundworms Reveal New Information about Dystonia Protein, TorsinA

What can you learn from a roundworm? A lot! Research supported in part by the DMRF has revealed important new information about torsinA, the cellular protein known to cause dystonia when abnormal due to a mutation in the DYT1 gene. A group of investigators led by Dr. Lesilee S. Rose at University of California, Davis studied the role of torsinA in a type of roundworm called C. elegans. The team recently published a very comprehensive paper describing their findings. In short, torsinA appears to be essential for the normal function and localization of nucleoporins, the nuclear membrane proteins that are part of the nuclear pore complex through which large molecules are transported in and out of the nuclear membrane. Additionally, the investigators discovered abnormalities in the nuclear envelope very similar to those found in flies and mice.

Understanding the role of torsinA in dystonia is one of the most active areas of dystonia research. When torsinA was first discovered in 1997, it was an unknown protein. Scientists have since been working to understand the role of this mysterious protein in cells. Understanding the role of normal torsinA will shed light on how and why mutant torsinA causes dystonia—and this will lead to new treatment strategies that have the potential to interrupt or correct the dystonia disease process. Current treatments for dystonia suppress symptoms without altering or curing the underlying problem.

A pair of outstanding, recently-published studies reveal research progress in understanding the genetic origins of primary torsion dystonia and myoclonus-dystonia.

A team of researchers led by Drs. Kailash Bhatia and Nicholas Wood of University College London have discovered two brand new dystonia genes. Reports of primary torsion dystonia inherited in an autosomal-recessive manner—often lumped together as “DYT2” dystonia—have been documented for decades, but until now no specific genetic cause was identified. Through a combination of gene identification techniques, the HPCA gene has been shown to cause DYT2 dystonia in two unrelated families. HPCA encodes the protein hippocalcin which plays a role in regulating calcium channels. Most identified genetic dystonias are inherited in an autosomal dominant manner, which means that only one parent needs to carry a mutation for a child to develop dystonia—an example of this is DYT1 primary torsion dystonia. In the case of DYT2, however, both parents must carry the mutation for offspring to develop neurological symptoms.

The same research group made an important discovery pertaining to myoclonus-dystonia. The team discovered that the KCTD17 gene, a potassium channel component, causes myoclonus-dystonia in a group of families who do not carry previously identified SGCE mutations known to cause this rare movement disorder. This discovery extends the genetic spectrum of myoclonus-dystonia and will likely inspire more mechanistic studies. DMRF grant recipients Drs. Ebba Lohmann and Thomas Gasser of University of Tübingen (Germany) co-authored the paper and acknowledged the Foundation’s support.

Genetic discoveries provide opportunities to better understand the biochemical origins of dystonia, which is critical to developing new treatment strategies.


From Inspiration to Discovery: 2015 Research Projects

The DMRF is committed to addressing the most pressing unresolved questions in dystonia research and inviting scientists from all over the world to apply for funding to advance progress in these areas. The Foundation also negotiates research contracts to enlist partners to work on specific projects or address a knowledge gap in the field. This would not be possible without the support of DMRF members and years of investment in basic science.

The DMRF is proud to share this year’s outstanding research investigations, some of which are continuing from 2014. Congratulations to this year’s award recipients, and infinite thanks to our supporters for making this research funding possible.

Development of Novel Reagents to Augment Cholinergic Signaling in Dystonia—2nd year
Randy Blakely, PhD, Craig Lindsley, PhD, J. Scott Daniels, PhD
Vanderbilt University Medical Center

Medications that impact the nervous system by acting on the neurotransmitter acetylcholine have proven helpful in the treatment of dystonia. A team of researchers at Vanderbilt University Medical Center are testing a group of molecules that could potentially be developed into drugs that reduce dystonia symptoms by affecting acetylcholine but with fewer side effects than existing drugs.

Interactions of M4 and D1/Gαolf Signaling in Striatum: Implications for Treatment of Primary Dystonia—2nd year
P. Jeffrey Conn, PhD, Vanderbilt University Medical Center
Jürgen Wess, PhD, National Institutes of Health
Denis Hervé, French Institute of Health & Medical Research (INSERM)

Recent genetic studies reveal that mutations in the GNAL gene cause primary torsion dystonia. The protein that GNAL encodes, called Gαolf, is present in an area of the brain known to be involved in normal movement and motor control called the striatum. Gαolf is specifically involved in communication between neurons and carrying messages transmitted by the neurotransmitter dopamine. Furthermore, another neurotransmitter called acetylcholine acts on specific neurotransmitter receptors to suppress Gαolf. A team of researchers led by Dr. Jeffrey Conn is examining how drugs that act on these receptors might restore the balance of dopamine and acetylcholine in the brain, and possibly provide a novel treatment approach.

A New Gene and a Novel Pathway Leading to Myoclonus-Dystonia—2nd year
Dennis Bulman, PhD, Children’s Hospital of Eastern Ontario Research Institute
David Grimes, MD, University of Ottawa
Anthony Lang, MD, University of Toronto
Christine Klein, MD, University of Lübeck, Germany

This project builds on Dr. Bulman’s discovery of a novel gene mutation that causes myoclonus-dystonia. Since nothing is yet known about the function of this gene, known as IncRNA, the study aims to clarify its role in regulating other genes including SGCE, the major gene associated with myoclonus-dystonia. The DMRF is collaborating with the Brown Family Foundation to support this work.

The Mechanism of Developmental Dysfunction in DYT6 Dystonia—2nd year
William Dauer, MD, University of Michigan

Dr. Dauer’s goal in this contracted project is to identify transcriptional targets of the THAP1 gene which when mutated has been found to cause DYT6 primary torsion dystonia. The biological pathways impacted by THAP1 will also be investigated to identify which key pathways may be disrupted by dystonia-causing mutations.
Integrating the Neurocircuitry of Cervical Dystonia across Mouse and Man
Michael Fox, MD, PhD, Veronique Vanderhorst, MD, PhD, Daniel Tarsy, MD, Ron Alterman, MD
Beth Israel Deaconess Medical Center

Dystonia is not due to problems in a single brain region, but a set of regions or brain circuit. Identifying these regions, their connections, and how they work together or fail to do so is critical for understanding dystonia. Using a collaborative and team approach with broad expertise in human and rodent neurocircuitry, the researchers propose to integrate a new mouse model of dystonia with advances in human brain imaging. They will determine how sites relevant for cervical dystonia in the mouse brain can be identified in the human brain and whether the brain connectivity of those sites is abnormal in patients with dystonia. The results will provide new insights in the neural circuitries underlying dystonia and guide the selection of novel sites that form targets for neuromodulation (such as deep brain stimulation) to treat dystonias.

Whole Exome Sequencing in Families with Myoclonus-Dystonia Syndrome without SGCE Mutations from Turkey and Germany
Thomas Gasser, MD, Saskia Biskup, MD, PhD, Ebba Lohmann, MD, University of Tübingen
Hasmet Hanagasi, MD, Istanbul University

This research aims to genetically analyze various families affected by myoclonus-dystonia in search of new genes associated with this disorder. These studies involve a collaboration among clinical research groups in Germany and Turkey as well as a genetics company based in Germany. The DMRF is collaborating with the Brown Family Foundation to support this work.

Effects of Novel GABA-A Receptor Subunit Selective Compounds on the Severity of Dystonia
Angelika Richter, PhD
University of Leipzig, Germany

In this contracted project the effects of novel GABA-A receptor subunit selective drugs are studied in a hamster model of paroxysmal dystonia. Two novel drug candidates have been provided by BioCrea and Merz Pharmaceuticals.

Evaluation of the Effects of a Novel Nicotinic Agonist, AZD1446, on Neurochemical and Electrophysiologic Endpoints in DYT1 Mouse Models
David Standaert, MD, PhD
University of Alabama, Birmingham
Antonio Pisani, MD, PhD, University of Rome

One of the current limitations of oral medications most commonly prescribed to treat dystonia is the incidence of intolerable side effects. A team of investigators is exploring whether a drug called AZD1446 could potentially provide relief for patients without the unintended effects frequently caused by these medications. The DMRF is collaborating with Cure Dystonia Now to support this investigation.

Role of Cerebello-Basal Ganglia Interactions in Primate Dystonia
Thomas Wichmann, MD and Yoland Smith, PhD
Emory University

Studies in rodents have suggested that interactions between the cerebellum and the basal ganglia may underlie some forms of dystonia. Given the fact that the brain anatomy differs substantially between rodents and primates, these results must be extended to non-human primates to further validate their relevance towards human dystonia. Thus, the researchers leading this study propose to address this issue by studying dystonia in Rhesus monkeys. They will study how symptoms in different body areas correlate to anatomical and functional interactions between the cerebellum and the basal ganglia. The significance of these experiments is that they may identify the first monkey model of dystonia of cerebellar origin, and may help examine the role of anatomical and functional interactions between cerebellum and basal ganglia in this disorder. The ultimate goal is to use the knowledge generated by these experiments to develop therapies for human dystonia of cerebellar origin, such as medications or new deep brain stimulation approaches.
The Dystonia Medical Research Foundation (DMRF) offers opportunities to connect with others impacted by dystonia and build networks for coping support.

**Impact of Dystonia on Social Connections**

There are numerous ways in which dystonia can make it more challenging to feel connected to other people. Tom Seaman knows this firsthand. Within less than a year, the active entrepreneur went from noticing a little stiffness in his neck to spending nearly 24 hours a day on the floor with relentless, chronic pain. Cervical dystonia turned his life upside down. Now a Professional Life/Health Coach, he uses his experiences to help others address the complexities of living with chronic illness, depression, anxiety, and coping.

“Dystonia creates challenges that can change the dynamics of a relationship,” explains Tom. “While dystonia can end friendships, it can also bring special people closer to us.”

Pain, fatigue, and other symptoms may make it difficult for individuals with dystonia to keep up with the activities and schedules of friends and family. Symptoms may be unpredictable and complicate making plans in advance or keeping social commitments. Friends and family may not fully grasp what it is like to live with dystonia. “Friends may want to ‘fix’ us,” says Tom. “They may back out of our lives, not knowing what to say or do. They may expect us to ‘get better’ or ‘get over it.’ This misunderstanding can cause friction and tension.”

Dystonia can also cause logistical complications in transportation, finances, and navigating public spaces. Living with dystonia requires frequent problem-solving and brainstorming creative new ways to accomplish tasks.

It is common for individuals with dystonia to withdraw and isolate from other people. Tom relates: “My depression about a life I thought I lost resulted in isolation and loneliness, which further fueled my depression. This also created anxiety. I didn’t care enough about myself to seek help and didn’t want people pushing me to get help, so I remained silent about my internal pain and as isolated as possible.”

**Health & Wellness**

The health benefits of a strong social circle are many. When comparing individuals who feel socially connected to those who do not, there are documented differences in important biological systems. Social isolation may weaken overall physiological processes necessary to repair and maintain the body. Social isolation is correlated to delayed wound healing times, increased pain, and poorer sleep quality. Adult friendships have been linked to lower risks of depression and premature death. Feeling isolated may be more detrimental than being isolated.

In a DMRF survey, 84% of people with dystonia stated they have felt isolated because of the disorder. Loneliness and feelings of isolation are also common among spouses, family members, and the parents of children with dystonia. There is a growing body of research to suggest feeling lonely can have a powerful effect on health. Physicians are increasingly recognizing friendships and social connections as important prescriptions for patients.

Tom adds: “We may begin to take worse care of ourselves. Lonely people are less likely to be physically active or eat properly. It’s important to take good care of ourselves and to know that our value and worth has not diminished due to dystonia.”

**A Friend Indeed**

*Dystonia, Social Isolation, & Friendship*

In a DMRF survey, 84% of people with dystonia stated they have felt isolated because of the disorder. Loneliness and feelings of isolation are also common among spouses, family members, and the parents of children with dystonia. There is a growing body of research to suggest feeling lonely can have a powerful effect on health. Physicians are increasingly recognizing friendships and social connections as important prescriptions for patients.
Inner Feelings & the Outer World
The interaction of loneliness and stress is complex. Loneliness is a stressor that can lead to anxiety, depression, irritability, distrust, hostility, and lowered self-worth. Individuals who feel lonely can find it more difficult to tolerate the ups and downs of daily living—minor negative experiences like traffic congestion, disagreements with other people, or issues at work seem more intense. Individuals who feel stressed may be more likely to engage in risky behaviors and not seek medical care when needed.

“We need to be careful how we treat people and not let the feelings and anxiety we have about our illness be taken out on others,” says Tom. “Be open with family and friends about how you feel. Ask them how they feel. Relationships are a two-way street.”

Support Systems
Friends often encourage healthier habits and advise each other to seek medical attention when sick or in distress. Individuals with strong social relationships tend to navigate stressful situations easier. They tend to know where to look and whom to ask for assistance when having difficulty. They are more likely to have multiple streams of information to inform decision making.

Loneliness can make it more difficult for individuals to feel positive and at ease in social situations. “Being different can feel embarrassing, making us self-conscious,” explains Tom. “We can feel as though everyone can see what we are feeling on the inside, causing great anxiety.” It can take time and practice to change the self-perpetuating cycles of loneliness, stress, and isolation.

Support Groups
“When re-engaging the world, do what is comfortable,” Tom suggests. “Join a group where you can connect with people who have the same interests—clubs, meet-up groups, and dystonia support groups. Make friends with similar challenges, but also be around people who are different so you can gradually expand your comfort zone.”

DMRF support groups plug you into a group of people who truly understand life with dystonia, and provide access to the latest information on research and treatment.

Most importantly, support groups provide an opportunity to receive and give support. Tom explains: “Sharing our feelings is a way to break isolation. When we focus on the needs of others, there is less attention on our lonely thoughts and feelings.”

If there isn’t a support group in your area, consider partnering with the DMRF to start one. For more info: www.dystonia-foundation.org/supportgroups.

Community Events
Jennifer Kassis of New York is amazed by the new connections she has made since forming the Southern Tier Dystonia Support Group and hosting the first-ever Binghamton Dystonia Zoo Walk in September: “A couple came to the Zoo Walk because their daughter has dystonia. They saw me promoting the event on TV and wanted to meet me and get information on my support group. We cried and hugged. Truly a beautiful moment.” Jennifer has had symptoms of generalized dystonia since childhood.

Katie Carlson traveled to attend the Zoo Walk from Connecticut to support Jennifer after the two women connected online. “When I was diagnosed with cervical dystonia I felt depressed and alone,” Katie explains. “One of the most uplifting connections I made was with Jennifer Kassis, who listened to me and brought me hope, even in her own time of struggle. I was able to meet Jennifer in person at the Zoo Walk and thank her for all she has done.”

Katie attended the Zoo Walk with her husband and two small children. “I was honored to be able to support Jennifer’s efforts to raise awareness and, for the first time in my life, meet others with dystonia. I can’t thank the DMRF enough for making connections like these possible.”

Continued on page 14
Continued from page 13

For a listing of scheduled DMRF events as dates are confirmed, go to: www.dystonia-foundation.org/events.

Online Forums
Online dystonia forums are support services that are available 24 hours a day, seven days a week, to people all over the world. Brandy Martin is a member of the Support4Parents of Children with Dystonia group on Facebook: “I’ve made many friends and received lots of information for my daughter as she started school this year. I don’t know what I would do without the support and encouragement I receive from this group.”

For information about the DMRF’s online groups, go to: www.dystonia-foundation.org/online.

Reaching Out
“We are social beings and require contact with others to be healthier,” concludes Tom. “Surround yourself with people who lift you up, believe in you and your dreams, and give you a spark. Reach out because loneliness is painful and can confuse you into thinking you are an outcast.”

Contact the Dystonia Medical Research Foundation (DMRF) to learn more about the resources available to help you feel more connected at 800-377-DYST (3978) or dystonia@dystonia-foundation.org. Or visit www.dystonia-foundation.org.

Tom Seaman earned a Bachelor’s degree in Education and Psychology, after which he pursued private business ventures in health education. While pursuing his Master’s degree in Counseling he developed cervical dystonia. After numerous ineffective treatments and years of physical and mental pain, Tom learned and applied a combination of techniques that greatly improved his quality of life. Tom is now dedicated to helping others with dystonia and chronic illness benefit from his experiences. He is a Certified Professional Life/Health Coach and author of the book, Diagnosis Dystonia: Navigating the Journey. He also co-leads the DMRF Cervical Dystonia Support Forum on Facebook.

Tom’s Tips for Nurturing Friendships & Making New Connections:

- When dystonia puts strain on a friendship, both parties need to accept that things have changed. Be open with your feelings. This can make relationships stronger and more meaningful than before.
- Take ownership of the qualities you have that will enhance other people’s lives. Don’t deprive others of your gifts and let others share their gifts with you.
- Show interest in others. Ask questions.
- Be careful about perceiving suggestions as judgments. People may be trying to help.
- Make an effort to joke about things, especially dystonia.
- Accept that some friendships simply fizzle out.
- When re-engaging the world, do what is comfortable. Maybe start online if you are hesitant to get out with people just yet.
- Go where there are people. Get used to being around others in a social setting.
- Stay in touch with people and make it easy for people to contact you.
- Open up to people. Acquaintances become friends when we are comfortable sharing intimate things with one another.
- Track down old friends you knew before your diagnosis.
- Create a social calendar and stick to it.
- With everyone: be authentic, friendly, understanding, flexible, a good listener, positive, supportive, dependable, and respectful.

Stay in Touch!
Sign up for the DMRF’s monthly e-newsletter for the latest updates and announcements: www.dystonia-foundation.org/email
Dystonia Advocacy Network Organizes First-ever Legislative Briefing on Dystonia

Dystonia Awareness Month drew to a close with a legislative briefing in Washington, DC on September 29. Congressman Chris Smith of the 4th District in New Jersey sponsored the briefing, helping to further dystonia awareness on Capitol Hill.

Beth-Anne Sieber, PhD of the National Institute of Neurological Disorders & Stroke at the National Institutes of Health (NIH) and Program Officer for dystonia presented a research overview. Dr. Sieber also noted the importance of research partnerships such as the Dystonia Coalition and collaborative relationships with patient advocacy organizations, academic institutions, and the NIH.

Janice and Len Nachbar, constituents of Congressman Smith, shared their daughter Joanna's experience with dystonia. They described how Joanna, like many members of the support groups they lead, was misdiagnosed for years. They spoke about the importance of dystonia awareness in the general public and in the medical community. “I found sharing our family’s journey with dystonia to be a very moving experience,” said Janice. “It was very empowering for Joanna. It was a wonderful meeting.”

Congressman Smith became aware of dystonia after meeting the Nachbars during Dystonia Advocacy Day in April. He has been an outspoken advocate for increased funding for medical research. Congressman Smith entered the Nachbars’ testimonies into the Congressional Record.

DMRF Executive Director Janet Hieshetter moderated the meeting and asked all present to support robust funding for NIH and for support to include dystonia in the Department of Defense Peer Review Medical Research Program for Fiscal Year 2016.

For more information on becoming a legislative advocate, contact dystonia_advocacy@dystonia-foundation.org or 800-377-3978.

Son Honors Mom with Dystance4Dystonia Walk

Brian Gardner organized the very first Dystance4Dystonia Putnam Walk to benefit the DMRF on October 17. Brian’s mother Susan Gardner has lived with dystonia since 1997. “After witnessing the challenges people living with dystonia go through on a daily basis,” he says, “I wanted to organize this event in honor of my mother and everyone who battles dystonia.”

Susan was diagnosed with cervical dystonia at age 31. Despite treatment, the disorder takes a toll. “There are activities I can no longer enjoy,” Susan explains. “I take one day at a time. I accept my bad days and enjoy my good ones. My family has stood by me.”

“My mother is the strongest woman I know,” says Brian. “When meeting her you would never know the constant pain she is in, dealing with dystonia. She’s cheerful, loving, courageous, and beautiful.”
PEOPLE ON THE MOVE

The DMRF is deeply grateful for our grassroots supporters who work year round to promote dystonia awareness and fundraise for medical research. Volunteers were especially active during Dystonia Awareness Month in September, creating a groundswell of awareness across the country. In addition to the events and activities included below, volunteers secured articles in the local press, used online social media, and found other creative ways to raise visibility of dystonia.

Hundreds of Dystonia Moves Me: Show Me Your Moves kits were distributed to support groups and individuals who worked to bring visibility to dystonia in their local communities.

The Dystonia Support & Advocacy Group of San Diego County presented Grammy Award-winning recording artist Juice Newton in a benefit concert in support of dystonia research and programs. Many thanks to Juice and her band, and event organizers Martha Murphy and Paul Fowler.

In commemoration of Dystonia Awareness Month, DMRF Past President and VP of Development Dennis Kessler was featured as a guest blogger by the Center for Jewish Genetics which he founded in the early 1990s. The Center’s mission is to educate Jewish families on the diseases for which they are at increased risk, for example DYT1 early onset dystonia.

Hundreds of prize baskets were raffled at the Chicago Basket Bash in October to raise funds in support of the DMRF’s mission to cure dystonia. Beth Farber, husband Steve Laser, and parents Joel and Harriett Farber host the annual event in honor and memory of Shari Farber Tritt who developed severe dystonia at age eight. Since 2005, the family has made the Basket Bash a family tradition, raising urgently-needed funds for medical research. Beth also organized “Fly for a Cure” an indoor cycling fundraiser held at Flywheel Sports.

Guido and Rita Battaglini of New Jersey hosted a multi-family yard sale in August to support the DMRF. L to R: Frank and Susan LoPresti, Barbara and Chick Leiby, Guido Battaglini, and Ron and Shelly Miller.

The DMRF partnered with Dr. Georgeta Varga of the Parkinson Disease Movement Disorder Clinic in Austin, Texas for an educational meeting on dystonia and the latest in available treatments.

Carol Bolz and her experience with dystonia were featured in the Success Stories section of the September 2015 issue of Toastmaster magazine.

Jason Dunn and Mike Delise of Michigan secured numerous news stories in the Detroit-metro press for their efforts to promote dystonia awareness, including efforts to petition the White House and Jason’s brief encounter with President Barack Obama.

Walter Cooper promoted dystonia awareness throughout Malmstrom Air Force Base in Montana, including adhering Dystonia Moves Me bumper stickers to military vehicles.
The 7th Annual Minnesota Dystonia Golf Classic organized by longtime DMRF supporter Wayne Erickson was held August 15 at Gopher Hills Golf Course. Wayne and his experience with blepharospasm were featured in a previous issue of the Dystonia Dialogue.

At the request of Travelers Rest, SC resident Kelley Rainey, Mayor Wayne McCall issued a proclamation to recognize September as Dystonia Awareness Month. Kelley also decorated a gazebo at the popular Swamp Rabbit Trail to promote awareness locally and was featured in local news media.

Thanks to Heather Connor, stories from members of the dystonia community were featured throughout September by The Mighty, a disability and chronic illness blogging site. Submitting writers included Jean Abbott, Rev. Mike Beck, Pat Brogan, Ginny Bryan, Caitlyn Connelly, Heather Connor, Nicole Dean, Jenelle Dorner, Whitney Jory, Matt Lawrence, Elizabeth Schultz, Tom Seaman, Rebecca Sharp, Michelle Shaw, Carrie Siu Butt, Pamela Sloate, and Lisa Zimmermann.

As team Beat Dystonia, Pat Brogan, Bobby Hutton, and Lisa Wentley placed second in overall team relays at the 19th Annual Dewey Beach Triathlon in Delaware. Pat also guest bartended at Whaleyville Oasis Bar and Grill during Bike Week for tips donated to the DMRF.

On September 12, Greg and Alicia Troy of California hosted the Dystonia Moves Me Barn Dance to raise dystonia awareness and funds to support the DMRF. The event included authentic traditional barn dancing, prizes, and refreshments.

A number of volunteers throughout the country obtained dystonia awareness proclamations from Governors or local governments: Miyoshi Brame (AZ), Ginny Bryan (NY), Debbie Bunch (MO), Ruth Curtis (ME), Mike Delise (MI), Val and Ernie Inman (FL), Johnny McCoy (TN), Martha Murphy (CA), Bob and Ginny Spencer (AZ), Linda Thompson (MS), Martha White (NY), and others.

Mother and daughter Debra Ronning and Annie Ritsch organized “A Night of Movements” on October 26 in Elizabethtown, PA featuring chamber music by Trio Caprice and guest speaker Barbara O’Connell, MD. The event brought visibility to dystonia and collected donations in support of the DMRF.

Gina Rosendall-Saucedo hosted a dystonia awareness party at a trampoline park in Lansing, MI on September 13 to bring visibility to dystonia and commemorate Governor Rick Snyder’s proclamation to recognize September as Dystonia Awareness Month. The event attracted the attention of the local news. Gina is pictured with sisters Breanna and Peyton Strange.

DYSTONIA MOVES ME ONLINE

For more dystonia awareness moments, be sure to check out the Dystonia Moves Me: Show Me Your Moves page online: www.dystonia-foundation.org/dystoniamovesme and the Dystonia Moves Me playlist on the DMRF YouTube channel: www.youtube.com/facesofdystonia
Fifteen-year-old Paige Kaiser assembled the largest team at the 2nd Annual Pittsburgh Dystance4Dystonia Zoo Walk in September. Team “Paige’s Peeps” had over 70 members who attended to support Paige at the Zoo Walk.

“I had a great time at the Zoo Walk,” she says. “It felt really good to have so many people there to support me. And it was exciting to meet people like me. I had never met anyone with dystonia before.”

Paige was mistakenly diagnosed with cerebral palsy at age eight after she began having trouble with her feet. At 13 she was diagnosed with dystonia after the symptoms had spread to her hands. Although she deals with daily pain and is treated with botulinum toxin injections in her legs, she remains as active as possible including in competitive color guard (flag spinning).

When asked what she hoped the public would understand better about dystonia thanks to awareness efforts like the Zoo Walk, she replied: “Just because you can’t always see the dystonia, doesn’t mean it’s not there. We are fighters. We have pain every day. We go through a lot.”
“My children warmed my heart when they asked if they could try to raise money for the DMRF. They love their baby sister so much and in watching her struggles they would give anything to help. Though we feel helpless a lot of the time, this is how we are turning our lemons into lemonade!”

Misty Boutin’s children Payton and Austin held a lemonade stand in honor of their youngest sibling.

Mulshine Girls Continue to Shine in Dystonia Awareness

Once again, 11-year-old Grace Mulshine and cousins Brooke Mulshine, Chloe Mulshine, and Leila Mulshine held a very successful lemonade stand to support the DMRF. The annual lemonade stand was started by Grace’s older sister Catherine, who is now in high school. The sisters’ father Brendan has dystonia and the girls are committed to raising research funds for a cure.

Catherine and Grace were especially inspired by meeting Libby Karns who has multi-focal dystonia and leads the South Bend, Indiana Dystonia Support Group. The family attended a support group meeting while passing through the area on the way home from volunteer camp. “What a special person Libby is,” said Catherine and Grace’s mother Suzanne. “She feels like an adopted grandmother. She moved all of us. Meeting her was the best thing we did.”
Weeding Through the Hype
American Academy of Neurology Reviews Use of Medical Marijuana

The latest National Survey on Drug Use reports that nearly 20 million people over the age of 12 in the United States have used marijuana in the last month. Recreational use is on the rise. Restrictions on its medical use have loosened in several states. New legislation in California promises to establish a regulatory framework for medical marijuana to begin in 2018. Weed may be easy to find, but it is becoming increasingly challenging to find credible information about its role in medicine.

The American Academy of Neurology (AAN) published a review of research studies on the medical use of marijuana to treat neurological disorders. The report was conducted to help neurologists make informed decisions about the use of medical marijuana in states that allow physicians to prescribe it. The review included studies on the use of medical marijuana in multiple sclerosis, levodopa-induced dyskinesias in Parkinson’s disease, chorea associated with Huntington’s disease, Tourette syndrome, cervical dystonia, and other conditions.

Part of the challenge of conducting a review of this kind is that the data available are limited. Furthermore, there are many preparations of medical marijuana available. These include synthetic drugs that mimic the effects of marijuana and whole plant marijuana or its extracts.

The Food & Drug Administration (FDA) has approved two cannabinoid drugs. Dronabinol and nabilone are synthetic drugs that mimic the chemical characteristics of marijuana. They have specific indications for use in AIDS and cancer chemotherapy patients. The FDA allows physicians to prescribe these drugs “off-label” under certain circumstances.

Growing, possessing, processing, distributing, and consuming whole plant marijuana are illegal at the federal level. A number of states have legalized medical use and a small number have legalized recreational use.

Whole plant marijuana can be consumed in a variety of ways that influence its chemical composition, dosing, and efficacy. It can be consumed orally, smoked, or vaporized. Marijuana products have varying ratios of specific compounds that have different physiological effects. There is no system in place for testing, labeling, or quality assurance in states that have legalized medical marijuana. These variables make it difficult to provide physicians or patients with objective information about the effects of medical marijuana.

The conflict between federal and state law complicates the legal and social ramifications of medical marijuana use and future medical research.

The AAN report concludes that, based on the limited data available, oral cannabinoid drugs are not likely effective for treating levodopa-induced dyskinesias in Parkinson’s disease. Oral cannabinoid drugs may be modestly helpful in treating chorea associated with Huntington’s disease. There is insufficient evidence to determine efficacy in Tourette syndrome. A study on the use of an oral cannabinoid in cervical dystonia, which the DMRF partially funded, did not produce data to support or deny efficacy—additional research is needed.

Medical marijuana is probably effective in reducing several symptoms of multiple sclerosis including spasticity and pain. Smoked marijuana is of uncertain efficacy and may worsen posture and balance. Cannabinoids probably do not improve tremor in multiple sclerosis.
Although the pleasant effects of using marijuana are well known, like any drug there can be side effects: dizziness, slowed reaction time, paranoia, anxiety, and short-term memory loss. The AAN review reported there was a 1% risk of serious psychiatric side effects including depression, hallucinations, and becoming suicidal.

As is the case with any non-traditional treatment approach, it is essential that patients with dystonia who consume or are curious about marijuana discuss the matter candidly with their physicians. Individuals who use marijuana need to inform their physicians; newbies need to have an open discussion with their doctors before lighting up.

Individuals with dystonia should also consider sources carefully when exchanging anecdotal information about marijuana use with others, especially on the internet. Online support groups, for example, are full of individual, unverifiable accounts of marijuana use that represent vastly different results—some beneficial, some detrimental, some neutral.

The AAN report stresses that more systematic research is needed on the use of medical marijuana in neurological disorders and that if the evidence suggests it is beneficial, it should be prescribed as other drugs are. A number of clinical trials are underway to investigate the use of cannabinoids in neurological disorders such as spinal cord injury, motor neuron disease, dementia, multiple sclerosis, and others.

**About Medical Marijuana**
- Marijuana (cannabis) contains numerous pharmacologically active compounds called “cannabinoids.”
- A prominent compound in marijuana called tetrahydrocannabinol (THC) can produce feelings of well-being and reduced anxiety as well as paranoia and psychosis.
- A compound found in marijuana called cannabidiol (CBD) limits the psychoactive effects of THC and may have greater therapeutic value.

**Talking to Your Doctor about Complementary Therapies**
A survey of individuals with adult onset dystonias revealed that over half reported using complementary or non-traditional therapies. Many reported using complementary therapies in combination with traditional treatments such as botulinum toxin injections. Complementary therapies may include acupuncture, biofeedback, chiropractic, exercise and massage therapy, vitamin/herbal supplements, and numerous other approaches.

The National Center for Complementary & Alternative Medicine suggests these tips for talking to your healthcare providers about non-traditional therapies.

- Be sure to include all therapies and treatments you use when completing patient history forms. It may be helpful to make a list ahead of time.
- Tell your health care providers about all therapies or treatments—including over-the-counter and prescription medicines as well as herbal and dietary supplements.
- Don’t wait for your providers to ask about your use or interest in complementary therapies. Bring up the subject proactively.
- If you are considering a new treatment approach, ask your health care providers about its safety, effectiveness, and possible interactions with medications (both prescription and nonprescription) and/or implanted medical devices.
- Talking openly with your doctor about complementary therapies is important to make sure your physician is fully informed about your condition and to avoid unsafe interactions.
How would you describe your symptoms?
It started when I was 15 years old with a tremor in my right hand. I started breaking pencils, having difficulty writing, holding things. About six months after that, I started having full-body dystonic storms. The doctors didn’t know what any of it was. It wasn’t for another 12 years that I was diagnosed with paroxysmal generalized dystonia.

When I was in college, I would wake up every morning and have a dystonic storm in bed. It’s so disorienting because you can hear, you’re completely aware of everything that is happening, but you’re convulsing all over the place. And it’s extremely painful. I would manage to get to my classes but there was so much stress with being in school and walking all over campus that I would get back to my dorm and have severe storms for five hours, every day. Now I’m very good at minimizing my symptoms. I make sure I get enough sleep. I have to seriously limit my physical activity. The main symptoms I have on a daily basis are trouble with my hands and feet—they want to curl. So walking can be challenging and tiring. It can be hard to write. I have neck and back pain. But as far as the storms, as long as I take care of myself and limit stress and activity, I’ve gotten to the point where I only have a bad one every couple months.

How did you get through so many years without a diagnosis?
At first, I saw a lot of doctors and felt like I had to figure out what this was. I was afraid. As the years passed with no answers, I just tried to hide the symptoms as much as possible, to not talk about it. Family members were completely stressed by the whole thing. I just tried to mask my symptoms as much as possible so it wouldn’t upset people.

What has helped you cope?
Initially I just believed it was going to get better. After I finished my Master’s degree I realized I needed to change my lifestyle. That was a really hard decision to make. Especially because it seems like every time you meet someone, the first thing they ask is “What do you do for a job?” That was always an uncomfortable moment. I’d explain that I have dystonia, it’s a movement disorder, and I’m not able to work. And people didn’t understand that. But I try to educate people about dystonia. Also, my husband is amazing—he’s a wonderful support and great problem-solver. I’m very lucky.

When you look back, how do you feel about those years you were not diagnosed?
At this point I am at peace with it. It’s more frustrating to me that so few people seem to know what dystonia is. When the doctors told me what it was, it was a huge relief. I could talk to other people; I could go to support groups. But at the same time, now that I have this word to describe what’s wrong, it doesn’t do that much good because so few people know what the word means.

Why is it important for you to support DMRF?
I really want to spread awareness and make sure other people don’t have to wait to be diagnosed. When you are in that period of time when you’re not diagnosed, you can’t do anything because you don’t know: Are you going to get worse? Is it going to kill you? Is it going to go away and you’ll get better? There is no way to live your life. It’s really important to increase awareness especially among the medical community. The more involved I become, the more rewarding it is. It makes you feel good and that what you are doing makes a difference.

To learn more about paroxysmal dystonia, visit www.dystonia-foundation.org

PERSONAL PROFILE
Treacy Henry

Treacy Henry is a longtime DMRF supporter and dystonia awareness advocate. She resides in Massachusetts and is an active member of local support groups including the Rhode Island Dystonia Support Group.

Treacy (R) and her aunt Carol McGrath attended the Providence Dystance4Dystonia Zoo Walk.
How did your symptoms begin and how were you diagnosed?
The spasmodic dysphonia (SD) and writer’s cramp started about the same time. In 1973 I was working and preaching in Australia. I had gone from preaching once a month in the States to preaching three times a week. My voice started to skip and strangle. By 1975 it was pretty bad and nobody in Australia had any clue what it was. Even when I got back here in ’77, no one had a clue what it was. In ’82 I was diagnosed with SD. I mostly ignored the writer’s cramp. It was the least of my problems. They diagnosed it at NIH [National Institutes of Health] when I was volunteering for SD studies in ’88. After the SLAD-R surgery, NIH called asking if I wanted to be in a writer’s cramp study and I said sure. Since March I’ve been getting botulinum toxin in the arm, and that’s made a big difference. It seems to be helping. My handwriting is improving.

Medically what has helped you the most?
In 1989 I started getting botulinum toxin for the SD. I would get six to 10 months on an injection with a very low dose. I was getting a good response. A year ago, I had SLAD-R surgery. And it’s been fantastic. At first you’re breathy, but the voice breaks are gone. It was a couple months before I had much volume, and before that three to four weeks of severe coughing. My voice now is good. It’s amazing how talking on the phone went from being my worst enemy to my best friend—it’s a piece of cake now.

I had a very interesting realization about a week ago. I realized that I was talking to people who for 40 years I wouldn’t have talked to. I was in a campground in South Carolina and there was a worker, a local man, cleaning the bathroom stalls. I got into this great conversation with him. I wouldn’t have initiated a conversation like that before the surgery, because of my voice. You really isolate, and this brought it home to me—the difference the surgery has made.

What has helped you cope?
My wife Ruth and my family have been so supportive. It brings me to tears when you talk to someone with dystonia and their spouse or family aren’t supportive. My friends have been great, church is great. Even when I felt I needed to leave the parish Ministry, they said don’t leave, we’re used to you—we listen, we understand. That was a good place to be, because people tended to be supportive. And then I became a psychotherapist for the last 25 years and that has been a great fit.

Why is it important to you to be involved with the DMRF?
The mutual support and hope for the future with research. When we have support group meetings, you see the same people—I’ve made lifelong friends now and there are always new people joining the family. Being able to welcome them and reassure them and encourage them is so important.

What guidance do you have for the newly diagnosed or those who are struggling?
Find or start a support group! I’m constantly referring people to the DMRF. With online support groups, they are there for everyone, worldwide. It’s wonderful. I’ve definitely made some great friends.

For more information about writer’s cramp, spasmodic dysphonia, and SLAD-R surgery visit www.dystonia-foundation.org
Hundreds of Dystonia Moves Me: Show Me Your Moves kits were distributed throughout Dystonia Awareness Month in September to support groups and individuals who worked to bring visibility to dystonia in their local communities.

For scenes from the campaign, visit www.dystonia-foundation.org/dystoniamovesme