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On the Cover:
For more than 40 years the Dystonia Medical Research Foundation (DMRF) has worked to put itself out of business. The mission of the organization from the beginning has been to advance research for more effective treatments and ultimately a cure and, because we recognize that the science can never move fast enough for those living with dystonia, to promote greater dystonia awareness, to provide up-to-date and accurate educational resources, and last, but certainly not least, to support the well-being of dystonia-affected individuals and their families. See page 10–11 for an overview of key programs in support of the DMRF mission.

The Dystonia Dialogue is the magazine of the Dystonia Medical Research Foundation (DMRF). It is published three times a year to provide information to individuals affected by dystonia, family members, and supporters of the DMRF.

The Dystonia Medical Research Foundation (DMRF) is a non-profit, 501c(3) organization founded in 1976. The mission is to advance research for more effective treatments and a cure, to promote awareness and education, and to support the well being of affected individuals and families.

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The Dystonia Dialogue reports on developments in dystonia research and treatments but does not endorse or recommend any of the therapies discussed. Individuals are urged to consult a physician with questions and concerns about their symptoms and care.

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Dear Friends,

You are critically important to the DMRF. The Foundation is supported by dedicated, generous individuals who believe a cure for dystonia is possible. And not just possible, but inevitable, if we work hard enough and smart enough. The DMRF’s goal is to find a cure for dystonia and, in the meantime, provide information and guidance to those affected. We know there is much work to be done, and that far too many of you are struggling day-to-day under the burden of this life-changing disorder.

Everybody has something to contribute to the DMRF’s success. The DMRF’s impact on dystonia research progress and service to the patient community are possible thanks to you. Whether you give financially, promote awareness, participate in legislative advocacy, or offer your friendship and compassion to others affected by dystonia, you have a role to play in getting us closer to a cure.

We are grateful for your concern for others affected by dystonia. We understand your desire to spare others any difficulty you may have faced in obtaining a diagnosis, finding treatment, or coming to terms with the realities of living with dystonia. You believe in awareness efforts to help quicken diagnosis, providing patients and families with the information they need to make treatment decisions, and programs to help train more movement disorder specialists. You want to know that the DMRF is making every effort to inspire new medical discoveries that improve lives.

On page 10, you’ll see an overview of the key programs you make possible when you make a gift to the DMRF. Please renew your membership or join the DMRF today. Your generous support goes a long way to help us get closer to a cure while providing individuals with all forms of dystonia the information and support they need. We can’t do this without you, and can’t thank you enough for your support.

Sincerely,

Janet L. Hieshetter
Executive Director

Art Kessler
President

RENEW YOUR MEMBERSHIP OR JOIN TODAY
dystonia-foundation.org/membership

Thank you for your support!
DMRF-Funded Discoveries Reveal Unexpected Drug Effects and Clarify Neurocircuitry Underlying Dystonia

Several recent science publications describe exciting discoveries made by a research team in Germany led by DMRF-funded investigator Professor Angelika Richter, DVM, Director of Institute of Pharmacology, Pharmacy, and Toxicology at the University of Leipzig.

Professor Richter and her collaborators have explored new GABA drug candidates in a dystonic hamster model. Novel drug candidates were provided by BioCrea GmbH and Merz Pharmaceuticals. The drugs did not work as expected, but the research team may have discovered an unknown mechanism of action of these drugs. It appears they act through very precise interactions with specific subunits of GABA receptors. GABA receptors respond to the neurotransmitter gamma-aminobutyric acid (GABA), an important messenger chemical of the central nervous system. Professor Richter and her team have also developed a new motor-behavioral test in DYT1 mice created by William Dauer, MD—also with DMRF funding—that demonstrates sensorimotor abnormalities in these mice.

These findings represent important progress in drug candidate exploration and in defining the neurological circuits and molecular mechanisms underlying dystonia, all of which are needed for progress toward new therapies.

DMRF Relationships with Federal Agencies Critical to Research Progress

As an internationally-recognized authority on dystonia, the DMRF maintains an ongoing dialogue with government research and medical agencies to represent the dystonia community. These conversations create opportunities to promote greater awareness of dystonia among federal policymakers and help advance research goals.

Late last year, the Food & Drug Administration’s Center for Devices and Radiological Health called upon the DMRF to help them better understand the role and impact of deep brain stimulation (DBS) as a dystonia treatment. The DMRF was represented by Chief Scientific Officer Jan Teller, MA, PhD and DMRF Support Leader Marcie Povitsky, who underwent DBS to treat severe generalized dystonia. They met with Division Director Carlos Peña, PhD and his team to talk about the current state of dystonia research, provide a patient perspective on DBS for dystonia, and learn about the device review and approval process.

The DMRF was also pleased to participate in a meeting in November at the National Institute of Neurological Disorders & Stroke (NINDS) in Bethesda, Maryland. DMRF Scientific Director Mahlon DeLong, MD and Chief Scientific Officer Jan Teller, MA, PhD met with NINDS Director Walter Koroshetz, MD and members of his team to discuss dystonia research funding.

The DMRF and NINDS have cooperated on a number of initiatives over the years, including groundbreaking dystonia workshops and program announcements. Mark Hallett, MD, Chief of the NINDS Human Motor Control Section, was interviewed in the Winter 2016 Dystonia Dialogue about his lab’s dedication to dystonia research and major discoveries in dystonia neurophysiology. Program Director in the Neurodegeneration Group, Beth-Anne Sieber, PhD, attends DMRF’s Medical & Scientific Advisory Council meetings.

The DMRF looks forward to continuing these conversations with the FDA and NINDS, and continuing to provide the dystonia community with a voice to influential federal agencies.

FIND THE DMRF ON

sharecare.com/group/dystonia-medical-research-foundation

Sharecare is a health and wellness social media platform. Create your profile and connect with the DMRF today.
2017 Mahlon DeLong Young Investigator Award to Focus on X-linked Dystonia-Parkinsonism

The DMRF has announced the recipient of the Mahlon DeLong Young Investigator Award. Aloysius Domingo, MD, PhD, a postdoctoral research fellow at Massachusetts General Hospital earned the $55,000 award for a project entitled, “Deriving Signatures in X-linked Dystonia-Parkinsonism through Integrative Genomic Studies.”

X-linked dystonia-parkinsonism (XDP) is a degenerative neurological movement disorder characterized by symptoms of dystonia in combination with symptoms of parkinsonism (tremors, bradykinesia, rigidity, balance instability, shuffling gait). It is believed to be caused by DNA changes in the TAF1 gene. XDP affects Filipino men almost exclusively. Women may be carriers but rarely become symptomatic. Only about 500 patients have been documented, and experts believe XDP is under-reported.

“As a Filipino researcher in the US, the search for the genetic underpinnings and definitive disease mechanism of XDP is not like any scientific endeavor for me,” says Dr. Domingo. “It represents a personal desire to help my countrymen by removing frustrating barriers that prevent the development of therapy for this rare genetic condition. The award from the DMRF is a step towards this goal, not merely by support through funds, but hopefully also by generating awareness and directing attention to this orphan disorder.”

The goal of Dr. Domingo’s project is to clarify the genetic architecture of XDP and begin to understand the brain pathways and networks that become abnormal as a consequence of the causative mutation. Improved treatment options are urgently needed, and the disease mechanisms in XDP may harbor clues into the mechanisms of other forms of dystonia and parkinsonism.

“Dr. Domingo is already well-known for his award-winning genetic and clinical studies on XDP and other movement disorders,” says Jan Teller, PhD, Chief Scientific Officer with DMRF. “He is exceptionally well-trained and prepared to carry out this work having received an MD degree in the Philippines and a recent PhD in Neurogenetics from the University of Lübeck in Germany. We hope that his long-term commitment to studying this form of dystonia will result in numerous breakthrough discoveries.”

The namesake of the award is DMRF Scientific Director Mahlon DeLong, MD, neurologist and professor at Emory University School of Medicine in Atlanta. His research has advanced the understanding and treatment of Parkinson’s disease, dystonia, tremor, and other neurological movement disorders. His pioneering discoveries contributed greatly to the resurgence of functional stereotaxic brain surgery, which has transformed the lives of thousands of patients affected by these debilitating disorders. His longtime leadership as Scientific Director at the DMRF transformed the Foundation’s science program and inspired many investigators and clinicians in the dystonia field. To honor Dr. DeLong’s lifetime achievements and service, the DMRF established The Mahlon DeLong Young Investigator Award to support an early career PhD or MD researcher pursuing ambitious research in dystonia.
Dystonia Advocacy in 2017

The Dystonia Advocacy Network (DAN) is a grassroots organization that brings people together to speak out with a single, powerful voice on legislative and public policy issues that impact individuals and families affected by dystonia.

Dystonia advocates from across the country are meeting in Washington, DC on March 21–22 for legislative training and meetings in Congressional offices to educate legislators about dystonia and issues that affect the dystonia community. A report from Advocacy Day will appear in the next Dystonia Dialogue.

Dystonia advocates develop relationships with their legislative leaders to help them understand the challenges of those living with dystonia. The DAN continuously works to adopt and advance a legislative agenda which raises awareness of dystonia, educates policymakers about dystonia, addresses patient care issues, and moves research forward.

How You Can Help
By becoming a legislative advocate, you can have a powerful effect on the laws and policies that affect your life and the lives of countless others in the dystonia community. Taking action is not as time-consuming or difficult as you may think. To get started:

- Visit dystonia-advocacy.org/agenda to read a simple summary of the dystonia community’s legislative agenda.
- Know who your legislators are and establish communication with them. The DAN can help you draft correspondence to your Members of Congress to articulate your dystonia story and ask them to support the dystonia community. This simple activity will introduce you to your congressional offices, and make them more likely to act when you reach out to them about specific legislative issues.
- Sign up to receive DAN legislative alerts via email at: dystonia-advocacy.org/contact
- When you receive a DAN legislative alert, respond promptly by calling or sending an email to your legislators. The DAN makes it easy. Every legislative alert includes simple instructions to take action.
- For more information on the DAN and becoming a dystonia advocate, contact the DMRF at dystonia@dystonia-foundation.org or 312-755-0198. The dystonia community needs you, and your participation will make sure your voice is heard.

DAN Responsibilities
- Develop, implement, and manage a legislative and policy agenda to meet the needs of the dystonia community.
- Develop and manage the communications and resources necessary to achieve legislative goals.
- Speak with a united voice for the dystonia community.
- Develop, implement, and manage a network of legislative advocates to achieve legislative goals.
- Monitor and track legislative issues relative to issues pertaining to the dystonia community.
- Develop and maintain Congressional relationships and identify dystonia champions.
- Develop and oversee the use of advocacy tools.
- Evaluate annually the effectiveness of actions and activities.
- Explore areas of collaboration.
21st Century Cures Act
Includes Programs to Benefit Dystonia Community

In December, then-President Barack Obama signed into law the 21st Century Cures Act, a bi-partisan, multiyear effort to support biomedical research and drug discovery. The law will provide $4.8 billion to the National Institutes of Health (NIH) for the BRAIN initiative, the Precision Medicine Initiative, cancer research, and regenerative medicine efforts. The legislation will also provide $500 million to the Food & Drug Administration (FDA).

The DMRF continues to work to implement programs positioned to benefit the dystonia community. These include the development of a National Neurological Conditions Surveillance System; provisions to the FDA for the authority to apply the humanitarian device exemption to devices that treat disorders that affect up to 8,000 individuals (up from current cap of 4,000); and additional provisions to support the work of young investigators and the sharing of research data. The Cures Act will support reducing barriers to researcher collaboration and support the FDA to further engage the patient perspective, modernize clinical trials, and develop new pathways to approve treatments and devices.

The DMRF applauds all who worked to support this landmark legislation and acknowledges Representatives Fred Upton of Michigan and Diane DeGette of Colorado for their tireless efforts to champion this Act.

ADVOCACY VICTORIES
Here are just a few of the DAN’s important accomplishments:

Congressional Briefing on Dystonia
The first-ever Congressional Briefing on dystonia was held to improve dystonia awareness among US Representatives, Senators, and staffers.

Protected Access to Deep Brain Stimulation
The DAN thwarted regulation proposed by the Centers for Medicare & Medicaid Services to limit deep brain stimulation access to volunteers enrolled in clinical studies. This would have prevented countless patients from having access to potentially life-changing treatment.

DOD Began to Support Dystonia Research
DAN advocates urged legislators to add dystonia to the list of conditions eligible for study through the Department of Defense Peer-Reviewed Medical Research Program in the FY 2010 Defense Appropriations Bill. Advocates have successfully kept dystonia in the program every year since, resulting in millions of dollars in additional funding awarded to dystonia investigators.

Protected Access to New Botulinum Neurotoxin
At the urging of DAN advocates, the Centers for Medicare & Medicaid Services issued a temporary unique billing code for a newly approved botulinum neurotoxin type A product. This ensured physicians and patients had timely access to all dystonia treatments approved by the Food & Drug Administration.

Safeguards for New Biologic Therapies
Dystonia advocates successfully urged legislators to establish an approval process for follow-on biologic products at the Food & Drug Administration with significant patient safety provisions. Botulinum neurotoxin is a biologic and a mainstay of treatment for many with dystonia.
What Dystonia Taught Me
Lessons Learned from 20 Years of Teaching Middle School

By Carl Luepker

The first day of every school year I had to introduce myself and then my generalized dystonia, as if it were a second identity, to a new group of 6th graders. I decided this was necessary when it became impossible to hide my dystonic speech. As soon as I talked or used my arms, the dystonia showed itself, and I was viewed as disabled by others. During my last year of teaching, the introduction for what was “wrong with me” became almost apologetic. My dystonia was winning the 35 year fight over ownership of my body.

There was a proverb I learned when I lived in Zimbabwe: *If you can talk, you can sing.* The proverb is meant to be empowering. The point at which, during October of last year, I decided the 2015/2016 school year was my last year of teaching was when my son asked me to sing my favorite song, and I couldn’t. This was a difficult loss. I thought, If I can’t sing, then I can’t talk… Additionally, last year I couldn’t write at all on student work, and typing was my only, albeit painful, means to communicate on written assignments (which students also had to type). My students needed more energy from me, and my nerves were fried. I cared deeply enough for my students that they needed a better version of Mr. Luepker.

Over my 20 years of teaching mostly at-risk middle school students, I chose to learn from my dystonia. An important value for me as a disabled person is to reach the same goals as my best colleagues, but to get there by creatively thinking outside the box. I would find and use any and all technologies to accommodate my professional and instructional needs. My dystonia forced me to put three of the four subjects I taught into Google Classrooms. These were virtual classrooms where students could type their work, watch instructional videos, and even create presentations. And because my district wanted immediate feedback for students and parents, I gave access to my principal so she too could view and even comment on student work as well as see my grades and communications. Google Classroom was so brilliant for me that I could technically be at home sick and see the work I assigned my students being done in real time with the substitute. Because necessity is the mother of invention,

I was the first teacher in my district to set this up. My students became familiar at an early age with an application that they would see more consistently in the future.

Writing on the board was an evolution in body posturing over the 20 years during which I taught. Using chalk in the late 90s was awkward and dystonic in my non-dominant left hand, which I had been forced to switch to when I was 12. As chalk boards were replaced with easier-to-use, less messy
white boards, it bought me another five years. Most recently I used an ActiveBoard, which projected my typed flip charts and provided a heavier pen with a magnet that was more manageable for writing. I had to hold the pen with a stiff underhanded grip, as if it were a large paint brush. By the end of last year, I typed more words on my flip charts than I spoke. My students were forced to read out of procedural necessity, and I was a good writer. I even used hand signals to prompt transitions in my classroom.

Students learn most from what they see and what they do. Trust is critical when working with at-risk kids. I talked 10% as much as my fellow teachers. A natural extrovert, it was awkward to listen first, but it helped me respond to my students’ social and emotional needs. Many of my students didn’t have an attentive adult listener in their lives, and I could be that for them. This ‘listen first’ relationship nurtured student/teacher trust, and I again thank dystonia for that. Orthodox teachers often fill the sound space with their voices, dominating the noise within the room, which can even drown out the incessant tangential conversations middle school students seek out. I became a more direct communicator, with no words to waste, and in a “Minnesota Nice,” occasionally passive aggressive common culture, this felt more genuine and courageous to me.

As dystonia began to waste away my realistic “out of the box” ideas, I needed to become a self-advocate. Asking for help meant becoming more vulnerable, but I still intended to meet my professional goals, so I had to be courageous. I was lucky to work with a staff that was used to understanding and providing accommodations. There was only one day that I did not receive work at a staff meeting electronically and thus couldn’t type my answers. Without accommodation, my dystonia was more exposed. I felt firsthand what some students may have experienced in my own classroom, perhaps when faced with a math test—an urgent desire to disappear. My colleagues knew about my dystonia. Whether from what my teammates shared with my permission, or through social media, my personal information became more transparent, and I was okay with that vulnerability.

There is a turning point, where disability overtakes more than half of your physical abilities, and you become over-defined by it. This turning point last October left me committed to my students first and fulfilling my contract second despite my daily struggles. I remember dozens of mornings waking up with a voice I knew wouldn’t work and wondering, How am I going to talk all day when I can’t even clearly place my order at Starbucks on my ride to work?

Throughout my 20 years of teaching I sought to maximize the strengths I had, and show my students and colleagues all the positivity I could muster. As a worker of 30 years, teaching was always the most challenging job I loved. What teaching with dystonia brought out from my students was a rare compassion only young people have, one un tarnished by real cynicism. I could relate to how some of my students got ignorant looks from strangers, due to certain stigmas and stereotypes, and they knew I shared at least a part of this—being perceived as someone or something you’re not.

I miss the energy and hope my students gifted me. I wanted to teach them by example to push back against life struggles, which are inevitable for all of us. I will miss telling them every day that it is their imperfections that make them rare and perfect.

Carl Luepker resides in Minnesota with his wife and two children. Carl developed symptoms of DYT1 generalized dystonia at age 10. A lifelong educator and writer, he has been published in The Washington Post and authored a novel, Spite: The Biding Time Series. He is currently featured in a series written by the Minneapolis Star and Tribune and has been interviewed for an upcoming piece on his journey by National Public Radio’s Morning Edition. He underwent deep brain stimulation surgery in January.
Research

The DMRF supports research projects while acting as a catalyst to grow the field of dystonia research. The DMRF is addressing the most pressing unresolved questions in dystonia by working with researchers and collaborators from all over the world.

Key Programs:

- **Research Funding:** DMRF has supported 500+ research investigations related to the causes, mechanisms, prevention, and treatment of all forms of dystonia.

- **Scientific Workshops:** Bringing researchers together generates new ideas, leads to collaborations, and attracts new experts to work in dystonia.

- **Pharma/Biotech:** The DMRF has formed alliances with pharmaceutical partners to speed-up the development of new therapies.

- **Clinical Fellowship Training Program:** DMRF has invested $2M in training physicians in the diagnosis and treatment of movement disorders with special expertise in dystonia.

- **Community Initiatives:** The DMRF provides staff support for collaborative research projects including the Global Dystonia Registry, Dystonia Brain Collective, and Dystonia Coalition.

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THENKS TO YOU: DMRF Supporters Power Our Progress

**Our Story is Your Story**

The Dystonia Medical Research Foundation was founded by a family blindsided by dystonia and eager for answers. 40+ years later, the DMRF represents a community of families bonded by dystonia and working together for the greater good.

**WHAT SUCCESS LOOKS LIKE:**

- A world in which all dystonia patients have increased and improved treatment options
- A world in which dystonia treatment is affordable and accessible to all who need it
- A world in which dystonia research is funded at increased levels commensurate with better-known diseases
- A world in which the needs of the dystonia community are reflected in government healthcare policies
- A world in which no one with dystonia is stigmatized or isolated
- A world without dystonia

**OUR VISION**

A cure is possible for everyone affected by dystonia.

**WHO WE ARE**

The Dystonia Medical Research Foundation is the leading dystonia not-for-profit organization, made up of the Board of Directors, Medical & Scientific Advisory Council, and professional staff. A national network of volunteers is a portal for support, awareness, advocacy and fundraising.
WHAT WE DO
For 40+ years, we have been of service to the dystonia community by:
• Providing global leadership on matters critical to dystonia
• Advancing medical research toward improved therapies and a cure
• Promoting greater awareness of dystonia
• Educating patients and families
• Providing support resources for patients and families

Advocacy
The DMRF works to improve quality of life of those affected by dystonia by educating elected officials on dystonia and the policy needs of the community.

Key Programs:
○ Dystonia Advocacy Network: The DMRF is a member and provides staff support for the DAN, which gives the dystonia community a voice to Congress and federal policymakers.
○ Collaboration with Government Agencies: The DMRF is in ongoing communication with federal research and medical agencies to represent the dystonia community and advance research goals.

Support
The DMRF reduces social isolation of patients and families by offering peer-to-peer support programs that help patients and families connect with one another.

Key Programs:
○ Individual Support: DMRF staff are available by phone, email, web, and social media to those seeking information and support.
○ Support Groups: Local support groups are resources for coping support and access to credible information.
○ Internet Support Forums: Online groups are available 24/7 and offer a sense of belonging regardless of geography or mobility.

Awareness & Education
The DMRF offers awareness and education programs to identify the mis/undiagnosed, improve timely diagnoses, improve societal understanding of dystonia, and further the Foundation’s mission.

Key Programs:
○ Dystonia Moves Me: DMRF’s annual awareness campaign mobilizes hundreds of volunteers across the country to promote dystonia awareness locally and via social media.
○ Dystonia Dialogue: The newsletter is distributed via mail, support groups, DMRF events, local community events, and medical offices.
○ Educational Resources: The DMRF offers printed educational materials, a detailed website, and online webinars.
○ Educational Meetings: Communities across the country host dystonia educational meetings every year.
○ Community Events: Volunteers across the country organize and participate in public events to represent dystonia and the DMRF.

50,000+
The number of people to which the DMRF provides direct contact every year through meetings, events, mailings, and office inquiries.

200+
The estimated number of general inquiries to DMRF staff per month via phone, email, web, and social media.
During Dystonia Awareness Month in September, former DMRF Medical & Scientific Advisory Council member and past grantee Dr. Mark LeDoux climbed and summited Mount Kilimanjaro in Tanzania to raise funds for dystonia research and advocacy. To celebrate his efforts to climb the highest mountain in Africa, donors made gifts in his honor to celebrate his successful climb of 19,341 feet.

Pat Brogan competed in the Berwick “Run for the Diamonds” 9-miler on Thanksgiving Day and Rehoboth Beach Half Marathon in December. Every donor who supported him with a donation to the DMRF had their name printed on his jersey at both races. Pat’s 12th Annual Help Find A Cure 4 Dystonia fundraiser took place December 17 at Smoke Rings Cigar Bar, Hazleton, PA. The event raised over $5,000 and the The Lagler Family/ADP of Allentown generously matched the amount, bringing the total to more than $11,000. Pat’s annual events have raised a total of $190,000+ in support of DMRF efforts to find a cure.

Principal Brian Cordeiro at Saint Philomena School in Portsmouth, RI proclaimed January 20 be “Dress Down for Dystonia Day.” Beth Paolero is employed at the school, and her daughter Maddie Paolero has generalized dystonia. To show their support, students made a donation to the DMRF in return for permission to wear “home clothes” instead of their school uniforms. Maddie and her service dog Reno visited the students to thank them.

The students raised over $1,100! The Paoleros have raised thousands of dollars in support of the DMRF, often in partnership with Saint Philomena School.

Team DMRF at the TCS New York City Marathon has raised $112,000 since 2013. In 2016, Mike House ran in honor of Clay Burns, a personal training client who has dystonia. Amber Wood ran in honor of her father Charles Foster who has cervical dystonia and blepharospasm, and mother Virginia Foster who is active in the Greater Washington, DC Dystonia Support Group.

A chamber music concert to benefit the DMRF and Rushmore Music Festival took place January 26 in Davie, FL. The event was organized by Brett Walfish, Missy Walfish, and Katie Smirnova. They were inspired by their friendship with Shari Farber-Tritt, who developed severe dystonia as a child and was an active dystonia advocate.
More than 40 people attended the January meeting of the Dystonia Support & Advocacy Group of San Diego County featuring a presentation by dystonia investigator David Peterson, PhD from the Institute for Neural Computation at UCSD and Computational Neurobiology Laboratory at Salk Institute for Biological Studies. See page 14 for an interview with Martha Murphy who founded and leads the San Diego support group.

Helen Capparell has honored brother-in-law Pat Brogan by participating in the Dystance4Dystonia program for four years. She has run multiple races a year in support of the DMRF. In 2016 she ran the Runner’s World Hat Trick which is three races in one weekend—a 5K, 10K, and Half Marathon. She also completed the Philadelphia Half Marathon, Rehoboth Beach Half Marathon, and the Bahamas Half Marathon.

Jen DeVore has run multiple races on behalf of the DMRF and dystonia community, including the TCS NYC Marathon (twice!) and the Aspire Harrisburg Marathon this November. She has also organized multiple fundraisers on behalf of the DMRF including “dress down” days through her employer. Jen was inspired to support the DMRF by her friendship with a local family and meeting others in the dystonia community through her involvement in the Dystance4Dystonia program.

Mike became passionate about dystonia advocacy after meeting DMRF member Jason Dunn, a friend of his daughters, who has lived with severe dystonia since childhood. Now in his 30s, Jason has undergone four brain surgeries and countless medications, injections, and medical consultations. While dystonia has taken his ability to speak and walk freely, he lives independently, travels, and is an active dystonia advocate, appearing on nationally broadcast television programs and in numerous media stories. Mike and Jason were presented with DMRF Star Awards for their tireless awareness efforts.

Many thanks to DMRF Community Leadership Council member and Support Leader Melissa Phelps and Yolanda Gould for organizing the Blue Christmas For A Cure Dystonia Pageant in support of the DMRF. Special thanks to the 80+ contestants who participated.

Science breakthroughs are unpredictable. What if your $5 donation made the difference to fund a cure for dystonia? This is the premise behind the $5DollarCure4Dystonia campaign launched by dystonia awareness activist Mike Delise. He is working to raise awareness and funds for urgently-needed research toward a cure. To join the campaign and help get the word out, visit 5dollarcure.com/.
Going the Extra Mile
Support Leader Martha Murphy Marks 30th Anniversary

Martha Murphy was 23 and newly married when she woke up one morning with a stiff neck. Over time, she began experiencing pain and involuntary movements in her neck.

The symptoms became so severe she was unable to remain employed at an administrative job she loved in the mental health field. By the time she was finally diagnosed with cervical dystonia in 1987, 12 years had passed.

The year she was diagnosed, Martha founded the Dystonia Support & Advocacy Group of San Diego County, which will mark its 30th anniversary in 2017. Martha has become a vital member of the DMRF family and exemplary support group leader.

“Martha has led her support group longer than any other leader affiliated with the DMRF. She has done a remarkable job,” says Paula Schneider, DMRF’s Leadership Chair. “Her efforts have been tireless. She is a tremendous resource, a great sounding board for people, and a role model in how she has handled her own dystonia. She always shows up, always goes the extra mile.”

Martha joined the DMRF staff in 2004 as part-time Brain Bank Liaison, but says she identifies most as a support leader. “I love the support group,” she says. “To meet people who have been alone and thinking they are the only person in the world who is dealing with these weird symptoms and movements, and for them to find a whole room full of people who have gone through similar experiences is really priceless. It can be life-changing.”

The support group meets every other month. Martha mails and emails a newsletter in advance of each meeting. The support group also organizes dystonia awareness activities and fundraises for research. To date the support group has raised over $50,000 in support of the DMRF and the National Spasmodic Dysphonia Association.

Martha encourages support group members to be as informed as possible about dystonia, treatment options, and resources for general wellness. Meetings frequently feature guest speakers, and presentations are often video recorded so those unable to attend have access to the information shared. She keeps her members informed of dystonia research advancements, often inviting investigators from nearby academic institutions to attend meetings as featured guests.

“Since I was diagnosed, there have been a lot of changes and improvements and breakthroughs.” At the time Martha was diagnosed, only one botulinum neurotoxin product was available to patients; now there are four. Deep brain stimulation surgery was not yet available. Multiple genes have since been discovered, and additional medications have come on the market. “But still I completely understand and can relate to the person who says It seems like nothing is being done and I don’t know how I’m going to make it with this disorder. Some people are terribly impacted by their dystonia. So you have to keep reassuring them that the research is happening. It doesn’t happen overnight, it’s slow, it’s tedious. But the research that the DMRF is involved in is very powerful and can be life-changing for people.”

Martha knows firsthand the difference medical research can make and the value of volunteering for clinical research studies. She participated in an important early study of Myobloc® (rimabotulinumtoxinB), a therapeutic botulinum neurotoxin developed as an option for patients who did not respond adequately to Botox® (onabotulinumtoxinA), the first therapeutic botulinum neurotoxin to be approved for use in dystonia. Myobloc remains the mainstay of Martha’s treatment to this day, 20 years later. “Volunteering for research is a win-win...
situation for the research community and for the patients because by and large you are getting a new form of treatment that is not available to the general public yet, and you can be on the cutting-edge of something that can make a big difference in people’s lives.”

Martha considers her symptoms well-managed, but cervical dystonia continues to take its toll. “The thing you have to remember, and I’m not always good about this, is that you have to be kind to yourself and pace yourself and not do too much. We have to find work-a-rounds for the things we can’t do, or can’t do as well. You have to give yourself permission to not be perfect.”

“For me, keeping a sense of humor is invaluable. We do actually a lot of laughing at our support group meetings, which may sound weird. It helps. I have to say we have some really, really wonderful people in our support group. And I really appreciate how much they embrace new people who come in and make them feel at home and comfortable, and go up to them and talk to them before and after the meetings. It feels like family.”

Members of her group frequently lend a hand with setting up for and cleaning up after meetings. These seemingly simple gestures take on greater significance when dealing with chronic dystonia symptoms and energy is at a premium. “We have a good team.” Martha insists she receives as much support as she gives. “If you can support other people, they support you. That’s the way it’s supposed to be.”

“I can’t quite imagine not doing the support group. I find people to be interesting, fascinating, complex creatures. I’m just interested in them. If you can do something to make someone’s journey a little less stressful or difficult than your journey was, why wouldn’t you do that?”

For more information about the Dystonia Support & Advocacy Group of San Diego County, visit dystoniaofsandiego.com, email leader@dystoniaofsandiego.com, or call 619-582-1961. To search for dystonia support groups near you, visit: dystonia-foundation.org/supportgroups. If you are interested in starting a dystonia support group in your community, contact the DMRF at 800-377-3978 or dystonia@dystonia-foundation.org

Get Social
Check Out DMRF Online Forums

If you do not have access to a local support group, you can still connect with others in the dystonia community. Check out the following online social forums.

Cervical Dystonia Support Forum
facebook.com/groups/dmrf.cervical/
Moderators: Denise Gaskell & Tom Seaman

Oromandibular, Blepharospasm & Cranial Dystonia Support Forum
facebook.com/groups/OMDBlep/

Generalized Dystonia Support Forum
facebook.com/groups/dmrf.gen/
Moderator: Paula Schneider

20/30 Dystonia Group - A Forum for People in Their 20s and 30s
facebook.com/groups/2030dmrf/
Moderators: Ginny Bryan, Chelsi Christman & Marcie Povitsky

Support4Parents of Children with Dystonia
facebook.com/groups/support4parents.dmrf/
Moderators: Carol-Ann Peralta and Dena Sherry

Parenting with Dystonia Support Forum
facebook.com/groups/dmrf.parenting/
Moderator: Jenelle Dorner

Dystonia Spouses & Loved Ones
facebook.com/groups/dmrf.lovedones/

DBSforDystonia Yahoo Group
health.groups.yahoo.com/group/DBSforDystonia/
Moderator: Dee Linde

Online Dystonia Bulletin Boards
dystonia-bb.org/
Moderators: Bob Campbell, Jeff Harris & Linda Walking Woman

For a complete list of DMRF’s online social forums, visit: dystonia-foundation.org/online
Occupational therapists work with clients to identify the barriers that interfere with everyday activities and develop solutions to overcome those barriers. Working with an occupational therapist can be an important part of a comprehensive treatment plan for dystonia and other movement disorders.

Breaking Down Barriers
An initial consultation with an occupational therapist will include a detailed discussion of the client’s daily life. “I really delve into finding out who is this person?” explains Professor Sabel. “What are the things they want to do, need to do, are expected to do.” Occupational therapists help clients with issues related to grooming, bathing, meal preparation, housekeeping, child care, shopping, and even money management. “I’m going to look at the physical barriers, the environments a client has to deal with, the behavioral issues of that person. Ultimately we will develop a plan to make it easier for them to accomplish their goals.”

Challenging the Body & Brain
Therapeutic exercise may be part of an occupational therapy treatment plan, but Professor Sabel stresses that the goal is more than physical fitness: “We all have experience with rote gym exercises. Those movements are specific, and they don’t always correlate to how we need to use our bodies doing every day activities: getting off the toilet, reaching down to clean the litter box, reaching into the cupboards to get something out. There are so many possibilities of movement required in real life, so the occupational therapist may recommend traditional therapeutic exercise, but it always relates to something functional.”

Many tenets of occupational therapy are rooted in the science of neuroplasticity, the nervous system’s capacity to adapt and learn. “There is this potential for creating new pathways by challenging the nervous system in different ways. We know from motor learning theory how the nervous system learns and integrates tasks, and the ability of the nervous system to generalize learning.” For instance, under normal circumstances, if a person reaches to drink from a cup, the nervous system has learned over time to complete the task whether the cup is a mug, a wine glass, or a little paper cup. The body applies the appropriate timing, speed, and coordination automatically. If someone is having difficulty using the hand and arm, and this is making it difficult to drink from a cup, an occupational therapist will help a client practice the movements with cups of various sizes, weights, materials, and textures. This challenges the nervous system to master the subtle motor and sensory adjustments necessary to complete the task under real world conditions.
Dystonia & Body Awareness

A concept that Professor Sabel has found especially important for clients with movement disorders is body awareness. He teaches clients to notice the subtle movements and sensations of the body that often go unnoticed due to behaviors and habits acquired over the years, sometimes to compensate for neurological symptoms.

“We’re familiar with using our senses: we smell, we see, we feel, we taste, we touch,” he explains. “But proprioception is an amazing skill that we don’t pay much attention to unless we are in a pitch black room and need to move around. We have all these receptors in our body that let our brain know exactly where our body is in space at any one time, and we can tap into that.”

As a popular guest speaker for the New York City Dystonia Support Group, Professor Sabel has introduced attendees to techniques that help people move with greater ease and control: “If I have cervical dystonia and I’m trying to turn my head, my habit may be to rotate my head and not let the rest of my body move at all. This makes those neck muscles that are already working too hard because of the dystonia become even tenser. But if I disperse the effort by letting other parts of my body participate in the movement—the pelvis, the spine, the shoulders—that takes some stress and strain off of those neck muscles.” A 15-minute demonstration led by a qualified professional can help individuals make subtle adjustments that have an immediate impact on function.

“If you are having trouble moving your body, if you have pain and discomfort, the more you can become aware of your body and understand your body, you then can have more control over things.”

Search for Meaning

Occupational therapy is an art as well as a science. “A vital part of working with somebody,” says Professor Sabel, “is tapping into their motivation, challenging them in the context of real life activities that are meaningful to them and, again, always relating back to something functional.”

One of Professor Sabel’s clients was an engineer recovering from a stroke. His balance was impaired, he could only stand for short periods of time, and he had trouble with fine motor coordination in his hand. The stroke also damaged vision in one eye, and his spatial perception was off; he had trouble differentiating top from bottom, back from front. These issues were creating many difficulties in his everyday life. In the initial interview and evaluation, Professor Sabel learned that one of the client’s passions was woodworking. The man had a workbench in his home and loved to build with his hands. So Professor Sabel developed a creative strategy to address all of his client’s issues in a single activity: building a birdhouse.

“I had a wooden birdhouse kit, and I had him standing at a height-adjustable table. I put all the pieces to make the birdhouse on the table with the instructions, and I spread them out across the whole table. He had to stand there, look at the instructions, locate the pieces, and figure out how to put the pieces together. He had to use these little nails and a hammer to put the pieces together. I was challenging him on many of the underlying issues that were making it hard for him to do lots of activities: balance, spatial relations, fine motor, vision. By incorporating an activity that he liked, that was meaningful to him, it kept him so engaged that I was able to have him stand for 25 minutes before he had to take a break. When we did the more traditional balance exercises, after 2-3 minutes he would say he was tired and ask to sit down. But this kept him engaged. If you walked by the room, and saw us standing there, you might say, Oh, how nice, they are doing arts and crafts, but it was more than that because it was challenging him and it was something that he cared about. And it really helped move him forward.”

Environment

Occupational therapists help clients thrive in different environments. There are often simple and inexpensive adjustments to be made in a home that make daily activities easier. Occupational therapists also help people function outside the home, not just physically but socially.

Professor Sabel recalls a client with dystonia who became withdrawn. “He and his wife used to love to go see movies, go to restaurants. It became really hard for this person to get out and move. He was self-conscious being in public and being seen by other people. This would create tremendous anxiety. So he disengaged.” A dear friend was getting married, and the man was unsure whether he would be able to attend the wedding. He and Professor Sabel talked through his concerns. The man’s biggest concern was wearing formal clothing especially the restroom. The second concern was being able to stand long enough to attend the wedding. He and Professor Sabel worked with this man to develop a plan that would prepare him to attend the wedding. Continued on page 18
Contribute to Research by Registering as a Brain Donor

The brain tissue of deceased individuals with dystonia is a precious and scarce resource that investigators value deeply in the pursuit of a cure. Registering in advance as a brain donor is a way to contribute to the field of dystonia research that assists researchers working today and in the future.

The DMRF works in partnership with the Harvard Brain Tissue Resource Center (HBTRC) at McLean Hospital in Belmont, Massachusetts to assist people interested in registering as brain donors. The HBTRC maintains a private collection of brain tissue from individuals with all forms of dystonia. The DMRF also serves as the administrative center for the Dystonia Brain Collective, a partnership among several dystonia patient groups to encourage and facilitate brain donation. Donated tissue is available to researchers for dystonia-related studies.

For more information on brain donation or to begin the registration process, visit dystonia-foundation.org/brain or contact the DMRF at 800-377-3978 or brainbank@dystonia-foundation.org.

About Brain Donation

- Brain donation does not alter the appearance of the donor or interfere with memorial services or remembrance practices.
- There is no cost to the donor or family.
- Brain donation does not violate the tenets of most religious or spiritual organizations.
- Donor information remains confidential. However donors must inform next of kin that they are registered to donate their brain.
- Donated brain tissue must be received at the HBTRC within 24 hours of the donor’s death. HBTRC staff are on call 24 hours a day.
- Individuals have the right to withdraw from the program at any time.
- By combining brain donation with monetary support, you immeasurably multiply your assistance to the progress of dystonia research.

Richard Sabel, MA, OT, MPH, GCFTP is a clinical Assistant Professor at SUNY Downstate Medical Center’s Occupational Therapy Program and Educational Director of East West Rehab. His clinical expertise is in adult physical rehabilitation. He is a certified Feldenkrais practitioner and incorporates complementary interventions such as Yoga and Tai Chi into occupational therapy practice. Professor Sabel teaches at several professional degree programs and divides his clinical time between private and community practice.

Professor Sabel will be a featured speaker at the NYC Dystonia Patient Symposium on May 7, 2017 at Friedmann Conference Center at Mount Sinai/Beth Israel.

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This included calling the venue in advance and asking about accessibility of the facility. A friend took the man shopping to purchase the outfit he planned to wear at the wedding, and they practiced the buttons, the belt, the zipper. When he arrived at the venue the day of the wedding, he immediately checked out the restroom to make sure there were no unforeseen problems.

“When I saw him at the next session,” says Professor Sabel, “he said he had an amazing time, and he had no barriers. He was able to negotiate everything. It was important for him to be there, and we made it happen by problem-solving together. This is what an occupational therapist will do.”

Working with an Occupational Therapist

Individuals with dystonia who are interested in consulting an occupational therapist are encouraged to discuss a referral with their doctor. It is recommended that the occupational therapist have experience working with clients with movement disorders.

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Drew Jackson is a former teacher who developed dystonia and other neurological symptoms suddenly in 2015, resulting in hospitalization and weeks in a rehab facility before he was diagnosed. He had previously recovered from a similar episode many years prior. He and his wife Jessica are members of the Western PA Dystonia Support Group.

How did your symptoms begin?
It started with a headache and drooping in my face. Within days, my speech was gone, I developed tremors, my walking deteriorated. I had horrible tremors in my left arm and head tremors. I couldn’t keep my head up, couldn’t keep my head straight. My neck muscles were so tight, everything was rigid. I would also have intense dystonic episodes. I was having issues communicating due to the hemi-facial spasm. The whole experience was difficult and humbling. In the back of my head, I was thinking a year from now I’ll be back walking, I’ll be out of the wheelchair, I’ll be fine. That kept me going. I was telling everyone, Oh we’re going to be good. A lot of it I was doing for other people so they wouldn’t be down. But I also had this creeping doubt like, man, this is worse than before and you’re older this time. This may be a permanent situation. Outwardly I tried to keep my sense of humor and tried to keep smiling, but there was a lot of doubt in my head.

How does dystonia affect you today?
Because I have gotten healthier physically, there are days I feel great. I feel like I am walking better. I don’t need my cane if I’m inside the house, just if I go out. But then if I have to rush to do something I’ll get reminded I’m still slower than usual. The bad days, I would say physically and mentally, are when I start doubting myself, thinking I can’t do something. For example, when I do my laundry I can’t carry the laundry basket down the steps any more. I have to go down the steps backwards and drag it down the steps. Things like that knock my head a little bit. I understand I am making progress. Some days I feel like I should be better or farther along.

What complementary therapies help you?
I do see a mental health therapist once a week, and she has taught me mindfulness meditation. I do breathing techniques to help me if I have an episode or am struggling physically or mentally. Also I had at one point gone to myofascial therapy and that really helped the tremor in my left arm. Boxing therapy has done wonders for me. It’s a program for people with PD [Parkinson’s disease], but it really is effective for me. I have been able to take some of the “regular” boxing classes in addition to the PD classes. We’re basically using all of the training techniques that someone training for a fight might use. We’re in the fight of our lives, against PD or dystonia. We’re using the heavy bag, speed bag, foot work, and all of those things help with PD and dystonia. There is no contact except for the bags or the trainers’ mitts. It’s helping my core strength and balance. The people there, they all have PD, but a couple of them have dystonia in a foot or hand as well. Some of these guys are a lot older than me—like my parents or grandparents age—but I consider them older brothers and sisters. They are in there banging away at these bags, and they are smiling, and they are loving life. That gives me strength.

What medical therapies help you?
I am on many medications—some of them I am still learning to pronounce. Botox® injections help as well in my neck and shoulder. It’s uncomfortable to get the shots, but it’s worked pretty well for me. The doctor will also give me a little bit around my eye to keep my eye open. I still have a little pulling on the right side of my face, but I’m ok with that.

Any advice you would give to others who are new to dystonia?
I would say make sure you have someone with you to ask your doctors as many questions has possible. My wife and family have been really supportive. My mom is a nurse. My wife was really determined to get answers. When we were finally told yes this is dystonia, and there is no cure, it was like a punch in the gut. Neither my wife nor I were prepared. Definitely ask questions—question everything.
Renew Your DMRF Membership Today

As a member of the DMRF, you will continue to receive the latest news and information.

- Go to dystonia-foundation.org/membership,
- Use the enclosed donation envelope,
- Complete and return the form on the back of this newsletter, OR
- Call the DMRF headquarters at 312-755-0198.

Thank you for your support!

Stay in Touch!
Sign up for the DMRF's monthly e-newsletter for the latest updates and announcements:
dystonia-foundation.org/email