



DYSTONIA
MEDICAL
RESEARCH
FOUNDATION

Dystonia Dialogue

NEWSLETTER OF THE DYSTONIA MEDICAL RESEARCH FOUNDATION

Winter 2018 | Volume 41 • No. 3

INVESTIGATORS & FAMILIES UNITED FOR A CURE



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Childhood Dystonia

Thank you...

...to the DMRF community for all you do to increase awareness of dystonia. DMRF supporters host local events, distribute awareness materials, and spread the word on social media. See page 17 for more in "People on the Move."

Mandi Sleeper organized the **Nashua, New Hampshire Dystonia4Dystonia Walk** on June 2.

The **3rd Twin Cities Dystonia Zoo Walk** was organized by Shanna and Brad Schmitt, Billy McLaughlin, and members of the Minnesota Dystonia Support Group. Guest speakers included Dr. Robert Raike.

The **Detroit Dystonia Zoo Walk** reached its fifth year on July 22. The event was organized by DMRF Community Leadership Council Member Rosemary Young. Guest speakers included Dr. Neepa Patel.

The **3rd Portland Dystonia Zoo Walk** was organized by Dee Linde and members of the Portland, Oregon & Southwest Washington Dystonia Support Group. The event took place August 19.

Sheila Killham, leader of the Cedar Rapids Dystonia Support Group, organized the **Dogs for Dystonia Dog Walk** in Marion, Iowa on August 25.



The **4th St Louis Dystonia Zoo Walk** organized by June Tritley took place August 26. Guest speakers included Pam Wheatley and Dr. Joel Perlmutter.

The **Temple Challenge Wacky 5K** is a free monthly event organized by Stacy Houser of West Virginia to encourage fitness. September's event was dedicated to dystonia awareness in honor of Temple Challenge participant Kristin Cinglie.

On September 8, the **6th Cincinnati Dystonia Zoo Walk** took place, organized by DMRF Community Leadership Council Member Melissa Phelps. Guest speakers included Drs. Donald Gilbert, Steve Wu, and Michael Schmerler.



The **4th Providence Dystonia Zoo Walk** took place September 8. The event was the joint effort of several families spearheaded by Sue Baron, Tricia Bono, Beth Paolero, Paula Schneider, and Steve Sowka. Guest speakers included dystonia advocate Mandi Sleeper and Dr. Umer Akbar.

DMRF Board Member Pamela Sloate organized the **4th Bronx Dystonia Zoo Walk** on September 16. Over 600 people attended. Guest speakers included Dr. Jesse Goldberg. Team



Ali's Zoo-Gooders had 140 members and raised over \$50K.

The Flanagan Family, who created the original Dystonia Zoo Walk, continued their support of the DMRF with the **7th Cleveland Dystonia Zoo Walk** on September 22. Many thanks to the organizing committee: DMRF Community Leadership Council Member Karen Flanagan, Jane Ann Flanagan, and Gale Flanagan. Dr. Aasef Shaikh provided remarks.

The **5th Pittsburgh Dystonia Zoo Walk** took place September 23 thanks to Western Pennsylvania Dystonia Support Group co-leaders MaryRae Nee, Ed Cwalinski, and their supporters, including Chris Mack of The Fan Morning Show on 93.7 Pittsburgh Sports Radio and Tara Sorley. Dr. Donald Whiting provided remarks. The Zoo Walk was preceded by **Dystonia Day with the Pirates** at PNC Park on September 4.

The **2nd Stomp Out Dystonia Run/Walk** took place September 22 in Beloit, Wisconsin. The event is spearheaded by Jessica Lawler.

Luanne Pinedo Madden, David Madden, and Holly Machado hosted

the **3rd Fresno Dystonia Zoo Walk** on September 29. Special guests included H. Spees from the City of Fresno.

500 people participated in the **2nd Philadelphia Dystonia Zoo Walk** on September 30 organized by Janice and Len Nachbar, Joanna Manusov, and Raman Patel. Special guests included US Congressman Dwight Evans, Dr. Pedro Gonzalez-Alegre, DMRF supporter Debra Ronning, and DMRF Leadership Chairperson Paula Schneider.

The **Indy Hunt for a Cure for Dystonia** took place September 30 in Garfield Park, Indianapolis. The organizing committee included Chelsi Christman, Sarah Ernstberger, and Sunshine Fox. DMRF Community Leadership Council Member Beth Farber and longtime Support Leader Joel Farber were in attendance.

Beth Farber, Steve Laser, and Joel Farber organized the **Chicago Basket Bash** in October. The family advocates for dystonia in honor and memory of Shari Farber Tritt who developed severe dystonia as a child and Harriett Farber who was a devoted dystonia advocate.

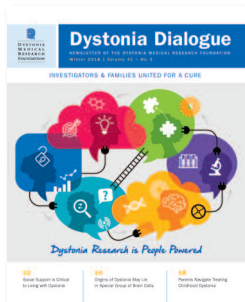
And still to come... Dystonia Zoo Days are planned for San Diego on November 3 and Phoenix on November 17. Find info at dystonia-foundation.org/events.

The DMRF thanks National Sponsors Allergan, Ipsen, Merz, and US WorldMeds for their generous event support.

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On the Cover:



DMRF is a partnership between research and patient communities. One of the things that makes the Foundation unique is the ongoing emphasis on researchers and families working together toward scientific progress and improved daily living for those affected. In this issue of the *Dystonia Dialogue* you'll find numerous examples of investigators and families working together, directly and indirectly. For example, patients are critical to research progress because

only individuals and families living with dystonia can take part in clinical research. See page 10 for an overview of ways to participate in clinical research and page 14 to read about novel research that may soon lead to innovative new treatments.

In turn, the medical and scientific community is turning out to support patients, not only in their tireless research toward the cure, but by participating in community events, promoting dystonia awareness, advocating to the federal government, and helping DMRF create educational resources to make sure people have access to the very latest information.

Partial support of the Dystonia Dialogue is provided by educational grants from Allergan Foundation.

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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Foundation Update



Art Kessler
President



Janet L. Hieshetter
Executive Director

The mission of the Dystonia Medical Research Foundation (DMRF) is broad, intentionally so. The needs of the dystonia community are great. For more than 40 years, the DMRF has stood up for those impacted by dystonia through awareness, education, advocacy, and support programs, while also investing in research that will lead to better treatments and ultimately a cure.

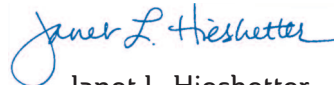
The DMRF's most important long-term goal is to go out of business, for all the right reasons, when there is no longer a need for what we do. To achieve that long-term goal, research is the key. In the remaining months of the year, we're organizing scientific meetings on drug discovery and myoclonus-dystonia, plus a workshop dedicated to dystonia in partnership with the National Institute of Neurological Disorders & Stroke. On page 9, you'll read about outstanding second year grant investigations exploring various aspects of dystonia and an exciting new research contract aimed at developing a radical new approach to neuromodulation therapy for dystonia and other neurological disease.

The DMRF's programs in awareness, education, support, and legislative advocacy are more important than ever. Increasing awareness of what dystonia is and how it changes lives are fundamental to everything we do. Volunteers across the country are promoting awareness of dystonia by organizing events and participating in the DMRF's annual Dystonia Moves Me campaign. Educational meetings for individuals with all forms of dystonia and their loved ones are taking place in cities throughout the coming months. Our legislative advocacy work ensures that the needs of the dystonia community are addressed by government leaders and that dystonia researchers have access to funding through all available federal agencies. On page 22 you can read about the transformative experience of participating in Dystonia Advocacy Day for one first-time attendee. The importance of support cannot be overstated because no one should go through the dystonia journey alone. On page 12 you'll find information about resources available through DMRF's support program.

Your support fuels scientific discoveries and sustains unique programs to benefit individuals and families impacted by all types of dystonia. You are the essential ingredient that holds the DMRF mission together. Thank you for your support. We cannot do it without you.



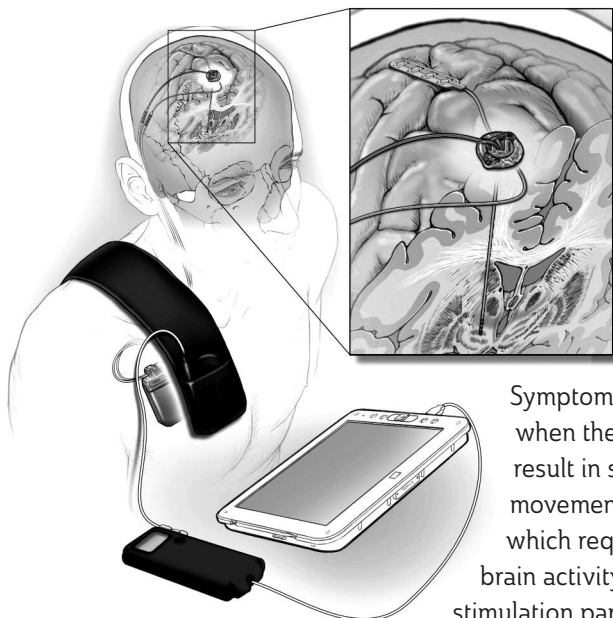
Art Kessler
President



Janet L. Hieshetter
Executive Director

Successful Adaptive DBS for Parkinson's Disease Offers Promise for Neurological Disorders

A team of investigators at University of California, San Francisco led by past DMRF Medical & Scientific Advisory Council Member Dr. Philip Starr has made a groundbreaking step toward a next-generation approach to deep brain stimulation (DBS).



They have successfully tested a fully implanted adaptive DBS system in Parkinson's disease patients that senses brain activity and automatically makes adjustments to stimulation parameters according to a patient's needs. This pioneering study was conducted in two patients.

DBS is a proven effective therapy for Parkinson's disease, dystonia, and a growing list of neurological and psychiatric disorders. In current DBS systems, an implanted medical device delivers continuous stimulation to the brain and adjustments to the stimulation must be made using a remote control device in the hands of a highly trained clinician.

Symptoms of Parkinson's disease can fluctuate dramatically, so there may be times when the DBS system delivers more stimulation than a patient needs. This can result in stimulation-induced adverse effects including abnormal, involuntary movements referred to as dyskinesia. By contrast, an adaptive DBS system, which requires implantation of an additional brain 'sensor,' can detect specific brain activity signatures associated with DBS-induced side-effects and adjust the stimulation parameters in real-time, without the need for an external device.

The goal of the study was not to demonstrate that adaptive DBS is therapeutically superior to current DBS, but to demonstrate that this new approach is feasible without losing efficacy. Both patients in the study retained the clinical benefits of their original DBS system. One of the advantages of adaptive DBS is energy savings to the stimulator battery, resulting in fewer stimulator replacement surgeries. Additional advantages are that adaptive DBS is adjusted without the need for a clinic appointment and/or the need for patients to adjust their devices manually.

This study is one example of ongoing research efforts to make DBS systems more customized to a patient's individual needs and to reduce the frequency of adverse effects.

Swann NC, de Hemptinne C, Thompson MC, Miocinovic S, Miller AM, Gilron R, Ostrem JL, Chizeck HJ, Starr PA. Adaptive deep brain stimulation for Parkinson's disease using motor cortex sensing. J Neural Eng. 2018 Aug;15(4):046006.



STAY IN TOUCH!

Join the DMRF email list by submitting a request at dystonia-foundation.org/contact or calling 800-377-3978.

Targeted Gene Panel May Provide Cost-Effective Diagnostic Tool for Movement Disorders



Movement disorders, including dystonia and parkinsonism, are a diverse group of diseases, often with varied and overlapping symptoms. These disorders can be acquired or inherited, with numerous genes identified, making diagnosis a challenge. A consortium of French researchers has

devised a tool to help clinicians diagnose complex movement disorders. The research group created a 127-gene panel and tested it in 380 patients with suspected movement disorders. The panel had a diagnostic yield of 22%, which is in accordance with other targeted genetic testing for neurological disorders. The pre-labor cost of the gene panel per patient is a fraction of the cost of whole-genome and whole-exome genetic sequencing, methods that have been growing in utility for research but remain largely impractical for widespread clinical use due to the high cost and reluctance of health insurance providers to reimburse.

The research group also uncovered several unexpected correlations between clinical signs and genetic results. For example, findings suggest that the PRKRA(DYT16) gene, associated with a rare recessive form of childhood onset dystonia-parkinsonism, should be added to the list of genes tested in patients with suspected myoclonus-dystonia.

The gene panel may be particularly relevant to patients with undiagnosed childhood onset or familial movement disorders. Looking forward, the researchers suggest that testing the gene panel in a larger group of patients would be revealing, as well as potentially adding genes to the panel.

Montaut S, Tranchant C, Drouot N, et al. French Parkinson's and Movement Disorders Consortium. Assessment of a Targeted Gene Panel for Identification of Genes Associated With Movement Disorders. JAMA Neurol. 2018 Jun 18.

Treatment with Cannabinoids Improves Dystonia in Huntington's Disease



Dystonia is a common motor symptom in Huntington's disease, a progressive brain disorder characterized by uncontrolled movements, loss of cognitive abilities, and emotional disturbances. A team of

clinicians from Germany and Austria reported striking results after treating individuals with early onset Huntington's disease with cannabinoids, synthetic drugs that are derivatives of natural compounds found in cannabis (marijuana).

In all seven patients who participated in the study, the Huntington's disease was advanced and dystonia symptoms were severe. Cannabinoid treatment reduced dystonia symptoms in every participant. In some cases, the positive impact on quality of life was profound. One patient regained the ability to lift his head, which allowed him to engage socially and improved dental hygiene. He also regained use of a clenched hand. Another patient who was wheelchair bound regained the ability to walk without assistance. Several participants showed improvements in mood and behavior.

The investigators acknowledge the limitations of the study due to a small sample size, limited follow-up, and lack of randomization of the trial. However, the results are so encouraging that a double-blind, placebo controlled trial is highly warranted to study in more depth the effects of cannabinoids on dystonia in Huntington's disease.

Reports on the use of cannabinoids in dystonia are very limited, but new data, often coming from studies like the one described here, increasingly suggest the need for more clinical trials.

Saft C, von Hein SM, Lücke T, Thiels C, Peball M, Djamshidian A, Heim B, Seppi K. Cannabinoids for Treatment of Dystonia in Huntington's Disease. J Huntingtons Dis. 2018;7(2):167-173.

Storm Reading Cast Reunites for 30th Anniversary

For decades, artist-activist Neil Marcus has challenged how society views individuals with disabilities. He became an iconic figure in the disability culture movement that coalesced in the late 1970s and 1980s by using theatre, dance, poetry, and contemporary art to express his unique vision: "Disability is not a brave struggle or 'courage in the face of adversity.' Disability is an art. It's an ingenious way to live."

Neil received a United Nations Society of Writers Medal of Honor for his play *Storm Reading*, a groundbreaking work that explored disability, human relationships, and the human condition. Neil performed alongside Matthew Ingersoll and Kathryn Voice. This year, the cast reconvened for a special 30th anniversary event at Lobero Theatre in Santa Barbara, California on September 21. Actor and director Anthony Edwards hosted. The leadership of the DMRF extends heartfelt congratulations to the cast and director Rod Lathim for this special production.

Neil lives and works in Berkeley, California. He began developing generalized dystonia at age eight. Neil has danced internationally since the 1980's, and is included as a seminal voice in the National Endowment for the Arts Oral History Project. He is frequently recognized for his appearance on the hit television show, *ER*. He has published numerous books and his poetry has been published internationally in diverse publications.

Top Dog

Dogs for Dystonia Raises Awareness & Research Funds



urgent need for research toward a cure.

Dogs for Dystonia is an annual campaign to raise awareness while celebrating the special bond between people and their dogs. Each February, dog lovers unite in a Virtual Dog Walk to raise visibility of dystonia and the

This year, a fluffy little guy named Zack was top dog for collecting donations, earning \$1,900. Zack belongs to Joyce Sederburg, who participated in the Zoo Walk in honor of DMRF Board Member Marilynne Herbert. Marilynne's two daughters have dystonia and have undergone deep brain stimulation surgery.

"We didn't know about dystonia until we met our friend Marilynne, who is one of the kindest, most loving and generous people that we know," says Joyce. "All of the support that people gave to Zack in the Virtual Walk campaign was really due to all of us who love and respect Marilynne and her family so much!"

To be notified when registration opens for the 2019 Virtual Dog Walk, send us a request to join our email list at dystonia-foundation.org/contact or call 800-377-3978.

High Five!

*\$5DollarCure4Dystonia Continues to be Force
for Dystonia Research*



Science breakthroughs are unpredictable. What if your \$5 donation made the difference to find a cure for dystonia? This is the premise behind \$5DollarCure4Dystonia launched by dystonia awareness all-stars Mike Delise and Jason Dunn. More than \$30,000 has been raised to date. Funds raised through \$5DollarCure4Dystonia support research.

For more information about \$5DollarCure4Dystonia and to donate, visit 5dollarcure.com. A Facebook group about the campaign can be found by searching keyword "\$5Cure4Dystonia."

Renewing Grants Explore Basic & Clinical Aspects of Dystonia

Decades of investing in dystonia research have produced a vibrant field brimming with new opportunities for medical discoveries to improve lives.

The DMRF Medical & Scientific Advisory Council is made up of distinguished clinicians and scientists whose expertise guides the Foundation's science programs and research funding. DMRF's science plan is reviewed annually to inform the topics and scheduling for research funding cycles.

The DMRF issued a request for research proposals inviting projects on any aspect of basic research on dystonia or clinical work directed at developing new and improved treatments. The Foundation is proud to renew the following outstanding projects.

Neuroanatomical Substrates for Disrupted eIF2-alpha Signaling in Dystonia - 2nd year
Nicole Calakos, MD, PhD, Duke University

Tremor, Oscillations, Synaptic Plasticity, and DBS for Dystonia - 2nd year
William Hutchison, PhD, Toronto Western Hospital (Canada)

Determining the Role of Torsin in Nuclear Pore Complex Assembly - 2nd year
Patrick Lusk, PhD, Yale University

Synaptic Plasticity in a Mouse Model of Paroxysmal Dystonia - 2nd year
Alexandra Nelson, MD, PhD, University of California, San Francisco

Investigation of Striato-Pallidal Connections in a Mouse Model of DYT1 Dystonia - 2nd year
Giuseppe Sciamanna, PhD, University of Rome tor Vergata (Italy)

Dystonia-associated Endoplasmic Reticulum Defects and the (De)regulation of Neurotransmission - 2nd year
Patrik Verstreken, PhD, VIB Leuven (Belgium)

For research project summaries, visit dystonia-foundation.org/funded_projects

New Project Combines Artificial Intelligence & Deep Brain Stimulation to Treat Dystonia

The DMRF is partnering with Jesse H. Goldberg, MD, PhD of Cornell University on a project to engineer a revolutionary new generation of deep brain stimulation (DBS) devices to treat dystonia and other neurological diseases.

Dystonia results from abnormal brain activity that can be corrected by direct electrical stimulation of dysfunctional brain pathways. In current DBS systems, an implanted medical device delivers continuous stimulation to the brain and adjustments to the stimulation must be made using a remote control device in the hands of a highly trained clinician. A major obstacle to providing patients with maximum benefit from this therapy is knowing where in the brain to stimulate and tailoring stimulation parameters to the unique needs of each patient. Dr. Goldberg proposes a radically new approach to DBS. He is using artificial intelligence to develop a system in which a computer, interconnected with the brain, figures out exactly how and where to stimulate to restore normal movement.

In this three-year project, Dr. Goldberg will establish the feasibility of this concept in mice. He is collaborating with Mert Sabuncu, PhD in the School of Electrical and Computer Engineering and School of Biomedical Engineering at Cornell University.

Dystonia Research is People Powered

Volunteers are Essential for New Discoveries

You don't necessarily need a science degree to contribute to dystonia research. Individuals with all types of dystonia, and sometimes family members, can actively participate in research in several ways.

Volunteer for Clinical Research

Clinical trials are research investigations that involve human volunteers. Participants help evaluate medications, medical devices, and other treatments in scientifically controlled settings. In the United States, clinical trials are required before a drug is approved by the Food & Drug Administration (FDA) and made accessible to the public. Trials may be designed to assess the safety and efficacy of an experimental therapy, to assess whether a new treatment is better than the standard approach, or to compare the efficacy of two therapies.

Clinical studies (sometimes called observational studies) gather information from volunteers through medical exams, tests, or questionnaires.

Patients play an invaluable role in these investigations. Clinical research is not possible without human volunteers.

"I volunteer for any and all dystonia studies," says Barb Johnson who has lived with dystonia for 40 years. "Knowing that I may be contributing for a cure brings great personal satisfaction." Barb participated in a clinical

trial for botulinum neurotoxin before FDA approval, an instructional demonstration for doctors-in-training learning to administer botulinum neurotoxin injections, a study in sensory tricks, a tolerance study for a new medication, and others. "The sensory tricks one was such a fun study. But it made for a bad hair day," she jokes, "because I wore a cap with lights on the rim to track the movements of my head."

If you are interested in learning more about participating in clinical research, consider asking your movement disorder specialist if they are aware of studies in need of volunteers. Additionally, search for clinical trials by keyword and location on these websites:

- National Institutes of Health Clinical Trials
nih.gov/health/clinicaltrials/
- Center Watch Clinical Trials
centerwatch.com

About Clinical Trials

- Participants may have access to treatments that are not otherwise available and may not be FDA approved for several years.
- Study participants receive treatment free of charge and may be reimbursed for travel expenses or paid for their time.
- Participants can leave a research study at any time.



Join the Global Dystonia Registry

Individuals with dystonia, from all over the world, can participate in research without leaving their homes by sharing their medical histories in a confidential database called the Global Dystonia Registry (GDR). Individuals with adult onset focal dystonia are especially encouraged to participate. The GDR complements the work of the Dystonia Coalition, a cooperative project between medical researchers and dystonia patient advocacy groups, for which the DMRF serves as an administrative center. The DMRF also provides administrative support for the Global Dystonia Registry. To date 5,200 people have joined in this critical research initiative.

The goal of the GDR is to support future dystonia studies, including clinical and research trials. Although the focal dystonias have many different manifestations, most experts believe they share a common mechanism that causes the disorder. The common causes may be a similar gene defect, similar lifetime experiences, or both. Collecting information from different patient populations may help identify the common features they share. For more information and to register visit globaldystoniaregistry.org.



Register as a Brain Donor

Studies that have made important contributions to dystonia would not have been possible if volunteers had not decided in advance to donate their brains to research.

"It's about giving something that only we can. That only people with our disease can," says Melody Probst, a registered brain donor. "Research brings all of us suffering from this terrible

About Brain Donation

- Brain donation does not alter the appearance of the donor or interfere with memorial practices.
- Donor information remains confidential. However donors must inform next of kin that they are registered to donate their brain.
- Individuals can withdraw from the program at any time.

disease a little closer to what we all hope for each day: a cure. By donating, I know I am doing all I can to help."

A first-of-its-kind study lead by Diego Iacono, MD, PhD of the Biomedical Research Institute of New Jersey performed sophisticated analysis on brain tissue samples from individuals with DYT1 dystonia, non-manifesting DYT1 carriers, and matched controls. Samples used in the study were obtained through the private collection of tissue made possible by brain donors registered through the Dystonia Brain Collective, a partnership among several dystonia patient groups to encourage

and facilitate brain donation. The findings elaborated on past research that detected specific differences in neurons from DYT1 dystonia brains. Although a larger scale study is needed to confirm the results from this study, the data supports the emerging concept that DYT1 dystonia is a neurodevelopmental disorder, meaning that there is an impairment in how the brain grows and develops. DYT1 dystonia is the most common inherited childhood dystonia.

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, which processes tissue requests from 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.

COPING

CONNECTIONS

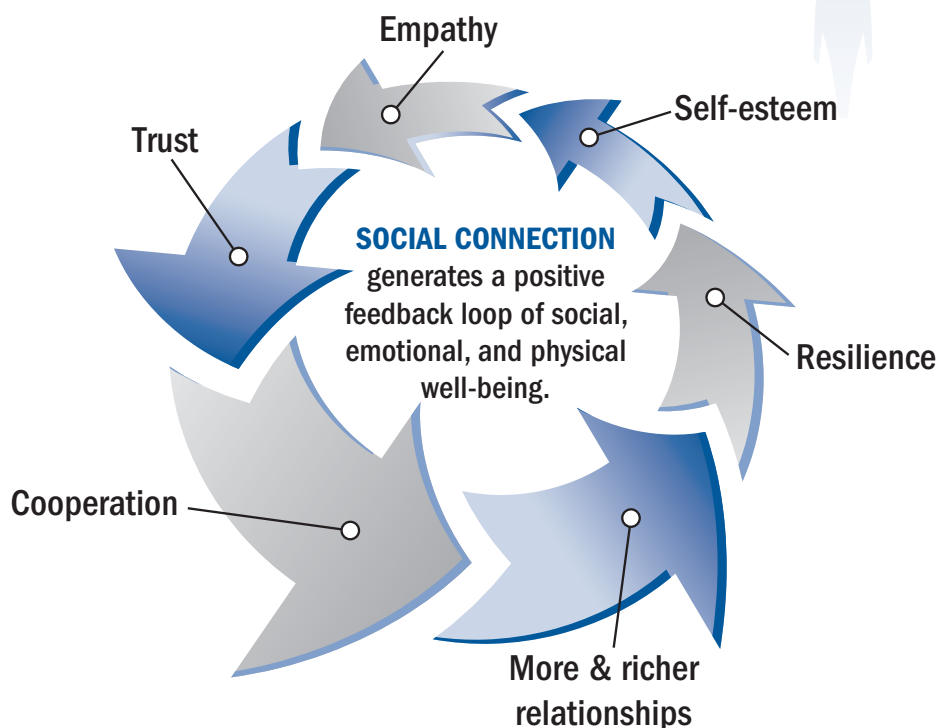
Social Support is Critical to Living Well with Dystonia



“Support groups provide a space for those of us with dystonia to share our stories and struggles in a safe and accepting environment. We feel less alone and more understood. We become better informed about our treatment choices and walk away with useful information to support us in our journey with dystonia.”

~ Alli Feeley, Founder & Leader of Greater Chicagoland Dystonia Support Group

- Feelings of loneliness are common among individuals with dystonia.
- Spouses, family members, and parents of affected children also feel isolated.
- The holiday season can be especially challenging.
- Feeling connected to other people can have a profound impact on your health, physically and emotionally.



Support groups also provide opportunities to volunteer for dystonia awareness, legislative advocacy, and fundraising.

Benefits of Peer Support

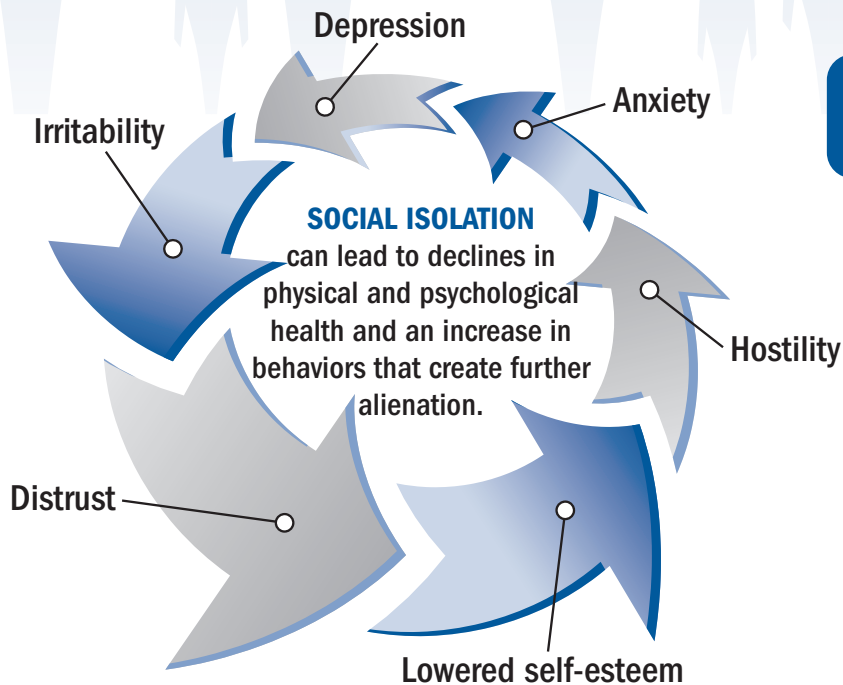
- + New friendships
- + Advice for daily living
- + Reduced stress
- + Greater acceptance of life with dystonia



DMRF connects you to people who understand life with dystonia.

WHY IS DYSTONIA ISOLATING?

- A dystonia diagnosis is often a shock, both to the individual and the family.
- Dystonia may create an unexpected, unwanted shift in family dynamics and responsibilities.
- Attending to health needs may be time-consuming and expensive.
- Individuals may feel a loss of control and loss of identity.
- Individuals may withdraw from social interactions due to pain, diminished self-worth, and logistical challenges.



SUPPORT RESOURCES ARE AVAILABLE TO HELP YOU FEEL CONNECTED:

✓ **SUPPORT GROUPS**

DMRF offers peer support groups in communities across the USA. Search support groups at dystonia-foundation.org/support_groups

✓ **ONLINE FORUMS**

Online groups are available for specific types of dystonias, parents, loved ones, young adults, and individuals who have had deep brain stimulation. Find links for online forums at dystonia-foundation.org/online

✓ **INDIVIDUAL SUPPORT**

DMRF is available by phone, email, web, and social media to those seeking information and support.

FEELING connected is what matters.
It's the **QUALITY** of relationships, not the **QUANTITY** that makes a difference.

**Can't find a dystonia support group in your community? Consider starting one.
Contact the DMRF at dystonia-foundation.org/contact or 800-377-3978.**

More than Meets the Eye

Origins of Dystonia May Lie in Special Group of Brain Cells

“I thought, let me take all my exposure and background in eye movements and control systems and put that into movement disorders and it will work like a charm—a goldmine to explore.”

Like many investigators working in dystonia, Aasef Shaikh, MD, PhD of University Hospitals, Cleveland VA Medical Center, and Case Western Reserve University is bringing a unique perspective to the study of movement disorders. He is an expert in the neurology of eye movement. He has earned several awards from DMRF including a Clinical Fellowship and a research grant. His current project funded by DMRF is exploring origins of dystonia in the brain and developing new treatment strategies.

From Eyes to Neck

One of the mysteries of dystonia is that multiple areas of the brain are involved: basal ganglia, cerebellum, midbrain, and others. For example, individuals who suffer a stroke in the basal ganglia develop dystonia symptoms. Meanwhile, a person with a brain disease affecting the cerebellum may develop dystonia, even when the basal ganglia are normal. Many individuals with dystonia have no detectable structural damage to their brains.

“We are looking at the same phenomenology—dystonia—and we are looking at different culprits, at different parts of the brain,” explains Dr. Shaikh. “We know different treatments, for example treating the neck with botulinum toxin or implanting a deep brain stimulator in the basal ganglia, help the disorder. So the fundamental question is how do we link everything together?”



“Dystonia is one of the network disorders, and I’m trying to understand how different nodes of the network talk to each other and how we can modulate the network in a counter-intuitive way. If something is broken, we don’t focus on what is broken. We make up for what is broken with something that is working.”

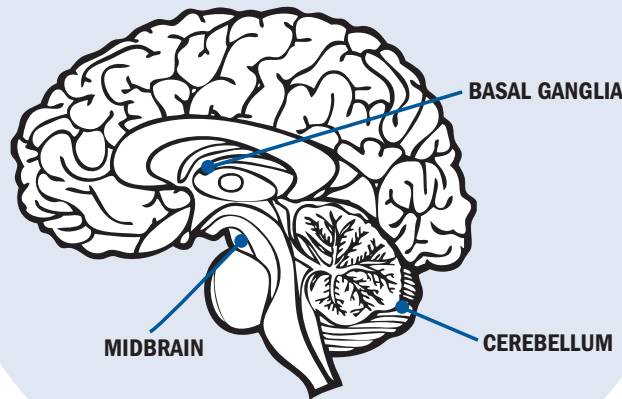
~ Aasef Shaikh, MD, PhD

While training with David Zee, MD and H. A. “Buz” Jinnah, MD, PhD at Johns Hopkins University School of Medicine, Dr. Shaikh began to look at cervical dystonia as a prototype for dystonia more generally: “We thought the way nature controls eye movement could probably be the same way nature controls the head or limbs. And the way eye movements go wrong could be the same way head movement or limb movement can go wrong, and that leads to different movement disorders.”

In order for the human eye to maintain a steady gaze, the brain must distill information from across the nervous system into clear instructions to the muscles. This is done through a neural integrator, a group of specialized brain cells.

Dr. Shaikh believes a neural integrator for head movement may be the common pathway where multiple contributing factors for cervical dystonia converge. The neural integrator translates information from the motor networks and the sensory system into a steady signal instructing the head muscles to stay in position. A problem in the neural integrator prevents the brain from maintaining control of the head, resulting in the jerky, shaky, involuntary movements characteristic of cervical dystonia.

THE HUMAN BRAIN



Exploring the Network

In this ambitious work, Dr. Shaikh is currently focusing on three specific elements that contribute to cervical dystonia: cerebellum, basal ganglia, and proprioception, which is the brain's ability to sense body position. "The neural integrator forms a network," he explains. "That neural integrator probably is linked to all these three areas and anything that goes wrong in any of these three areas can have ultimate repercussions that manifest in the form of a dystonia." His research suggests that cerebellum, basal ganglia, and proprioception are key sources of feedback to a neural integrator located in the midbrain.

The midbrain has been implicated in cervical dystonia for decades. In the 1970s, before the availability of botulinum neurotoxin therapy, surgeons treated cervical dystonia by creating lesions in the midbrain. The location of the lesions was guided by the latest brain recording technologies for the time. The results of the surgeries were unpredictable, and the procedures were eventually abandoned. However, those patients continue to assist dystonia investigators to this day. Dr. Shaikh explains: "I was fortunate to collaborate with Alexey Sedov, PhD who had access to magnetic tapes from these patients who had recordings from the midbrain. We took all those tapes out of storage, and we re-analyzed the recordings with modern technology. And that data

from their tapes, that was collected decades ago, showed there was a signature of a failure of neural integration—what we predicted from previous studies with Dr. Jinnah."

That's not all: "We also found that the impairment is not within this circuit, the impairment is somewhere in the feedback which goes into the circuit. So the part of the brain which does this function [the neural integrator] is completely normal, but it is getting abnormal signals from somewhere. That somewhere could be either cerebellum, basal ganglia, or proprioception. So that fits with our integrator model and that's what we are looking at now, how that network is connected."

Vulnerability as Opportunity

The neural integrator is susceptible to dysfunction from any point along the network. This vulnerability may, paradoxically, provide an opportunity for correction: If dysfunction in one part of this network can cause dystonia, intervening at different points in the network may compensate for the

dysfunction. "Of course I'm not going to answer this in a two year grant period," says Dr. Shaikh, "but what we want to show is that when we do intervention at one part of this network, we can see repercussions of that intervention in the other part of the network."

Dr. Shaikh was introduced to sophisticated single-cell brain recording techniques while training with DMRF Scientific Director Mahlon R. DeLong, MD at Emory University. He is using that expertise to understand how botulinum neurotoxin changes the basal ganglia-cerebellum-proprioception network: "Right now, as part of the funded DMRF grant, we are intervening at the neck and we are recording from basal ganglia, looking for the same type of signature that we found in the midbrain."

Botulinum neurotoxin, the primary medical treatment for cervical dystonia, is injected directly into dystonic muscles. Even though botulinum neurotoxin is injected far away from the brain, the effects of therapy influence how the brain manages head position. "We want to show that botulinum toxin, we believe, changes the muscle spindle in the neck," says Dr. Shaikh. Muscle spindles are receptors in muscle that detect changes in stretch and tension. Because the effects of botulinum neurotoxin injections normally take a week or more to show up, ironically, using

Continued on page 16

botulinum neurotoxin is not useful for these experiments. Instead, Dr. Shaikh is using muscle vibration to mimic certain qualities of injected botulinum neurotoxin. Vibration has been a research tool for decades, and the physiological effects in muscle are well understood.

Dr. Shaikh elaborates: “Vibration also activates muscle spindles, and the response is instantaneous, so what we find in the operating room is that when we are isolating a single cell and then we start vibrating the muscle we see the changes in the activity of the single cell and that’s what is telling us that by changing the muscle spindles we are changing the output of the basal ganglia.”

Toward New & Improved Treatment

The ultimate goal of Dr. Shaikh’s work is to improve treatment for dystonia, including the development of original new therapies. Here are just a few examples of the outcomes he is working toward.

☑ Improving Botulinum Neurotoxin Therapy

Understanding how the brain controls head position and how that impacts the dystonia circuit may help refine therapies such as botulinum neurotoxin injections. This understanding may ultimately help explain why botulinum neurotoxin works so well for some people and less well, or not at all, for others. Dr. Shaikh explains: “Getting to the bottom of how proprioception changes the whole circuit will give us a better insight about how we can direct the therapy, whether we want to target the muscle first or we want to target the proprioception.”

☑ Vibration Therapies

Many individuals with dystonia use vibrating massage devices to relieve muscle pain and tension. Vibration has long been used therapeutically. In an additional series of experiments, Dr. Shaikh is demonstrating that vibrating the neck in cervical dystonia patients diminishes neck tremors. This discovery is informing the design of wearable devices that deliver therapeutic vibration to the neck. “There are a few prostheses like that around,” Dr. Shaikh explains, “but they are developed without rigorous scientific basis. Hopefully our work will lead to scientific, evidence-based prostheses development which will instantaneously help some patients with neck tremors.”

☑ Smarter Deep Brain Stimulation

The next generation of implanted neuromodulation systems such as deep brain stimulation are “smart” devices that sense brain activity and adjust therapy to the patient’s needs in real time. Part of the challenge of developing these devices is knowing the specific brain activity signatures that indicate movement symptoms. Dr. Shaikh’s work intends to uncover a brain signature for dystonia: “We are measuring output of the basal ganglia. So, when there is more dystonia there is different output and when you vibrate the neck and reduce dystonia there is different output. So these differences will allow us to modulate DBS to get better outflow from basal ganglia.” (You can read more about advancements in DBS on page 6.)

☑ Alternate Targets for Therapy

Dr. Shaikh’s efforts to discover novel treatment approaches to reduce dystonia symptoms is especially

relevant to surgical procedures. “When somebody gets dystonia because of stroke in basal ganglia our hands are tied—we cannot go in there and stimulate basal ganglia with DBS because it’s damaged,” he explains. “What I am interested in doing is finding an alternate target. If I miss the boat at basal ganglia can I go into the thalamus and stimulate there, or can I stimulate the cerebellar outflow fibers and get meaningful improvement from that? Alternate targets is something which excites me the most and it is directly relevant to my research which is currently being funded by DMRF.” This work is also directly relevant to individuals who have undergone DBS but not experienced much benefit.

Patient Partnerships

Science depends on a constant flow of new ideas and innovative collaborations among experts from diverse disciplines. This is true at every level in the field of dystonia, from basic research to clinical trials. Progress also depends heavily on individuals with dystonia and their families. Dr. Shaikh’s work would not be possible without the volunteers who generously make themselves available to participate in research. “We are learning from the patients,” he explains. “The OR is my lab. The clinic is my lab. And whatever we are learning from patients, we are directly implementing to help them.”

To learn about opportunities to participate in dystonia research, see pages 10–11.

People on the Move

The DMRF is grateful for the grassroots volunteers across the country working to improve dystonia awareness in their communities. Every volunteer makes a difference!



Martha Murphy, Mary Friedel, and Susi Pensel of the Dystonia Support & Advocacy Group of San Diego County were presented with a Dystonia Awareness Month proclamation from the San Diego City Council, represented by City Councilmember **Georgette Gomez**.

June is Dystonia Awareness Month in New Jersey thanks to the teamwork of dedicated volunteers. **Marguerite Weiss** obtained Dystonia Awareness Proclamations from Brick Township and Ocean County. DMRF Support Leaders **Janice and Len Nachbar** received proclamations from Governor Phil Murphy, Monmouth County, and Freehold Township plus a joint Resolution from the New Jersey Senate and Legislature.

Many thanks to **Linda Davis** and the Medford, New York Chamber of Commerce for organizing a Dystonia Awareness Night and concert on August 29. Special thanks to **ErinKelly Connell** for her efforts toward the event. Guests included Assemblyman **Dean Murray** who presented Linda with a Dystonia Awareness Proclamation.

Thanks to **Mike Delise**, Governor **Rick Snyder** of Michigan issued a proclamation once again recognizing Dystonia Awareness Month in September.



In October, **Allison Botti** ran the Steamtown Marathon in north-eastern Pennsylvania to raise dystonia awareness and funds for research.



Thanks to **Carrie Speckman**, Governor **John Kasich** and Lieutenant Governor **Mary Taylor** of Ohio issued a proclamation recognizing Dystonia Awareness Month in September. Carrie also made a number of social media videos to promote awareness.

Wanda McCune secured a Dystonia Awareness Proclamation from West Virginia Governor **Jim Justice**. Wanda's young son **Dylan** is diagnosed with generalized dystonia.



Asheville, North Carolina Mayor **Esther Manhaimer** presented DMRF Board Member **Marilynne Herbert** with a proclamation recognizing September as Dystonia Awareness Month.

Sarah Pirio Richardson, MD, from University of New Mexico, spoke at a Dystonia Educational Meeting in Albuquerque on September 9. This was the first of a series of meetings scheduled throughout the fall in cities across the country, supported by an educational grant from Allergan.



Robbie Nabors and members of the Mobile, Alabama Dystonia Support Group met with Mayor **Sandy Stimpson**, and he presented the group with a Dystonia Awareness Proclamation.

Finding a Path Forward

Parents Navigate the Twists & Turns of Treating Childhood Dystonia

Parents of children with dystonia face weighty decisions about the care and treatment of their kids. Although there are general treatment guidelines for childhood dystonia, each child is unique and therapy must be customized to their needs and the needs of the family.

Peer support among parents is often critical to nurturing healthy, resilient families. DMRF provides opportunities for parents and families to connect with others in the dystonia community.



The Sorleys

“That year from the onset of symptoms to getting the genetic test results back is a blur,” recalls dad Chris Sorley. “We were like a cat with a shiny object, every time a possible diagnosis popped up we would turn in that direction and say, well maybe this is it.”

Two years ago, Chris and Tara Sorley’s now five year old son Braden was diagnosed with myoclonus-dystonia, a movement disorder characterized by rapid, jerking muscle contractions (myoclonus) and/or twisting, repetitive

movements and awkward postures (dystonia). A percentage of cases are caused by changes in the DYT11/SGCE gene, which can be confirmed through testing.

The relief of a diagnosis was quickly replaced by the challenge of treatment. Although research in myoclonus-dystonia is ongoing, the disorder is notoriously difficult to treat. Response to treatment can vary from patient to patient, so therapy is largely a process of trial and error.

“Just because you found the doctor who is willing to help and willing to listen, that doesn’t mean they have a magic solution,” says Chris.

“I lived on Google,” says Tara. She scoured the Internet for studies and treatments to discuss with Braden’s doctors. “Chris would say to me, you need to go to sleep, and I’d say I cannot sleep!” she says with a laugh. Tara’s online searches have been the driving force behind finding the medications that have helped Braden so far. However, despite consulting numerous experts, they have yet to find a truly effective treatment strategy. “It’s very hard to decide what to do for a little person who can’t vocalize for themselves,” says Tara. “You question everything. You question

yourself. You always want what’s best for your child.”

• • •

Oral medications have been the mainstay of Braden’s treatment. Many of the medications used to treat movement disorders can cause side effects such as sedation, mood changes, and even increased movement symptoms. In a child as young as Braden, whose brain and body are rapidly developing, it can be difficult even for his parents to sort out what signs or behavior are normal five-year-old conduct, and what might indicate a medication side effect. It can be difficult to decipher whether a medication is helping at all. Several drugs initially reduced Braden’s symptoms but were discontinued after losing efficacy over time.

“I wanted to be able to say ok we found out what’s wrong, let’s fix it now,” says Chris. “But it doesn’t work that way. You often feel like there is no finish line, no end in sight. And that becomes very trying and frustrating.”

The dystonia affects Braden’s right leg, making it difficult for him to walk without assistance. The myoclonus causes jerking movements in his limbs. He is maxing out on his medication dosage and the myoclonus symptoms are increasing. There are times he cannot physically keep up with his peers.

"He is hyper aware of his body. He will outright say, 'I can't run because I have dystonia,'" explains Tara. "He will remove himself, for example, at recess, he will sit and play with chalk while the other kids are running around playing tag. He's very adaptive."

Braden has grown up with his symptoms. He doesn't know any differently. He climbs, he swims, he loves riding his bike. He has neighborhood friends. He started all-day kindergarten this fall with a full-time aide.

"There are times I truly think in his little mind he doesn't want us to worry about him," says Tara. "I'll be holding his hand and I'll feel him struggling, and I'll say, buddy, did you get a jerk? And he'll say yeah. I'll ask if he's ok, and he'll say yeah. He just tries to act like everything is ok."

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The Sorleys credit their support system of family, friends, and neighbors for helping them cope with the challenges and pressures of life with dystonia. "Connecting with other people via the Western PA Dystonia Support Group and finding out about the Zoo Walk, that was huge because it's something to focus on," says Chris. Tara adds: "Something we are passionate about."

The Pittsburgh Dystonia Zoo Walk is an annual event spearheaded by local volunteers. It has become an activity the whole Sorley family participates in. Chris co-hosts the "The Fan Morning Show" on 93.7 Pittsburgh Sports Radio and has elevated visibility of dystonia throughout the metro area. Tara assembled Braden's Brigade, the Zoo Walk's highest earning team this year. Promoting dystonia awareness



has also become important for the Sorleys' nine-year-old daughter Ava. Tara explains, "With the Zoo Walk, her friends and their parents come, and that excites her because it's a fun event but it's also for our family, and she's proud of that. They did a dystonia awareness day at her school and it made her feel really special because she got to be on the morning announcements and talk about dystonia."

Just as there is no magic pill to relieve Braden's symptoms, there is no set road map for families impacted by dystonia to follow. "We are by no means any closer to him being better, we're just at a totally different place," says Tara. "You can't give up."

The Sorleys take one day at a time, each challenge as it comes. They are determined to make life better for Braden and to make a difference for the greater dystonia community, while growing stronger as a family. "It's important for parents to not lose sight of each other and the rest of your family, which can be hard because you're so focused on the dystonia and finding answers. You have to have some balance in your life," says Chris.

"You have to be ok for your kids to be ok," adds Tara. "You have to stay strong."

The Rudolphs

David Rudolph was eight years old when he was diagnosed with DYT1 dystonia in 2003. The symptoms began with an odd posture in his left foot. David's parents, Diane and Mark Rudolph, quickly connected with the DMRF, learning as much as they could about dystonia and treatment options. David was prescribed a combination of oral medications clinicians often recommend for childhood dystonia.

"I was also doing meditation with him, trying foot reflexology—anything we could think of," says Mark. Diane was taking David to physical therapy three times a week, with five-year-old daughter Rachel in tow.

David's feet began to turn inward so severely he walked on the outside edges of his soles. His toes curled. It was difficult to write. His medications had to be carefully timed to avoid making him drowsy in school. "We didn't know how well the meds were working or if he stopped the meds whether he would get worse a lot faster, and there was no way we were going to experiment with that," says Mark. By the start of sixth grade, David was using a wheelchair part-time because walking was so difficult.

David's favorite activities were becoming too physically demanding to be fun. Although he had to quit Cub Scouts and Little League Baseball, the community welcomed him to participate in numerous recreational programs at whatever level he was

Continued on page 20

able. He was invited to shoot hoops during school basketball games even though he was unable to run the court. A martial arts instructor invited him to try karate from his wheelchair.

One day, Diane noticed her daughter Rachel walking with an odd gait. “I thought, oh no, why is she walking like that, because I knew she did not have the DYT1 dystonia gene,” says Diane. She asked Rachel if she injured her foot. “Rachel said, ‘I don’t want David to feel badly about how he walks so I’m walking like he does.’ It’s very difficult for siblings, but I think dialogue is probably what saved us through a lot of those difficult times.”

• • •

By 2004, Diane and Mark had joined the DMRF Board of Directors. David was nine years old. The Rudolchs had learned about deep brain stimulation (DBS) as a therapy for dystonia and met numerous people who underwent the procedure. DBS involves implanting electrodes in the brain and a battery-powered stimulator in the chest wall. In 2006, the Rudolchs inquired whether this treatment could help David. Although multiple experts concluded David was a candidate for DBS, they first recommended increasing the doses of his medications. “That got him out of a wheelchair,” says Mark. The physical improvements were so profound he was able to play on the school basketball team with the help of braces on his legs.

“My parents involved me in my treatment as much as was appropriate for my age,” recalls David. “It wasn’t just them talking to the doctors and me sitting there listening. I felt comfortable with the treatment I was getting because I was involved in the

decision-making to an extent.” High school was challenging, but manageable. There were times the higher doses of medication made it difficult for David to keep up with his schoolwork due to side effects like sedation and memory difficulties. He had a number of orthopedic surgeries on his feet to protect his joints from the constant dystonic muscle contractions. “David wasn’t that cut and dry of a case where nothing else is working so we have to try DBS,” explains Mark. DBS was emerging as a very promising treatment for DYT1 dystonia, but physicians could not predict the degree of benefit David could expect.

As David shifted into adulthood, so did the dynamics surrounding decisions about his care. “Once he turned 18, DBS was really going to be his choice,” says Mark. “We were not going to push him into anything.”

By his sophomore year of college, David reached a tipping point. The brain fog from the medications had become unbearable. “Physically I was doing ok,” David explains, “but it’s very hard not being able to remember 24 hours ago. I couldn’t remember what I learned in class the day before.” He chose to have DBS after he graduated from college.

Today, David is 23 years old. He had DBS about a year ago. Physically and cognitively, he has exceeded all expectations. “Every month got a little better,” recalls Diane, “there was more light in his eyes. His humor seemed to come back.”

“Within about three months of the surgery I was totally off meds,” says David. “The distance I am able to walk has increased astronomically. I can

walk at least a couple miles, and before it was hard to walk a block.”

David recently began post-graduate studies in Israel, half a world away from the family’s home in Los Angeles.

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The Rudolchs could not be happier with how David is doing today. “I’m not sure I want to call it a regret,” says David, “but considering DBS earlier, at least from my perspective, would have been good for me. I wish I had done it, not necessarily in sixth grade when we first started discussing it, but definitely maybe at the end of high school.”

Diane agrees: “If there was a chance to do some things differently probably we would, but we are where we are. I think David is pretty happy and thankful that he really drove the decision. It’s been life-changing for our whole family.”

Diane and Mark continue to serve on the DMRF Board of Directors. They fundraise to benefit DMRF and support research grants in David’s name. They are a point of contact for families considering DBS.

“Any person or family has to make their own decisions as to what is best for their family,” says Mark, “but talking to people and knowing what options are out there is really helpful.”

“The DMRF has been a wonderful community of parents who have walked the path,” says Diane. “We followed those who went before us, and now there are people following us. We pay it forward through the DMRF.”

For more information about childhood dystonia, and DMRF resources for parents, see page 21.

Dystonia in childhood is a movement disorder that causes uncontrollable muscle contractions. These muscle contractions result in twisting, repetitive movements and abnormal postures. The movements and postures may be chronic or occur in episodes. Symptoms can vary with body position, specific tasks, emotions, and state of consciousness.

Diagnosing dystonia in children is complex and requires careful evaluation by experts in pediatric movement disorders. Childhood dystonia often occurs as a symptom of an underlying brain disorder.

Dystonia can be the only movement symptom a child has or occur along with other movement symptoms—for example, myoclonus (jerking movements).

Dystonia in children is diagnosed primarily by expert observation of the physical symptoms. The diagnostic work up may include blood tests, genetic testing, testing for metabolic disorders (lumbar puncture), magnetic resonance imaging (MRI), and electroencephalography (EEG).

Cerebral palsy is among the most common causes of dystonia in children. Additional causes can include hypoxic brain injury, infections, autoimmune disorders, metabolic disorders, stroke, toxins, and certain medications.

There are a number of inherited dystonias that affect children, many of which are attributed to a single gene variant.

Treatment may include physical therapy, occupational therapy, adaptive equipment and devices, speech therapy, behavioral strategies such as relaxation

Signs of Dystonia

- A body part is flexed or twisted into an abnormal position.
- Repetitive and patterned body movements, which may resemble tremor.
- Movement symptoms worsen with voluntary action. This can cause the incorrect perception that the child is ‘faking’ the symptoms.
- Symptoms may be present while walking but not running or swimming.
- Symptoms diminish or disappear during sleep.
- Attempting a movement task on one side of the body may activate dystonia symptoms on the opposite side.
- Dystonic movements and postures may be temporarily relieved by a gentle touch or specific action called a sensory trick.

techniques, oral medications, botulinum neurotoxin injections, and/or deep brain stimulation surgery.

Treatment for dystonia in children tends to focus on reducing the movement

symptoms, but there are non-movement aspects of dystonia can have a significant impact on overall quality of life and should be considered as part of a comprehensive treatment plan. These may include pain management, difficulties with daily tasks, trouble using the hands, difficulty with seating, mobility challenges, sleep difficulties, and sometimes communication issues. Children with dystonia, especially inherited dystonias, may have higher rates of anxiety and depression than children overall.

Resources for Parents

- **Support4Parents of Children with Dystonia**
A closed peer support group on Facebook for parents.
facebook.com/groups/support4parents.dmrp/
- **Dystonia in Children & Adolescents**
A webinar recording with pediatric neurologist and movement disorder expert Jonathan Mink, MD, PhD.
dystonia-foundation.org/families
- **Never Look Down by Zachary Weinstein & Alyssa Dver**
An illustrated book for children with dystonia.
dystonia-foundation.org/shop

PERSONAL PROFILE

Mike Cahall

Mike Cahall of Kentwood, Michigan is a former Communications Supervisor in the U.S. Army, Minister, and Educator. He developed generalized dystonia following a head injury sustained while serving in the military.

How did your symptoms start?

It took over 20 years before I was properly diagnosed. When my disease first began to manifest, the changes and challenges it caused me physically, mentally, and emotionally were hard for others to notice. I began to notice some very strange things that would affect me off and on. For example, back in the 1980s, I was still a smoker and sometimes I couldn't "flick my Bic!" I couldn't spit without it running down my face. In class, when I was taking notes, my fingers cramped up. Whenever I found myself under a tremendous amount of physical or emotional stress, my eyelids would close involuntarily. Well, when you go to a doctor and tell them you're concerned because you can't light your lighter or that you can't spit, you end up getting a lot of strange faces looking back at you! I was told there was nothing wrong with me physically. They told me that my problems were much more likely to be psychological and I needed to see a psychiatrist.

When I was living in Brighton, Michigan my neurologist told me that I had a terminal disease. And that I only had a limited number of years left to live and that the last years would be too difficult for anybody but professional health-care personnel to take care of me. So I left the only woman I ever loved and moved into the Home for Veterans in Grand Rapids. There I was referred to a pain specialist. He and I had an immediate rapport! My trust in him was "marrow deep!" (Abraham Lincoln's words about General Ulysses S. Grant.) I had finally found a doctor who was willing to listen to me. And who believed me. With his help and guidance, my pain



Mike Cahall attended his first Dystonia Advocacy Day on Capitol Hill. He is pictured (R) with US Senator Gary Peters of Michigan.

level became acceptable. I'm still his patient. After working together for over 14 years, I know that having him in my life has been one of the greatest blessings I've ever received!

How would you describe how dystonia affects you on a daily basis?

I'm a person whose entire body is affected. My legs and feet were the last parts of my body to develop contractions and pain. My feet can be difficult to handle. There's two kinds of feelings. The first isn't really painful but it's really annoying. It feels like there are rubber bands attached to each toe and my toes are being

pulled under. It takes a lot of effort to bring the toes into a position that I can tolerate. The second problem is I'm unable to stand up straight. The dystonic contractions have caused severe scoliosis and that has caused problems with pain running down my legs and into my feet. I usually avoid situations where other people can see the difficulty I experience when I eat. I have trouble speaking clearly enough so that other people can understand what I am trying to say. It's kind of difficult, but I refuse to allow this disease to keep me from doing and experiencing everything that life has to offer.

How would you describe the changes deep brain stimulation (DBS) made for you?

I was so lucky because I was chosen to be part of a clinical study that was investigating the effects of deep brain stimulation on people who had severe cases of generalized dystonia. The study was underwritten by the Department of Defense [DOD]. The doctors were very honest with me

What is Dystonia Advocacy Day?



**Dystonia
Advocacy Network**

Each spring, dystonia advocates gather in Washington, DC for a two-day advocacy event. Participants receive comprehensive advocacy training and the opportunity to network with others impacted by dystonia. Dystonia Advocacy Day culminates with a trip to Capitol Hill for meetings in the offices of Members of Congress and urging their support on key legislative issues.

The event is organized by the Dystonia Advocacy Network (DAN), a grassroots organization that brings dystonia-affected individuals together to speak out with a single, powerful voice on legislative and public policy issues which impact the dystonia community. To learn more, visit: dystonia-advocacy.org

because they told me they would consider a 25-30% improvement a successful outcome. I told them that any improvement would be a great outcome, as far as I was concerned! The results were far beyond anybody's expectations. Thank God for the DOD sponsorship of the clinical study that gave me my life back.

What was your experience attending Dystonia Advocacy Day in Washington, DC?

In many ways my dystonia has helped me to become a better person. One of the things that changed was the way I channeled my anger. Before my condition, I allowed my anger to control my actions. I owe so much to my therapist who, during the seven years that we spent together, helped me understand that when I felt angry I needed to channel it into positive actions that might help bring about the desired outcome. That simple lesson has been the driving force that has empowered me to never give up. Even when everything in my life might appear hopeless, I never let go of my hope!

Being a part of the Dystonia Advocacy Network helps me turn my anger into positive action. I came to understand that my anger was the result of feeling powerless. Being with such a dynamic group of people showed me that there's strength in numbers. I also learned that I had the power to influence our lawmakers. The time I spent with the Dystonia Advocacy Network was a tremendous turning point for me.

Being with other people who had been through similar experiences made me feel like I wasn't alone anymore. I found this exhilarating! I think it was the greatest gift I've ever been given.

The other thing I realized much more fully was, knowledge is power! I had information that turned out to be beneficial for our cause. The fact that I was a beneficiary of a clinical study that was funded by the DOD made an impression on the legislators and their aides. I was also able to soak up so much information. When I finally left Washington DC, I left feeling empowered by all the knowledge that I'd been able to take with me.

What advice do you have for others living with dystonia? What keeps you going?

There were many times over the years when the only thing that enabled me to make it through different experiences was hope. Hope is that "something" that enables us to keep going, even when it appears as if we're standing in front of an immovable object. Maybe, by sharing my experiences, I'll be able to lead someone else to that place within where hope always abides.

Dystonia Dialogue

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