



DYSTONIA  
MEDICAL  
RESEARCH  
FOUNDATION

# Dystonia Dialogue

NEWSLETTER OF THE DYSTONIA MEDICAL RESEARCH FOUNDATION

Summer 2022 | Volume 45 • No.2



**A MOMENT in science can launch a LIFETIME of DISCOVERIES**

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Dystonia Journal  
Publishes Inaugural Issue

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Paralympian Sydney  
Peterson Triple-Medals  
in Beijing

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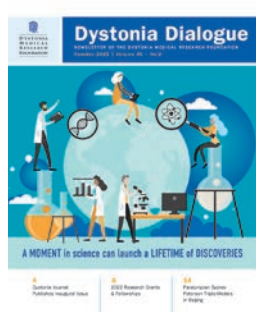
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Dystonia investigators around the world are expanding the boundaries of what is known about dystonia and implications for novel treatments. See page 6 to read about the latest research projects supported by DMRF.

*The Dystonia Dialogue is supported by Ipsen Biopharmaceuticals.*

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, 501(c)(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.

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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### Dystonia Medical Research Foundation

One East Wacker Drive  
Suite 1730  
Chicago, Illinois 60601-1980  
**Phone** 312 755 0198  
800 377 DYST (3978)  
**Web** [dystonia-foundation.org](http://dystonia-foundation.org)

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# Foundation Update



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Certain moments in dystonia research are flashpoints for accelerating progress. They illuminate new research directions and attract additional experts to the field. A moment in science can launch a lifetime of discoveries and generations of progress.

One such flashpoint was the discovery of the TOR1A/DYT1 gene by DMRF-funded investigators in 1997. 2022 marks the 25th anniversary of the announcement from Xandra Breakefield, PhD, Laurie Ozelius, PhD, and their collaborators that they identified a gene variant for early-onset torsion dystonia, a debilitating childhood form of dystonia.

Identifying the TOR1A/DYT1 gene was not an overnight accomplishment. Investigators worked for years to locate affected families, collect blood and biosamples, and use the limited technology available to hunt the gene. DMRF leaders at the time knew identifying the gene could crack open new possibilities in diagnosis and treatment and supported the cutting-edge equipment the investigators needed.

The gene identification resulted in a target for new dystonia therapies, testing to aid diagnosis and identify gene carriers, more predictable success from deep brain stimulation, reproductive medicine options to protect future generations of affected families from developing TOR1A/DYT1 dystonia, new investigators joining the dystonia field, and an explosion of data about the biological mechanisms underlying all types of dystonia.

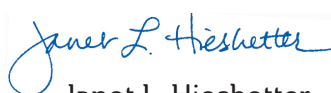
Several DMRF Board Members who supported the gene identification remain on the Board today and are now witnessing their grandchildren growing up without the risk of inheriting dystonia, thanks to that hard-earned moment of discovery.

Drs. Breakefield and Ozelius were incredibly dedicated in their pursuit of TOR1A/DYT1 and have made additional momentous contributions to dystonia research. DMRF is proud to currently support Dr. Breakefield's work to develop gene therapy for dystonia (see page 7). Both investigators as well as Susan Bressman, MD, another key contributor to the gene discovery, will be honored with a Changing Lives Through Science Award at upcoming Dystonia Zoo Days (see page 12 for more).

The DMRF invests in research because we know it has the power to transform lives. We will not stop looking for answers until no individual or family is ever again burdened by dystonia. Thank you for your support of DMRF.



Mark Rudolph  
President



Janet L. Hieshetter  
Executive Director

***DYSTONIA ZOO DAYS ARE BACK! See page 12 for information on in-person AND virtual events.***

## In Memoriam: Joel Farber



Joel Farber and daughter Beth Farber at the Chicago Basket Bash.

The DMRF regrets to share that extraordinary dystonia volunteer Joel Farber died on May 31, 2022. Joel had endless enthusiasm for connecting individuals and families affected by dystonia and educating federal policy makers about the urgent need for dystonia research. He hardly met a stranger without educating them about dystonia and frequently used public platforms and social connections to promote dystonia awareness.

“Joel’s passing leaves a hole in the dystonia community,” said DMRF Executive Director Janet Hieshetter. “The DMRF benefitted greatly from his tireless enthusiasm for the mission to find a cure and his genuine affection for individuals with dystonia and their families. Joel touched the lives of so many, and he will be very much missed.”

Joel’s late daughter Shari Farber-Tritt developed generalized dystonia as a young child, which inspired him and his late wife Harriett to work tirelessly on behalf of the dystonia community. Shari and the Farber family were featured in the documentary, *Twisted*. The DMRF extends our deepest condolences to Joel’s surviving loved ones including his daughter Community Leadership Chair Beth Farber and her husband Steve Laser.

## Dystonia Highlights Research Advancements



The inaugural issue of *Dystonia*, the DMRF’s official scientific journal, was published in April 2022. Two additional special issues are planned, titled *Motor Circuits and Motor*

*Symptoms in Dystonia: Translational Approaches from Animal Models to Patients and Models, Mechanisms, and Maturation in Developmental Dystonia*. Each are accepting submissions until September 15, 2022.

*Dystonia* aims to be the leading journal in the field by publishing open access premier research on all basic, clinical, and translational aspects of the dystonias. Articles published to-date include a feasibility study for providing yoga instruction via video conference to individuals with cervical dystonia (focal dystonia of the neck), a deep dive into the clinical characteristics of blepharospasm (focal dystonia of eyelid muscles), examination of tremor in focal hand dystonia, and more.

*Dystonia* provides online, free gold open access to the journal and all of its research publications to the general public. Access the journal at: [frontierspartnerships.org/journals/dystonia](https://frontierspartnerships.org/journals/dystonia)

*Dystonia* is partially supported by the Joan Miller Young Investigator Fund and Tuft Family Foundation.

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# Pamela Sloate Bronx Dystonia Zoo Day Named for Late Dystonia Advocate

DMRF Board Member Pamela Sloate died from cancer on April 12, 2022. In her honor, the newly named Pamela Sloate Bronx Dystonia Zoo Day to benefit DMRF will take place Sunday, September 10, 2022 at The Bronx Zoo. The event is in its sixth year, after a two-year hiatus due to the Covid-19 pandemic, with Pam previously serving as Chair of the planning committee. Clinician-scientist Susan Bressman, MD of Mount Sinai will be presented with the DMRF's Changing Lives through Science Award for her contributions to the discovery of the DYT1/TOR1A gene responsible for early onset dystonia 25 years ago.

Mark Rudolph, DMRF President, said, "Pam was an inspiration to me. The Foundation benefited greatly from her example and many accomplishments. Under her leadership, the Bronx Dystonia Zoo Day became one of DMRF's most successful events."

Pam began developing dystonia at age eight, and the condition progressed into adulthood. She was a passionate legislative advocate on behalf of the dystonia community, leader of a dystonia support group, and author of the blog *Chronicles of a Dystonia Muse*.



Pam Sloate organized the 2016 Bronx Dystonia Zoo Day in between deep brain stimulation surgeries.

Pam's loved ones are forming team "Dystonia Muse" in her memory. All team contributions will be matched by an anonymous donor.

For more information about Dystonia Zoo Days, see page 12.

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## Dystonia Strong Celebrates Community



The new Dystonia Strong campaign highlights the strength and resilience of individuals with dystonia and their families while empowering the community to promote awareness. September may be Dystonia Awareness Month, but increasing awareness is essential all year long.

DMRF support groups across the country are hosting Dystonia Strong-themed events to help the community re-connect after the intensified social isolation of the Covid-19 pandemic. Dystonia support groups are invaluable sources of information, understanding, validation, and friendship. Visit the DMRF website to connect with your nearest DMRF support group.

DMRF has also launched a line of Dystonia Strong awareness merchandise including a baseball style shirt, yard, flag, notecards, magnets, and more. For more information, visit: [dystonia-foundation.org/strong](https://dystonia-foundation.org/strong)



## DMRF Research Awards Advance Deeper Understanding of Dystonia & Novel Treatment Approaches

*The ultimate goal of the DMRF's science program is to support the development of improved dystonia therapies and a cure. The DMRF is dedicated to stimulating the field of dystonia research and supporting the collaborations and projects necessary to accelerate progress.*

Currently funded projects are furthering our fundamental understanding of the underlying biological causes of dystonia, exploring novel new therapeutic approaches including gene therapy and “smarter” deep brain stimulation, understanding how dystonia-causing gene variants ultimately result in symptoms, and uncovering targets for new and improved therapeutics.

“The pace of dystonia research has been accelerating faster and faster, and DMRF is proud of its contributions to keeping this momentum going,” said DMRF President Mark Rudolph. “It’s always gratifying to see a new cycle of research projects announced and to anticipate the important new discoveries we can expect from these outstanding investigators.” Congratulations to the newest award recipients, and infinite thanks to DMRF supporters for making this research funding possible.

## GRANTS

*Research grants are available in support of hypothesis-driven research at the genetic, molecular, cellular, systems, or behavioral levels that may lead to a better understanding of the pathophysiology or to new therapies for any or all forms of dystonia.*

## New in 2022

### GABA Abnormalities and Stability in Cervical Dystonia

*Brian Berman, MD, Virginia Commonwealth University*

**Quick Summary:** The goal of this study is to understand the underlying brain changes in cervical dystonia and the role the neurotransmitter GABA plays in the disorder.

**More Detail:** One of the neurological mechanisms that contributes to dystonia is altered inhibition, which means the brain is unable to adequately suppress certain functions that need to be dialed back for the body to move normally. This proposal will lead to an increased understanding of the role that altered inhibition plays in cervical dystonia. This study will further help establish whether GABA levels in the brain change when dystonia is treated with botulinum neurotoxin injections and whether measurement of GABA levels is stable over time. If positive, the findings could lead to a reliable imaging test to aid in the diagnosis of cervical dystonia and/or to an objective way to track responses to therapies and thereby provide a very useful tool for clinical trials and treatment development. Findings from this study could further provide the preliminary data needed to apply for a larger federal grant to investigate the role abnormal inhibition plays in other types of dystonia as well as in the progression and spread of dystonia in affected individuals.

### Protein Kinase R Dysfunction in Dystonia

*Stephanie Moon, PhD, University of Michigan, Ann Arbor*

**Quick Summary:** The goal of this research project is to understand how changes in Protein Kinase R cause dystonia and possibly identify new drug targets.

**More Detail:** This project aims to understand why and how changes in a protein linked to dystonia affect neurons, both in normal and stress conditions. Researchers have found that mutations in the DNA that codes for Protein Kinase R causes an early-onset dystonia in children. Some children with this form of dystonia had worse symptoms after viral infection or surgery, conditions that can stress the body. These obser-

ventions suggest that small changes in Protein Kinase R could interfere with the cells' ability to react to stresses and perhaps contribute to dystonia. Dr. Moon's lab is interested in how Protein Kinase R helps neurons grow and respond to stresses in order to allow the brain to maintain function and control body movement. Understanding why changes in Protein Kinase R affect neurons will hopefully provide new targets to develop novel drugs and additional treatments.

### Molecular Mechanisms of Dysmyelination in Dystonia

*Dhananjay Yellajoshyula, PhD, Case Western University*

**Quick Summary:** White matter abnormalities in the brain contribute to dystonia, and this investigation will explore how a specific gene variant may lead to such abnormalities.

**More Detail:** Recent studies demonstrate white matter abnormalities from human individuals and mouse models with THAP1/DYT6 early-onset dystonia as well as additional forms of the disorder. White matter is a large intertwining system of neural connections across the brain. Additionally, several gene variants implicated in dystonia including THAP1 and YY1 have an established function in generating white matter during brain development. These findings suggest that white matter abnormalities contribute to dystonia pathology. In this proposal, researchers will investigate the cellular mechanism by which the THAP1 dystonia mutation results in white matter abnormalities. They hypothesize that the THAP1 mutation disrupts cellular pathways commonly regulated by the two dystonia genes THAP1 and YY1 in the glial cells necessary for generating the white matter. They will also identify proteins that bind with THAP1 and define how these interactions are disrupted by the THAP1 dystonia mutation.

## 2nd Year Funding

### Normalizing DYT1 Cholinergic Neurons by CRISPR

#### Disruption of Mutant TOR1A Allele- 2nd Year

*Xandra Breakefield, PhD, Massachusetts General Hospital*

**Quick Summary:** This project seeks to investigate the development of gene therapy for dystonia.

**More Detail:** This proposal represents a step in evaluating whether gene therapy could be effective in TOR1A/DYT1 dystonia. Dr. Breakefield's lab has shown that selective disruption of the mutant TOR1A gene can normalize biologic

*Continued on page 8*



cell functions in patient skin cells. Since dystonia is a neurological disease, the next step is to evaluate whether this approach can normalize function in TOR1A neurons (brain cells). Through the work of D. Christopher Bragg, PhD and Nutan Sharma, MD, PhD, the investigators have access to stem cells from TOR1A/DYT1 patients, which can be turned into neurons. If successful in rescuing neurons, the lab will work with David Standaert, MD, PhD to translate the technology into a mouse model which would provide some of the data needed for the Food & Drug Administration to allow a clinical trial. Ultimately, Dr. Breakefield envisions a clinical trial in which children carrying the mutant TOR1A/DYT1 gene and manifesting symptoms at an early age are administered gene therapy in a single dose. This could be done at the same time as deep brain stimulation (DBS), with the intent to eventually turn off the DBS device to assess if it remains needed. The ultimate goal of this effort is the development of better therapies for TOR1A/DYT1 dystonia.

#### **A Next Generation Sensing Neural Interface Study for Adaptive DBS in Dystonia**

*Simon Little, MBBS, MRCP, PhD*

*University of California, San Francisco*

**Quick Summary:** Investigators are examining unique brain activity patterns found in dystonia patients treated with deep brain stimulation.

**More Detail:** Deep brain stimulation (DBS) has revealed a pattern of brain activity in individuals with dystonia that is linked to muscle activity. However, investigators do not yet know the significance of this signal, for example, whether it causes muscles to contract or is simply a marker that they have done so. To answer these questions, Dr. Little and team are using new sensing-enabled DBS devices that can record brain signals as well as provide stimulation therapy. They have implanted the device in a small group of patients and found that the dystonia signals are present in all recorded so far. They are also investigating how this signal relates to muscle activity and sensory processing. They will test this new adaptive stimulation to see if it is more effective and causes fewer side effects than standard continuous DBS. This study will further understanding of how brain signaling goes wrong in dystonia, knowledge that could potentially lead to the design of improved therapies.

#### **Role of Cerebellar Network Excitability and Plasticity in the Pathophysiology of Dystonia**

*Antonio Pisani, MD, University of Pavia*

**Quick Summary:** This project seeks to advance understanding of the underlying neurology of dystonia by focusing on two specific types of inherited dystonia.

**More Detail:** Dr. Pisani and team are studying brain circuits in two types of genetic dystonia: DYT1 and DYT25. They are testing the idea that loss of these genes leads to changes in brain plasticity, which is how the brain learns motor tasks. They will study two animal models, one with the TOR1A/ DYT1 gene mutation and the other with loss of GNAO1/DYT25. They hope to learn about the effects of the loss of these genes on brain plasticity. One of the features of abnormal movements in dystonia is that once the symptoms develop they may become permanent. This is a kind of dysfunctional plasticity. Therefore, if investigators can understand the mechanisms and control the abnormal plasticity, they might be able to “undo” the changes in the brain that cause these movements.

#### **Targeting the cAMP Pathway in the Striatum to Treat Dystonia**

*Emmanuel Roze, MD, Paris Brain Institute*

**Quick Summary:** This project investigates how disruptions in neuron signaling in the striatum lead to dystonia.

**More Detail:** The striatum is a structure deep in the brain that plays a critical role in the control of movements. cAMP is a molecule that regulates many cell functions, including in neurons (brain cells). The cAMP signaling pathway controls processes important for the function of neurons in the striatum and the control of movements. Various genes that encode proteins involved in this cAMP pathway can cause dystonia when mutated, particularly GNAL/DYT25 and ADCY5. Mutations of GNAL/DYT25 lead to *reduced* cAMP production while mutations of ADCY5 lead to *increased* cAMP production. To better understand how disruptions in the cAMP pathway produce dystonia, investigators are characterizing movement dysfunction and striatal biochemical abnormalities of genetic mouse models. They will correct the abnormal cAMP pathway in the mouse models using drugs and investigate whether treatment improves the biochemical abnormalities and movement dysfunction. Finally, they will evaluate the effect of caffeine in ADCY5-related dystonia patients which has been helpful in some patients.



## RESEARCH FELLOWSHIPS

*The DMRF has attracted new researchers to the field by creating funding awards to support young investigators at different stages in their scientific training. Postdoctoral fellowship awards support outstanding young scientists who have earned a doctoral degree and have embarked on a period of mentored research. DMRF is supporting postdoctoral fellows who are working to fundamentally improve our understanding of brain dysfunction and molecular mechanisms underlying dystonia.*

### New in 2022

#### **Beyond Theta: Analyzing Oscillations Across the Frequency Spectrum in Patients with Dystonia Implanted with Sensing-Enabled Pulse Generators**

*Stephanie Cernera, PhD, The Regents of the University of California, San Francisco*

**Quick Summary:** This study will examine individual patients' brain signals while they are performing different movements and use what is learned to improve therapeutic deep brain stimulation results.

**More Detail:** New deep brain stimulation (DBS) devices are capable of recording data from deep brain structures and the surface of the brain, giving researchers insights into how these brain regions communicate when dystonic movements occur. However, most studies reporting brain patterns in dystonia patients have been short recordings during DBS surgery, in which the patient's movements are limited. These studies have detected a distinct pattern of brain activity in deep and surface brain structures in patients with dystonia. However, this pattern is not noticeable in every dystonia patient's brain, and the significance is unknown due to short recording time during surgery.

DBS therapy is moving towards "smart" stimulation, in which therapy can be delivered or adjusted based on a patient's personal brain activity signatures. The challenge is, how would a physician know when a patient with dystonia needs stimulation delivered, or how DBS is changing someone's personal brain patterns? Within this proposal, investigators want to explore a patient's personal brain patterns while they are performing different movements in the comfort of their own homes. This will allow them to learn more about various brain patterns and relate these brain patterns to different behavioral states such as walking (a "good brain state") or when a patient is experiencing symptoms of

### **DMRF Clinical Fellowships Train Young Movement Disorder Specialists**

In response to the widespread need for additional dystonia clinical experts, the DMRF sponsors one-year clinical fellowships to help train physicians in the diagnosis and treatment of movement disorders with special competence in dystonia. The fellowships focus on training in both the clinical evaluation and care of patients with dystonia and clinical research on dystonia. The training is patient-oriented and includes hands-on experience in clinics as well as participation in professional meetings and workshops.

#### **Congratulations to our 2022 DMRF Clinical Fellow and mentor:**

**Jun Yu, MD, University of Florida**

**Mentor: Michael Okun, MD**

In 2022, the clinical fellowship program is supported by grants from Ipsen and Merz.

dystonia (a "bad brain state"). Additionally, they can study how these personal patterns change when stimulation is on versus off. Finally, once investigators understand how a personal brain activity signature changes with stimulation, they can use this pattern to program the patient's DBS device so that they are receiving the best treatment. The methods used in this study will be converted to a sensing-enabled DBS device that new DBS patients can be implanted with in order to maximize treatment benefit.

#### **Investigating Sensory Dysfunction in a Mouse Model of Early-Onset Generalized Dystonia**

*Amanda Pocratsky, PhD, University College London*

**Quick Summary:** Results of this study will provide key insights into the underlying biological mechanisms of sensory dysfunction in dystonia.

**More Detail:** While the characteristic features of dystonia are movement related, there is also a widely recognized sensory component to this movement disorder. For example, individuals often use "sensory tricks," such as lightly touching

*Continued on page 10*

the affected body region, to temporarily alleviate dystonia symptoms. These alleviating effects can be reproduced by applying sensory stimulation to the affected regions. These, and many other observations, collectively implicate a role for the sensory system in dystonia, but the underlying biological mechanisms remain unknown. Dr. Pocratsky will investigate how sensory dysfunction occurs in a new TORIA/DYT1 animal model. The experiments outlined in this project will provide scientists with a solid understanding of where and how sensory dysfunction occurs in dystonia from transduction (how sensory information gets turned into a nerve signal) to conduction (how the signal transmits across the nervous system) to transmission (how the signal communicates with other nerve cells). These data will provide key insights into the underlying biological mechanisms of sensory dysfunction in dystonia. Not only could these findings transform understanding of dystonia, but they may also reveal key sensory-based targets for new therapies.

## 2nd Year Funding

### Investigating Abnormal Neurodevelopment in a Novel *in vivo* Model of Inherited Dystonia

*Simon Lowe, PhD, University College London Institute of Neurology*

**Quick Summary:** Several types of dystonia develop during specific age windows of childhood or adulthood. This project seeks to better understand why and how certain types of dystonia develop during specific stages of brain development.

**More Detail:** Some disease-causing mutations act acutely, which means they cause a disorder by directly altering the function of the brain, affecting its ability to perform tasks. Other mutations act developmentally, which means they alter the way the brain develops, causing lasting alterations in the way the brain works. Knowing which is happening is key to understanding and treating a disorder. Dr. Lowe is investigating a form of dystonia caused by a single mutation in the gene *KCNMA1*, which has a number of important roles in neurons (brain cells). Dr. Lowe and team developed the first animal model of this disorder in the fruit fly. Using advanced genetic techniques, he is able to turn the mutation “on” and “off” at different stages of the flies’ lifecycle. Preliminary data show that turning the mutation on in the adult fly has no effect but turning the mutation on and then back off again during its development causes severe, lasting movement defects in the adult fly. These defects very much resemble the movement dysfunction seen in humans. This is

a clear demonstration that the mutation causes movement dysfunction in the fly by altering nervous system development. Dr. Lowe aims to confirm this and identify the key developmental stage with additional experiments, and then ask the question how this mutation affects development.

### Neural Signals in the Cerebellar Nuclei Gate the Manifestation of Dystonia-like Symptoms

*Meike van der Heijden, PhD, Baylor College of Medicine*

**Quick Summary:** This study is investigating the cerebellum as a target for new treatments to alleviate dystonia symptoms.

**More Detail:** Recent studies have suggested that the cerebellum may be a central node in a brain network that triggers dystonia in humans and mouse models. One specific area of the cerebellum, the cerebellar nuclei, sends neural signals to other regions of the brain and spinal cord that are involved in motor control. Imaging studies in dystonia patients and electrical recordings in dystonia mouse models have shown that these neuronal signals are different from people without symptoms and control mice, respectively. Interestingly, therapeutic stimulation of the cerebellar nuclei using deep brain stimulation (DBS) alleviates symptoms in some people with acquired dystonia and in a mouse model with severe dystonia.

Dr. van der Heijden hypothesizes that the cerebellar nuclei act as a fulcrum in the expression of dystonia symptoms. On the one hand, abnormal neuronal signals in the cerebellar nuclei can cause dystonia-associated symptoms. On the other hand, stimulating these nuclei with DBS can alleviate dystonia-associated symptoms. However, to fully understand how to best optimize DBS treatment, it is necessary to know precisely what the balanced state neuronal signaling is in the cerebellar nuclei, and in what direction these communication signals are skewed in mouse models of dystonia. To answer this question, the investigators are recording brain activity profiles in multiple mouse models of dystonia with different severities of dystonia-associated symptoms. They will use mathematical computations to determine what aspect of the neural signals are abnormal and cause dystonia-associated movement impairments. They hope to find precisely how cerebellar signals contribute to dystonias with different causes. This knowledge will be an important step for optimizing cerebellar DBS to become a first-line treatment for patients with dystonia.

# Dystonia Coalition Awards

The DMRF is proud to partially support two Dystonia Coalition Pilot Project Awards. The goal of the Pilot Projects Program is to foster the most promising clinical and translational studies on dystonia.

## **Moodscreen for Cervical Dystonia: A Diagnostic Accuracy Study of Depression and Anxiety**

*Davide Martino, MD, PhD, University of Calgary*

**Quick Summary:** This study aims to improve care for individuals with dystonia by identifying the most useful methods to screen individuals for co-occurring depression and anxiety.

**More Detail:** Depression and anxiety frequently negatively impact how individuals with dystonia feel and function—and often cause more disability than involuntary movements and postures. This is especially the case in cervical dystonia (CD) and additional forms of adult-onset, isolated idiopathic dystonia. There is a need to update the model of care for dystonia patients to detect and treat disabling non-motor symptoms. Selecting the most accurate, feasible, and cost-effective screening system for depression and anxiety will be essential to this care model upgrade and is the core intention of this project. The primary aim is to assess different clinical rating instruments for depression and anxiety in patients with cervical dystonia compared to a gold standard assessment. The second aim is to measure whether these instruments are universally useful across age, educational level, body spread of dystonia, and ongoing therapies.

### **What is the Dystonia Coalition?**

The Dystonia Coalition is a collaboration of medical researchers and patient advocacy groups focused on accelerating clinical research in the field. Fifty-six research centers



in North America, Europe, Asia, and Australia are participating. Program Director is H. A. Jinnah, MD, PhD of Emory University School of Medicine. DMRF Scientific Director Joel S. Perlmutter, MD of Washington University in St Louis is Co-Director. The DMRF supports the Dystonia Coalition by providing staff and organizational support at no cost.

An “Update on Dystonia Coalition” webinar is available for viewing at: [dystonia-foundation.org/research/dystonia-coalition/](https://dystonia-foundation.org/research/dystonia-coalition/)

This will be a cross-sectional, multi-center observational study to collect general demographic and clinical data and perform psychiatric assessments.

## **The Effect of Multi-Day Continuous Theta-Burst Stimulation on Symptoms of Cervical Dystonia**

*Daniel Corp, PhD, Deakin University*

Non-invasive brain stimulation techniques, such as theta-burst stimulation (TBS), hold potential as a safe and effective treatment for dystonia, especially for less severe cases where deep brain stimulation surgery may not be recommended. Dr. Corp previously localized a brain network showing specific abnormality in cervical dystonia, with the key nodes located in the somatosensory cortex and the cerebellum. The primary aim of this project is to employ a randomized controlled trial to investigate whether stimulation of this location can improve symptoms of cervical dystonia. A secondary aim is to test whether stimulation results in changes in brain structure or physiology and whether these changes are associated with symptom improvement. If the present study can demonstrate efficacy of this protocol, the investigators plan to use these data to apply for funding to perform a larger trial that could lead to the approval of this treatment in cervical dystonia patients. In addition, they have now completed lesion network mapping analyses in additional forms of dystonia (e.g. hand dystonia, blepharospasm, etc.). Therefore, the present study will indicate whether these networks are also likely to respond to similar brain stimulation protocols.



## ***Join us for In-Person Events & Virtual Dystonia Zoo Day***

The DMRF is pleased to return to in-person events in select cities to re-unite the dystonia community, promote awareness, and support the mission to find a cure.

We're especially excited to announce this year's Dystonia Zoo Day season, with in-person events and TWO virtual opportunities to participate regardless of geography.

***Register at [dystonia-foundation.org/events](https://dystonia-foundation.org/events)***

**NATIONAL  
SPONSORS**







## VIRTUAL KICK OFF CELEBRATION

**August 18, 7PM Eastern / 6PM Central / 4PM Pacific**

Join us for the launch of our Zoo Day season and make sure you're up-to-date on all the ways you can connect with others in the dystonia community and support the mission to find a cure. Program will include special guests including a zoo keeper chat.

Registration for the Kick-Off Celebration is free and includes registration for the 3rd Virtual Dystonia Zoo Day & Closing Ceremony. You can show your support by ordering a t-shirt and/or making a donation. For details and to register, visit: [dystonia-foundation.org/letszoothis](https://dystonia-foundation.org/letszoothis)

### MAXIMIZE YOUR SUPPORT:

- All team donations will be matched, doubling your support.
- All registration fees for teams of 25+ will be matched.
- For every 10 registrations, DMRF gets \$100 from the match.



## IN-PERSON EVENTS

***Local Dystonia Zoo Days are back!***

Paid registrations for any in-person Dystonia Zoo Day (\$25 adults/ \$15 children) includes zoo admission, refreshments, and event t-shirt.

- Portland – August 20
- Toledo – August 27
- Bronx – September 10
- Boston – September 17
- Pittsburgh – September 18
- Cleveland – September 25
- Los Angeles – October 1
- Twin Cities – October 2



## 3RD VIRTUAL DYSTONIA ZOO DAY & CLOSING CEREMONY

**October 8, 12PM Eastern / 11AM Central / 9AM Pacific**

No one has to face dystonia alone! Join us for a virtual program to unite the dystonia community, support research, and increase dystonia awareness. The program will feature special guests including a zoo keeper chat, inspiring stories, research Q&A, and more.

***Sign up at [dystonia-foundation.org/letszoothis](https://dystonia-foundation.org/letszoothis)***

# Nordic Skier Sydney Peterson Triple-Medals at Beijing Paralympics

In March 2022, 19-year-old cross-country skier Sydney Peterson competed for the first time with Team USA at the Paralympic Winter Games in Beijing. She won a medal of every color: gold, silver, and bronze. She is tied for the second most decorated US athlete at the Beijing Games.



Sydney Peterson skis for her college team and Team USA.

“It was a pretty amazing experience,” said Sydney, who skis for St Lawrence University in New York while double-majoring in neuroscience and biophysics. “There was, I wouldn’t say ‘pressure,’ but [at the Paralympics] more people are watching, and people want you to do well. Getting 1st through 3rd place is a big deal but, after that, if you are 4th or 30th, no one really cares.”

## AT A GLANCE

- Cross-country skier Sydney Peterson won three medals at her first-ever Paralympic Games.
- She developed hand and leg dystonia at age 13.
- She is tied for the 2nd most decorated US athlete at the Beijing Games.

She won gold in mixed relay cross-country, silver in long-distance cross-country, bronze in sprint cross-country, and placed 6th in middle-distance cross-country.

• • •

Growing up in Minnesota, Sydney began skiing around age five. By seventh grade she was on the high school skiing team. At 13, she began developing pain and movement difficulties in her left arm. She was ultimately diagnosed with dystonia affecting her left arm, hand, and foot. “I was in pretty

severe pain that limited everything I could do. Thankfully I have found better ways to deal with that pain so now I won't say pain is a limiting factor." Sydney experiences persistent discomfort from tight muscles in her limbs, and she has limited use of her left arm and hand.

Sydney qualified for Junior Nationals in high school, despite regularly having to adjust her technique to accommodate dystonia. "Some days, I just couldn't use my arm at all, so I'd either ski with one pole or just switch to running for my training," she said. Ultimately, she found one-armed skiing with a splint for her pole hand and a brace in her boot was the best arrangement for her performance.

"There were definitely difficult moments, leading up to the Games and at the Games, where I really wish I could just ski with two poles again, but I can't," she said. "In college skiing, everyone's technique generally looks the same because that's the 'best' technique, but in paraskiing you have to play to your individual strengths more. For me, that's leaning onto my right side rather than skiing more symmetrically."

One of the mainstays of Sydney's treatment is botulinum neurotoxin injections every 90 days. She intentionally scheduled the injections so she would be at the peak of benefit while at the Paralympics. However, she then decided to skip the injections because of the risk of coronavirus exposure to herself and the team by attending the appointment. "If I got Covid-19, we wouldn't be able to go to Beijing at all, so I didn't want to risk that," she said. Her performance at the Games was all the more remarkable given she went without such an important component of her treatment.



Since competing in Beijing, Sydney has been featured in national and local news media, met President Joe Biden at the White House, thrown out the first pitch at a Minnesota Twins MLB baseball game, and earned thousands of fans in the dystonia community.



Her accomplishments of the last several months have been extraordinary, and yet she also appreciates the sweet satisfaction of everyday victories: "Part way through last semester I switched my shoelaces out to stretchy ones that you don't have to tie, and that saved me so much time every morning!" She also discovered a clever way to put her hair in a ponytail without help by bracing her arm against a towel rack in her bathroom.

Sydney trains year-round and has not given up on one day skiing with two hands. "Skiing is like a fun version of physical therapy, and it's been a great way to keep my body healthy

and strong," she said. "It is one of the best things I can do for myself."



The drive that makes Sydney a star athlete motivates her to push back against the challenges of dystonia. "I'm going to doctors' appointments, trying to figure out how can I get this better while still knowing there's not a cure and there's not a perfect solution," she explained. "I've always thought, okay, I want to figure this out, but I am not necessarily at peace with it." The sense of community and friendships that come with being part of a team also help keep her going: "You don't always have good races, but generally someone on your team had a good race that day. So, then you can be happy for them rather than being sad about not having a great race."

As she enters her junior year of college in the fall, she is eyeing the possibility of climbing the ranks of elite student athletes in the National Collegiate Athletic Association (NCAA).

*Recaps of Sydney's races in Beijing are available for viewing on the Paralympics YouTube channel: [youtube.com/c/paralympics](https://youtube.com/c/paralympics)*

# What Causes Dystonia?

The biological origins of dystonia are not yet fully understood. However, for some individuals, it is possible to identify a cause.

## INHERITED DYSTONIA

Some cases of dystonia are genetic. Numerous gene variants are known to cause dystonia. Below are some examples:

### DIAGNOSIS

Early onset torsion dystonia

### GENE

TOR1A/DYT1

Dopa-responsive dystonia

GCH1/DYT5, TH

Rapid onset dystonia-parkinsonism

ATP1A3/DYT12

Wilson disease

ATP7B

Myoclonus-dystonia

SGCE/DYT11

Leigh syndrome

MT-ATP6

## ACQUIRED DYSTONIA

Some cases of dystonia occur due to a specific external cause. Below are some examples:

### CAUSE

Birth injury

### DIAGNOSIS

Dystonic cerebral palsy

Infection

Viral encephalitis, HIV infection, tuberculosis

Medication exposure

Tardive dystonia, tardive dyskinesia

Toxins

Poisoning

Blood supply conditions

Stroke

Injury

Traumatic brain injury, head trauma

## IDIOPATHIC DYSTONIA

Many, if not most, cases of dystonia occur without any apparent cause. Many cases of adult-onset focal dystonia fall into this category.

**Sporadic** – Individuals may develop dystonia without an identifiable cause or family history.

**Familial** – Families may have multiple individuals diagnosed with dystonia, strongly suggesting a genetic cause, but the individuals do not test positive for any known dystonia-causing genes.

## IMPLICATIONS FOR TREATMENT

The underlying cause of dystonia may influence the treatment plan. For example, TOR1A/DYT1 dystonia and tardive dystonia tend to respond positively to deep brain stimulation. If an individual develops dystonia following a stroke or traumatic brain injury, there may be additional neurological and cognitive symptoms that require treatment, depending on the extent of injury to the brain.





## DYSTONIA 101

Dystonia is a complex disorder to understand. Here are the basics:

- Dystonia is a neurological disorder. It affects the brain's ability to control voluntary muscle movement.
- Dystonia does not affect vital organs such as the heart.
- There are numerous types of dystonia. It can affect a single body area or multiple muscle groups.
- Dystonia may occur with other movement symptoms such as tremor, myoclonus, or parkinsonism.
- Inherited dystonias are those with a known genetic origin, for example, gene variants such as TOR1A/DYT1, THAP1/DYT6, and SGCE/DYT11.
- Dystonia sometimes results from birth injury, drug exposure, brain injury, infection, and other identifiable causes.
- Treatment options include oral medications, botulinum neurotoxin injections, deep brain stimulation, and less invasive methods such as physical or occupational therapy.
- Individuals with dystonia may experience depression and anxiety disorders, including social anxiety.
- Stress does not cause dystonia, but symptoms may worsen in stressful situations.

For more information, visit [dystonia-foundation.org](http://dystonia-foundation.org)

# People on the Move

The DMRF is grateful for the volunteers across the country working to improve dystonia awareness and support medical research. Every effort makes a difference!

## PET PALOOZA Raises Dystonia Awareness & Research Funds



Linda Davis and Charlie



Terri Chapman and Sydney



Sarah Adams with Molly and Spencer

In February, Dystonia Pet Palooza brought animal lovers together to raise awareness while celebrating the special bond between individuals with dystonia and their pets. The campaign raised \$20,000.

Congratulation and many thanks to DMRF supporter Terri Chapman. Her dog Sydney was the top earning pet, raising over \$4,100. Sadly, Sydney died the day Terri registered for Pet Palooza, and all gifts received were made in her memory. We are also saddened to report that since Pet Palooza, Sarah Adams' dog Molly has also passed away.

These top dogs raised more than \$250:

- Attila - Teagan Maurer
- Benny – Nancy Adams
- Blue – Gina Paolo
- Charlie – Linda Davis
- Fiona – Lucinda Mayer
- Ginger – DeAnne Bean
- Jasper - Lindsay Hudson-Austin
- Lulu – Dee Linde
- McKinley & Carter – Eric Greenberg
- Mishka – Bailey Taylor
- Molly - Emily McNaughton
- Molly & Spencer – Sarah Adams
- Nugget & Lady D - Pamela Kinzler
- Snoopy – Virginia Bryan
- Sydney – Terri Chapman
- Tiger, Olive & Cassie – Kari Youngberg
- Tovah – Betsy Cohen
- Yuki – Sarah Tremblay

The DMRF express deep gratitude to all the generous people and creatures who participated in this important and fun campaign.



The Nashua Distance4Dystonia 5K Walk/Run organized by Amanda Lockhead took place in May to raise dystonia awareness and funds for urgently needed research toward a cure. More than 100 people attended to share their support, after the event had been paused in recent years due to the pandemic. The event raised over \$5,000.

**DYSTONIA  
ZOO DAYS  
ARE BACK!**  
**See page 12 for  
information on  
in-person AND  
virtual events.**

## THANK YOU Social Media Fundraisers



Thank you to everyone who generously collects donations to benefit DMRF on social media. Media platforms like Facebook and Instagram provide DMRF with limited information about these fundraisers, and we wish to acknowledge all of our supporters.

To collect donations for DMRF through Facebook's fundraiser tools, go to [facebook.com/fundraisers](https://facebook.com/fundraisers) to get started.

Learn how to use Instagram donation stickers at [help.instagram.com](https://help.instagram.com)

Be sure to notify DMRF of your social media fundraiser to be eligible for the match. Tag or DM us on Facebook or Instagram (@dystoniarmf), or send a message via [dystonia-foundation.org/contact](https://dystonia-foundation.org/contact)

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## YOU ARE NOT ALONE

### Peer Support Offers Understanding & Connection

A dystonia diagnosis can feel isolating and overwhelming. Connecting with peer support resources means access to information and validation.

The DMRF partners with volunteers to sponsor local support groups and online support forums.

Support groups provide local opportunities for information, camaraderie, and community activism. Many support groups are meeting by video conference or hybrid in-person meetings with the option to participate virtually.

Private Facebook groups are available 24/7 and connect you to others in the community throughout the country and all over the world.

To locate local and online support groups, visit: [dystonia-foundation.org/support/](https://dystonia-foundation.org/support/) or reach out for assistance at 800-377-3798 or [dystonia@dystonia-foundation.org](mailto:dystonia@dystonia-foundation.org)

## FINANCIAL SUPPORT

Americans with disabilities live in poverty at more than twice the rate of people without disabilities. Sixty-five percent of adults with disabilities participate in at least one safety net or income support program. These resources may help fortify financial security:

### Pharmaceutical Assistance

Drug manufacturers often offer financial assistance programs to help individuals who meet specific criteria afford treatment. This includes the companies that manufacture botulinum neurotoxins, one of the most common dystonia treatments. Information about these programs is available at [dystonia-foundation.org/botulinum-toxin-injections](https://dystonia-foundation.org/botulinum-toxin-injections)

### Employment Resources

The Job Accommodation Network (JAN) offers guidance on workplace accommodations and job protections for employees with disabilities, including dystonia. Go to [askjan.org](https://askjan.org) and search "dystonia."

JAN can also direct you to state vocational rehabilitation agencies that help individuals with disabilities obtain and/or maintain employment.

### Disability Benefits

The US Social Security Disability Insurance (SSDI) and Supplemental Security Income (SSI) programs provide assistance to people with disabilities. Information about eligibility and the application process is available at: [ssa.gov/benefits/disability](https://ssa.gov/benefits/disability)

DMRF offers a fact sheet of Frequently Asked Questions about Social Security Disability Benefits at [dystonia-foundation.org/print](https://dystonia-foundation.org/print)

### Local Resources

States and municipalities may offer assistance programs for local residents. Contact your state Department of Human Services to make sure you are not missing out on programs for which you may be eligible.



# DMRF Formalizes Commitment to Mental Health

## Conversation with New VP of Mental Health Programming & VP of Support

The hallmark symptoms of dystonia are involuntary movements and postures, but this only tells part of the story.

While the DMRF has long recognized the need for emotional and mental support for individuals and families, the Foundation is taking added steps to address mental health in the dystonia community. Earlier this year, the DMRF formalized this priority by creating a Mental Health Programming Committee. Past Leadership Chairperson Paula Schneider is stepping into the role of Vice President of Support to replace Karen Ross, PhD, who is now the first Vice President of Mental Health Programming.

Karen has served on the Board of Directors since the 1980s. Her adult son developed generalized dystonia as a child. She is a retired Clinical Psychologist and Family and Marriage Therapist. She authored the book *Holding the Hope: A Parent's Guide to Living with Dystonia*.

Paula developed dystonia in her 30s, and it quickly became severe. She was one of the first dystonia patients in the country to undergo deep brain stimulation (DBS). She has served on the DMRF Board of Directors since 2009 and is a liaison between the Board and DMRF peer support groups.

...

Karen and Paula recently met for a conversation about mental health and dystonia and how the Covid-19 pandemic has made it all the more important to ensure the emotional and mental health needs of the dystonia community are being addressed.



Karen Ross, PhD and Paula Schneider are guiding DMRF efforts to address mental health.

### AT A GLANCE

- **Untreated mental health disorders reduce quality of life for individuals with dystonia.**
- **The Covid-19 pandemic has taken a toll on many people's mental health.**
- **DMRF created a Mental Health Programming Committee to formalize efforts to address mental health needs.**

"It's very stressful for everybody," said Karen. "Everyone has been touched in some way by the effects of the pandemic. But for people that have dystonia, or pre-existing conditions, it's a double whammy. The anxiety is heightened by so much more right now."

The Covid-19 pandemic has exacerbated issues the dystonia community has long struggled with including healthcare and treatment access, accessibility of public spaces, social isolation, lack of social services for individuals with disabilities, and mental health disorders including depression and anxiety.

Echoing the various types and severity of dystonia, as well as individual life experiences, mental health experiences also tend to be varied.

"It's hard to describe what people are experiencing in a general way. It's very individual," said Karen. "I know for me, when our family was going through this, and for others that I know, trying to get a diagnosis, trying to get treatment, just going through that whole process—it's traumatic. Dystonia is a huge traumatic experience for everyone in the family."

"The isolation with this condition is just unbelievable," added Paula, "and that was the case before Covid."

Paula underwent deep brain stimulation (DBS) in 2001 after more than a decade of severe disability. Ironically, it was the success of the procedure, and the startling absence of pain and dystonia symptoms, that made her realize how profoundly dystonia affected her physically, emotionally, and mentally.



"I didn't realize how sick I was and the effect it was having on other people," she said. "It's very hard to describe what the pain of dystonia is like, with the pulling and the twisting and all of that, until you don't have it anymore. It creeps up on you." She recalled that friends confessed, years after the fact, that they felt uneasy spending time with her when they sensed she was in pain. They didn't know if they were helping by staying to visit and socialize, or whether they should leave and let her rest.

There was a time Paula didn't realize she was presenting "classic" signs of clinical depression until, to her surprise, her doctor brought it up.

"You were suffering," offered Karen. "I think that would be a way to describe how much suffering you were going through, that you had no idea how much you were suffering until you weren't. Those are coping mechanisms because it allows you to survive the best you can, but it takes a toll when you have suffered and suffered and suffered. That too is traumatic."



Karen stressed how intertwined social connection is to mental health: "If you don't have family members or a therapist or somebody you can talk to about any of these things, it makes it so much more isolating and depressing and painful. It's reaching out that is usually hard for people, maybe the hardest part, reaching out to say I need to talk for 10 minutes or to get on one of the private social media groups and say this is what's going on with me and I don't even know what to do. Taking that first step is difficult. The listening part is the healing part. When you're being listened to, and you're able to truly express what's going on, you feel better afterwards."

Paula agreed: "Just having somebody listen and acknowledge that the pain is real is so important."

The goals of the Mental Health Programming Committee are to identify needs in the dystonia community related to mental health and to assemble experts and resources to address those needs. The DMRF's efforts include research as well as patient outreach. In the fall, DMRF is planning the first-ever scientific workshop on non-motor aspects of dystonia. DMRF is also co-funding a Dystonia Coalition Pilot Projects grant on improving methods for screening individuals with dystonia for mental health concerns. See more on page 10.

Karen said she looks forward to keeping the dystonia community informed of the Mental Health Programming Committee's activities as they progress: "We are in the process of trying to discover what people need most and finding ways to help people get what they need most. We're doing everything we can to help."

"This is an organization that really cares about people," said Paula. "We try to show every day that we care."

For more information about dystonia and mental health, visit:  
[dystonia-foundation.org/mental-health/](https://dystonia-foundation.org/mental-health/)

## DYSTONIA & MENTAL HEALTH

- Individuals with dystonia frequently experience debilitating issues such as depression, anxiety, pain, and sleep difficulties.
- Research is showing that untreated mental health disorders among individuals with dystonia are one of the most powerful predictors of reduced quality of life.
- Lessening dystonic symptoms without treating co-existing mental health disorders may not necessarily help someone function or feel better.

## RESOURCES

- Project Healthy Minds  
[projecthealthyminds.com](https://projecthealthyminds.com)

## IF YOU ARE IN CRISIS, HELP IS AVAILABLE:

- **Call** the National Suicide Prevention Lifeline at 800-273-8255/En español 888-628-9454
- **Chat** online at [suicidepreventionlifeline.org/](https://suicidepreventionlifeline.org/)
- **Dial 988.** This is a new three-digit dialing code that will route US callers to the National Suicide Prevention Lifeline.
- **Text** "HELLO" to 741741

# Personal Profile

## Kathy Ray

Kathy Ray resides in Atlanta and is retired from a career in hospitality and marketing. In addition to leading the Atlanta Dystonia Support Group she volunteers with multiple local organizations.

### How did your symptoms begin, and how were you diagnosed?

I am 75, and I've had blepharospasm a quarter of the time I've been alive. Like most blepharospasm patients that I've talked to, women particularly, when that twitch starts, at first we think it's our makeup. So, you throw out that mascara. Then I thought it was the laundry detergent, my skin cleanser, in my attempt to find out what this was. What's interesting about my blepharospasm, certainly not everybody's, but my blepharospasm reacts to 'adrenaline,' I'll call it. When I'm 'on' and geared up and talking, it's gone. When I'm by myself and I haven't spoken out loud that day, it's all over the place. So that made it even more confusing because, the job I was in, I would make a speech in front of 500 people, and it didn't happen. So, I thought, well, it's not a nervous tic. If I was going to be nervous, that would probably be when I was nervous. I prowled through doctors and finally it was my GP who noticed it. I said, what do you think it is? She said, I think it's neurological. You think it's something wrong with my brain? That's a scary thing to hear at first. I made it over to a movement disorder clinic in Atlanta, I've been with them for 25 years. I don't like to complain about my ailment because when I go to the movement disorder clinic for my injections, I see a lot of stuff. A lot of stuff that I don't have. With the botulinum injections and meds, it is a relatively minor thing in my life. I never decline a social invitation, so it doesn't impact my social life, the problem is getting to the social life because when I'm in the car by myself, it's just going a mile a minute if I don't take my medicine. The biggest thing is reading, which I almost can't do now. Reading, driving, and watching TV are when it acts up.

### Talk about your experiences as a support group leader.

We had about 80 people come to our first meeting. It was really good. Our second meeting was supposed to be in March 2020, but everything shut down. By April we were



Kathy Ray is diagnosed with blepharospasm, a focal dystonia of the eyelid muscles.

meeting every month by Zoom. I get something out of our meetings every time. There's a little light in me that really flames up when I can see that somebody was helped by something I tried to do. It makes me so happy, and it gives me so much energy. When we have new people, I love to see the way the group surrounds that person. We all go around and introduce ourselves and provide this cocoon for them to hear all these things and then we say, are you comfortable to tell us about what's going on with you? And these sweet people will ask gentle questions so that person can spill out more stuff if they want. It's really therapeutic for everybody involved.

### What advice or what encouragement do you find yourself giving the most often?

The advice we as a group give most often to a new person is: Find the right doctor. Just stay after it. If it's not working, get out and try somebody else. I say two times, two chances. If you don't feel something for that person and you don't feel like they feel something for you, move on. Since the audience for this [interview] is people who have dystonia, I think the main thing that I would say is that I believe that it is our responsibility, those of us who are afflicted—if that's the right word—to do what we can: answer every survey, sign up for research studies, give a little money. I think we owe it to the next generation to do what we can now to help our researchers. Because there are things that are happening, these sparks here and there, and that new thing probably won't be ready for me because I may not be around, but it'll be ready for somebody I know. And I believe that our organization is one of the reasons that people are more familiar with dystonia and more research is happening. Something in me, a part of my soul, is fed when, at the end of the day, I feel like I have at least done something to give back. My big thing in life is joy. I want to have and feel and create joy.

# Personal Profile

# Fabio Salazar

Fabio Salazar is a mechanical engineer working in the energy industry. He lives in Katy, Texas and leads the Houston Dystonia Support Group.

## How did your symptoms begin, and how were you diagnosed?

I started having neck pain in 2014, a pain doctor diagnosed it was degenerative disc disease. In 2016 I started feeling some tightening of a muscle going through my neck and about six months after that, the neck spasms started, my head was moving to the right side. My wife has had cervical dystonia since she was 27, so we knew about dystonia already. I went to a movement disorders specialist, he confirmed the diagnosis and started treating me with botulinum neurotoxin injections. The diagnosis was straightforward for me. The injections have not been as effective as other cases that I know of, I feel it helps a little by making the movements less strong; additionally, stretching helps me a lot. In the morning, as soon as I wake up, I feel really great, my neck is relaxed, I can move my head everywhere, but then after a few seconds or minutes, the spasms start again and the tightening builds up through the day, especially if I have a busy day, or have to walk a lot, or do much physical activity standing up. I get tired and at night it is usually more difficult, so stretching really helps me when I do it.



Fabio Salazar was diagnosed with cervical dystonia in 2017.

## How would you say dystonia has impacted your life overall?

There are things that I do not do any more, like running, which was my main hobby and exercise. I felt more alive when I used to run, especially outdoors, I miss that a lot. It takes more time to do things because I have to continue to hold my head with my hand, trying to keep my head in position, unless I can sit with a headrest supporting my head. Dystonia affected my work initially, I did not drive for a year because of the head movement. But it became more under control later. Now I can do mostly everything that I did before, except running. It was hard at the beginning, but it's better now. When dystonia started, I assume most people diagnosed with dystonia go through this, you feel

your whole life changing. At the beginning, I wanted to be just on the floor. Whenever I came back from work, I laid on the floor. I did not want to be standing up or sitting. I've seen that in other cases, it seems like laying on the floor is what gives you some relaxation or maybe just less effort with your muscles. When you are healthy, you can think in terms of weeks and months, but when you have dystonia, especially if it is a hard case, you think in terms of minutes and seconds. You wonder if you will be able to continue to do what you need to do. It can seem unsurmountable. As time

goes by, even though it is affecting you, you find a way to continue.

## Why was it important to start a support group?

It's difficult to understand someone having a problem unless you go through it and feel it. My wife suffered for years and I was there to help her, I accompanied her to botulinum injection appointments. Nevertheless, when I had it in my body, I realized just how difficult it was. I learned from my wife. I thought I could help others based on what I knew. Helping others also helps you, somehow. You are not focused on yourself but on helping someone else.

## What advice do you have for others with dystonia?

Don't let dystonia define who you are as a person. I know this can be difficult to do. Try to keep doing what you like to do, even if it's not perfect. Try not to isolate, especially from people who care about you and want to help you. Find out what works for you. Some doctors focus on the medical treatment only, but other things may help as well, like exercise, dancing, even listening to music. It's good to know what doctors and organizations like DMRF are doing in research. The main thing is to not isolate yourself and do not let dystonia take over your life. Maybe it's an opportunity to grow, it not a punishment from life or God. Don't put too much stress and pressure on yourself to recover, that can help too.



## Dystonia Dialogue

Dystonia Medical Research Foundation  
One East Wacker Drive · Suite 1730  
Chicago, Illinois 60601-1980

PHONE 312 755 0198 · 800 377 DYST (3978)

WEB <https://dystonia-foundation.org>



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## Upcoming Dystonia Events—Join Us Virtually & In-Person



The DMRF is returning to in-person events in select cities to re-unite the dystonia community, promote awareness, and support the mission to find a cure.

**DYSTONIA ZOO DAY SEASON IS AUGUST 18–OCTOBER 8,**  
and includes both local events and virtual opportunities.

***Learn more at [dystonia-foundation.org/events](https://dystonia-foundation.org/events)***