



DYSTONIA
MEDICAL
RESEARCH
FOUNDATION

Dystonia Dialogue

NEWSLETTER OF THE DYSTONIA MEDICAL RESEARCH FOUNDATION

Summer 2023 | Volume 46 • No.2

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Maddie Paolero sits with her service dog, JJ. Learn if a service dog may be right for you and what it takes to find and care for one (p.9).

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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.

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



Mark Rudolph
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Janet L. Hieshetter
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We are thrilled to share with you some of the highlights of the recent Samuel Belzberg 6th International Dystonia Symposium, which took place in Dublin from June 1–3, 2023. This seminal event gathered experts, researchers, and clinicians from around the globe, who united to exchange knowledge and advancements in the field of dystonia research. The discussions continued over coffee breaks and in between meetings, creating an environment conducive to fruitful exchanges and deeper connections among attendees. The passion and unwavering dedication displayed by the participants was evident in every session.

The journey leading up to this symposium stands as a testament to the steadfast commitment of the organizers. The planning for this event began in late 2017, with the original date set for June 2020. Despite the challenges and uncertainties faced along the way, the symposium came together beautifully. We extend our heartfelt gratitude to Drs. Buz Jinnah and Antonio Pisani, the Co-Chairs of the symposium, for their tireless efforts in orchestrating this exceptional event.

One of the highlights of the symposium was the profound impact of DMRF Co-founder, Frances Belzberg's opening remarks. Fran spoke of her gratitude to all for their work and reminded everyone that people are waiting for the answers that only research can provide. Fran was joined in Dublin by her daughter, Cheri, who served as the inspiration for the formation of the DMRF and inspires so many to this day with her courage. Art Kessler, immediate Past President of the DMRF, eloquently expressed the gratitude felt by everyone present. His words encapsulated the collective sentiments of appreciation, admiration, and hope for the future.

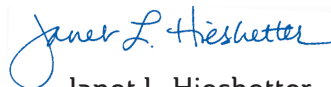
We extend our sincere gratitude to all the speakers, presenters, and attendees who contributed to the resounding success of this symposium. Your unwavering dedication and tireless efforts have significantly advanced the field of dystonia research, bringing us ever closer to the realization that a cure for dystonia is not an unattainable dream, but a tangible possibility.

We also want to thank the young investigators who traveled to Dublin early for the DMRF Emerging Leaders meeting. We had the opportunity to share a day with some of the brightest minds in the field to talk about their research, their interest in dystonia and the challenges they currently face. Witnessing the next generation of dystonia researchers forging connections and fostering a sense of camaraderie was truly inspiring. These interactions and collaborations will undoubtedly shape the future of dystonia research, fueling progress in the field.

A lay summary of the meeting is under development and will be available in the fall. Stay tuned.



Mark Rudolph
President



Janet L. Hieshetter
Executive Director



Filipa França de Barros, PhD

DMRF Awards New Grants and Fellowships to Uncover Dystonia Mysteries

DMRF's new research grant and fellowship awards aim to expand our knowledge of dystonia, develop novel therapies, and provide hope for those living with dystonia.

The Dystonia Medical Research Foundation (DMRF) supports research aimed at expanding the knowledge of the genetic, molecular, physiologic and pathologic basis of dystonia. The Foundation is proud to announce the latest research grant and fellowship awards to advance progress toward dystonia treatment options and ultimately a cure.

"The DMRF prides itself in funding the most promising research and are gratified to see important discoveries as a result," said DMRF President Mark Rudolph. "It's exciting to see these new research projects and imagine what new insights will emerge."

This year's newly funded projects cover a range of research studies that may one day lead to clinical applications that include, but are not limited to:

- **Better understanding of the brain function for more precise targets for brain stimulation methods**
- **Safer, less costly, and more accessible alternatives to deep brain stimulation**
- **New drug discoveries for DYT1 dystonia**

Congratulations to the newest award recipients and thank you to DMRF supporters for making this research funding possible.

GRANTS

DMRF's research grants support 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.



Neuroanatomical Isolation of Networks in Dystonia Through Analysis of Causal Brain Lesions

*Daniel Corp, PhD,
Deakin University, Victoria, Australia*

RESEARCH GOAL:

Dr. Corp will study the brain anatomy of dystonia by analyzing connecting links between symptoms and brain structures affected by lesions of various origins. The goal is to identify novel targets for new brain stimulation methods.

More Detail:

The brain regions responsible for dystonia remain unknown, making it difficult to use targeted treatments, such as non-invasive brain stimulation, which has been effective in other neurological disorders. While traditional neuroimaging methods can show the brain areas that are different in dystonia, they cannot show which of these actually drive dystonia symptoms. This research will use lesion network mapping to gain a better understanding of the different regions of the brain that are involved in the different types of dystonia. The goal is to provide new targets for brain stimulation with potentially better outcomes for all forms of dystonia.

This project will utilize existing brain lesions that cause dystonia to find the regions responsible for dystonic symptoms. The location of the brain lesion is extremely valuable information because it allows causal links to be drawn between that brain region and dystonia symptoms. Investigators then use a method called 'lesion network mapping' to take this a step further and map not only the location of the lesions, but also their connections in the brain. This isolates the brain regions driving dystonia symptoms with far greater accuracy.

Lesion network mapping has been applied to cervical dystonia to demonstrate that there were only two specific brain regions that were connected to all lesions causing cervical dystonia: the cerebellum and the somatosensory

cortex. As a result of these findings, researchers are conducting clinical trials testing whether 2 weeks of daily non-invasive brain stimulation to a particular somatosensory cortex site relieves cervical dystonia symptoms.

This research will expand to apply lesion network mapping to all other types of dystonia to see whether there are a core set of brain structures that are always damaged in all types of dystonia, or alternatively, whether different types of dystonia involve quite distinct brain structures. This has important implications given that treatments, such as deep brain stimulation, do not work in all types of dystonia.



Theta Burst Transcranial Focused Ultrasound as a Novel Treatment for Cervical Dystonia

*Jean-François Nankoo, PhD, University
Health Network, Toronto, Canada*

RESEARCH GOAL:

A novel non-invasive brain stimulation technique, the transcranial focused ultrasound, has the potential to be a safer, less costly, and more accessible alternative to deep brain stimulation. This project aims to explore the effectiveness of focused ultrasound in cervical dystonia.

More Detail:

Dr. Nankoo's research will investigate whether a new treatment, transcranial focused ultrasound (TUS), might reduce dystonic symptoms in patients with cervical dystonia. Previous studies suggest that cervical dystonia is a disorder involving multiple key brain structures in brain areas known as the basal ganglia and the cerebellum. Current treatment options include local injections of botulinum neurotoxin to the affected muscles and oral medications, and both have side effects. Deep brain stimulation (DBS) can be effective for relieving symptoms of dystonia in some severe cases. However, DBS involves an invasive brain surgery and further surgery for battery replacement with significant risks and costs.

There is a need for new treatments, specifically non-surgical alternatives to DBS. Transcranial magnetic stimulation (TMS), a non-invasive neuromodulation method, has been shown to improve dystonic symptoms. However, TMS cannot penetrate deep within the brain, which severely limits its efficacy for dystonia treatment. Dr. Nankoo's study will investigate the

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use of a novel form of non-invasive brain stimulation known as low-intensity transcranial focused ultrasound (TUS) on the basal ganglia and the deep part of the cerebellum to normalize brain activity and reduce dystonic symptoms in patients with cervical dystonia.

TUS applied to the scalp uses acoustic waves that can easily and painlessly penetrate deep and selectively within the basal ganglia and cerebellum to modulate brain activity. This study will carry out a treatment protocol of 5 consecutive days of TUS of the basal ganglia and the deep part of the cerebellum and evaluate the effects on brain connections and dystonic symptoms. It is the first to use TUS for non-invasive neuromodulation in dystonia, and will provide crucial information to plan larger and longer-term treatment studies using TUS for different types of dystonia.



Identify Lead Compounds That Can Rectify the Aberrant Chaperone Localization to Restore Cellular Quality Control

*Christian Schlieker, PhD,
Yale University, New Haven, Connecticut*

RESEARCH GOAL:

Dr. Schlieker has been developing advanced molecular and cellular methods to develop new pharmacological approaches that may prevent neuronal dysfunction in DYT1 dystonia.

The aim of his current study is to select specific compounds that may potentially be developed into dystonia drugs.

More Detail:

Only limited treatment options are presently available to alleviate symptoms of DYT1 dystonia, the most severe and most common form of congenital dystonia. And there is no drug on the market that reduces the risk of disease onset in carriers of the mutation. Only about 30% of allele carriers develop the disease, usually in childhood or early adolescence. Dr. Schlieker's research will offer insight into the cellular pathology resulting from the DYT1 (also known as Torsin) mutation to better understand which defects trigger the disease. Investigating the cellular pathology is essential for developing pharmacological strategies that mitigate the risk of disease onset.

Investigators have identified the molecular defect underlying DYT1 dystonia—the mutation results in a defective TorsinA protein. On a cellular level, Torsin activity is required for

building nuclear pore complexes (NPCs), essential channels that are responsible for transporting cargo in and out of the cell nucleus. In cells with mutations in Torsins, NPC assembly is reduced, particularly in neurons. Thus, defective transport is one problem in neurons in which Torsin function is compromised.

Investigators have found a second and likely more severe consequence of this failed assembly, in which NPC subunits accumulate in small herniations in nuclear envelopes (NE) and develop highly toxic properties. These structures have spongelike properties in many dystonia models. Defective proteins, which normally degrade, are instead sequestered in the herniations, leading to aberrant protein accumulation in the cell. Also, molecular chaperones essential in safeguarding balanced protein development in the cell are also captured by these toxic structures, preventing them from performing their critical cellular roles.

Dr. Schlieker's research team will use tools for monitoring NPC assembly, chaperone localization, and protein accumulation to develop new pharmacological approaches that disrupt the cascade leading to neuronal dysfunction. Investigators' immediate goal is to identify lead compounds with drug-like properties that can be further improved to reduce toxicity. The long-term goal is to develop drugs that reduce the risk of disease onset in DYT1 dystonia by counteracting hallmarks of cellular dystonia pathology.

RESEARCH FELLOWSHIPS

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.



The Role of Direct and Indirect Basal Ganglia Pathways Engaged in Skilled Forelimb Movement in a Mouse Model of DYT1 Dystonia

*Filipa França de Barros, PhD,
Fundação D. Anna de Sommer
Champalimaud, Lisbon, Portugal*

RESEARCH GOAL:

The aim of the project is to quantify and manipulate the brain activity during dystonic movements in genetically engineered

mice. The results should enable targeting specific neurons in the basal ganglia to develop more efficient therapies.

More Detail:

Dystonic patients exhibit abnormal movements that worsen when movement is initiated. The striatum, a major input structure of the basal ganglia, plays a central role in the control of voluntary movement. Inputs provided to striatal medium spiny neurons (MSNs) are integrated by two parallel pathways—the direct pathway (D1-MSNs) and the indirect pathway (D2-MSNs).

Both pathways are complementary and simultaneously active during movement initiation, but their exact contributions to normal movement remain a topic of debate. Using a mouse model modeling DYT1 dystonia, Dr. França de Barros is currently studying the in vivo activity of striatal MSNs during spontaneous behavior. Preliminary results show that, after a peripheral nerve lesion that induces dystonic movements, DYT1 mice exhibit a significant increase in the activity of D1-MSNs when initiating a movement. However, it is still unknown if this imbalance is general or only present during dystonic movements. A potential causal role of this change in the activity of D1 and D2-MSNs in dystonia remains to be assessed. The main aim of the project is to quantify and manipulate the striatal activity underlying a dystonic forelimb movement in DYT1 and wild-type mice. Determining the changes that occur in the striatal activity will bring investigators closer to revealing DYT1 dystonia's pathophysiology. It will also open new scientific questions regarding how basal ganglia pathways could be targeted to produce more efficient therapies for dystonic patients.



***Mahlon DeLong Young Investigator Award
Spike-Triggered Adaptive Closed-Loop
Cerebellar Deep Brain Stimulation (DBS)
For Dystonia**

*Linda Kim, PhD,
Baylor University, Houston, Texas*

RESEARCH GOALS:

Dr. Kim will study the unique dystonia-initiating neural signals in the cerebellum. Such signals can serve as biomarkers for triggering an adaptable closed-loop deep brain stimulation response to restore normal movement with high precision.

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Glossary of DMRF Grant Research Terms

Basal ganglia – The basal ganglia are a group of neuronal structures deep in the brain known to be critical for movement control and other executive functions, behaviors, and emotions.

Cerebellum – The part of the brain in the back of the head between the cerebrum and the brain stem. The cerebellum controls balance for walking and standing, and other complex motor functions.

DYT1 dystonia – DYT1 dystonia is one of the most severe and most common forms of congenital dystonia. It is caused by a mutation in a gene encoding TorsinA, a protein that is important for the normal function of nerve cells in the brain. In patients with DYT1 dystonia, TorsinA does not function properly, which can result in involuntary muscle contractions, twisting of specific body parts such as an arm or a leg, tremors, and other uncontrolled movements.

Lesion – A type of damage or abnormal change in the brain, caused by a disease or injury. The abnormality is typically seen on a brain-imaging test, such as magnetic resonance imaging (MRI) or computerized tomography (CT).

Somatosensory cortex – A region of the brain that is responsible for receiving and processing sensory information from across the body, such as touch, temperature, and pain.

Transcranial focused ultrasound – Non-invasive neuromodulation technology that uses ultrasonic waves to heat or ablate tissue and can focus on deep brain regions.

Visit dystonia-foundation.org for more.

More Detail:

Conventional deep brain electrical stimulation (DBS) treatments in people with dystonia target brain regions that are not optimized for dystonia-specific networks. These treatments fail to consider how abnormal cerebellar neural signals influence the onset, progression, and severity of dystonic motor symptoms in real time.

With dystonia encompassing multiple motor dysfunctions, such as twisting postures, co-/over-contractions, abnormal gait, and initiation and progression of tremors, it is unclear how dystonia-inducing cerebellar miscommunications progress over time to result in multiple symptoms of varying intensity and affecting different body parts. Different aspects of motor symptoms in dystonia likely involve distinct miscommunication that arises from particular neural signatures in the cerebellum. Dr. Kim's research will explore exactly how these miscommunication errors in the cerebellum drive dystonic postures and/or tremor with different onset and severity. This information is needed to correctly dose DBS treatment in real time according to symptom(s) that are present. An ideal treatment would kick in only when symptoms are present and terminate when it is no longer needed. Such an approach would help to minimize any potential side effects of having unnecessary overdose of treatment.

To address this gap in knowledge, Dr. Kim will be leveraging the sensitivity and location of the cerebellar nuclei in a mouse model of dystonia to improve the effectiveness of cerebellar deep brain stimulation treatment. By artificially activating cells using light, researchers can start disentangling the intertwined neural miscommunications with more refined precision. Understanding the relationship between abnormal cerebellar miscommunication and dystonia onset, progression and severity over a long-term observation can help define biomarkers for optimizing treatment options for people with dystonia.

Current DBS treatments are continuously applied and often lead to side-effects that result from being "overdosed". Instead, Dr. Kim proposes to use a real time, progressive dosing of cerebellar electrical stimulation that monitors dystonic symptoms with millisecond precision. This research will address a major gap in establishing dystonia-specific neural signals that drive different motor symptoms in dystonia and allow for a customized, self-controlled treatment.

Dr. Mahlon DeLong, DMRF Scientific Director emeritus, has made pioneering discoveries in the neuroscience of movement and movement disorders. To honor Dr. DeLong's lifetime achievements and service, the DMRF established **The Mahlon DeLong Young Investigator Award, which recognizes and supports the outstanding research of an early career PhD or MD investigator.*

DMRF Clinical Fellowships Support Training Movement Disorder Specialists

Despite advances in clinical management and research, there is an urgent need for better understanding of dystonia in its various clinical manifestations and for the development of better treatments. To help meet the demand for more experts in dystonia, DMRF offers one-year clinical fellowships to support the training of exceptionally qualified physicians in preparation for their clinical and/or research career in movement disorders with emphasis on dystonia and a special competence in the use of botulinum toxins.

The 2023 DMRF Clinical Fellow



Mariel Pullman, MD
Icahn School of
Medicine, Mount Sinai,
New York, NY
Mentor:
Susan Bressman, MD

Dr. Pullman's fellowship will include mentored involvement in research, extensive clinical exposure to the broad range of dystonia subtypes and all aspects of diagnosis and clinical management to establish her as a clinician and clinical-translational researcher specializing in dystonia.

In 2023, the clinical fellowship program is supported by grants from Merz Therapeutics and Ipsen Biopharmaceuticals.

Service Dogs Can Change Lives For Those With Dystonia

Maddie Paolero and Dee Linde share how their dogs, JJ and Murphy, bring independence and companionship to their lives. Learn if a service dog may be right for you and what it takes to find and care for one.

At age 5, Maddie Paolero was diagnosed with early-onset generalized dystonia. “I started noticing when she was 2 or 3 years old that she was tiptoeing a lot,” said her mother, Beth Paolero. “When her tiptoeing didn’t improve with orthotics, they first said she had a mild case of cerebral palsy, and it wasn’t progressive. They were wrong.”

The Paolero family was introduced to the idea of getting a service dog for Maddie at a DMRF family symposium in Chicago when they saw a demonstration by Janelle Dörner and her service dog, Kramer. “Maddie wasn’t in a wheelchair full time yet, but we knew her dystonia was progressing and thought a service dog might help,” Beth said. Little did they know that Maddie would one day be the one demonstrating her service dog at dystonia conferences and bringing her dog to Washington, D.C., to meet with senators and advocate on behalf of the dystonia community.

Maddie got her first dog, Reno, a Labrador/Golden Retriever mix, when she was 13 through Canine Companions. At 29, she’s had her current dog, JJ, also a Lab/Golden mix for about four years. Maddie credits much of her independence to Reno and JJ. JJ even attended Rhode Island Community College with Maddie, who recently earned her medical insurance coding certificate.

His ability to “get” is the most important task JJ performs for Maddie who uses a wheelchair and struggles with fine motor control. When she drops things, JJ is there to pick them up for her. He performs many other physical tasks for Maddie, but his ability to make her smile is a bonus benefit. “He’s a good snuggler, and he’s funny,” Maddie said. “He likes to play and sing with a ball in his mouth.”

What to Expect with the Application Process

Service dog organizations have similar application processes and can take up to two years from initial application to bringing a service dog home. (See accompanying article for a list of certified service dog organizations.) The first step is to



JJ assists Maddie Paolero during her physical therapy sessions.

submit an online interest form through the organization’s website. If you qualify, they will send a more detailed application, which asks for information from your doctor, inquires about the home setting, and other details.

When Maddie and Beth applied with Canine Companions, they conducted a phone interview to learn more about Maddie’s specific needs, tasks they’d like the dog to perform, and the kind of temperament they want in the dog. “Do you want it to be goofy and playful or more reserved? Maddie wanted a dog that licks a lot, but not everyone wants that,” Beth said.

“You have to wait to see if you’re going to be invited to team training, which happens four times a year for Canine Companions,” she said.

Because they live in Rhode Island, Beth and Maddie were invited to Canine Companion’s Northeast Training Center in Medford, NY, to meet and train with their dog. Team training lasts for two weeks, and Canine Companions, like most reputable service dog organizations, covers all accommodations.

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"The first few days you work with a few different dogs, but by day three you get your pre-match," Beth said. The newly matched service dog then goes everywhere with their person, including dorm rooms at night so they can bond with the dog. During the training, Beth and Maddie learned 50 different commands and went on field trips with the dog to learn how to work with him in public. "We went to a mall food court, and the dog is not supposed to eat the food that is thrown under the table," Beth said. "You learn that dogs aren't robots and what to do when they do something wrong. You really feel confident when you leave there after the two weeks of intense training."



Dee Linde got her service dog, Murphy, through America's VetDogs.

A Service Dog's Life

Dee Linde, who has tardive dystonia, went through a similar process with her service dogs from America's VetDogs. She had her first service dog, a black Labrador, Violet, for five years until the dog had to retire early because of allergies. Dee got her current dog, a Golden Retriever, Murphy, in April last year.

Canine Companions and America's VetDogs, like many service dog organizations, breed and train the dogs from puppyhood. They typically use Labradors, Goldens and often a mix of the two. "Goldens have a really soft mouth and are a little goofier, and Labs are very focused," said Beth, who has become a volunteer puppy raiser for Canine Companions.

Puppy raisers train the dogs general obedience and socialization in their homes for the first 1.5 years. The dogs then go through more advanced training for another six months before being matched at about age 2. They usually start thinking about retiring the dog around age 10, younger if the dog can no longer do the tasks for various reasons.

"It's a big responsibility. It's not like having a pet where you can just have it out in the yard and let it wander off and do whatever it wants to," Dee said. "You have to be with the

dog at all times, take him out on leash when he needs to go to the bathroom. You need to keep up his daily routine and feeding schedule. It is an important part of being a team."

Reputable service dog organizations keep close tabs on the dog's health and provide ongoing support and training to service dog families throughout the life of the dog. Owners are expected to take their dogs to regular veterinarian checkups where the vet fills out a report for the organization ensuring the dog is a proper weight, nails trimmed, coat groomed, and generally healthy.

Physical Tasks and Emotional Benefits

Murphy does many physical tasks for Dee, like picking things up, getting her phone, and pushing the handicap buttons to open doors. She no longer needs to use a cane or walker because Murphy helps her balance, especially when she goes up and down stairs.

"It's hard to say what he does for me because I don't think about it anymore. He just does it. He's an extension of me," Dee said. "He's so bonded to me and so dedicated. He just looks at me like, 'What can I do for you next?' He's wonderful."

JJ is particularly helpful for Maddie when it's shower time. The dog turns on the bathroom light, helps her remove her clothes, and even puts them in the hamper. "He makes me more independent," Maddie said. JJ provides peace of mind for Beth too because she can leave Maddie with JJ if she needs to leave the house or be in separate meeting room at church, for example.

Both JJ and Murphy know some extra commands to assist in high-anxiety situations for Maddie and Dee. When JJ hears the command "cover", he lays across Maddie's body to provide comfort with his body weight. This also helps when she's on the ground doing physical therapy.

Murphy understands “center” and will stand in front of Dee between her and another person to give her some personal space when they’re talking. One of Dee’s favorite commands is “rest”, which she often uses at medical appointments to help ease her anxiety. The dog will rest his head on Dee’s leg or hand, so she can stroke his soft ears. “It’s extremely calming,” she said.

Along with the physical help a service dog provides, the companionship and responsibility of handling a service dog have tangible benefits for people with dystonia. “It’s a distraction, something else to think about and care for,” Beth said. “The social crutch of the dog is nice sometimes too. Instead of being seen as ‘the girl with the wheelchair,’ she’s ‘the girl with a really cool dog.’”

Maddie and Dee’s service dogs have become an invaluable part of their lives, but there is much to consider. They offer these tips for others with dystonia who might benefit from a service dog:

Do your research. Make sure the organization is accredited by Assistance Dogs International (ADI). Their website, assistance-dogsinternational.org, has a member search that can help you narrow down options according to your specific needs and location. These are nonprofit organizations that should not charge you for the dogs or their training. In some cases, even medical bills for the dog may be covered. For Dee, Murphy is registered with the U.S. Department of Veterans Affairs as a dog of record and is considered a prosthetic. The VA pays for insurance that covers all medical bills for the life of the dog.

Start the application process early. It can take 6 months to two years to get a service dog, depending on the organization. “If you’re even thinking about getting a service dog, go ahead and sign up,” Beth said. “It’s a long process, long waiting lists, and you can always back out or postpone if circumstances change.”

Know your ADA rights. Because these dogs are officially certified as service dogs, they should be allowed to go anywhere with you. “Even restaurants,” said Dee who had a bad experience with a restaurant owner not wanting her to bring Murphy in. You need to fill out a special form 48 hours before air travel, so planning is key. “Most airlines are very agreeable as long as it’s a legitimate service dog,” she said.

The physical tasks a service dog can perform for someone with dystonia can open a whole new world of independence and possibility. The emotional benefits are a very welcome side effect too. “Murphy’s always smiling,” Dee said. “He fills my heart with so much joy and love. You look at him and can’t help but feel happy.”

EXPLORE SERVICE DOG ORGANIZATIONS

Many reputable service dog organizations can assist people living with dystonia, but here is a short list of the more commonly used programs. There should be no cost to be paired with a service dog from these organizations.

America’s VetDogs | vetdogs.org

America’s VetDogs provides service dogs to enhance mobility and renew independence to veterans, active-duty service members, and first responders with disabilities. The training center is based in Smithtown, NY.

Canine Companions | canine.org

The largest service dog organization, Canine Companions has provided more than 7,300 highly trained service dogs to people with disabilities. The organization has six regional training centers throughout the U.S.

Dogs for Better Lives dogsforbetterlives.org

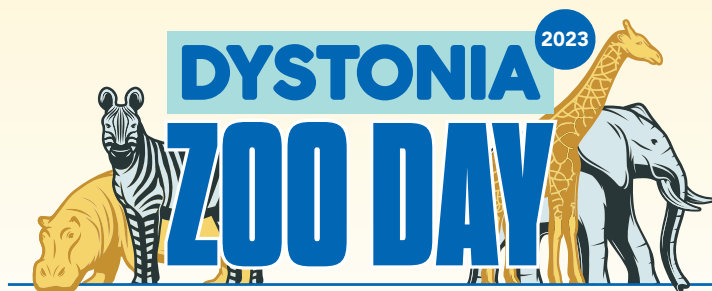
Dogs for Better Lives is one of the few national organizations that trains shelter dogs to become service dogs—in addition to utilizing purpose-bred dogs. Campuses are in Central Point, OR and East Falmouth, MA.

Freedom Service Dogs freedom servicedogs.org

Founded in 1987, Freedom Service Dogs has grown to become one of the leading service dog organizations in the country. The training and breeding facility is in Englewood, CO.

NEADS® World Class Service Dogs NEADS.org

NEADS began as the original hearing dog program and expanded to service dogs in 1987. Its facility is based in Princeton, MA, and 95% of puppies are trained by prison inmates.



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



IN-PERSON ZOO DAYS

Congratulations to Shanna and Brad Schmitt for kicking off the 2023 Zoo Days season with a successful Twin Cities event on June 24, 2023! We thank Dee Linde, Erinn Stimson, and Judith Mason for organizing Portland's Dystonia Zoo Day on July 29. More information is on page 17.

Get ready to embark on a wild experience as Dystonia Zoo Days comes to a zoo near you! Bring your family, friends, and a sense of adventure as we invite you to join us for an extraordinary event that combines the power of community, the wonders of nature, and the drive for a cure. It's going to be a roaring good time!

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

- Fort Wayne, IN – August 6
- Long Island, NY – August 19
- St. Louis, MO – August 27
- Bridgeport, CT – August 27
- Indianapolis, IN – September 9
- Pittsburgh, PA – September 17
- Toledo, OH – September 23
- Boston, MA – September 30
- Chicago, IL – October 7



4TH VIRTUAL DYSTONIA ZOO DAY

**OCTOBER 14, 12 PM Eastern
11 AM Central • 9 AM Pacific**

Join us for Virtual Dystonia Zoo Day, an online gathering that will unite the dystonia community from near and far. Together, we'll raise awareness, support groundbreaking research, and pave the path towards a cure for dystonia. The program will feature special guests including a zookeeper chat, inspiring stories, research Q & A, and more. This event is free for all to attend, but for those who want to showcase their support, we offer a limited-edition t-shirt for just \$15.

Sign up at dystonia-foundation.org/letszoothis

MORE DYSTONIA EVENTS

Don't miss these opportunities to re-unite the dystonia community, promote awareness, and support the mission to find a cure.

- Dystonia Awareness Month – September
- Angels Walk for Dystonia, Sewaren, NJ – September 23

***For more information or to register for an event, please visit
dystonia-foundation.org/events***

SEPTEMBER IS DYSTONIA AWARENESS MONTH

What are you doing for Dystonia Awareness Month?

Share your photos, videos and ideas with the DMRF at awareness@dystonia-foundation.org

**There are various ways you can contribute to promoting recognition
during Dystonia Awareness Month this September:**

☒ **Share your story**

Whether you've experienced dystonia or know someone who has, sharing your personal journey on social media, through interviews or at local events can make a powerful impact.

☒ **Wear blue for dystonia**

Encourage others to participate in "Wear Blue for Dystonia" campaign by donning blue clothing or accessories throughout Dystonia Awareness Month. Share pictures on social media using relevant hashtags to increase visibility and engagement. #DystoniaAwareness #DystoniaAwarenessMonth #DMRF

☒ **Educate others**

Spread knowledge about dystonia by sharing informative resources, facts, and articles on social media. Help dispel misconceptions and promote accurate understanding of the condition. The DMRF provides free educational publications, newsletters, fact sheets, and brochures. Email us at awareness@dystonia-foundation.org to order materials.

☒ **Connect with support groups**

Engage with local dystonia support groups to foster a sense of community. Attend meetings, share experiences, and provide support to individuals and families affected by dystonia. Visit the dystonia-foundation.org for information about joining a support group in your area or online.

☒ **Advocate for dystonia**

Contact the DMRF to receive information on advocacy issues and then reach out to local representatives, healthcare providers, and policymakers to raise awareness about dystonia and advocate for increased funding, improved access to treatment, and better support systems for those with dystonia.

Remember, even small actions can make a significant impact in raising awareness about dystonia.

Together, we can spread knowledge, support research, and build a supportive community for those affected by this condition. For more information, visit dystonia-foundation.org/get-involved/dystonia-moves-me



Mental Health Care Provider Overview

It is not uncommon for some people with dystonia to also suffer with depression and/or anxiety. Knowing where to seek help can be overwhelming and sometimes a hindrance to getting help. To break down that barrier, DMRF spoke with Laurie Strawgate-Kanefsky, PhD to explain the various types of mental health providers and offer some critical first steps for those living with dystonia who may be facing these health concerns.

Dr. Strawgate-Kanefsky has a personal history with dystonia, and she's been a mental health professional for decades. She's the oldest of four sisters and is one of three who have the dystonia gene. While she's not symptomatic, she knows the struggles of living with dystonia through her nephew who was diagnosed at age 7. Dr. Strawgate-Kanefsky has a master's degree in social work from Columbia University and her doctorate from NYU. As a medical social worker, she helped dialysis and kidney transplant patients deal with the psycho-social side effects of chronic illness and medical trauma, very familiar territory to many with dystonia.

Whole Health Approach

Dr. Strawgate-Kanefsky is a big proponent of the whole health approach—treating patients' physical well-being along with their mental health. "If you

can support the patient's mental health, they're more likely to do what they need to do to address their dystonia symptoms," she said.

In an ideal world, doctors would take the initial step to address a dystonia patient's mental health by screening them during their regular appointments, asking them questions, and referring them to a mental health provider. Unfortunately, this isn't always the case, and individuals with dystonia may need to initiate the process. With this in mind, Dr. Strawgate-Kanefsky offers a few tips dystonia patients can take to advocate for their own mental health.

Share your feelings and ask for help. "You need to communicate. You need to be open, and you need to share information," she said. Whether you talk

to a family member, a friend or your doctor, you may need to be the one to first recognize that you need mental health assistance and ask for help.

Look for a mental health provider in a pain management department.

Pain, a common problem for many with dystonia, may contribute to mental health concerns. Dr. Strawgate-Kanefsky says most teaching hospitals have pain management departments. She suggests asking your neurologist to refer you to a mental health professional in the pain management department because they specialize in dealing with chronic pain.

Mental health professionals may not know about dystonia, but help them learn. Don't expect therapists to know what dystonia is at first, but you can usually tell in your first consult if they

seem open to learn and help you with the challenges. “Once they know what it is and what the issues are, any competent therapist would be able to partner in the work that person needs to do,” Dr. Strawgate-Kanefsky said.

The DMRF has a dystonia resource packet you can share with your mental health provider available on its website: dystonia-foundation.org under the tab *Living With Dystonia*.

Find a therapist you trust. Meeting with a mental health professional should feel comfortable and is an ongoing relationship that develops over time. “It’s all about the relationship with your therapist. If that person doesn’t feel right, find someone else,” she said.

Start with talk therapy. For dystonia patients, Dr. Strawgate-Kanefsky recommends starting with talk therapy first with a counselor, social worker, therapist, or psychologist. Although some psychiatrists offer talk therapy, they are more geared toward prescribing psychotropic medications. “Especially with dystonia, adding more drugs is probably not what you want to do first,” she said. However, if behavioral therapies don’t seem to be moving the needle, then medications prescribed by a mental health professional you trust may help.

Approach the right type of mental health provider. There is currently a high demand for mental health assistance and a shortage of professionals in this post-pandemic era. Having a basic knowledge of various types of mental health professionals can help you narrow down options and avoid sitting on waiting lists with the wrong type.

TYPES OF MENTAL HEALTH PROFESSIONALS

This list of mental health professionals should help you navigate which type of provider may be the best fit for your mental health needs. It is always important to inform your physicians of all your healthcare providers and medications you are using for all health conditions, including over the counter and herbal remedies.

The following mental health professionals can provide counseling and with proper training, assessments; however, cannot prescribe medication. They are trained to provide individual and group counseling:

Clinical Social Worker – Counselor with a master’s degree. Trained to make diagnoses, provide counseling, and provide case management and advocacy; usually found in the hospital setting.

Licensed Professional Counselor – Counselor with a master’s degree in psychology, counseling, or a related field.

Mental Health Counselor – Counselor with a master’s degree and several years of supervised clinical work experience.

Nurse Psychotherapist – Registered nurse who is trained in the practice of psychiatric and mental health nursing.

Marital and Family Therapist – Counselor with a master’s degree, with special education and training in marital and family therapy.

Pastoral Counselor – Clergy with training in clinical pastoral education.

Peer Specialist – Counselor with lived experience with mental health or substance use conditions. Assists clients with recovery by recognizing and developing strengths and setting goals.

The following mental health professionals have doctoral degrees:

Clinical Psychologist – A psychologist is trained to make diagnoses and provide therapy. However, they cannot generally prescribe medications in most states.

Psychiatrist – A medical doctor with special training in the diagnosis and treatment of mental and emotional illnesses. A psychiatrist can prescribe medication, but they often do not counsel patients.

Child/Adolescent Psychiatrist – A medical doctor with special training in the diagnosis and treatment of emotional and behavioral problems in children.

Source: Mental Health America, mhnational.org



CLC Chair Beth Farber Carries on Family Legacy of Dystonia Advocacy

Beth Farber, DMRF Community Leadership Council chair, has known dystonia for as long as she can remember. Beth was 5 when her sister, Shari, who was 7 at the time, began walking with a limp because her right foot turned inward. Her parents took Shari to multiple doctors all over the United States to try to figure out what was wrong.

Being the early 1970s, most doctors were unaware of dystonia and misdiagnosed her with everything from “attention seeking” to cerebral palsy. One even blamed Shari’s dystonic movements on the water in Florida where they lived at the time.

Within a year, Shari was wheelchair bound because the entire right side of her body began to twist inward and spasm. The family had moved back to the Chicago area and continued to try to find help for Shari. Finally, they met with a doctor in Bronx, N.Y., who diagnosed Shari with dystonia. “My parents were so relieved to finally have a name for this,” Beth said.

They had a name for her condition, but they found themselves with more questions, needing support, and nowhere to turn. Determined to help others in the dystonia community with very similar stories, Beth’s parents, Joel and Harriett Farber, founded the Dystonia Foundation in Illinois, which later merged with the Dystonia Medical Research Foundation.

From a very early age, Beth was involved alongside her parents to raise dystonia awareness, fundraise for research and help other families dealing with dystonia. Beth was a founding member of DMRF’s Community Leadership Council (CLC) and has been chair for the past three. Sadly, Beth’s sister, Shari, and parents have passed, her father most recently in May last year. “I have to continue what my parents started. Whatever it takes to help other families like mine,” Beth said.

The CLC is a group of community leaders from across the United States who work with the DMRF Board of Directors and staff to achieve the foundation’s mission. Like Beth, CLC

DMRF Community Leadership Council

Beth Farber, Chair
Ginny Bryan
Kristin Cinglie
Jacquelyn Coello
Karen A. Flanagan
Jillian Johnson
Dee Linde
James Metherell
Melissa Phillips
Shanna Schmitt
Rosemary Young

members have a personal connection to dystonia and are committed to raising funds for DMRF programs; raising awareness of dystonia; advocating for legislation or policy changes that reflect the needs of the dystonia community; and working to support those who are affected by dystonia and their families. Currently, 11 CLC members meet virtually quarterly to discuss fundraising ideas and how to continue to raise awareness about dystonia and support the foundation’s mission. “We all have different perspectives on dystonia,” Beth said. “When we meet together, we learn from each other’s experiences.”

The CLC is central to the success of DMRF network fundraising events, like Zoo Days and Petpalooza, and each CLC member manages events in their own communities. Beth’s family organized annual charity prize raffles (Chicago Basket Bash) for 14 years and two scavenger hunt fundraisers (Hunt for a Cure), with funds going to the DMRF in honor of Shari.

Between the ages of 10 and 13, Shari had five brain surgeries. She was able to walk without a wheelchair but had to sacrifice her speech because of the surgeries. Shari attended mainstream school, went to college, became an avid horse rider, traveled internationally, and got married. She and her husband, Ira, were featured in a PBS documentary, *Twisted*, which premiered in 2007. “My sister got her toughness from my mother. She had to fight for everything,” Beth said. Beth understandably gets choked up talking about her parents and sister, who died tragically in an accident in 2010. As CLC chair, she continues to fight for a cure for dystonia hoping one day others won’t have to deal with the same pain and hardship her sister did.

People on the Move

The DMRF is grateful for the generosity, hard work, and commitment of each volunteer involved in these events. Their efforts have not only raised funds for medical research but also raised awareness and brought communities together to support individuals with dystonia. Thank you for making a difference and being champions in the fight against dystonia.

Twin Cities Dystonia Zoo Day

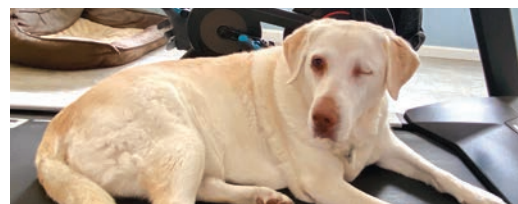
The DMRF applauds Shanna and Brad Schmitt for their dedication in spearheading the highly successful Twin Cities Dystonia Zoo Day as part of the Zoo Days kick-off event. Additionally, the DMRF expresses its gratitude to Governor Tim Walz for proclaiming September as Dystonia Awareness Month as part of the festivities. Moreover, the DMRF extends thanks to Dr. Stephanie Standal for graciously sharing her expertise and speaking at the event. The collective contributions of Shanna, Brad, and their collaborators have made a significant difference in raising awareness about dystonia in the Twin Cities area.

Portland Dystonia Zoo Day

The DMRF congratulates Dee Linde, Erinn Stimson, and Judith Mason for their exceptional work in organizing the Portland Dystonia Zoo Day. Through their efforts, they have made significant contributions to educating the community and fostering support for individuals affected by dystonia.

Nashua Dystance 4 Dystonia

The DMRF extends its heartfelt appreciation to Amanda Lockhead and her family for their exceptional organization of the Nashua Dystance 4 Dystonia event in Nashua, New Hampshire. The unwavering dedication and commitment displayed by Amanda, her husband Jim, and their two children in raising awareness for dystonia is truly inspiring.



10th Pet Palooza Anniversary

The DMRF extends its thanks to Ginny Bryan, Jacquelyn Coello, Denise Gaskell, and Dee Linde for their efforts in planning the 10th Pet Palooza Anniversary. Their hard work and dedication have resulted in an impressive fundraising total of over \$24,000, which will greatly contribute to dystonia research and improving the lives of those affected by this condition. The DMRF would also like to give special acknowledgment to Haley, Terri Chapman's remarkable pet, for raising over \$5,000 as the "Top Dog." As part of the month's festivities, Janice and Len Nachbar were recognized by the Pet Palooza Committee for their efforts in starting Dogs for Dystonia in honor of their daughter Joanna Manusov and her beloved pets Nicki and Princess.

Dystonia Editorial Board Meets in Dublin

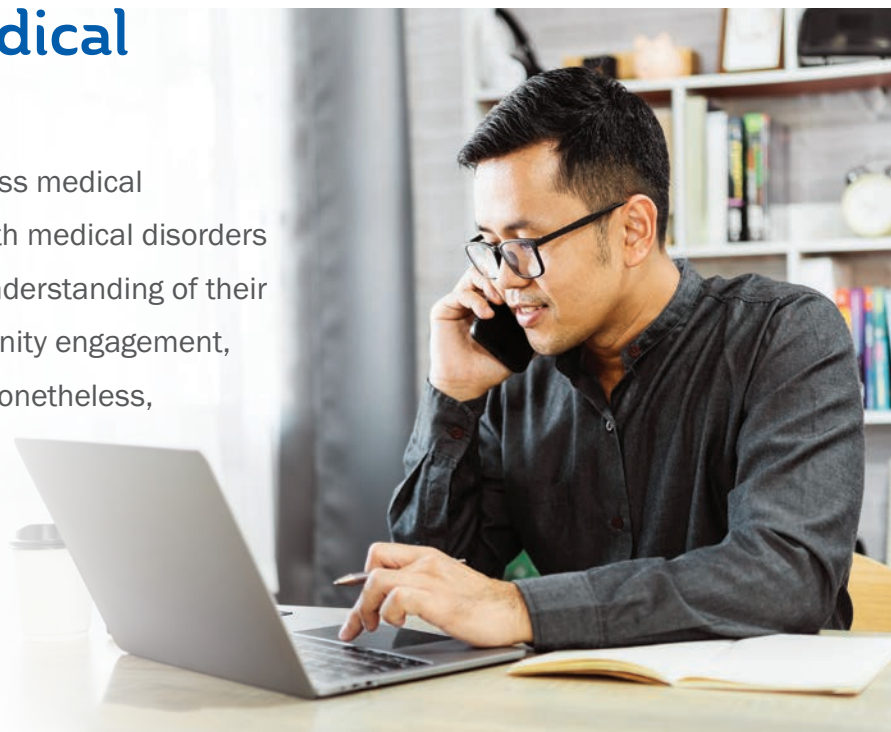


Anthony Lang, MD; Joseph Jankovic, MD; Alberto Albanese, MD; Cynthia Comella, MD; Alfonso Fasano, MD, PhD; Sanjay Pandey, MD; Roy Sillitoe, PhD; Kristina Simonyan, MD, PhD; Jan Teller, PhD

The Editorial Board of the DMRF's journal, *Dystonia*, convened in Dublin on June 1, 2023, for its inaugural in-person meeting. The purpose of the meeting was to review the great progress made by this scientific publication in its first year. The enthusiasm among all attendees was palpable, reflecting their strong dedication and support for the journal. *Left to right:* Mark Hallett, MD; Aasef Shaikh, MD, PhD; Amy Field, *Frontiers*; David Standaert, MD, PhD;

Safely Search for Medical Information Online

The internet has revolutionized how we access medical information, providing individuals dealing with medical disorders with invaluable resources to deepen their understanding of their condition. It serves as a platform for community engagement, support, and the exchange of information. Nonetheless, it is crucial to develop effective strategies to discern reliable medical information from the abundance of misinformation available online. Below is information to help you identify sources of accurate, balanced information.



Ask yourself these questions:

1. What is the source of the information and is it reliable?
2. Are there references or citations provided to support the information presented?
3. Is the information up-to-date and in line with current medical knowledge?
4. Are there multiple reliable sources that corroborate the information?
5. Does the information come from healthcare professionals or reputable medical organizations?
6. Are there any potential conflicts of interest, such as sponsored content or advertisements that may influence the information provided?
7. Does the website or platform have a moderation or fact-checking process to ensure the accuracy of the information?
8. Are the claims made supported by scientific evidence or research studies?
9. Are there any red flags or sensationalized language that may indicate unreliable information?
10. Does the website provide information on the author's qualifications or expertise in the relevant medical field?

Find Familiar Names

Follow the organizations or groups you're familiar with as a starting place for seeking out reliable materials, up-to-date and accurate information, treatment options, and ongoing research. Noteworthy examples of reliable organizations with online presences include the Dystonia Medical Research Foundation, and the National Institute of Neurological Disorders and Stroke. Online medical libraries, such as PubMed or Google Scholar, offer access to peer-reviewed scientific studies and research articles. These sources provide reliable and evidence-based information on dystonia.

Scrutinize the Sources

When evaluating online information, it is important to recognize that it may be influenced by personal, financial, or ideological factors. It is advisable to be cautious of exaggerated claims and clickbait-style headlines, which can often mislead. One helpful strategy is to examine the website's URL, as it can provide valuable insights into the source responsible for the content. URLs ending in ".gov" indicate websites owned by United States government agencies, while those ending in ".edu" belong to educational institutions. Additionally, URLs ending in ".org" typically represent nonprofit organizations. Taking these factors into consideration can aid in assessing the reliability and credibility of the information you encounter.

Helpful resources and information are available at these websites:

► **Dystonia Medical Research Foundation (DMRF)**

A leading organization dedicated to advancing research, providing education, and raising awareness about dystonia. Their website offers comprehensive information about dystonia, treatment options, and support resources.

Website: dystonia-foundation.org

► **National Institute of Neurological Disorders and Stroke (NINDS)**

A division of the National Institutes of Health (NIH), NINDS provides reliable information about various neurological disorders, including dystonia. Their website offers resources on diagnosis, treatment, and ongoing research.

Website: ninds.nih.gov

► **Dysphonia International**

The National Spasmodic Dysphonia Association (NSDA), now Dysphonia International, is dedicated to improving the lives of people affected by spasmodic dysphonia and related voice conditions through research, education, awareness and support.

Website: dysphonia.org

► **Dystonia Europe**

An alliance of national dystonia patient associations across Europe, Dystonia Europe aims to improve the lives of people with dystonia. They provide information, support, and advocacy for patients and their families.

Website: dystonia-europe.org

► **Dystonia Medical Research Foundation Canada (DMRFC)**

DMRFC is a Canadian organization committed to funding research, raising awareness, and supporting individuals with dystonia. Their website provides information on local resources, research updates, and patient stories.

Website: dystoniacanada.org

► **Dystonia Society UK**

Dystonia Society UK is a charity that offers support and information to people affected by dystonia in the United Kingdom. Their website provides resources on different types of dystonia, treatment options, and practical advice.

Website: dystonia.org.uk

Stay Current

It is essential to check the date of online medical information, outdated information may not reflect the most current scientific consensus and could potentially be misleading or incorrect.

Cross-reference Information

It's always beneficial to cross-reference health claims found online with reputable sources. Either check websites you already trust or confirm information with professionals.

Be Mindful on Social Media

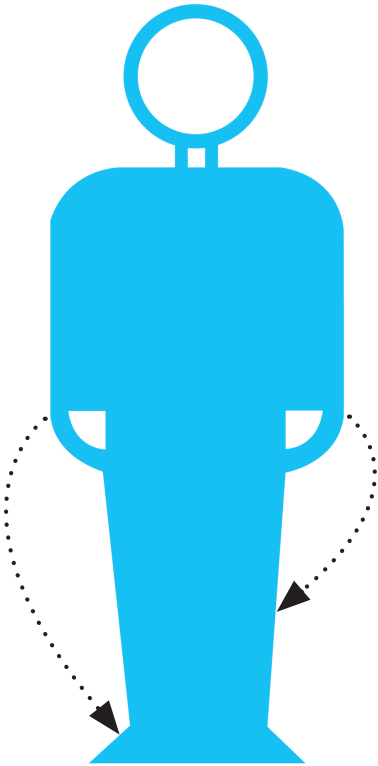
Proceed with care when engaging with social media platforms, as personal stories shared online, while insightful, may not represent the experiences of the general population

and may lack scientific evidence. Specifically, when relying on “expert” information from platforms like TikTok, exercise caution, as they may lack scientific rigor and verification. A recent study revealed that 40% of medical videos on TikTok contained misinformation. It is crucial to remain skeptical of claims promoting “miracle” cures. To ensure accuracy and reliability, it is advisable to stick to verified accounts of reputable medical organizations or professionals when seeking medical information.

Individuals with dystonia should always consult their healthcare professionals or specialists to validate and interpret the information they find online. Remember that online information should not replace consultation with healthcare professionals.

FOCUS ON

Lower Limb Dystonia



Common Symptoms of Lower Limb Dystonia

- Foot rolls outward so that the sole faces inward
- Foot or leg turns inward
- Foot points downward
- Extension of big toe
- Toe curling
- A sense of tightness or stiffness in the limb
- Changes in walking or running gait

Lower limb dystonia refers to dystonic movements and postures in the leg, foot, and/or toes. Individuals often have to adapt their gait while walking or running to compensate for the dystonic symptoms, and/or may require the assistance of mobility devices such as crutches, walkers, or wheelchairs.

Ironically, symptoms of lower limb dystonia may occur while walking forwards but improve or disappear when walking backwards. Some individuals may be able to temporarily lessen symptoms with a 'sensory trick' to another part of the body, such as gently touching the face or head.

Dystonia affects men, women, and children of all ages and backgrounds. In children, lower limb dystonia may be an early symptom of an inherited dystonia. In these cases, the dystonia may affect additional areas of the body. Children with cerebral palsy may have limb dystonia, often with spasticity (muscle tightness and rigidity). Foot turning can be the first symptom of dystonia in children and may be misdiagnosed as club foot, leading to unnecessary orthopedic procedures.

Foot turning can be the first symptom of dystonia in children and may be misdiagnosed as club foot, leading to unnecessary orthopedic procedures.

When seen in adults, lower limb dystonia seems to affect women more often than men. Age of onset is typically in the mid-40s. Symptoms may develop after periods of prolonged exercise, for example long-distance running or walking. Symptoms are often triggered by running ("runner's dystonia"), walking, or standing but may be absent at rest. Electrolyte disturbances and disorders of energy metabolism must be ruled out if symptoms occur only with exertion and fatigue.

For many adults who develop lower limb dystonia, there is no identifiable cause, and it is not associated with additional neurological disease. Known causes include brain injury, stroke, or injury to the limb. A fixed position of the foot may suggest that the dystonia was acquired because of an injury or underlying cause. Some patients develop a pain syndrome called complex regional pain syndrome (CRPS) or reflex sympathetic dystrophy (RSD) after an injury with associated dystonia.

In some cases, fixed foot dystonia is a symptom of a functional (psychogenic) movement disorder or functional neurological symptom disorder (conversion disorder). Many people with functional movement disorders experience a mix of motor symptoms including dystonia, tremor, gait disorders, and tics. They may also

experience weakness, sensory symptoms, non-epileptic seizures, and other functional neurological symptoms.

Dystonia is not a typical early symptom of Parkinson's disease but, in rare cases, lower limb dystonia appears to be an early sign of certain parkinsonian conditions. For example, toe pointing or curling triggered by exercise has been documented in individuals ultimately diagnosed with young-onset Parkinson's disease under the age of 50. Lower limb dystonia may also occur in the context of generalized dystonia, for example inherited disorders such as early-onset (DYT1) dystonia and dopa-responsive dystonia.

Treatment to lessen dystonia symptoms may require a combination of approaches. Botulinum neurotoxin injections are among the most effective therapies to help relax dystonic muscles and alleviate the abnormal movements and postures. Oral medications, such as anticholinergics, baclofen, and/or benzodiazepines may also be used. Deep brain stimulation surgery may be an option for select patients, especially for inherited dystonia which can progress to generalized dystonia. Physical therapy and/or occupational therapy may improve overall daily functioning. Part of the treatment plan may involve using orthotics to help lessen symptoms and make it easier or more comfortable to walk.

Highlights from the Samuel Belzberg 6th International Dystonia Symposium

The Symposium took place June 1–3 in Dublin, Ireland. Look for a lay summary report this fall!



Jan Teller, PhD; Joel Perlmutter, MD; Fran Belzberg; Buz Jinnah, MD, PhD; and Rich Lewis, MD



Francesca Morgante, MD, PhD and Victor Fung, MBBS (MD), PhD, FRACP



Meike van der Heijden, PhD



Selma Aybek, MD



Antonio Pisani, MD, PhD

Personal Profile Kristin Cinglie

Kristin Cinglie lives in Monroe, Michigan with her husband, Paul, and son, Dylan (27), who is studying bioengineering. With her late onset of dystonia, she's had to adjust to life not going to plan.

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

I was an avid runner prior to dystonia. While training for my second half marathon, I started experiencing issues during some of my training runs. I would get these really strange cramps in the back of my left leg. On other days, my shoulders would lift up, and I couldn't get them to relax. My head started pulling to my left shoulder, and after a while, I lost the ability to turn my head right and left. I went to the doctors, they ran tests, and everything came back fine. As you know, that's one of the frustrations with dystonia.

There was a lot going on in my life at the time, so it was easy for me to make excuses for how I was feeling. I had just gone through a divorce. I was a single mom raising my son and working full-time as a chemical engineer. Since the doctors hadn't been able to find any definitive answers for my symptoms, it was suggested that it may be due to stress or over training. At the time, that seemed like a reasonable answer.

After months of finding no relief, I continued to search for other possibilities. I probably saw 8 or 10 different types of doctors before seeing a movement disorder neurologist at the Cleveland Clinic in Columbus, Ohio. I was initially diagnosed with cervical dystonia in 2012 since most of my symptoms were in my neck and shoulders at that time. I have since been diagnosed with generalized dystonia and experience symptoms in most of my body. It is progressive, so I continue to have new symptoms all the time.

How are you managing now?

I have good days and bad days. It is challenging to live with a disorder that is so unpredictable. It makes it difficult to commit to things or make plans. I am fortunate to have a wonderful support group of people who are understanding and always willing to help. My problem is that I am not good at asking for help, so that is something I need to continue to work on.

As far as my medical care is concerned, I am being treated by the neurology team at Henry Ford in Detroit, Michigan. I receive a botulinum toxin injection every 90 days and take



Kristin Cinglie (center) with her father, Kim, and mother, Joyce Cinglie

oral medications daily to help with muscle spasms. I had DBS in October of 2020, and it has improved my balance and mobility, so I no longer rely on a cane, which is great. Unfortunately, it has not been as helpful in controlling my headaches. I still struggle with terrible headaches most days.

What has been your biggest challenge since the diagnosis?

Accepting and being at peace with the "new" me has been my biggest challenge since being diagnosed with dystonia. It was difficult to move back home to Michigan where I grew up. It was a constant reminder of how much things had changed. I relied on a cane to walk and struggled with daily tasks. I was no longer working, as I had medically retired from my career due to my illness. I tried to be the best wife and mom I could be with all that was going on, but none of what was happening was part of my plan. I felt like a shadow of my former self and constantly felt like I was letting everyone down. It was all so overwhelming. That was when the mental health aspect really kicked in for me.

How did you take control of your mental health?

It took me a long time to understand that to manage the disorder and have quality of life for myself and my family, I needed to take care of my mental health as well as my physical health. I sought out a therapist who prescribed anxiety medication for a short period of time. I needed the medication in the beginning so that I could have purposeful conversations and apply the things they were trying to teach me.

I attended therapy twice a week at first and then less frequently as I was able to better manage my anxiety. I have continued to include mental health care as part of my treatment plan over the last several years. I am thankful for the support my family has given me through this part of my journey. It can be a difficult step to take, but I would encourage anyone going through a significant life change to seek out the help they need to manage their situation. For me and my family, mental health care was a necessary part of learning to cope with the changes dystonia has brought to our lives.

How did you connect with DMRF and what advice do you have for others with dystonia?

I connected with the DMRF when I started attending Dystonia Zoo Days. I had chosen to keep my dystonia journey private for a long time, so I waited years before getting involved with the dystonia community. My advice to others would be to get involved sooner rather than later. Dystonia can be a very isolating diagnosis, and having a community of people who can relate to what you are going through can be very helpful. Attending the zoo event for the first time was an eye-opening experience. I met people with different types of dystonia and in different stages of their journey. I spoke to drug reps and medical professionals about treatment options and had the opportunity to ask questions. I looked around at a room full of people who understood what we were dealing with, and I realized that we didn't have to do this alone...I had been choosing to do it alone!

Once I decided to get more involved, the DMRF has been so supportive. With their help, I have hosted a Wacky 5K and several Dystonia Zoo Days at the Toledo Zoo. They have welcomed my son and me to participate in Virtual Zoo Days, and I recently joined the DMRF Community Leadership Council. The DMRF and the activities I am involved in have helped to give me purpose again, and I will forever be thankful for that.

Living with dystonia is a day-to-day adventure. I wish I could tie my story up with a pretty bow and say that things are great and I am living pain-free, but that just isn't my reality. I guess the "bow" for my story is that I have learned to be more present and appreciative of the good things that happen each day, and I try not to worry about the future and the things I can't control.



Discover the Power of Dystonia Support Groups

Connect, Learn, and Thrive Together!

Dystonia, a condition unfamiliar to many until their own diagnosis, doesn't have to be faced alone. The Dystonia Medical Research Foundation (DMRF) understands the importance of support, which is why we partner with dedicated volunteers to sponsor local support groups and provide online support forums.

Our support groups offer invaluable opportunities for information sharing, camaraderie, and community activism. Connect with others who understand the challenges of living with dystonia, whether through face-to-face or virtual meetings in your local area or through our 24/7 accessible online groups. No matter where you are, you can find support, understanding, and friendship with others who are going through similar experiences. Join a support group today and experience the transformative power of connection, shared knowledge, and a supportive network. Together, we can navigate the journey of dystonia with strength and resilience.

To connect with a DMRF support group, visit: dystonia-foundation.org/living-dystonia/support

Don't have a dystonia support group in your community? Consider starting one. Contact the DMRF at dystonia-foundation.org/about-us/contact or 800-377-3978.

Dystonia Dialogue

Dystonia Medical Research Foundation

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September is Dystonia Awareness Month!

Turn to page 13 to see how you can help raise awareness.

Advancing Research, Raising Awareness and Supporting Individuals and Families Affected By Dystonia

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