



DYSTONIA
MEDICAL
RESEARCH
FOUNDATION

Dystonia Dialogue

NEWSLETTER OF THE DYSTONIA MEDICAL RESEARCH FOUNDATION

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Jon Davis bikes more than 4,000 miles a year to raise awareness about dystonia and funds for research.

The Dystonia Dialogue is supported by Ipsen Biopharmaceuticals.

Correction:

The Focus On Lower Limb Dystonia article on p.20 of the *Dystonia Dialogue*, Summer 2023, incorrectly printed the symptoms headline as spasmodic dysphonia. The correct headline should be **Symptoms of Lower Limb Dystonia**.

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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Mark Rudolph
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Janet L. Hieshetter
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As we reflect on the productive year we've had at the DMRF, we are filled with gratitude to all in the dystonia community for their support of the important work of the foundation.

We could not do all that we do without the generous support of our donors. Thank you for the confidence and trust you have placed in us. Your support is essential to the DMRF's funding of crucial research, allowing for the exploration of innovative hypotheses and for the engagement of young investigators. Additionally, your giving allows the DMRF to provide the important programs of support, education, and advocacy relied on by the community.

Thank you to the many volunteers who worked to host Dystonia Zoo Days that have brought communities together to celebrate one another and send the strong message that no one needs to go through dystonia alone. You'll find some of the highlights from this year's Zoo Days beginning on p.12.

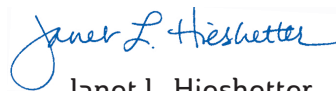
We especially appreciate the ongoing efforts of our support group leaders who work tirelessly to provide information and resources to all who reach out for assistance. These leaders inspire us to do more to improve the lives of all affected by dystonia. To our awareness warriors and tireless advocates: We say thank you for your willingness to share your personal stories to increase awareness and help us meet the needs of those affected by dystonia. One awareness warrior, Jon Davis, shares his story about biking across Scotland and the #Riding4Us campaign on p.4.

In June, the Samuel Belzberg 6th International Dystonia Symposium was a momentous achievement, and we are indebted to the Co-Chairs, Drs. Hyder Jinnah and Antonio Pisani, for their work in developing such a robust program, and to all of the speakers who contributed significantly to the success of the conference. The symposium drew people from 32 countries to discuss hot areas in dystonia research. Attendees left energized and enthusiastic about the potential for breakthroughs that we all await. What came through loud and clear is that dystonia research is a global effort, and that the DMRF is leading this effort. Learn about some of the latest on-going dystonia research on p.20.

Every member of the dystonia community has an important role in our achieving our ultimate goal—to find a cure and eliminate the need for an organization like the DMRF. As we prepare for the new year ahead, we are resolute in our resolve to reach this goal. We are not going anywhere until there is no longer a need. Thank you for your support and for joining us on this mission. Let's keep moving forward!



Mark Rudolph
President



Janet L. Hieshetter
Executive Director

Why Jon Davis Rides for Dystonia

Jon Davis has two passions in life: riding his bicycle and advocating for dystonia awareness. Davis cycled 4,000 miles last year, is on track to reach his 4,000-mile goal this year, and recently completed a 6-day bike journey across Scotland to raise awareness about dystonia and funds for research.



Photo by Mark Donovan

Jon Davis, here in the operating room with his surgeons, had deep brain stimulation (DBS) surgery in 2011.

His #Riding4Us campaign has raised more than \$17,500 for the DMRF, and he's only just begun. He plans to bike more than 4,000 miles again in 2024 and continue his #Riding4Us campaign—simply because he can.

There was a time when Davis couldn't ride a bike and even struggled to walk due to the progression of his generalized DYT1 dystonia. (See article on p.7 for more on DYT1 dystonia.)



#Riding4Us

He was diagnosed at age 13 after his teacher sent a note home concerned that she could not read his handwriting. Dystonia had affected the fine motor skills needed for writing with his left hand, so he learned to write with his right hand.

Writing with a different hand was the first of many trade-offs Davis would make because of dystonia. "Rather than fight with my body, I became a righty. A few years later, I was unable



Jon Davis (right) and his good friends, Mark Donovan (left) and Tim Nicholson, celebrate the success of his surgery every year by biking 100 miles.



Davis #Riding4Us in Scotland.

to play the cello, so I took up singing,” Davis said. “Constantly making sacrifices for this disorder defined my life as dystonia chipped away at my ability to function.”

Davis has loved biking since childhood. In his 20s, he cycled across France and Nova Scotia. At 31, he married his wife, Evelyn, and they had a son, Jack, several years later. For much of his life, he treated his dystonia with medications and physical therapy and tried to ignore his symptoms. He remembers a doctor suggesting that he might be a good candidate for deep brain stimulation (DBS), but he initially saw it as too invasive.

“I saw DBS surgery as drilling holes in my head and putting batteries in my chest, something I laughed at when my doctor mentioned it as an option when I still had the luxury of laughing,” he said.

As his dystonia symptoms continued to worsen, his treatment regimen wasn’t providing relief and medication side effects had become unbearable. After years of taking trihexyphenidyl, Davis asked his doctor to take him off the medication. He stopped taking the drug, he says, because he didn’t like the negative side effects, and he wasn’t experiencing any positive benefits of taking it for his dystonia symptoms.

“Life got bleak,” Davis remembers. “When you can’t bike, walk for much of any distance, or stand for any length of time—and you have a child to keep up with and a life to live—what seemed crazy when I was younger [DBS] became my last hope.”

A life transformed

In 2011, Davis decided to undergo DBS surgery, and he says it transformed his life. The gains post-surgery were small and incremental at first. Over time and with adjustments to the stimulation settings by his doctor, Davis regained the ability to stand with ease, walk, and write with his left hand. In 2013, he regained his lifelong joy of riding his bike.

“Despite dystonia—or because of my successful DBS treatment—I’ve been able to run my business and watch it grow several times over, ride a bike (my goal is to ride 4,000 miles each year), and live a life without the never-ending mental calculus of, ‘How do I get from point A to point B by taking the fewest number of steps?’”

Davis is incredibly grateful for his successful DBS treatment and is cognizant that his recovery is better than most. He says his body has recovered about 99% of its functionality. “Today, if I meet someone for the first time, they have no idea I have dystonia. When I tell them about my journey,

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I watch for that moment when they realize that what I can do is as close to a miracle of science as there is. I am thankful every day for what I've got," he said.

And this is why he rides for dystonia awareness and research funding—because he can. He rides for others who can't. He has been on the DMRF Board of Directors for five years and is always willing to talk with others who are considering DBS for dystonia.

Davis credits the DMRF for connecting him with others with dystonia who have undergone DBS and shared their experiences and medical contacts. The DMRF assisted with setting up his fundraising website and promoted his #Riding4Us campaign on social media. Davis put in the miles across Scotland and shared a daily video diary of his ride so the DMRF community could follow along. He also shared his story (and the QR code to his fundraising page) with fellow riders.

He rode 55 miles on the longest day of his Scotland trip and says conquering the big climbs was the most memorable. "The kind where you only see the road ahead, and ahead, and ahead, and realizing there is a loooooong way to go to get to the top. Then, at the top, the satisfaction of bombing down the hill I just conquered. One of my favorite signs was a '20% grade warning' sign. That is the kind of hill that is a steepness considered *hors catégorie* in the Tour de France."

The English translation for *hors catégorie* is "beyond categorization," which is a fitting description of Davis's dystonia journey. He has difficulty explaining the gratitude he feels after being given back something he had lost for so long—the free movement of his body. Getting this gift back is the motivation behind how Davis lives and why he rides for others with dystonia.

"I want to take advantage of the second chance I've been given. I didn't have this hardware inserted in my body only to sit on the couch," Davis said.



Photo by Mark Donovan

During DBS, the neurosurgeon asked Davis to perform certain tasks, like pointing his finger, to determine if the leads were in the right position.



Davis rode past Lochindorb Castle ruins located on an island in Lochindorb near Grantown on Spey in the Scottish highlands.

FOCUS ON DYT1 Dystonia

A form of hereditary, generalized dystonia

DYT1 dystonia, also known as early-onset, torsion dystonia, is a severe form of hereditary, generalized dystonia. Generalized dystonia refers to dystonia that is not limited to a single part of the body but affects multiple muscle groups throughout the body.



Generalized dystonia typically affects muscles in the torso and limbs, and sometimes the neck, face, and vocal cords. Patients have difficulty moving their bodies freely and controlling their body movements.

DYT1 dystonia typically begins around age 10 with the twisting of a foot or arm. Symptoms tend to begin in one body part and progress to involve additional limbs and the torso. Symptoms tend to be less severe the later in life they start and if they start in a hand or arm.

Most cases of early onset torsion dystonia are directly associated with a mutation that affects the DYT1 gene, which scientists discovered in 1997 with the support of the DMRF. In its normal state, the gene is responsible for providing the body with the genetic instructions to produce a protein called torsinA. The mutated DYT1 gene results in an abnormal form of torsinA, and this abnormal protein starts a chain reaction of biochemical processes that somehow disrupts the communication between the brain and muscles. This disruption of the nervous system leads to the debilitating physical symptoms of DYT1 dystonia.

Symptoms of DYT1 Dystonia

- Twisted postures, for example in the torso or limbs
- Turning in of the foot or arm
- Muscle spasms, with or without pain
- Unusual walking with bending and twisting of the torso
- Rapid, sometimes rhythmic, jerking movements (often myoclonic jerks)
- Progression of symptoms leading to areas of the body remaining in sustained or fixed postures

DYT1 dystonia is dominantly inherited, meaning that only one parent needs to have the mutation for a child to inherit the disorder. However, the DYT1 mutation also exhibits reduced penetrance, which means that not every person who inherits the mutated gene will develop symptoms. Only about 30% of individuals who have the DYT1 genetic mutation will develop dystonia. Research is ongoing to better understand this phenomenon.

If a person does not manifest symptoms before the age of 28 years, they will usually remain symptom free for life—even if they have the DYT1 mutation. The DYT1 mutation is responsible for about 90% of early onset generalized dystonia in individuals of Ashkenazi Jewish ancestry and up to about 50% of early onset generalized dystonia in other ethnicities.

Treatment

Individuals with dystonia are encouraged to seek treatment from a movement disorder neurologist or child neurologist with special training in movement disorders. A multidisciplinary team of expert medical professionals may be appropriate to tailor treatment to the needs of the patient.

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Genetic counselors are trained to help people decide whether getting a genetic test is right for them. They will also help interpret test results and offer counsel on the significance of the results for that family.

–Deborah Raymond, MS, genetic counselor at Mount Sinai Health System, New York City

A movement disorder specialist will develop a treatment plan that is customized to each patient. Most individuals require a combination of therapies. Treatment to lessen dystonia symptoms may include oral medications such as anticholinergics, baclofen, and benzodiazepines combined with botulinum neurotoxin injections, and/or surgical procedures such as deep brain stimulation (DBS). Research suggests that individuals with DYT1 dystonia tend to have the best outcomes from DBS compared to other forms of generalized dystonia.

Genetic testing is available to test for the DYT1 gene mutation. Individuals and families who wish to learn more about genetic testing for DYT1 and other genetic mutations associated with certain dystonias are advised to consult a genetic counselor. (See accompanying article.) “Understanding the genetic implications of DYT1 dystonia can be overwhelming,” said

Deborah Raymond, MS, genetic counselor at Mount Sinai Health System, New York City. “Genetic counselors are trained to help people decide whether getting a genetic test is right for them. They will also help interpret test results and offer counsel on the significance of the results for that family.”

Specific treatment may be needed to prevent loss of motion in the joints and/or curvature of the spine due to the dystonic postures. Complementary therapies to support overall functioning and wellness may include occupational therapy, physical therapy, speech/voice therapy, and other interventions depending on a person’s symptoms. Individuals with childhood onset generalized dystonia may have increased risk for depression, so monitoring and addressing emotional and mental health is often an important part of the treatment strategy.

Sources:

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What is a Genetic Counselor?

The first step for individuals and families who wish to learn more about genetic testing for dystonia is to consult a genetic counselor who is trained to educate families and prospective parents on the likelihood and risks of inherited diseases. Genetic testing is only available for specific forms of dystonia for which a gene mutation has been identified, like DYT1.

A genetic counselor can help individuals and families understand what testing may be appropriate for them. Testing may be suggested for adults and children who exhibit clinical symptoms that resemble the forms of dystonia for which gene tests are available. Adult biological relatives of individuals who have been identified as having a genetic form of dystonia may also be eligible for testing. Genetic testing in children who do not exhibit symptoms is generally not recommended, even if a biological relative tests positive for a dystonia mutation.

Individuals can locate a genetic counselor by consulting their movement disorder specialist or health insurance provider. **The National Society of Genetic Counselors, [nsgc.org](https://www.nsgc.org)**, can also provide contact information for genetic counselors.



DMRF Community Rocks Blue for National Dystonia Awareness Month

While the Dystonia Medical Research Foundation works to increase awareness for dystonia year-round, during National Dystonia Awareness Month in September, the DMRF intensifies its efforts with a variety of impactful campaigns. This past September, “Shine a Light on Dystonia,” “Wear Blue for Dystonia,” and Dystonia Zoo Days helped to bring attention to those living with the disorder.

This year, the “Shine a Light on Dystonia” campaign saw buildings from California to Massachusetts turn their lights blue in a show of support for the dystonia community. Iconic landmarks, including Niagara Falls; Willis Tower (formerly known as the Sears Tower), Tribune Tower, and other buildings on the Chicago skyline; Buffalo’s Peace Bridge; Market Square in Rapid City, S.D.; bridges across Massachusetts; the San Diego Convention Center; and city halls in Houston, Baltimore, and Warren, Mich., turned their lights blue at various times during the month.

With the new “Wear Blue for Dystonia” campaign, community members wore blue clothing and accessories throughout September to raise awareness. Conversations about dystonia were sparked by wearing stickers that said, “Ask me why I’m wearing blue.” The initiative saw participants share photos of themselves wearing blue with hashtags #DystoniaAwareness and #DystoniaAwarenessMonth to help increase visibility.

Dystonia Zoo Days kicked off in June and ran through October, showcasing a total of twelve events, including one virtual event and eleven in-person gatherings, with five of these coinciding with Dystonia Awareness Month. Be sure to check out the photos from Dystonia Zoo Days on p.12.

By sharing photos of buildings illuminated in blue lights, by wearing blue and inspiring conversations, and by attending events and making new connections in the community—you can make a difference. Dystonia Awareness Month 2023 was a notable success. We look forward to September of 2024 and making even greater strides in advancing dystonia awareness, research, and support. To learn more about our ongoing awareness initiatives, please visit the DMRF’s website.



Mental Health Support May Be as Close as Your Computer

Living with dystonia presents unique physical and emotional challenges. For many, finding the right mental health support can make a significant difference in improving their quality of life.

Fortunately, the digital age has made it easier than ever to access online therapy suitable to the needs of individuals with dystonia. However, it is important to know exactly what you want, need, and expect before you begin. Additionally, many new online services are emerging, and critically assessing the benefits of these services is key.

There are many benefits to online mental health treatment. Meeting with therapists virtually means that you can have a therapy session from the comfort of your home. This is especially valuable for those with mobility issues, transportation issues, or those who live in rural areas. In addition, many online platforms include messaging with a therapist—something that might be beneficial to many in the dystonia community.

To get the most out of your online therapy experience, there are important steps to take. You may want to talk with your physician or a trusted friend or family member as you begin this process.

Assess your needs:

Reflect on your emotional well-being and identify the specific challenges you'd like to address. Are you looking for a neutral third-party to help identify coping strategies to reduce stress? Do you need help navigating the changes in your life brought on by your dystonia? Identifying your goals can help as you begin the search for the right mental health provider for you.



Research and explore:

Look for therapists who specialize in chronic conditions and ideally have experience with movement disorder patients. Many providers have experience in chronic conditions and chronic pain and can use that expertise to help guide you. The DMRF has an informational packet for mental health providers that will help familiarize them with dystonia as they prepare for your session. This can be found on the website at dystonia-foundation.org/living-dystonia/mental-health-pros/ or by calling the DMRF office.

Utilize online therapy directories, search engines, or mental health platforms to find potential therapists. The DMRF has a resource library with materials to help you make informed decisions. In recent years, many online mental health companies have emerged—make sure you do your research to verify if these will be a good fit for you. Questions to ask may include: Do they take your insurance, or are they subscription services? What level of experience do the therapists have—are they students or have they been practicing for some time? Are you able to pick a provider, or are they assigned to you?

Verify licensing and security:

Ensure that your chosen therapist is licensed to practice in your state or country. Licensing is meant to protect both you and the mental health provider. Licensed mental health providers have received the proper, graduate-level training to help patients. This training includes supervised clinical

experience and that they've passed state and national licensing exams. Making sure that you are talking with a provider licensed in your state is critical—many states have online directories to check if your provider has the necessary qualifications.

In addition, make sure the platform you are using is secure. Therapy works when patients have a private space to share deeply personal experiences and emotions. If you're using video appointments, make sure the feed is secure. Like telehealth appointments with a physician, the system should be encrypted and HIPAA-compliant. This also goes for messaging with a therapist. Secure portals are essential and should be able to verify your identity and that of your provider.

Discuss compatibility:

Finding a therapist can take time—not every provider is a good fit for every patient. Many providers offer free initial consultations, which allows you the opportunity to assess their approach and personality. Don't be discouraged if the first person you talk to isn't right for you—it can take time to find the right fit, and finding the right person is important. Building a strong rapport with your therapist is essential for effective online therapy, so take the time to find someone you're comfortable sharing your concerns with.

Navigate financial considerations:

Online therapy is often more affordable and accessible, but it's crucial to explore payment options. Contact your insurance provider to inquire about coverage for online therapy. Some online providers do not accept insurance and instead operate as a subscription service. This might be right for some patients, but not for all. It's worth checking with your insurance provider to see if they cover teletherapy appointments and what costs they'll cover.

Research companies or providers may offer competitive fees or sliding-scale payment options for out-of-pocket costs. Many of the companies that do not accept insurance do offer sliding-scale payment options or financial assistance.

Seek additional support and resources:

Therapy may not be necessary for everyone. Mental health is an ongoing journey, and support beyond therapy can make a significant difference. (See accompanying article.)

Online therapy can be a powerful tool to help navigate the complexities of dystonia and provide the support and guidance you need to live your best life. By assessing your needs, researching qualified therapists, addressing financial considerations, and seeking additional support, you can embark on a journey toward emotional well-being and resilience.

Explore Dystonia Support Groups and Coping Strategies

Sharing your experiences and insights while learning from others in similar situations can be tremendously beneficial. In addition, speaking about these issues can help reduce the stigma around both mental health and dystonia. Join online support groups or communities specifically tailored for dystonia patients.

Online Support Groups:

The DMRF has many local support groups that can provide a sympathetic and understanding ear. In addition, the DMRF has online Facebook groups to provide community support. It is important to remember that support groups are not professional mental health services, but rather groups of people who are also living with dystonia and can empathize, provide practical coping strategies, and provide peer support. Information about these offerings can be found online or by calling the DMRF office.

Coping Strategies:

Explore coping mechanisms to manage the emotional impact of dystonia. Incorporate mindfulness, relaxation techniques, and self-care into your routine. Breathing exercises, working on your hobbies, or being in nature can provide significant stress reduction in your life. Similarly, building a support system with your friends, family, and community can be extremely beneficial.



Dystonia Zoo Days 2023

The Dystonia Medical Research Foundation would like to express its gratitude to all the volunteers who played instrumental roles in making the Dystonia Zoo Day events a tremendous success! With a total of 12 in-person events and one virtual event, this year's Zoo Days saw significant growth and outreach. Notably, five of the Zoo Days were hosted for the first time in Lansing, Mich.; Bridgeport, Conn.; Fort Wayne, Ind.; Indianapolis; and Long Island, N.Y. The volunteers worked tirelessly to organize these events, raising awareness and funds for dystonia research. The DMRF is exceptionally grateful to everyone involved.

The success of the DMRF's Dystonia Zoo Days events would not have been possible without the support of all our incredible local volunteers. We are looking to expand to new cities in 2024. Please contact events@dystonia-foundation.org if you are interested in planning an event in your hometown.

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The **6th Twin Cities Dystonia Zoo Day** was held on June 24 and was led by Shanna and Brad Schmitt, who also lead the Minnesota dystonia support group. This event featured special guest appearances by DMRF Awareness Ambassador and musician, Billy McLaughlin, along with Stephanie Standal, MD, from M Health Fairview, and DMRF Board Member, Dan Lewis.

The **6th Portland Dystonia Zoo Day** took place on Aug. 29 and was co-hosted by Dee Linde, Judith Mason, and Erinn Stimson. Dee Linde, a tireless advocate, leads the Portland and Southwest Washington Support Group.

The **1st Long Island Dystonia Zoo Day** took place on Aug. 19 in Holtsville, N.Y., and was organized by Linda Davis. The event featured Legislator Dominick Thorne, State Senator Dean Murray, Assemblyman Joe DeStefano, and Councilman Neil Foley.

The **6th St. Louis Dystonia Zoo Day** event was held on Aug. 27 and was organized by June Tritely in honor of her sons, Scotty and Quintyn.

The **1st Connecticut Zoo Day** event was held on Aug. 27 at the Beardsley Zoo in Bridgeport, Conn. The event was hosted by Janice and Len Nachbar, DMRF Vice President of Support Paula Schneider, and attended by Vice President of Development Ron Hersh. The highlights of the event were a presentation from Michelle Dagostine, MD, Hartford Healthcare, and a performance by Harmony on the Sound Chorus. State Representative Laura Dancho and Beardsley Zoo Director Gregg Dancho were special guests.

The **1st Lansing Dystonia Zoo Day** was held on Sept. 9 and was organized by Lisa Breedveld and Barry Casler. Lisa hosted the event in honor of her son, Keagan. The event included a presentation by Steven Mitchell, DO, from Bronson Healthcare in Kalamazoo, Mich.

Expands Outreach

The **1st Indianapolis Dystonia Zoo Day** was held on Sept. 9 and was organized by Sunshine and Darrick Fox, Sally and Sarah Ernstberger, and attended by DMRF Treasurer John Downey. Sunshine and Sarah co-lead the Central Indiana Support Group.

The **8th Pittsburgh Dystonia Zoo Day** was held on Sept. 17 and was organized by Chris and Tara Sorley, Ed Cwalinski, Jillian Johnson, and MaryRae Nee. One of the highlights of the event was a presentation by Timothy Lechlitter, MD, from Allegheny Health Network. Ed and MaryRae are co-leaders of the Western Pennsylvania Support Group.

The **1st Fort Wayne Dystonia Zoo Day** was held on Sept. 23 and was organized by MacKenzie Prater and members of Team #CARSONSTRONG.

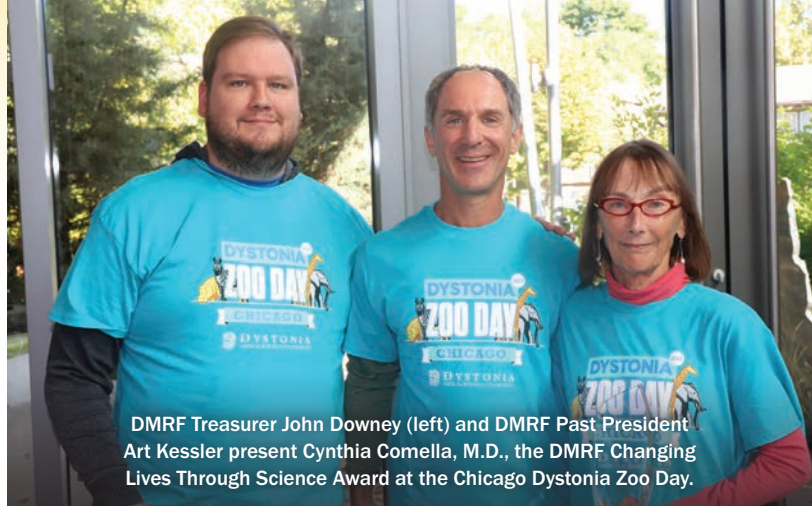
The **3rd Toledo Dystonia Zoo Day** event was held on Sept. 2 and was led by Kristin Cinglie. Peter LeWitt, MD, from Henry Ford Health participated in an education presentation for the guests.

The **2nd Boston Zoo Day** was held on Sept. 30 and was organized by DMRF's Vice President of Support Paula Schneider, Linda Cline, and Mary Letson. A special guest, Todd Herrington, MD, PhD, from the Mass General Brigham Dystonia Clinic addressed the crowd.

The return of the **Chicago Dystonia Zoo Day** event saw DMRF Board member Art Kessler and Treasurer John Downey present the Changing Lives through Science award to Cynthia Comella, MD, for her outstanding contributions to dystonia clinical research.

The **Virtual Dystonia Zoo Day**, co-hosted by DMRF President Mark Rudolph and DMRF Executive Director Janet Hieshetter, united the dystonia community from all over the country to learn from experts, boost dystonia awareness and raise funds for medical research.

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More Highlights from Dystonia Zoo Days



Thank you to our attendees, team leaders, speakers, and sponsors!



How to Talk with Your Doctor About Dystonia

A dystonia diagnosis can be life changing. To get the best outcome from your treatment, you should be actively involved in the development and management of your care plan.



Research has shown that people who have strong partnerships with their healthcare providers do better both physically and emotionally. Central to developing a strong partnership is good communication with your doctor, which sounds simple but has its challenges. The time limitations of medical appointments, stress brought on by waiting for answers, and simply not knowing what questions to ask are a few examples. In addition, identifying a doctor who shares your communication style—who you are in sync with—contributes a great deal to effective communication.

To achieve effective communication, some planning is necessary and starts with finding the provider that is a good fit for you. It is important that you feel comfortable talking with your doctor about how you feel, how your treatment is going, and about your questions, concerns, and expectations from your

treatment plan. Don't be afraid to recognize that the doctor you are currently working with may not meet your needs. Always remember that the No. 1 expert on your dystonia is you.

Preparing for your appointment

- 1. Questions:** Make a list of the questions you want your doctor to answer. It is easy to forget what questions you have when you are waiting for your appointment. Having a list will help you focus on the most important questions and help your doctor respond to them.
- 2. Bring another person if you can:** If possible, bring someone with you to take notes and retain the information that is shared. A family member or good friend will also be there to assist and support you.
- 3. Medications:** Bring a list of your medications to the appointment. It is important for your doctor to know all the medications you are taking, including dietary supplements.
- 4. Learn about dystonia:** Inform yourself about dystonia and the treatment options available. Stay informed about what is happening in dystonia treatment, so you can talk with your doctor about whether a change in your treatment plan will benefit you.

During your appointment

During your appointment, share your list of questions with your doctor. Be mindful of their time limitations and try to ensure your most important questions get addressed by letting your doctor know your most pressing questions first. Don't be surprised if it is recommended that you make another appointment just to speak with your doctor about all your questions. You can also ask if you can speak with the nurse regarding your questions.

Some practical steps to consider:

- Repeat the answers back to your doctor to make sure you got them right. Take notes and ask if you can record the responses to your questions if you are not able to have someone with you.
- If the responses are not clear or if you still do not understand, ask your doctor to explain it more simply or perhaps to show you a diagram to better explain.
- If your doctor uses an online patient portal to communicate with you and you need help in understanding how to use the portal, ask if there is someone in the office who can assist you in learning how to use the portal.

Questions Patients Commonly Ask Healthcare Providers

- ▶ What **form** of dystonia do I have?
- ▶ How will my dystonia affect my **day-to-day** life?
- ▶ Is my dystonia **genetic**? Should I be genetically tested? Should my children be tested?
- ▶ Can I expect my dystonia to get **worse**? Will it spread?
- ▶ What are my **treatment options** and what do you recommend?
- ▶ What can I **expect from my treatment**? When will it start to work? How do I know if it is not working?
- ▶ What are the potential **side effects**?
- ▶ What is the **cost** of the proposed treatment?
- ▶ What is the brand and generic names of the **drugs** being prescribed?
- ▶ If you have **pain**, ask how your pain will be treated.
- ▶ What is the **best way to reach you** with questions?

Having your questions answered and your concerns addressed can help reduce the stress and anxiety of living with dystonia. Being an equal partner in your healthcare and having productive conversations with your doctor can make a big difference in getting the most of your treatment.

DMRF's Monthly Donors Club: *A Special Group of Supporters*



It would be impossible to sustain the Dystonia Medical Research Foundation's vital research programs and community outreach without our Monthly Donors Club. The generous support from our monthly donors touches every aspect of the DMRF's activities from events to the free resources and education programs for individuals living with dystonia and their families.

While the Monthly Donors Club is entirely flexible, with donors able to choose the amount they give every month and the duration of their support, monthly donations allow the DMRF to plan ahead. Even just \$5 a month makes a significant difference, and the impact of your gift is felt throughout the dystonia community.

Becoming a monthly donor is simple. To learn more about how you can join this special group of supporters and make a lasting difference in the fight against dystonia, visit the DMRF's dedicated page for monthly donors or scan the QR code above. As a token of appreciation, any donor who signs up before the end of the year will receive a dystonia awareness magnet.

Joining the DMRF's Monthly Donors Club is an opportunity to have a consistent and meaningful impact on dystonia research, support the programs you love, and bolster awareness efforts. Your contributions sustain the critical work being done by the DMRF, and your dedication is deeply appreciated. Join this special group of supporters today and be a part of the ongoing fight against dystonia. Together, we can make a world of difference for individuals and families across our community.

Monthly donation website: dystonia-foundation.org/donate-other-ways/donate-online/monthly

People on the Move

The Dystonia Medical Research Foundation is grateful for the generosity, hard work, and commitment of each volunteer who made this Dystonia Awareness Month such a success. Their efforts have not only raised funds for vital medical research, but also raised awareness and brought together communities across the country to support individuals with dystonia. Thank you for making a difference and being champions in the fight against dystonia.

Congratulations and many thanks to Melissa and Michael Caravello who spearheaded the inaugural **Angels Walk for Dystonia** in Middlesex County, New Jersey. Angels Walk was scheduled to take place on Sept. 23, but had to be cancelled because of the severe weather tropical storm Ophelia brought to the area. The annual event, organized as a tribute to their daughter, Angelina, and in memory of Joanna Manusov, a champion for those affected by dystonia who passed away in 2021, has raised more than \$18,000. The Caravello family was bolstered by the advocacy work set in motion by Joanna's devoted parents, Janice and Len Nachbar. Melissa and Michael are already busy planning the 2024 event.



Congratulations to Janice and Len Nachbar for receiving a proclamation for **Dystonia Awareness Month** from **Mayor Laura R. Hoydick of Stratford, Conn.** Len was also interviewed by TV News 12 to promote Connecticut Zoo Day and Dystonia Awareness Month.



Our heartfelt gratitude goes out to Tracey DeYoung, Victoria Katz, and Cary Weigert, the organizers behind the **Dystonia Warrior Ride**. The Dystonia Warrior Ride is a charitable motorcycle ride that begins in Woodstock, Illinois, and makes several stops in Wisconsin. This July, Beth Farber, Chair of the DMRF's Community Leadership Council, served as a guest speaker for the event. The Ride not only generated funds for dystonia research but also raised awareness at every stop across Northern Illinois and Southern Wisconsin.

Thank you to Evelyn Rubak, Amy and Seth Gordon, and DMRF Board member, Jon Davis, for hosting a **"Party with a Purpose."** This festive night of live music, food, and drinks, as well as games, raffles, and giveaways raised funds for the Dystonia Medical Research Foundation's programming and research.

We commend Dystonia Awareness Warrior Jason Dunn and Mike Delise for receiving **Dystonia Awareness Month proclamations from both Mayor James R. Fouts of Warren, Michigan, and Governor Gretchen Whitmer of Michigan.** Their dedication was highlighted during an appearance on FOX2 Detroit, where they discussed their awareness initiatives. Furthermore, they successfully championed the lighting of Warren City Hall in blue, in support of Dystonia Awareness Month.



Thank you to Crystal Edmonds, co-leader of the North Carolina Dystonia Support Group, for receiving a proclamation for **Dystonia Awareness Month from the Fayetteville, North Carolina City Council.**

Tips for Managing Holiday Stress

Living with dystonia is challenging year-round, but the holiday season can pile on additional stress that can affect your health and dampen the spirit of the season. DMRF spoke with Mary and Mark Letson of Melrose, Mass., to find out how they manage the holidays as a person with dystonia and as a caregiver.

Mary has familial tremors and cervical dystonia. She's been a hairdresser for 40 years and says the holidays can be particularly stressful both on the job and at home. "When I get stressed, I get very shaky. As a hairdresser, that's a problem," Mary said.

Her husband, Mark, says he can sense when the stress is getting to Mary, especially when she's tired and over-extended. "I try to help out where I can—make meals, help decorate, be there for the boys when they were little," Mark said. "We've only accomplished what we have because we've worked together as a team all our years."

He's retired and their two boys are adults, so life during the holidays is much easier than it used to be. They remember a time when balancing family and work life with holiday gift giving, decorating, party planning, and hosting was overwhelming.

"Every year I would have a meltdown, especially when the kids were little. I put so much pressure on myself between work craziness and holiday preparation. I thought I was going to be the next Martha Stewart," Mary said. "I've let go of that pressure now. That's my biggest piece of advice. Don't put so much pressure on yourself."

Here are some practical tips for taking the pressure off during the holidays:

- **Be realistic**

Forget Martha Stewart and Norman Rockwell. Perfection isn't achievable during the holidays and only brings stress trying to achieve it. Aim for enjoyable, not flawless, celebrations.

- **Maintain your treatment plan**

Prioritize your health. Don't let additional holiday tasks derail your doctor visits, physical therapy and/or other scheduled treatment for dystonia.

- **Limit social commitments**

Special events and invitations multiply during the holiday

season. You don't need to attend them all. Choose the most important ones to you and decline the others.

- **Simplify traditions**

Modify holiday traditions to suit your health demands. For example, downsize the decorating, go potluck for big family meals, and focus gift-giving on a select few.

- **Delegate**

Don't try to do everything on your own. Ask for help with holiday preparations, such as cooking, cleaning, or decorating.

- **Listen to your body**

Be aware of your dystonia symptoms and any potential triggers that may worsen them. Avoid situations during the holidays that may exacerbate your symptoms and maintain proper diet and exercise.

- **Plan to rest**

Incorporate rest into your holiday plans. Take breaks to recharge during the day and prioritize getting a good night's sleep.

- **Seek professional help if needed**

An inordinate amount of stress can lead to anxiety and depression. Seek treatment from a professional who can provide talk therapy and medication if warranted.

- **Enjoy the season**

Remember the true meaning of the holidays and enjoy your family and friends. Your close family and friends are not going to judge you, Mary says. They may not know how it feels to live with dystonia, but they're at least aware of some of the challenges.

The holiday season should be a time to celebrate and be with family. Taking steps to manage stress and prioritizing your health will help you truly enjoy the season.

For more tips on how to manage holiday stress, visit DMRF's YouTube channel to view a webinar recording, *Self Care for the Holidays*.

Dystonia Research Watch

The DMRF is dedicated to supporting research on all aspects of dystonia to improve treatment and ultimately find a cure. DMRF's scientific leaders keep close tabs on emerging research that may affect the dystonia community. Two research areas have recently gained momentum:

1.) Non-motor symptoms of dystonia

A recent DMRF workshop was dedicated to non-motor aspects of dystonia; a summary paper will be published soon in *Dystonia*. Interestingly, another study published in *Parkinsonism and Related Disorders* inquired about the possibility that injected botulinum neurotoxin (BoNT) may also reduce anxiety in patients with cervical dystonia. DMRF promotes and supports interdisciplinary studies of the non-motor symptoms of dystonia.

Dystonia is an open access journal

Dystonia aims to be the leading journal in the field by publishing research results on all basic, clinical, and translational aspects of dystonia. You can freely access the journal at: frontierspartnerships.org/journals/dystonia

To assess the severity of motor symptoms, researchers used the Toronto Western Spasmodic Torticollis Rating Scale (TWSTRS), and to assess the levels of anxiety, they used State-Trait Anxiety Inventory (STAI). The STAI scale allows researchers to separate state from trait anxiety. State anxiety is the anxiety a person feels when faced with a threat or frightening situation and will pass when the threat or frightening situation has passed. Trait anxiety appears to be ever-present general tendency to worry, be fearful and perceive events as threatening.

2.) Non-invasive neuromodulation as a potential therapy for certain types of dystonia

DMRF issued a call for research proposals that focus on non-invasive brain stimulation therapies, and several projects are ongoing. To those living with dystonia, progress in novel therapies can seem slow and there are many reasons for that. A recent article in DMRF's scientific journal, *Dystonia*, discusses the factors hindering research specifically regarding transcranial magnetic stimulation (TMS).

Here is more detail from two recent studies:

Study Explores Botulinum Neurotoxin's Effect on Anxiety in Cervical Dystonia Patients

It is well documented that as many as 30 to 40% of people with cervical dystonia also experience increased anxiety. In a recent study published in *Parkinsonism and Related Disorders* (www.elsevier.com/locate/parkreldis), researchers from Rush University, Chicago, report the results of a 12-week prospective, observational study of the effect of botulinum neurotoxin (BoNT) injections on the state of anxiety in 60 people with cervical dystonia (CD). They tested the hypothesis that BoNT has a beneficial effect on anxiety that is independent of its effect on motor severity.

Participants were evaluated at the time of BoNT treatment and six weeks later, the time considered to have the maximum benefit for motor symptoms. These evaluations were conducted via telemedicine. Participants were asked to complete the Generalized Anxiety Disorders-2 item (GAD-2) survey every two weeks using a special app until the conclusion of the twelve-week study. All participants had been treated with botulinum neurotoxin previously for an average of 9.5 years.

Other published studies have been conducted in CD and depression and mood. One study showed that improvement in depression symptoms was correlated with pain lessening, and that mood and CD motor symptom improvements were also positively correlated. The current study assessed BoNT treatment's impact on improving anxiety symptoms independent of the state of motor symptoms.

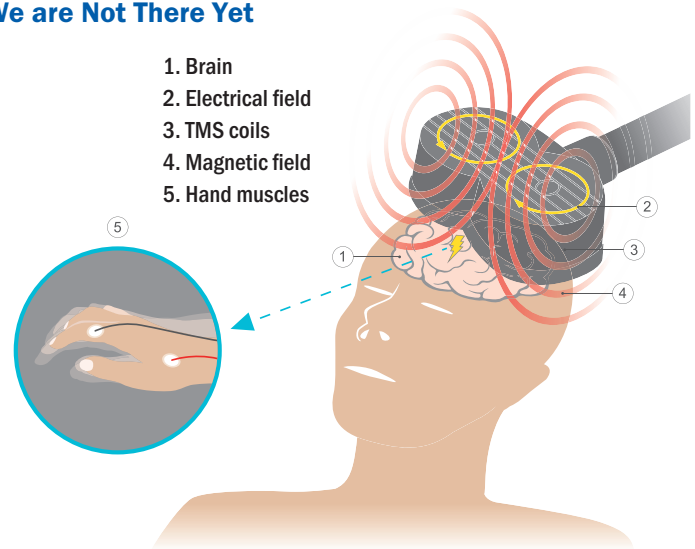
Investigators concluded that improvement in anxiety is unlikely to be only secondary to the improvement in the motor symptoms of CD. This may indicate a potential direct benefit of BoNT on anxiety. They acknowledged that the mechanism for this is not known, and that more investigation is needed to better understand this relationship.

Transcranial Magnetic Stimulation for Dystonia: Why We are Not There Yet

Transcranial magnetic stimulation (TMS) has emerged as a non-invasive treatment of some brain ailments. Considered noninvasive because it is done without any surgery, this form of brain modulation uses changing magnetic fields to induce an electric current at a specific area of the brain through electromagnetic induction. An electric pulse generator, or stimulator, is connected to a magnetic coil attached to the scalp.

Initially used as a research device, TMS has shown great promise in treating many neurological and psychiatric conditions. However, despite numerous studies, it has not been approved for dystonia. Why is that? A group of researchers from Duke University tried to answer this question in a review article recently published in *Dystonia*, DMRF's scientific journal.

Authors of the review article, *Transcranial magnetic stimulation: the road to clinical therapy for dystonia*, identified four major roadblocks that thwart adoption and approval of TMS for treatment of dystonia. These include limited and inadequately designed clinical trials; insufficient and sketchy evidence that TMS objectively affects behavioral measures; observed variability in response to TMS due to complex technical issues and selection of patients; and difficulties in optimizing TMS parameters to achieve the desired therapeutic effect. The article presents progress



TMS, not yet approved for dystonia, delivers stimulation to targets in the brain through the skin and skull.

achieved over the last few decades and identifies clear goals for future studies, such as comprehensive identification of reliable behavioral measures for various types of dystonia, precisely testing a range of stimulation parameters, using different modes of stimulation, and applying computational methods and models to optimize TMS efficacy.

Time will tell if these goals can be achieved to further research of TMS as a potential treatment for various types of dystonia.

Register with the Dystonia Brain Collective



The DMRF, in collaboration with other dystonia organizations, has formed the Dystonia Brain Collective to work with the Harvard Brain and Tissue Resource Center to advance dystonia research by providing critically needed brain tissue. So, when you no longer need your brain, we want it. Becoming a brain donor will help expedite dystonia research. Giving dystonia researchers the opportunity to study brain tissue is a crucial step toward developing improved therapies and a cure for dystonia in all its forms. Through the study of donated brain tissue, important clues about dystonia can be discovered. Your brain may help to unlock the mysteries of dystonia. The process to register is easy: **please call 800-377-3978 or email brainbank@dystonia-foundation.org**. The process is made as easy as possible for your family, and there is no cost. Please consider registering today.

DMRF Staff Spotlight

Meet Veronica Diaz, Manager of Community Engagement

We're spotlighting DMRF staff who are making a difference in the dystonia community, and we're delighted to kick off our series with Veronica Diaz.



Veronica Diaz

Veronica's journey with the DMRF began in December 2014 when she assumed the role of information coordinator. In this capacity, she served as the initial point of contact for individuals seeking information and resources related to dystonia. It was during these early days that Veronica formed deep and meaningful connections with those who reached out to the foundation. This experience not only provided her with a comprehensive understanding of dystonia but also allowed her to witness the powerful impact of empathy and support.

Reflecting on those initial interactions, Veronica recalls the many concerned mothers, fathers, husbands, and wives who were desperate for information to support their loved ones and to better comprehend this relatively unknown disorder.

In November 2017, Veronica transitioned into the role of operations coordinator. Her responsibilities expanded, and she began working closely with the development team. One of her key tasks was entering gifts into the DMRF database, a role that provided her with a comprehensive overview of the foundation's operations, further deepening her commitment to its mission.

In April 2019, Veronica took on her current role as manager of community engagement. In this capacity, she oversees more than fifty dystonia support groups and ten Facebook groups. Her dedication to these communities is unwavering, and she understands the profound impact support groups and online communities have on the lives of those living with dystonia.

When asked about the importance of support groups, Veronica said, "Dystonia can be an incredibly isolating disorder, and one of the most important things that the support groups provide is the knowledge that you are not alone. For individuals facing the challenges of dystonia, these groups represent safe spaces where they can connect with others who share their journey." It's within these communities that they find not only valuable information and resources but also crucial emotional support and encouragement.

Beyond her professional accomplishments, Veronica Diaz is also known for her love of animals, particularly her beloved dog, Oliver. Oliver, a 13-year-old miniature pinscher/chihuahua mix, has been her loyal companion through thick and thin. Veronica admits that she never considered herself a dog person until her youngest brother unexpectedly dropped Oliver off at her



The Colorado Dystonia Support Group, led by Paul Kavanaugh

house when he was just two months old. Uncertain about what to do, Veronica made the life-changing decision to keep Oliver, a choice she now describes as one of the best she ever made. Oliver is more than just a pet; he is her constant companion and source of comfort during life's ups and downs.

"Dystonia can be an incredibly isolating disorder, and one of the most important things that the support groups provide is the knowledge that you are not alone. For individuals facing the challenges of dystonia, these groups represent safe spaces where they can connect with others who share their journey." —Veronica Diaz

Veronica's journey from information coordinator to manager of community engagement has not only enriched her understanding of dystonia but has also allowed her to build invaluable connections within the dystonia community. For those seeking support, information, or a sense of belonging within the dystonia community, Veronica encourages exploration of the resources available on the DMRF website. She welcomes individuals to reach out to her to discuss how they can initiate a support group and learn about the ways in which the DMRF collaborates with support group leaders and online moderators. Veronica concludes with heartfelt gratitude, saying, "I am deeply grateful to the support group leaders and online moderators for their dedication to our community." Together, they continue to make a profound and meaningful difference in the lives of those affected by dystonia.

Finding Strength and Support: the DMRF's Support Groups



While many people join to find answers to questions, participating in a Dystonia Medical Research Foundation support group is about more than just acquiring information.

Dystonia support groups allow members to discover a safe and knowing community. Connecting with people who fully understand the distinctive challenges of dystonia can be significant in successfully managing the disorder. Sharing strategies for successful treatment outcomes and finding ideas for things that might help overcome the potential limitations that can come with a dystonia diagnosis can make a big difference in daily activities. Additionally, knowing one is not alone in the dystonia journey is really helpful. There is a community there to call upon when needed.

DMRF support groups, most often led by people who are also living with dystonia, are for patients as well as caregivers, friends, and family. If you or a loved one is living with dystonia, you don't have to face it alone. To connect with a DMRF support group, visit the DMRF website at **dystonia-foundation.org/living-dystonia/support**. There, you'll find information on local support groups and online forums.

If there isn't a dystonia support group in your area, you can be the first to get one started. To learn more about launching a support group, contact the DMRF at **800-377-3978** or **dystonia@dystonia-foundation.org**.

By joining a DMRF support group, you can experience the transformative power of connecting with the community and face the challenges of dystonia with strength and resilience. Remember, you don't have to go through this journey alone; help and support are just a click or call away.

Dystonia Dialogue

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More Highlights from Dystonia Zoo Days 2023!

