

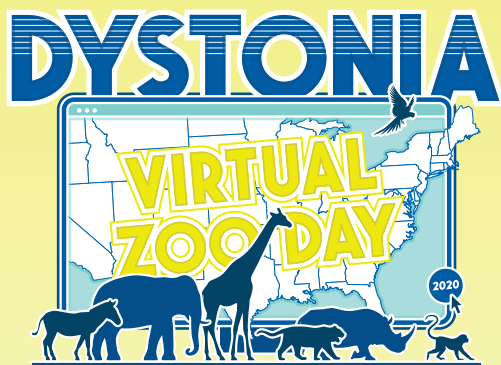


DYSTONIA
MEDICAL
RESEARCH
FOUNDATION

Dystonia Dialogue

NEWSLETTER OF THE DYSTONIA MEDICAL RESEARCH FOUNDATION

Winter 2020 | Volume 43 • No. 3



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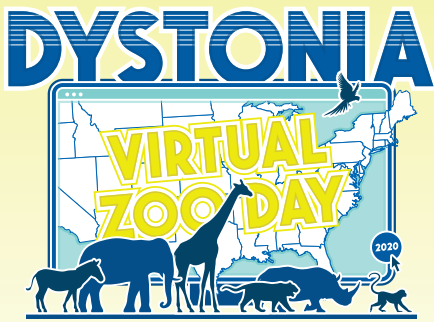
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Dystonia Community Unites for Virtual Dystonia Zoo Day

50 States & 9 Countries Represented

“Today is a demonstration of the powerful unity of the entire dystonia community coming together online to work towards a cure,” said DMRF President Art Kessler in his remarks to officially kick-off Virtual Dystonia Zoo Day on Saturday, September 12, 2020.

The program began with a lively opening from Lolly Lardpop, a puppet performed by Emmy-nominated entertainer Leslie Carrara-Rudolph, best known for her performance of Abby Cadabby on *Sesame Street*.

Executive Director Janet Hieshetter echoed the significance of the event: “The silver lining of having to cancel 12 local zoo walks is that for the first time ever the DMRF community—and really the entire dystonia community—is acting on the same day, at the same time, doing the same thing to generate awareness for dystonia. This is truly a national, and even international, event.” Virtual Dystonia Zoo Day attracted participants from 50 US states and nine countries. The Covid-19 pandemic caused DMRF to cancel its 2020 schedule of local zoo walks across the country and pivot to a virtual event.

Virtual Dystonia Zoo Day raised over \$265,000 in support of the DMRF mission. The National Planning Committee included: Jacquelyn Coello, Ed Cwalinski, Beth Farber, Karen Flanagan, Shanna Schmitt, Paula Schneider, Pam Sloate, and Brian Smuda.

Virtual Dystonia Zoo Day featured scheduled sessions throughout the day. Participants posted photos throughout social media using the hashtag *#LetsZOOthis*.

DMRF Community Leadership Council member Beth Farber moderated “Welcome to the Jungle: Live Zoo Keeper Chat.” Alex Burris and Katelyn Babcock from Toledo Zoo & Aquarium introduced viewers to two red pandas. DMRF event planner Kristin Cinglie and son Dylan Altman joined zoo keepers Alex Case and Kailey Doherty for an exposé on the two-toed sloth.

DMRF supporter Brian Smuda gave viewers a “Dystonia Research Back Stage Pass” with Medical & Scientific Advisory Council Member Christian Schlieker, PhD and DMRF Barbara Oliver Research Fellow Anthony Rampello, PhD of Yale University. The investigators shared highlights from their work to understand the changes in brain cells that underlie dystonia and that may ultimately point the way toward new treatments. The session was moderated by DMRF Vice President of Development Ron Hersh.

DMRF event planner Jacquelyn Coello moderated “Cubs Kingdom: Kids Zone,” a segment of the program for children. Highlights included a visit from Lolly Lardpop and Leslie Carrara-Rudolph, art with Ellen Cornett, tips for becoming a zoologist with DMRF support leader Maureen Lehon and Marie Bacher of Walden West, greetings from Beyond Imagination Alpacas, and a “DMRF Virtual Zoo” animated video by prodigy filmmakers Vivian and Sabine Harper.

“Float like a Butterfly: Chair Yoga & Mindfulness” with Richard Sabel MA, MPH, OTR, GCFP of SUNY Downstate Health Sciences offered a 30-minute relaxation demonstration moderated by Community Leadership Council member and support leader Shanna Schmitt.

Janet Hieshetter moderated the “Q&A with Movement Disorder Experts,” featuring Joanna Blackburn, MD of Northwestern Medicine, Rachel Saunders-Pullman, MD, MPH, MS of Mount Sinai, and Michele Tagliati, MD of Cedars-Sinai.

THANK YOU to Our National Sponsors

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THANK YOU to Our Elephant Sponsors

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Virtual Dystonia Zoo Day ended with “Croon Like a Baboon: Closing Ceremony & Sing-A-Long” with Awareness Ambassador Billy McLaughlin and Lisa “Z” Chesnutt performing “All You Need is Love” by The Beatles. During the Closing Ceremony, Art Kessler and Janet Hieshetter extended their gratitude to everyone who contributed to the program. “It’s been an awesome day,” said Art. “I really don’t want it to end, but I guess it’s that part of the day. Thank you to all of our participants and donors—we couldn’t do everything we do at the DMRF without your support.”



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What is Dystonia?

Dystonia is a disorder that affects the nervous system. Abnormal signaling from the brain causes muscles to contract excessively. This results in involuntary body movements and postures. Dystonia can affect a single body area or multiple muscle groups. There are numerous types of dystonia, and dozens of diseases and conditions may include dystonia as a symptom. For more information visit: www.dystonia-foundation.org



On the Cover:

The first-ever Virtual Dystonia Zoo Day on September 12, 2020 was a national—and international—day of dystonia awareness and fundraising for research. Participants represented all 50 US states and nine countries. More than \$265,000 was raised in support of the DMRF mission. Read more on pages 2–3.

The Dystonia Dialogue is supported by educational grants from The Allergan Foundation.

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

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

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



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President



Janet L. Hieshetter
Executive Director

As the end of 2020 comes into view, we have little doubt that it will be a memorable year for many of us. As we reflect on the work of the DMRF during this unusual time, we have learned from challenges as well as from unexpected new opportunities. We are inspired by how the dystonia community has rallied together to strengthen the ties that unite us, and DMRF remains committed to encouraging and supporting this sense of unity.

Virtual Dystonia Zoo Day on September 12, 2020 was a tremendous success. We were thrilled by the response in support of this unique, online event. From one coast of the country to the other, individuals with all types of dystonia, family members, friends, health care providers and institutions, researchers, pharmaceutical companies, and businesses across industries supported this awareness and fundraising effort. See pages 2–3 for more on Virtual Dystonia Zoo Day.

To address the feelings of isolation that the pandemic has caused or worsened for many, dystonia support groups are meeting by video conference to offer peer support. It is now possible to participate virtually in DMRF support group meetings, from any location across the country. Online dystonia forums continue to provide opportunities to connect and communicate on the internet. The information shared and friendships made through these peer support connections are invaluable, especially now.

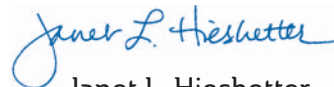
Movement disorder clinics are adapting to provide patient care in the new pandemic climate. Dystonia investigators are making adaptations to ensure research progress continues. Chief Scientific Advisor Dr. Jan Teller shares his thoughts on the impact of Covid-19 on dystonia research on page 12.

In all of this uncertainty, it remains certain that the DMRF is working for you. The DMRF remains available to the community, as reliably as ever. We are excited to explore new ways to adapt our programs and patient resources, including expanding the availability of online programs while making sure that those without easy access to internet technology are not left out.

If nothing else, 2020 has been a year to lean into the power of gratitude to fortify us in trying times. Now and always, the leadership of DMRF remains thankful for your generous support. It is a privilege to work on behalf of the individuals and families that make up the dystonia family.



Art Kessler
President



Janet L. Hieshetter
Executive Director

HOW HAVE YOU BEEN IMPACTED BY COVID-19?

Please share your feedback in a brief survey at dystonia-foundation.org/covid-survey

New Artificial Intelligence Platform Diagnoses Focal Dystonia with High Accuracy in Less Than One Second

Researchers at Massachusetts Eye and Ear in Boston have developed a unique diagnostic tool that detects dystonia from MRI scans, the first technology of its kind to provide an objective diagnosis of the disorder.

In a newly published study, researchers developed an AI-based deep learning platform called DystoniaNet to compare brain MRIs of 612 people, including 392 patients with three different forms of isolated focal dystonia and 220 healthy individuals. The platform diagnosed dystonia with an astonishing 98.8% accuracy. During the process, the researchers identified a microstructural neural network biomarker for dystonia. With further testing and validation, it may be possible for DystoniaNet to be implemented by movement disorder clinics to make high probability diagnosis of dystonia by MRI. In such cases a physician will be able to use this information to more confidently and quickly confirm the diagnosis and recommend treatment without delay.

“There is currently no biomarker of dystonia and no ‘gold standard’ test for its diagnosis. Because of this, many patients have to undergo unnecessary procedures and see different specialists until other diseases are ruled out and the diagnosis of dystonia is established,” said senior study author Kristina Simonyan, MD, PhD, Dr med, Director of Laryngology Research at Mass Eye and Ear, Associate Neuroscientist at Massachusetts General Hospital, and Associate Professor of Otolaryngology-Head and Neck Surgery at Harvard Medical School. “There is a critical need to develop, validate, and incorporate objective testing tools for the diagnosis of this neurological condition, and our results show that DystoniaNet may fill this gap.” Dr. Simonyan is a former member of the DMRF Medical & Scientific Advisory Council.

The study included three of the most common types of focal dystonia: laryngeal dystonia, characterized by involuntary

AT A GLANCE

- **A new diagnostic tool detects dystonia from MRI**
- **DystoniaNet uses artificial intelligence algorithms to detect subtle brain changes**
- **Researchers discovered a microstructural biomarker**

movements of the vocal cords that can cause difficulties with speech (also called spasmodic dysphonia); cervical dystonia, which causes the neck muscles to spasm and abnormal movements and postures in the neck; and blepharospasm, a focal dystonia of the eyelids that causes involuntary blinking and/or forceful eye closure.

Traditionally, a dystonia diagnosis is made based on tedious clinical observations.

Previous studies have found that about 50% of cases go misdiagnosed or underdiagnosed at a first patient visit.

DystoniaNet utilizes deep learning, a particular type of AI algorithm, to analyze data from individual MRI and identify subtler differences in brain structure. The platform simultaneously detects clusters of abnormal structures in several regions of the brain that are known to control processing and motor commands. These small changes cannot be seen by a naked eye in MRI, and the patterns are only evident through the platform’s ability to take 3D brain images and zoom into their microstructural details.

DystoniaNet is a patent-pending proprietary platform trained using Amazon Web Services computational cloud platform. The technology interprets an MRI scan for microstructural biomarker in 0.36 seconds.

Future studies are needed to examine additional types of dystonia and will require trials at multiple clinics and hospitals to further validate the DystoniaNet platform in a larger number of patients.

Valeriani D, Simonyan K. A microstructural neural network biomarker for dystonia diagnosis identified by a DystoniaNet deep learning platform. Proc Natl Acad Sci U S A. 2020 Oct 1;202009165.



RESEARCH BITS

New dystonia studies are published daily.

Below are a tiny sample of recent data and discoveries.

PLAN B.

TorsinA is a protein found in neurons (brain cells) that causes dystonia when it becomes abnormal due to small genetic changes. When TorsinA is abnormal, it cannot function properly in the cell. This loss of function ultimately leads to the development of dystonia symptoms. A related protein, called TorsinB, performs similar roles in neurons as TorsinA. A team of investigators recently demonstrated in mice that dystonia symptoms and related neuron dysfunction resulting from a loss of function of TorsinA can be corrected by increasing levels of TorsinB. These findings suggest that TorsinB may be mobilized to slow or prevent development of dystonia symptoms.

Li J, Liang CC, Pappas SS, Dauer WT. TorsinB overexpression prevents abnormal twisting in DYT1 dystonia mouse models. Elife. 2020;9:e54285. Published 2020 Mar 23.

DYSTONIA GENES & MENTAL HEALTH.

There are many types of dystonia, often presenting with very distinct symptoms. There is also a great deal of overlap in the physical manifestation of symptoms across the different types. Non-motor symptoms are common, including psychiatric disorders such as depression and anxiety. There have been numerous genes identified to cause dystonia, and it is not yet clear what these genes have in common.

A group of researchers set out to help disentangle the complex genetic landscape of dystonia by systematically analyzing genetic data from mice and humans. They set out to learn whether dystonia-causing genes can be linked to shared brain pathways and, if so, where in the brain this convergence takes place. Plus, whether there are underlying genetic links to the psychiatric disorders frequently seen in dystonia. Using new and well-established genetic approaches, the researchers tested 28 dystonia genes for increased expression in specific brain cell types. They found that multiple dystonia-causing genes interact and contribute to dysfunctional signaling in specific groups of neurons in the brain. Furthermore, commonalities have been found

in the underlying genetic basis of dystonia and a range of psychiatric disorders often seen in dystonia patients including anxiety, depression, and obsessive-compulsive disorder. These findings suggest that certain mental health disorders may be part of the underlying genetics of dystonia, thereby increasing an individual's risk of developing psychiatric symptoms. The report reflects the increasing call from experts to recognize and address non-movement symptoms in dystonia patients. The results also represent an important step in better understanding the brain pathways involved in dystonia and examining relationships among dystonia-causing genes and among dystonia types.

Mencacci NE, Reynolds R, Ruiz SG, et al. Dystonia genes functionally converge in specific neurons and share neurobiology with psychiatric disorders. Brain. 2020;143(9):2771-2787

Busch H, Klein C. 'Moving genes': how dystonia genes functionally converge on the transcriptome. Brain. 2020 Sep 1;143(9):2631-2634.

DRIVER'S ED.

A research team in the Netherlands set out to explore driving performance and driving safety in individuals with cervical dystonia compared to individuals without cervical dystonia. All participants in the cervical dystonia group participated in the study 4-8 weeks after botulinum neurotoxin treatment, which tends to be the time of maximum benefit. Volunteers completed a simulated driving assessment that included lane tracking, intersections, and highway merging. In the individuals with cervical dystonia, there was no indications that driving performance or safety were significantly different than the group without cervical dystonia. However, cervical dystonia patients notably reported higher levels of fatigue before and after driving.

Van den Dool J, Visser B, Huitema RB, Caljouw SR, Tijssen MAJ. Driving Performance in Patients With Idiopathic Cervical Dystonia: A Driving Simulator Pilot Study. Front Neurol. 2020;11:229.

Making Dystonia History

Dystonia Coalition Natural History Study Sheds Light on Dystonia Symptom Spread

The Dystonia Coalition is a global research network funded by the National Institutes of Health that has engaged investigators and patients on an unprecedented scale to improve dystonia treatment. Since 2010, the Dystonia Coalition has included a Natural History of Isolated Dystonia study aimed at better understanding how dystonia may progress over time and the underlying causes.

Does dystonia change over time? What genes might be involved in the development of dystonia? How often does dystonia disappear, and does it come back? How does dystonia impact quality of life? These are the types of questions the Natural History of Isolated Dystonia study seeks to answer.

“We want to figure out what happens over time when there’s not any interventions or treatment, so that we can understand the typical course of the disorder. It’s really important to understand that thoroughly if we want to be able to test new treatments,” explained Dystonia Coalition Co-Director and DMRF Scientific Director Joel S. Perlmutter, MD, who is the Elliot Stein Family Professor of Neurology, Professor of Radiology, Neuroscience, Occupational Therapy and Physical Therapy, and Head of Movement Disorders at Washington University in St Louis.

Investigator-Patient Partnership

The Natural History of Isolated Dystonia aims to recruit large numbers of dystonia patients and study their long-term dystonia experience. To date, more than 3,200 volunteers with dystonia have participated from all over the world—North America, Europe, the Middle East, Asia, and Australia. One of the biggest upfront challenges to a project of this kind is locating and engaging dystonia patients, and not only those who have regular contact with movement disorder clinics. Patients who regularly visit movement disorder clinics for treatment with botulinum injections and tend to be those

AT A GLANCE

- **Natural history studies examine the characteristics and progression of a disease**
- **A dystonia natural history study has been underway since 2010**
- **Adults with isolated focal dystonia are invited to participate**
- **The study has led to new data on dystonia spread**

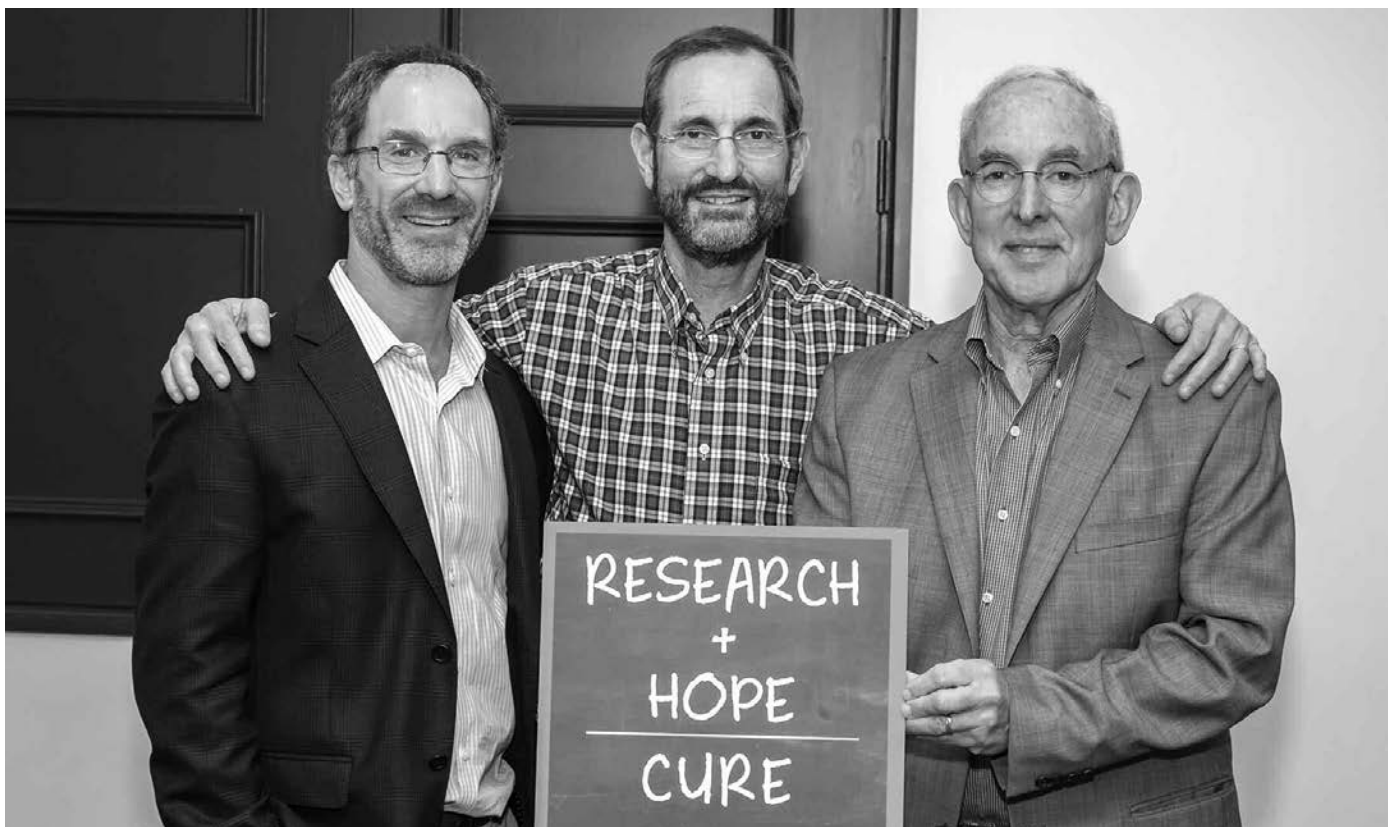
who respond well to treatment, can skew the data by not fully representing the patient population. Another challenge is retaining patients over the years for follow up study visits. “A very important way of retaining participants in these kinds of studies is to report back to them what we’ve been learning,” explained Dr. Perlmutter, “so the participants are enfranchised. Clearly they are the critical part of the research team, but it’s important to make them feel like they’re part of the research team.”

Discoveries & Impact

Dystonia Coalition studies have resulted in more than 100 study publications in the medical literature, an impressive measure of the discoveries and advancements being made. “This large number of publications demonstrates the substantial progress in understanding the natural history of these types of dystonia,” said Dr. Perlmutter. One of the critically important areas of focus is understanding how and when dystonia progresses.

Dystonia symptoms typically begin in one part of the body and may spread to other body areas. This is seen most dramatically in childhood onset dystonias, which may ultimately affect many parts of the body and particularly the limbs. Symptom spread is a possibility in adults, though the frequency and patterns are less well documented.

Scott Norris, MD, who trained with Dr. Perlmutter, led a study to examine the risk of symptom spread in cervical dystonia.



DMRF Scientific Director Joel S. Perlmutter, MD (C) is Co-Director of the Dystonia Coalition and Principal Investigator of the Natural History of Isolated Dystonia study. He is pictured with DMRF President Art Kessler (L) and Vice President of Science Richard Lewis, MD.

Dr. Norris is a past DMRF clinical fellow and current grantee, who has devoted much of his career to studying dystonia and treating patients. Understanding the frequency with which cervical dystonia may spread and patterns of spread is essential for developing strategies to minimize or prevent progression. Previous studies to understand spread in dystonia have tended to be small studies from individual clinics that examined data in retrospect. The Natural History of Isolated Dystonia study collects data prospectively, in real time, at multiple movement disorder clinics around the world. Looking at more than 1,400 cervical dystonia patients, Dr. Norris' analysis demonstrated that in approximately 80% of cases, focal dystonia that begins in the neck will not spread to other body areas. Spread to other body areas did occur in approximately 20% of cases, most often to the hand, facial muscles, and voice box.

The natural history data was also analyzed to examine what adult onset focal dystonias were most likely to spread, and in what patterns. An investigation led by Brian Berman, MD examined nearly 500 volunteers with blepharospasm, cervical dystonia, hand dystonia, and laryngeal dystonia (spasmodic

dysphonia). The study found that the body area where symptoms begin strongly predicts both the risk of spread and the body areas to which symptoms spread. Patients with blepharospasm experienced spread in 50% of cases, which is much more frequently than those with cervical (9%), hand (17%), or laryngeal (16%) dystonia. The most common sites of spread for blepharospasm patients are the oromandibular region (lower face and jaw). Patients with cervical dystonia most commonly experienced spread to the hand. Patients presenting with hand and laryngeal dystonia, as well as those with blepharospasm onset, who experienced spread most commonly experienced symptom spread to the neck. Those patients experienced spread within the first several years after diagnosis.

Understanding how and in what manner dystonia symptoms spread may lead to new strategies to anticipate which patients are at increased risk and to slow or stop progression in those at risk. "Using this kind of information is absolutely critical for designing a study if we wanted to test a drug to see if it could

Continued on page 10

Continued from page 9

slow the spread of dystonia in an individual,” explained Dr. Perlmuter. “To plan such a study, we have to know the risk of spread, and that tells us how many people we need for a study and how long we need to do the study. If we wanted to test a new therapy that could dramatically slow or even stop progression of dystonia, we couldn’t test it without this kind of data.”

Numerous lines of study using the natural history data from the Dystonia Coalition are tackling important clinical issues that profoundly impact patient quality of life including the complex relationship between dystonia and tremor, non-motor symptoms of dystonia such as depression and anxiety, and determining what types of dystonia respond best to available therapies. This kind of information is critical for patients and clinicians alike to more fully understand what to expect from the disorder long-term and to develop effective treatment strategies that target the manifestations or accompanying problems that impair quality of life.

Volunteers Needed

The Natural History of Isolated Dystonia study is actively recruiting individuals with isolated focal dystonia at Dystonia Coalition participating centers. Each patient who participates has a full assessment for dystonia and is asked to participate in return visits. Each patient is asked to complete questionnaires to assess impact on life activities. A blood sample is collected and stored in a biorepository to share among researchers doing genetic studies. Patients may withdraw from the study at any time.

Dr. Perlmuter assures that there are processes in place to keep study participants safe during the Covid-19 pandemic, which include thorough screening before entering a study facility and use of face masks upon entering: “It’s very safe, and lots of precautions are taken.”

The natural history study is currently funded until 2024. There may be an opportunity to extend the study in the future. Dr. Perlmuter encourages patients to consider volunteering, emphasizing their essential role in dystonia research: “If we want to develop new treatments for dystonia we cannot do that without people participating because we don’t know whether our interventions work unless we know the natural progression of the condition. We really need people to participate. The research is for all of us.”

To learn more about this study and/or to inquire about volunteering, visit: rarediseasesnetwork.org/dystonia



What is the Dystonia Coalition?

The Dystonia Coalition is a collaboration of medical researchers and patient advocacy groups that is working to advance the pace of clinical and translational research in dystonia to find better treatments. The Dystonia Coalition is funded by the Office of Rare Diseases Research within the National Institutes of Health. DMRF is proud to serve as an administrative center.

Current Dystonia Coalition Studies

Natural History of Isolated Dystonias

Aim: To learn more about how dystonia may progress over time and what causes dystonia

Biobank

Aim: To create a collection of blood samples for analysis

Patient-Centered Outcome Measures

Aim: To develop an app to monitor symptom severity

Objective Measures

Aim: To develop tools to measure the severity of symptoms objectively

Dystonia Coalition Achievements

- Developed a global network of investigators that includes 49 sites in 7 countries
- Established international consensus for the diagnosis and classification of all dystonias
- Collected clinical data from more than 3,200 volunteers for natural history studies
- Collected DNA samples from more than 3,200 volunteers
- Developed and tested rating scales for common types of dystonia
- Published more than 100 research papers

People on the Move

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



Mike Delise continues to champion \$5Cure4Dystonia, an awareness and fundraising campaign he started in partnership with **Jason Dunn**. Over the summer, Mike created a series of limited edition t-shirts for purchase, with all proceeds supporting medical research toward a dystonia cure.



Ricky Shomin of Michigan honored family member **Sherri Merrifield** by supporting \$5Cure4Dystonia and promoting awareness through his Warrior class stock car racing team.



"Hi, my name is **Hailey** and my mom has dystonia. I made bracelets and sold them to my friends and family to raise money to help research to make my mom feel better. I raised \$100!"

Hailey's mom is DMRF event organizer **Amanda Sleeper**.

The Singing Chef Signature Sauces to Benefit DMRF

The Singing Chef **Andy LoRusso** is donating a portion of proceeds from his upcoming Signature Sauces collection to support DMRF.



With thousands of international performances over 30 years, Andy LoRusso blends food, song, and dance to bring people together. He has partnered with celebrities and top chefs around the world. He is the author of the best-selling cookbook and album combo, *Sing and Cook Italian*.

Growing up in Newark, New Jersey, Andy discovered his passion for authentic Italian food and music in the kitchen with his Grandmother Grace. His grandmother, who was from a small town in Sicily, would play the arias of great Opera singers while she cooked. These fond memories and her recipes inspired Andy to launch a line of sauces to honor his Nona.

The Singing Chef Signature Sauces are available for purchase at singingchef.com. Stay tuned for announcements about a virtual cooking demonstration event to benefit DMRF in the coming months.



Dystonia Research Continues despite Covid-19 Pandemic

By Jan Teller, MA, PhD, Chief Scientific Advisor

The unexpected and sudden arrival of Covid-19 has disrupted the lives of everyone, including researchers. The death toll and infection rates continue to rise around the world. Societies look to health and medical scientists to conquer the virus. The pandemic also impacts scientific research itself in an unprecedented way.

Laboratory work was put on pause. Only some researchers can continue operating their labs and projects. The financial ramifications have been severe. Academic and scientific institutions have lost revenue normally used to fund research; some people lost their jobs or simply cannot continue their regular duties. Many long-term experiments will need to re-start later. Doctoral students and postdoctoral fellows have been most severely affected. Social distancing prevented not only normal work but also scientific meetings, established collaborations, or other interactions that are typically necessary for conducting clinical trials or research involving patients. Recruitment of new study participants has slowed or stopped. Relatedly, the impact on patient care has been profound. Many clinicians—including movement disorder specialists—were assigned to

other, Covid-19-related emergency duties. Dystonia patients have experienced widespread and prolonged appointment cancellations, in many cases disrupting botulinum neurotoxin injection schedules or deep brain stimulation procedures.

AT A GLANCE

- Covid-19 has impacted scientific research
- Dystonia research has not been dramatically affected
- Movement disorder clinicians are recognizing the need for novel approaches to patient care

At the same time, unforeseen opportunities have been revealed. Investigators who were forced to stay home had to come up with creative solutions to maintain research productivity. Paradoxically, the pace of publishing has not dropped. Unable to access their labs and places of work, scientists have used this time to analyze the data they already had, devoting more time to thinking about new ideas or future projects. Interestingly, the number of grant applications to the National Institutes of Health has not dropped significantly.

Crisis drives change, shifting focus to finding new, unexpected opportunities.

Collaborations within research institutions may strengthen, many internal policies and procedures will evolve, long-term support within organizations may stabilize institutional research by reducing reliance on external income sources.

Fortunately, dystonia research has not been dramatically affected. More papers are being published, grants submitted. Research that relies more on theoretical and conceptual approaches is flourishing. Some labs have begun to re-open. Although many meetings have been postponed, dystonia scientists are in constant touch by video conference and other communication means. The virtual annual meeting of the Dystonia Coalition in June attracted an unprecedented number of first-time attendees due in part to ease of participation without travel.

Movement disorder clinicians are calling attention to the urgent need for novel approaches to patient care, recognizing that the pandemic has had a distressing impact on many individuals and families impacted by chronic diseases and disorders,



including dystonia. Many movement disorder clinics have begun using telehealth and telemedicine technologies. Ironically this shift begins to finally address challenges many in the dystonia community have faced for decades, in terms of the frequent travel and mobility obstacles to visiting movement disorder clinics for treatment. Many health care providers across disciplines are mobilizing online platforms to help patients access not only important medical services but also supportive therapies such as physical and occupational therapy, and mental health services. While telemedicine consultations and assessments have their limitations, the pandemic may permanently and positively change the care of dystonia patients by making it easier to have access to doctors and health care providers outside in-clinic visits.

It will take time before we can fully comprehend the impact of Covid-19 on science and research. It has been, and will remain, profound—and hopefully, in numerous ways, for the better.

A DMRF webinar entitled “Telemedicine & Dystonia,” with movement disorder specialist Harini Sarva, MD is available for viewing at dystonia-foundation.org/treatment

Registering as a Brain Donor is Investment in Future Research



Registering in advance as a brain donor is a way to contribute to the field of dystonia research that benefits researchers working today and in the future.

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

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

About Brain Donation

- Brain tissue samples are a rare and valuable resource for dystonia investigators.
- Certain types of research studies can only be done with post-mortem brain tissue samples. Brain donors are essential to future scientific progress.
- Brain donation does not alter the appearance of the donor or interfere with memorial services or remembrance practices.
- There is no cost to the donor or family.
- Brain donation does not violate the tenets of most religious or spiritual organizations.
- Donor information remains confidential. However donors must inform next of kin that they are registered to donate their brain.
- Donated brain tissue must arrive at the HBTRC within 24 hours of death. HBTRC staff are available 24 hours a day.
- Individuals can withdraw from the program at any time.
- By combining brain donation with financial support, you multiply your contribution to the progress of dystonia research.

What is a Movement Disorder Specialist?

Dystonia is a disorder that disrupts the body's ability to move normally. It may come as a surprise to new patients that there is a subspecialty of medical doctors who have dedicated their careers to caring for people with movement disorders and researching these specific neurological problems.

A movement disorder specialist is a neurologist with intensive training and experience specifically in movement disorders. Although movement disorders have been documented for thousands of years, movement disorders as a field is relatively new in neurology.

Following medical school, a neurologist's training includes a one-year internship in internal medicine (or two years of pediatrics for child neurologists) and at least three years of specialized residency training in neurology. Many neurologists invest one or two additional years of fellowship training in a wide variety of subspecialties—movement disorders is one example. Many fellowships involve a combination of both patient care and research.

A movement disorder specialist is an essential member of the health care team for an individual with dystonia. Primary care providers, neurologists, physical or occupational therapists, mental health professionals, and additional specialists may also have important roles in contributing to and implementing a comprehensive treatment plan.

"I have had good treatment from a regular neurologist but my movement disorder neurologist is more knowledgeable about dystonia and does a better job injecting botulinum neurotoxin. I switched to the specialist and have been happy with the change," explained Carol Flynn, Leader of the Fairfield Dystonia Support Group in California. "Side effects of head drop and trouble swallowing were what encouraged me to switch. Those side effects immediately improved with the movement disorder specialist." Carol was a Registered Nurse for 40 years and was diagnosed with dystonia in 2005.

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

Tips for Locating a Movement Disorder Specialist

- A credential to identify a movement disorder specialist is that the physician has completed clinical fellowship training specifically in 'movement disorders.'
- DMRF offers a voluntary directory of physicians who treat dystonia at dystonia-foundation.org/find-a-doctor
- Child neurologists with expertise in movement disorders may be challenging to locate. Some movement disorder clinics treat children while others may refer young patients to an affiliated child neurology program.

Examples of Movement Disorders

- Ataxia
- Dystonia
- Huntington's disease and chorea
- Parkinson's disease and parkinsonism
- Myoclonus
- Restless legs syndrome
- Stiff Person Spectrum Disorder
- Tics and Tourette syndrome
- Tremors

"It took me 17 doctors to find my movement disorder neurologist, and she is everything the 16 others are not. I felt better as soon as I met her. She confirmed things that I already kind of knew about myself but didn't know why. I consider not only my neurologist who is focused in dystonia, but my DBS programmer, to be part of my 'team.' They genuinely care about me and show it. My advice to everyone still searching is don't give up. There are doctors out there who care. All the doctors who are not right for you will teach you what you want and need in a practitioner."

Lori Feldman was diagnosed with generalized dystonia in 2019 and underwent successful deep brain stimulation surgery (DBS)

In recent years, there has been tremendous advancements in new diagnostic information, pharmacological and neuro-surgical treatments for movement disorders as well as a greater understanding of the underlying causes. General neurologists, or neurologists in other subspecialties, cannot necessarily be expected to have background in movement disorders or be immersed in the latest dystonia research advancements. Some otolaryngologists, speech-language pathologists, neuro-ophthalmologists, ophthalmologists, and other health care providers may be qualified to diagnose and/or treat specific types of dystonia, depending on their training and experience.

The DMRF Clinical Fellowship Training Program is made possible by grants from Merz Pharmaceuticals, The Allergan Foundation, and Ipsen. For more information, visit:
dystonia-foundation.org/research

Wellness Resources



Living well with dystonia requires treating the physical symptoms, tending to emotional and psychological health, and accommodating your unique needs as an individual.

The DMRF offers a number of self-care resources intended to help support the well-being of individuals with all types of dystonia as well as loved ones and caregivers.

Living Well with Dystonia

dystonia-foundation.org/living-dystonia

"Dystonia & Covid-19: Your Questions Answered" with Mark Hallett, MD & Robert Chen, MB BCh, MB BChir
dystonia-foundation.org/covid-19-webinar

"Coping Strategies for Managing Stress During Covid-19" with Mary Hudson-McKinney, PT, MS, DPT, NCS
dystonia-foundation.org/wellness-webinars

"Self-Care for the Holidays" with Margie Frazier, PhD, LISW-S
dystonia-foundation.org/wellness-webinars

"Dystonia & Telemedicine" with Harini Sarva, MD
dystonia-foundation.org/treatment

Mindfulness Meditation Exercises with Karen K. Ross, PhD
dystonia-foundation.org/mindfulness

"Sleep & Movement Disorders" with Danette C. Taylor, DO, MS, FACN
dystonia-foundation.org/dystonia-sleep

Tips for Exercise

dystonia-foundation.org/benefits-of-exercise

More than a Movement Disorder: Emotional & Mental Health
dystonia-foundation.org/living-dystonia/mental-health/

"At a Loss: Dystonia & Grief"
dystonia-foundation.org/living-dystonia/mental-health

Make a Planned Gift, Make a Difference

Legacy Society Honors Visionary Donors

The DMRF was only a few years old in the late 1970s when Susan Aguilera's seven-year-old daughter Emma was diagnosed with dystonia by a special panel of neurologists in San Diego. Finally there was an explanation for why her little girl had lost the ability to write, struggled to walk, couldn't feed herself, and wasn't sleeping.

Susan recalls being "crushed" by the diagnosis and the shock of the lead physician informing her she would "have to find someone else" for treatment because no doctor in the area was familiar with dystonia.

Fatefully, Susan was made aware of John Menkes, MD, renowned child neurologist and DMRF's founding Scientific Director, practicing in nearby Los Angeles. Dr. Menkes confirmed the diagnosis and prescribed oral medications, hoping to slow the progression of the disabling symptoms. "He said the goal is to keep her on her feet, but I don't know if she will be able to write," recalled Susan, "but two years later she was writing."

Emma is now a mother and full-time social worker who works with older adults. "Emma has had long struggles, but she's still on her feet," said Susan. "I hope her story gives hope to someone. These kinds of little miracles do happen."

Over the years, Susan looked to the DMRF for information and support. She and her husband Augi became loyal donors and members of the Legacy Society. The DMRF established the Legacy Society to honor members who have generously made a lasting commitment to supporting dystonia research by naming DMRF in their estate plans. Donors who make such a pledge, at any level, are Legacy Society members.

"As a parent, I am totally grateful," said Susan "I think DMRF is outstanding. I know you are always out there. I call if I have an issue or a question, and knowing you are working on



Susan and Augi Aguilera (top) joined the Legacy Society in honor of their daughter Emma Chess, who is pictured above with daughters Maia and Qwynn.



Sheri Grube joined the Legacy Society to support dystonia research toward a cure.

Examples of Planned Gifts

Wills & Bequests

**Life Income Gifts:
Charitable Trusts
& Gift Annuities**

**Qualified
Retirement Plans**

Real Estate

dystonia has provided a level of support for me. It helps to know others are out there and that you're not alone."

Generous planned gifts have allowed the DMRF to fund crucial research aimed at developing new treatments while also supporting programs for families affected by all types of dystonia waiting for a cure.

"I'm sick of dystonia," said Sheri Grube with a laugh. "I would love a cure any time and the sooner we get more research funded the sooner that can happen." She too is a generous supporter of DMRF and member of the Legacy Society.

"Our family is blessed to be able to contribute financially to research," she explained. "My husband and I grew up poor farm kids, so we save everything for a rainy day, and I wanted to pass it on somehow. When it came to estate planning, I decided I'm doing it."

Sheri resides in Minnesota and has two adult children. She worked in the construction industry as a mechanical designer before being forced to retire due to multiple medical issues, including transverse myelitis, an inflammation of the spinal cord. She then began experiencing tremors and involuntary movements, and after five years of seeking answers was diagnosed with dystonia.

Since 2005 she has worked as a part-time visual artist and "full-time dystonia awareness advocate."

"It's really important to get the word out," she said. "People may think it's just my head shaking a little. Until they see me with my cane, they think it's invisible. They don't understand that a good day may be four hours of feeling ok, and if you overdo it those days, you might be laid up for a week to recover. Sometimes you are just trying to make it through the day, and distract yourself from the pain."

Planned gifts are contributions that are committed in the present, but given in the future. Planned gifts may offer a variety of benefits to donors, including flexibility and tax savings. They often enable individuals or families to contribute more generously to the DMRF than traditional donations. For example, one of the easiest options is to remember the DMRF in a will.

If you would like more information on planned giving options, complete and return the mailer enclosed in this newsletter, email the DMRF at legacysociety@dystonia-foundation.org, or call 800-377-3978.



Support Resources to Feel Connected

Feeling connected to others in the dystonia community can have a profound impact on health, physically and emotionally. DMRF connects you to people who understand life with dystonia:

✓ Support Groups

DMRF offers peer support groups in communities across the USA, many of which are meeting virtually by video conference. Search support groups at dystonia-foundation.org/support-groups

✓ Online Forums

Online groups are available for specific types of dystonias, parents, loved ones, young adults, and more. Find links for online forums at dystonia-foundation.org/online

✓ Individual Support

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

Benefits of Peer Support:

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

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

Update on Musician's Dystonia



Professional musicians are susceptible to a number of occupational conditions, including task-specific focal dystonia. The first signs of musician's dystonia are lapses in the usually instinctive ability to perform on the instrument. The top musical instruments associated with musician's dystonia are piano, guitar, and brass. Musician's dystonia is triggered by playing the instrument and does not typically affect other activities.

Steven Frucht, MD provided insights into recent developments in the musician's dystonia field. Dr. Frucht is Professor in the Department of Neurology at NYU Grossman School of Medicine & Director of the Division of Parkinson's and Movement Disorders. He and French Horn Player Glen Estrin founded The Leon Fleisher Foundation for Musicians with Dystonia (formerly Musicians With Dystonia) in 2000.

DD: How prevalent is focal dystonia among musicians?

SF: Several large epidemiologic studies have shown that the prevalence of musician's dystonia in professional musicians is as high as 2%. Likely, the risk to a performing musician during their lifetime of developing the condition is 1/200. This is substantially more common than the most common form of focal dystonia seen in clinic, cervical dystonia.

DD: Are musician communities more aware of focal dystonia?

SF: The Leon Fleisher Foundation for Musicians with Dystonia, and Leon and Glen's efforts have dramatically changed the way patients have been diagnosed. Also, the rise of the internet has helped patients around the world identify their problem. It has been more than a decade since I have introduced the idea of dystonia to an affected patient in the office—they come in for evaluation and already have read extensively about it. One benefit of this is that patients are diagnosed earlier, and it is easier to help them when the dystonia is less advanced. Leon's generosity in sharing his story has certainly decreased the stigma of acknowledging the diagnosis. In the US, the lack of clear protections for performing artists and absence of a financial safety net is still a big problem.

DD: How would you describe the significance of Leon Fleisher's return to two-handed playing?

SF: One could make the argument quite easily that Leon was the most important American pianist of the 20th century. His iconic recordings of the major concertos will not likely be equaled. As important, his 50 year career as a teacher, nurturing the brightest pianistic talents, will continue on for generations. His return to two-handed playing was an inspiration for every musician affected with dystonia, and for non-musician dystonia patients as well.

DD: What is the first step to developing a treatment plan for a newly diagnosed musician?

SF: The evaluation and treatment of a musician with dystonia needs to be individualized for the patient. Some patients adapt without treatment; others are in desperate need of botulinum toxin. Most important, the psychological and practical needs of the patient need to be addressed.

DD: The Leon Fleisher Foundation for Musicians with Dystonia set a path for musicians who are diagnosed and navigating life with dystonia. What accomplishments to date make you most proud?

SF: It has been the professional privilege of my career to care for more than 300 musicians, to partner with Glen and Leon to bring attention to this condition, and to meet such fascinating patients. I am most proud of the fact that patients are no longer suffering in silence, and that there is hope for meaningful recovery in many patients.

Musicians With Dystonia Renamed to Honor Leon Fleisher

By Glen Estrin, President, The Leon Fleisher Foundation for Musicians with Dystonia

The Musicians With Dystonia program of the DMRF was initiated in 2000 by movement disorder neurologist Dr. Steven Frucht and me, a professional French Horn player whose illustrious career was ended by focal embouchure dystonia at the age of 42. We established our Foundation to create awareness and provide information, support, and medical referrals throughout the world. Our first task was to assemble two Boards, a Musical Advisory and a Medical Advisory. Our search for the most appropriate Board members led first and foremost to Leon Fleisher.

Leon Fleisher, who sadly left us August 2 at age 92, was the most celebrated American born pianist of the 20th century, and was renowned internationally for his superb talent for eight decades. In 1964, at age 36, the fourth and fifth fingers of his right hand began to mysteriously curl.

The perplexing malady afflicting Leon was given dozens of diagnoses, all of them incorrect. He started performing with strictly his left hand, and turned his musical focus to conducting and teaching. After searching tenaciously for almost 25 years for an answer, Leon was finally diagnosed with focal dystonia.

Adult onset, task-specific writer's cramp dystonia was a diagnosis for the strangely thwarted and impeded performance of any fine motor skill, from surgeons with scalpels to golfers putting. The diagnosis had not been applied to musical motor skills until the relentless search by Leon and his good friend Gary Graffman, a much-admired American pianist who also exhibited the strange affliction. Maestros Fleisher and Graffman are known as the "Grandfathers of dystonia as a musician diagnosis."



Glen Estrin (L), the late Leon Fleisher (C), and Steven Frucht, MD partnered with DMRF to bring visibility to musician's dystonia.

Leon Fleisher was one of the most famous classical musicians on the planet, and the leader in the diagnosis of dystonia for musicians, so it was quite natural to ask his participation in our musician program in 2001. This warm, wonderful, generous man wholeheartedly agreed.

In 2004, Leon's tremendous support of the dystonia cause led to the phenomenal *Freedom To Play* awareness program. This 16-month long dystonia awareness initiative was worldwide, and featured

Leon as the greatest public spokesperson for dystonia ever known. In conjunction with the program, Leon received awareness awards for his significant contributions to the disorder at the annual meetings of The American Academy of Neurology, the American Neurological Association, the Movement Disorders Society, and the Society for Neuroscience.

The pharmaceutical public relations agency that implemented the program estimated the word dystonia was heard or read, amazingly, by over 250 million people worldwide due to massive print and broadcast coverage.

Not only was the *Freedom To Play* program responsible for the greatest awareness creation in the history of dystonia, it also stimulated enormous donations to the DMRF and for research. Leon Fleisher will forever be remembered as one of the greatest advocates for awareness and financial contributions the dystonia world will ever know.

Dr. Frucht and I are now extremely pleased to announce with the DMRF, and the gracious permission of Leon's widow Katherine Jacobson Fleisher, the renaming of our well-established 20-year old Foundation to honor Leon. Musicians With Dystonia will now proudly be known as The Leon Fleisher Foundation for Musicians with Dystonia.



Dystonia: Frequently Asked Questions

What is dystonia?

Dystonia is a brain disorder resulting in involuntary, abnormal postures or movements of the body due to excessive muscle contractions. Dystonia may be the only neurological disorder a person has, or the dystonia may be part of a medical condition or disease with additional neurological findings.

How is dystonia diagnosed?

At this time, there is not a single test to confirm or rule out a dystonia diagnosis. Instead, the diagnosis depends on a physician's ability to observe symptoms and obtain a patient history. Medical tests may be ordered to try to identify the cause for the development of dystonia, but a cause is often not found.

The dystonia diagnostic process may include:

- Patient history
- Family history
- Physical examination to assess functioning of the brain and nervous system
- Laboratory studies such as blood and urine tests, and analysis of cerebrospinal fluid
- Electrical recording techniques, such as electromyography (EMG) or electroencephalography (EEG)
- Genetic testing for inherited forms of dystonia
- Additional tests and screenings to uncover other possible causes for the dystonia

A movement disorder specialist is a neurologist with qualifications specifically in the diagnosis and treatment of movement disorders such as dystonia.

Do specific foods or vitamins affect dystonia?

There is no known correlation between dystonia and diet. Generally speaking, individuals with dystonia should have a nutritious diet sufficient in calories, considering the amount of muscle activity experienced

throughout the day. Stimulants such as nicotine and caffeine may make symptoms worse. Alcohol may make symptoms temporarily better (or worse) in some cases. Nutritional supplements and vitamins have not generally been shown to cause dramatic improvement in movement disorders. Substances that make a person more relaxed, calm, or sleepy may non-specifically improve symptoms. It is important for individuals to discuss any vitamins and supplements taken with their movement disorders specialist to guard against potential interactions or safety concerns. Certain foods can interfere with the absorption of specific medications.

Is tremor a symptom of dystonia?

Tremor is frequently, but not always, seen with dystonia. Differentiating between the two can be difficult in some individuals with dystonia. Tremor is an involuntary, rhythmic muscle contraction leading to shaking movements in one or more parts of the body, and dystonia is characterized by excessive muscle contractions causing abnormal, often repetitive, muscle movements and/or postures. Dystonic movements can be patterned and twisting and may resemble tremor. Individuals with cervical dystonia may have a head or hand tremor. Individuals with focal hand dystonia may have an associated writing tremor of the hand. Researchers continue to examine the relationship between tremor and dystonia.

Many thanks to past DMRF Clinical Fellow Christopher Groth, MD for reviewing the content of this article.

Look for additional FAQ in future issues of the Dystonia Dialogue.


If you have a question you wish to see addressed, email us at: contact@dystonia-foundation.org

DMRF Face Masks Promote Awareness



Help raise awareness of dystonia and follow public health recommendations when you wear DMRF's cloth face mask. Fighting for everyone with dystonia to have the freedom to move (safely) is at the heart of our mission. These masks are 100% cotton jersey, three-ply construction with two layers of jersey with a non-woven center for additional filtration. \$10 each. For more information and to order visit: dystonia-foundation.org/awareness-face-mask

Dystonia Moves Me Bingo Challenge



Dystonia Awareness BINGO

Take the Dystonia Moves Me bingo challenge!
Complete a row or the whole card.

D	M	R	F
WILDCARD Do Something Creative for Awareness	Read Article in Dystonia Dialogue	Share DMRF Post on Social Media	Join DMRF Email List
Sign-Up for DAN Alerts	Pass Along Your Dystonia Dialogue	Start a Facebook Fundraiser	Use Donate Sticker on Instagram
Share DMRF Website on Social Media	Watch Video on DMRF YouTube Channel	Order Info Cards	Give Info Card to Family Member
Post Awareness Selfie	Join Global Dystonia Registry	Give Info Card to Stranger	WILDCARD Do Something Creative for Awareness

Visit <https://dystonia-foundation.org/bingo> for hints and tips for completing each awareness action.

September was Dystonia Awareness Month, but promoting dystonia awareness is essential all year round. If you missed the opportunity to take this year's Dystonia Moves Me Bingo Challenge, any time is a good time to participate. The bingo card above contains 16 ways to promote dystonia awareness. How many can you complete—a row? The whole card?

DMRF makes it easy by providing tips and tricks to compete each dystonia awareness action at dystonia-foundation.org/bingo



Daria is a 2020 Douglas Kramer Young Advocate.

PERSONAL PROFILE

Daria Dragicevic

Daria Dragicevic resides in the San Francisco Bay Area of California. She is a university student studying linguistics and computer science, in the process of applying to graduate programs in neuroscience.

How have you been doing during the pandemic?

I was supposed to quit the spring semester because I got approved for DBS [deep brain stimulation] surgery. Then my DBS surgery got delayed, so that was pretty heartbreaking, but I ended up finishing the spring semester, which is great. And then I was the first person to get in for DBS at my hospital. So, it's kind of been a blessing to have the virus happen during my DBS because I've been able to recover at home where I can do research and do school work without having to get around my huge college campus in San Diego.

How did your dystonia symptoms start and how were you diagnosed?

I didn't have symptoms until my freshman year of college. I was 18. I started having extreme tremors in my hands and arms. I put it off, thinking I pulled a muscle. After three weeks I went to urgent care to get it checked out, and a unique thing happened. The doctor had essential tremor. He was insistent that I see a neurologist. I saw a movement disorder specialist and that's when my journey started. After genetic testing, it was discovered that I have an extremely rare genetic form of dystonia called dystonia 27 [DYT27/COL6A3] with only six other individuals currently identified with that type of dystonia. My symptoms affect my whole body now, but at first I just had tremors and cramping in my hands and arms. I wasn't able to hold a pen, put on makeup, and eventually I wasn't able to cut my own food or do any fine motor control tasks. I started experiencing jaw and neck dystonia. It constantly feels like my teeth are chattering. My neck is constantly spasming, so it's painful in that area. Then about a year after my diagnosis I started experiencing numbness in my legs and soon my right foot became contorted. It was

stuck inverted. Within three months, I went from playing collegiate water polo to needing a cane, to arm crutches, and then eventually a wheelchair. Really quick and very scary. I just thought I was going to have tremors for a while but it got worse so quickly.

How did you deal with the social aspect at school?

I have a really supportive group of friends in San Diego, which is fantastic. Even though we all became friends through water polo, they were very supportive when I had to stop playing, and my coach told me I could help coach our team instead of playing on the team. So I felt still connected. There's 36,000 students on campus, and at first I felt really awkward walking around with a cane or my forearm crutches. Once I got comfortable with it, I stopped noticing the looks people would give me. I'd have random friends, who didn't know I had dystonia at that point, say, oh you hurt your foot. At first I just said yeah, I hurt my foot, not wanting to talk about it. Eventually I realized it could be a good platform to explain, no, I have this neurological disorder called dystonia and this is what dystonia is like. It became a good way to talk about it.

How did you come to the decision about DBS?

We became very aware of DBS before I even was considering it. I knew it was something we were going to have to turn to unless I find a perfect medication. I was having horrible medication reactions. I was getting worse so quickly, and my doctor was pretty confident that surgery would help me, which it has.

I first had my left side of my brain done, my left electrode. The scariest part was, because it was during the pandemic, I couldn't have anyone there with me. I went into the hospital

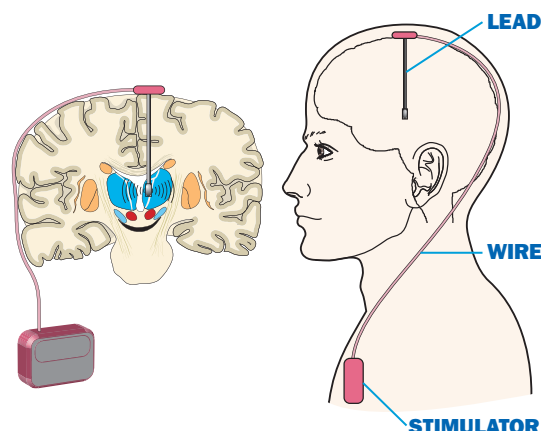
by myself. They let my mom stay for when I got my head shaved, which was really nice. I had never had surgery before. Everything went well. I called my mom 20 minutes after my surgery. I was awake through all of it. Obviously, it was a really intense experience, but I'm so interested in the science behind DBS that it was less scary because I was constantly asking my surgeon questions. He said I've never had someone talk so much during surgery. But I was so interested in what was going on and what they were doing. Two weeks later I came back, got my head re-shaved, and they did the right side electrode. Then I got put to sleep and they connected everything and placed my battery.

The recovery was pretty easy. Maybe after two weeks I was back doing my regular thing. Having the battery placement in my chest was weird because I'm still not fully used to it. I've only had a couple of programming sessions. I'm walking. My team is so happy with my progress. I'm the first patient with dystonia 27 to have this surgery. So they're planning on hopefully publishing a paper about it so other people know how well this form of dystonia, or at least mine, has reacted. My tremors are gone a lot. I'm still taking medications but probably once we get my foot a little bit more dialed in it'll be pretty great. It's been the best decision of my life so far. It's helped me so much in such a short period of time. I'm so happy to not be in pain every single day. It's much easier to do school, do research, feel like I'm living more of a normal life. There's some pain still, and there are things to work on, but the progress I've made helps me have a positive mindset.

What has been the greatest challenge of dystonia and how did you get through it?

Obviously it's a huge change in lifestyle. But the greatest challenge that I faced is the lack of awareness around this disorder. So many times I went to the hospital in such a state of despair, by myself, and had doctor after doctor say things like, *How do you want me to treat you? Should I admit you? What is dystonia?* I don't mind explaining the condition, but there's such a huge lack of knowledge behind dystonia that results in poor treatment. That is one of the issues that drives me to want to advocate and be a researcher in movement disorders because if I wasn't an expert on my own disease, I wouldn't be able to get proper treatment. The majority of the time, even though I'm only 21, I'm my own doctor in certain cases. I think a lot of people with dystonia feel this way. If you don't become an expert in your disorder, no one else around you will.

What is Deep Brain Stimulation?



Deep brain stimulation (DBS) is a surgical procedure used to treat a variety of neurological disorders, including dystonia. A battery-powered stimulator is implanted in the body and delivers electrical stimulation to the areas of the brain responsible for causing dystonia symptoms. The stimulator is implanted in the chest, and extension wires connect the stimulator to leads (electrodes) deep in the brain. In most cases, both sides of the brain are treated. The stimulation to the brain is adjusted by remote control to achieve the appropriate settings for each patient.

Adult patients are typically awake during the process of implanting the leads. Patients may be asked to respond to questions and instructions from the surgical team in the operating room. The stimulators and wires are implanted under general anesthesia. After a brief period of healing, the stimulator settings are activated and adjusted over a series of appointments. It can take weeks or months for individuals to achieve full benefit.

More information is available at dystonia-foundation.org/treatment

Dystonia Dialogue

Dystonia Medical Research Foundation

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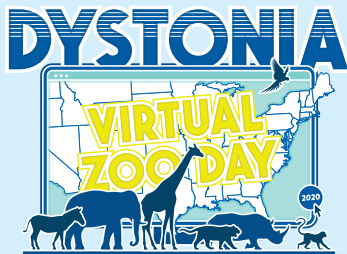


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