



DYSTONIA
MEDICAL
RESEARCH
FOUNDATION

Dystonia Dialogue

NEWSLETTER OF THE DYSTONIA MEDICAL RESEARCH FOUNDATION

Spring 2022 | Volume 45 • No.1

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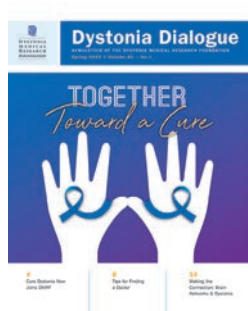
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researchers, uniting people of all backgrounds and abilities to better serve those living with this life-changing disorder.

The DMRF is governed by a volunteer Board of Directors led by the President. The Medical & Scientific Advisory Council (MSAC), led by the Scientific Director, is comprised of pre-eminent scientists with expertise across multiple medical and scientific disciplines. Read more about the role of the MSAC on page 6.

The *Dystonia Dialogue* is supported by an educational grant from Ipsen Biopharmaceuticals.

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

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

The *Dystonia Dialogue* reports on developments in dystonia research and treatments but does not endorse or recommend any of the therapies discussed. Individuals are urged to consult a physician with questions and concerns about their symptoms and care.

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Foundation Update



Mark Rudolph
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The best interest of individuals with dystonia and their families is at the heart of everything the DMRF does. Our efforts are led by people who know dystonia personally—the daily challenges and life-altering experiences. The DMRF is committed to uniting the community so that no one has to face dystonia alone and everyone has the opportunity to be part of the mission to find a cure.

Several organizational changes at DMRF are positioning us well for a productive year. We extend our deep gratitude to Immediate Past President Art Kessler for 12 years of superb leadership. Art will fortunately remain on the Board of Directors. We are looking forward to returning to in-person Dystonia Zoo Days and additional community events, Covid-19 conditions permitting. Similarly, some of our support groups are returning to meeting in person. DMRF will continue to offer virtual opportunities and online webinars so that anyone can participate, regardless of their location.

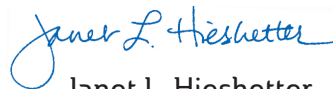
We are also pleased to announce that Cure Dystonia Now has joined DMRF, and we are combining forces to aggressively and strategically advance dystonia research that improves people's lives. Read more about this partnership on page 4.

In the meantime, as new medical and research discoveries accumulate, DMRF remains steadfast as a source of information and support for individuals with all types of dystonia. Access to expert medical care is critical to living well with dystonia, and attending to emotional and mental health is equally important. In 2022 DMRF will continue to highlight topics that encourage a holistic approach to treating dystonia, with an emphasis on mental health. In this newsletter, you will find tips for overall emotional wellness on page 10, and we look forward to bringing you additional relevant content throughout the year.

Thank you for being part of the DMRF.



Mark Rudolph
President



Janet L. Hieshetter
Executive Director

P.S. An interview with DMRF's newly elected President Mark Rudolph is available for viewing at dystonia-foundation.org/new-bod

SIGN UP FOR DMRF'S MONTHLY E-NEWS: dystonia-foundation.org/email

First Issue of *Dystonia* Planned for Spring



As previously announced, the DMRF has partnered with Frontiers Media to launch *Dystonia*, an open access journal. The journal will bring visibility

to the growing dystonia field and highlight advancements in science and clinical practice. This is the first scientific journal of its kind and a major milestone for dystonia research. The inaugural issue is scheduled for publication in the spring of this year.

Up until now, dystonia studies have been published in numerous available journals. *Dystonia* provides a centralized, go-to publication by and for dystonia investigators. It is Gold Open Access, which means the content is available to the medical community and public at no charge to readers.

More information is available at frontierspartnerships.org/journals/dystonia

The journal is partially supported by the Joan Miller Young Investigator Fund and Tuft Family Foundation.

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Cure Dystonia Now Joins DMRF



Cure Dystonia Now Founder Marc Miller (L) and DMRF Board Members President Mark Rudolph and Director Diane Rudolph.

Cure Dystonia Now (CDN) has joined the DMRF to further the organizations' mutual goals to benefit the dystonia community. CDN Founders Robin and Marc Miller were elected to the DMRF Board of Directors in February. The Millers established the DMRF Cure Dystonia Now Fund in continuation of their commitment to advance outstanding research by leading dystonia investigators.

"The Millers have been generous supporters of DMRF for years, and together we've funded critically important research investigations. We are excited to welcome them to our Board and begin this new chapter of collaboration," said DMRF President Mark Rudolph.

"This partnership will reduce redundant efforts and get us closer to what we all want—better treatments and ultimately a cure for dystonia. We're very much looking forward to continue working with DMRF, now as a part of this great organization," stated Marc Miller.

For more information about DMRF research efforts, visit: dystonia-foundation.org/research

New Consensus Guidelines for Botulinum Neurotoxin Therapy for Dystonia & Spasticity

Botulinum neurotoxin (BNT) therapy is one of the most common treatments to reduce dystonia symptoms. BNT therapy involves injecting appropriate doses of BNT into the appropriate selection of muscles. Both the muscles to be treated and the specific doses determine the dosing regimen for an individual patient. Developing the unique dosing scheme for each individual patient requires considerable experience on the part of the physician, making BNT therapy both a skill and an art. Dosing tables are available for physicians to provide a range of optimal doses for each target muscle.

An international working group of movement disorder experts recently published new and improved dosing tables for physicians with the goal of making BNT therapy more effective for patients. For the first time the dosing tables are based on statistical evaluation of real-life data from a large BNT clinic. "So far dosing tables have only been reporting dose ranges coming out of group discussions. Usually, they covered enormous ranges making them virtually useless for practical considerations," explained Dirk Dressler, MD, PhD,

first author of the study and Head of the Movement Disorders Section in the Department of Neurology, Hannover Medical School in Germany. "We give typical doses for each target muscle, we give variability, and we give limits so that the [physician] has all the information to decide the best target muscle dose." The statistical analysis was based on treatment data from 1,831 BNT injections in 36 different target muscles in 420 dystonia patients plus 1,593 BNT injections in 31 different target muscles in 240 spasticity patients. The investigators differentiated between dosing for dystonia and spasticity, which existing dosing tables do not offer. "We are extremely grateful to our panel of 26 world class experts in BNT therapy from all over the world. We are confident that this publication will be the key source for planning and dosing BNT therapy in dystonia and spasticity for the years to come," said Dr. Dressler.

Dressler D, Altavista MC, Altenmueller E, et al. Consensus guidelines for botulinum toxin therapy: general algorithms and dosing tables for dystonia and spasticity. J Neural Transm (Vienna). 2021;128(3):321-335.

Community Mourns Disability Activist Neil Marcus

DMRF regrets to report that beloved disability activist and longtime member Neil Marcus died in late 2021. For decades, Neil used his life and his art to challenge how society categorizes and reacts to individuals with disabilities. He became an iconic figure in the disability culture movement that coalesced in the late 1970s and 1980s by using theater, dance, poetry, humor, and visual arts to express his unique vision: "Disability is not a brave struggle or 'courage in the face of adversity.' Disability is an art. It's an ingenious way to live." He danced internationally since the 1980s, received a United Nations Society of Writers Medal of Honor for his play *Storm Reading*, and is included as a seminal voice in the National Endowment for the Arts Oral History Project. He appeared on the hit television show, *ER*. He published numerous zines, books, and academic works. His poetry has been published internationally in various media including in the prelude to *A Disability History of the United States*. Neil's extensive body of work leaves a rich legacy that will influence generations to come. His obituary appeared in *The New York Times*. Photo by Gary Ivanek.



Leading Experts Guide DMRF Research Strategy

Role of the Medical & Scientific Advisory Council

The DMRF Medical & Scientific Advisory Council (MSAC) provides advice and recommendations to the Board of Directors on research strategy for the science program. The MSAC is made up of distinguished experts in neurology, neuroscience, biochemistry, microbiology, genetics, epidemiology, and related fields. They continually assist DMRF in identifying new research avenues, conducting peer-review of research proposals, strategizing how to attract other experts to the dystonia field, and co-organizing and participating in DMRF scientific meetings.



The MSAC is made up of scientific and clinical experts who volunteer their time and expertise to guide DMRF research efforts.

“One of the greatest strengths of the DMRF over the years has been an active and engaged MSAC,” said DMRF President Mark Rudolph. “These experts are leaders in their fields, and we’re grateful for their insights and ideas on how the DMRF can be the most efficient, effective engine for outstanding dystonia research.”

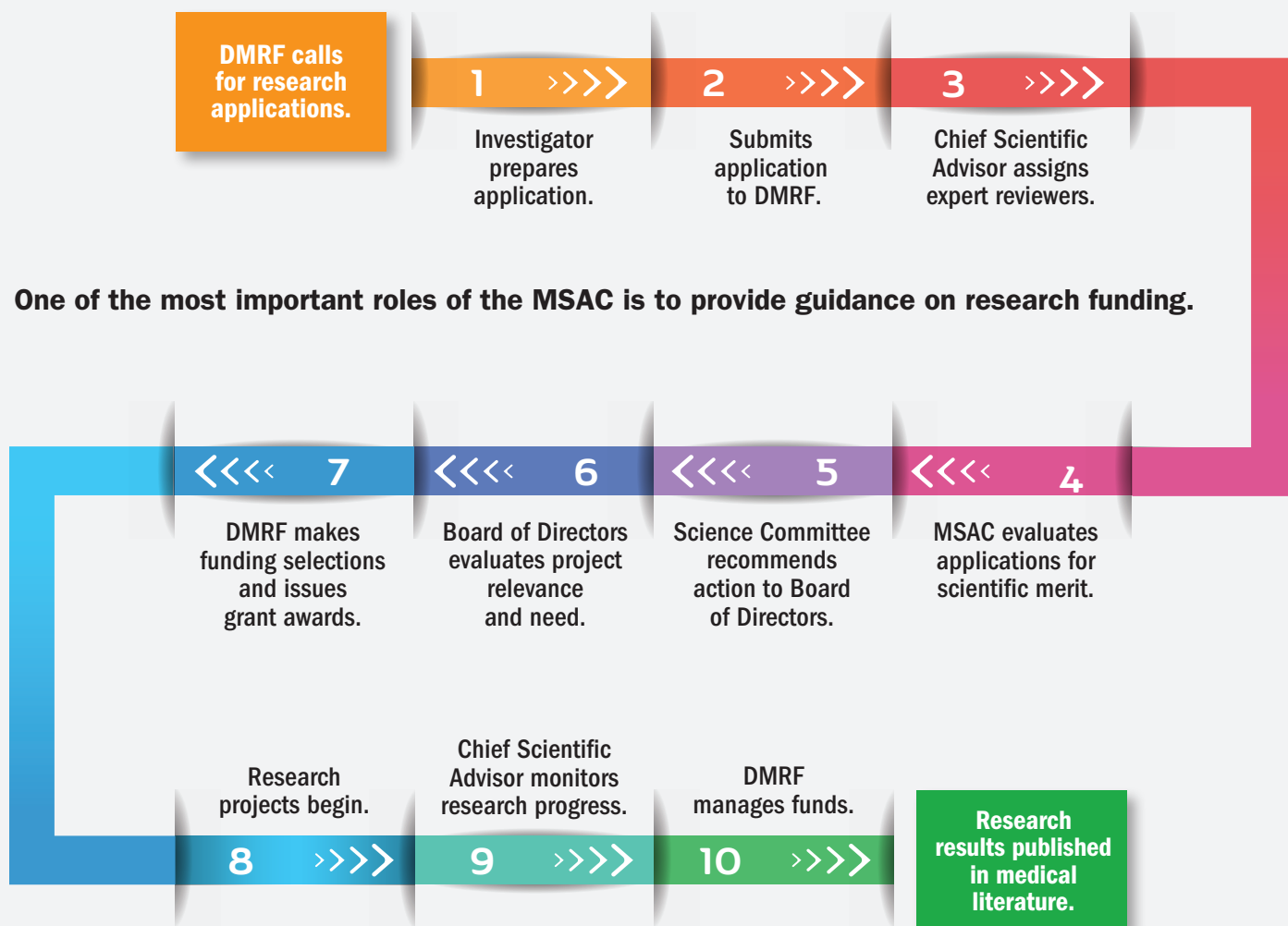
The MSAC is led by professionals of the highest caliber: Scientific Director Emeritus, Mahlon R. DeLong, MD of Emory University School of Medicine, winner of a 2014 Breakthrough Prize in Life Sciences; Scientific Director Joel S. Perlmutter, MD, Elliot Stein Family Professor of Neurology at Washington University in St Louis; Chief Scientific Advisor Jan Teller, MA, PhD, who works round-the-clock to help manage the Foundation’s research program; and DMRF Vice President of Science and Chair of the Science Committee

Richard Lewis, MD, Professor Emeritus in the Department of Orthopaedics, University of Rochester.

One of the qualities that makes DMRF unique among health advocacy organizations is the degree to which the MSAC and Board of Directors interact. The Board of Directors get to know the leading experts in dystonia research, and the scientists get to know the families whose experiences with dystonia embody why the work of DMRF is so important. For many basic researchers, whose work takes place in the lab rather than the clinic, the Foundation provides them with their first and unique opportunity to meet someone diagnosed with the disorder they study.

[Learn more about DMRF’s science and research efforts at: dystonia-foundation.org/research](https://dystonia-foundation.org/research)

OVERVIEW OF DMRF GRANTS PROCESS



One of the most important roles of the MSAC is to provide guidance on research funding.

The DMRF was proud to welcome four new members to the MSAC at the annual meeting in February 2022:

Scott Norris, MD

Washington University in St. Louis

Dr. Norris is a movement disorder specialist whose research interests include the pathophysiology of dystonia and the mechanisms of deep brain stimulation. He is a past DMRF Clinical Fellow and grant recipient.

G. W. Gant Luxton, PhD

University of California-Davis

Dr. Luxton is an expert in molecular and cellular biology with research interests in dystonia-causing proteins. He is a past DMRF grant recipient.

Andrea Kühn, MD

Charité University Medicine, Berlin

Dr. Kühn is a movement disorder neurologist whose research interests include deep brain stimulation and neuromodulation for movement disorders. She is a past DMRF grant recipient.

Sarah Pirio-Richardson, MD

University of New Mexico

Dr. Pirio-Richardson is a movement disorder neurologist who research interests include non-invasively altering plasticity in the brain and adult onset focal dystonia.

For a complete list of MSAC members, visit dystonia-foundation.org/people



Tips for Finding a Dystonia Doctor

One of the most common inquiries the DMRF receives is for assistance locating a physician who is qualified to diagnose and treat dystonia. They may have received a tentative diagnosis from a general neurologist and need a movement disorder specialist to confirm or rule out dystonia.

Their doctor may have relocated or retired. Or maybe they are interested in getting an additional medical opinion on treatment options. Because treating dystonia is so specialized, seeing the appropriate medical professional can make a dramatic difference in treatment options and results from treatment. Helping individuals and families locate qualified doctors is one of the most important services DMRF provides.

In August, Judy Draper contacted DMRF via Facebook for assistance locating a doctor and was provided options near her home in Texas. She said, “I looked them all up on the internet and selected several to call. I could not get an appointment with the doctors from the list immediately, but I did get an appointment for that week with a new member in one of the practices. After three years and seeing seven other doctors and having no answers, my husband and I sat and talked with him for over an hour. He answered our questions and has become a very valued member of my healthcare team.” Judy was diagnosed with focal foot dystonia associated with Parkinson disease.

Suggestions for locating a qualified doctor include:

Reach Out to DMRF for Assistance

It may come as a surprise that not all doctors can be expected to diagnose and treat dystonia. The DMRF maintains a voluntary directory of doctors who treat dystonia at dystonia-foundation.org/find-a-doctor. Also, DMRF staff is available by phone, email, and social media to provide individual assistance with finding a doctor.

AT A GLANCE

- Not all doctors can be expected to diagnose and treat dystonia.
- Seeing a qualified doctor can make a big difference in treatment options and outcomes.
- DMRF can help individuals locate doctors in their area.
- Support groups can also help identify doctors.

Consult a Movement Disorder Specialist

Dystonia is a disorder that disrupts communication between the brain and muscles, and this prevents the body from moving normally. 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 including

dystonia, Parkinson disease, essential tremor, tics, and more. Learn more about movement disorder specialists at: dystonia-foundation.org/movement-doctor/

Check Credentials

One credential to identify a movement disorder specialist is that the physician has completed clinical fellowship training specifically in movement disorders. Some movement disorder specialists may have clinical fellowship training in neurophysiology, but this sometimes indicates more of a specialty in epilepsy and seizure disorders rather than movement disorders. 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. Information about a doctor's training and credentials may be available on the medical office's website.

Consider a Physical Medicine Specialist

Depending on the type of dystonia, additional medical professionals may have appropriate credentials to offer

diagnosis and treatment. An increasing number of physical medicine and rehabilitation (PM&R) doctors (sometimes called “physiatrists”) treat dystonia, especially dystonia associated with cerebral palsy, stroke, and brain injury. Credentials that may indicate a PM&R doctor has expertise in dystonia include certification in electrodiagnostic medicine (EMG) and advanced training in neurorehabilitation. A small number of PM&R physicians have advanced fellowship training specifically in movement disorders neurorehabilitation. Learn more about PM&R doctors at: dystonia-foundation.org/pmr-physician/

Special Considerations for Focal Dystonia

Additional medical specialties may treat focal dystonia that affects a specific area of the body. For example, neuro-ophthalmology is an ophthalmic subspecialty that addresses the relationship between the eye and the brain. This can include blepharospasm, a focal dystonia of the eyelid and brow muscles. Although experience with blepharospasm may vary because it is a rare disorder compared to more common eye conditions, neuro-ophthalmologists are often very skilled in administering botulinum neurotoxin injections for eye conditions, including blepharospasm.

Laryngeal dystonia (also known as spasmodic dysphonia) is a focal dystonia of the vocal cord muscles. Treatment often involves collaboration between a speech-language pathologist and otolaryngologist (ear, nose, and throat specialist). An otolaryngologist who has further subspecialized in the larynx (voice box) and voice is a laryngologist.

Additional Sources

Your health insurance provider is an important source for identifying doctors and clarifying whether they are covered by your plan.

DMRF support groups and online forums may also help identify doctors. You may be able to connect with individuals who have firsthand experience with a particular doctor, the medical institution, and administrative staff—all of which may affect selecting a doctor that is right for you.

To locate support groups and online forums visit: dystonia-foundation.org/support/

How Can I Participate in Dystonia Research?

Research progress toward a dystonia cure cannot happen without volunteers. You can support dystonia research discoveries in more ways than one.

Consider these opportunities to have an impact:



Ask your Doctor: Your movement disorder specialist may be participating in a clinical trial that needs volunteers.



Search Online for NIH Studies: Search for dystonia clinical studies supported by the National Institutes of Health at [nih.gov/health/clinical trials](http://nih.gov/health/clinical-trials)



Register as Brain Donor: Individuals with all types of dystonia are invited to register in advance as a brain donor. For more information visit dystonia-foundation.org/brain



Join the Global Dystonia Registry: Registries help researchers better understand dystonia by collecting information directly from patients. The Global Dystonia Registry is confidential and you can un-enroll any time. For more info: globaldystoniaregistry.org



Donate: Financial contributions to the DMRF support cutting-edge research by the world’s foremost thought leaders and up-and-coming experts. Use the envelope enclosed in this newsletter to make a gift or donate online at dystonia-foundation.org/donate

DMRF extends sincere thanks to all those who contribute to research.

Emotional Wellness Toolkit

How you feel can affect your ability to carry out everyday activities, your relationships, and your overall physical and mental health. How you react to your experiences and feelings can change over time. Emotional wellness is the ability to successfully handle life's stresses and adapt to change and difficult times. The National Institutes of Health offers these tips for protecting and improving emotional health.

LOOK FOR THE POSITIVE

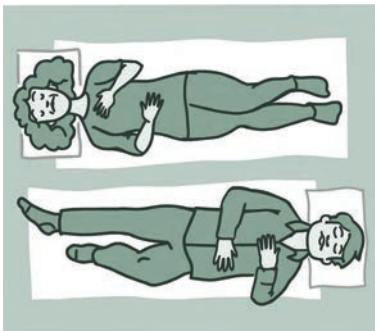


People who are emotionally well have fewer negative emotions and are able to bounce back from difficulties faster. Another sign of emotional wellness is being able to hold onto positive emotions longer and appreciate good times.

To develop a more positive mindset:

- Remember your good deeds.
- Forgive yourself.
- Practice gratitude. Being thankful stirs up positive emotions.
- Spend time with your friends.
- Explore your beliefs about the meaning and purpose of life.
- Develop healthy physical habits.

MANAGE STRESS



Living with dystonia can be stressful, even under the best of circumstances. Stress can give you a rush of energy when it is needed most. But if stress lasts a long time—a condition known as chronic stress—staying on high alert can become harmful rather than helpful. Learning healthy ways to cope with stress can also boost resilience.

To help manage your stress:

- Get enough sleep.
- Build a social support network.
- Show compassion for yourself.
- Ask for help.
- Exercise regularly.
- Set priorities.
- Try relaxation methods.

GET QUALITY SLEEP

Dystonia can make it difficult to sleep. Plus, to fit everything we need to do in a day, we often sacrifice sleep. But sleep affects both mental and physical health. It is vital to your well-being. Sleep helps you think more clearly, have quicker reflexes, and focus better. Take steps to make sure you regularly get a good night's sleep.



To get better quality sleep:

- Go to bed and get up each day at the same time.
- Sleep in a dark, quiet place.
- Exercise daily.
- Limit the use of electronics before bed.
- Relax before bedtime.
- Avoid alcohol before bedtime and stimulants like caffeine or nicotine.
- Talk to your doctor if you have ongoing sleep problems.

BE MINDFUL

The concept of mindfulness is simple: this practice is about being aware of what is happening in the present moment—notice all that is going on inside and all that is happening around you. It means not living your life on autopilot. Becoming a more mindful person takes time and practice. Here are some tips to help you get started.



To be more mindful:

- Take moments throughout the day to pause and notice the sights and sounds around you.
- Take some deep breaths in through your nose to a slow count of 4, hold for 1 second, and then exhale through the mouth to a slow count of 5. Repeat often.
- Practice mindful eating. Be aware of each bite and feel when you are full.
- Be aware of your body. Do a mental scan from the top of your head to the bottoms of your toes, bringing your attention to how each part feels.
- Find mindfulness resources to help you practice, including online programs.

GRIEVE LOSS



Loss can include a death, divorce, or any circumstance in which your world changes—including the lifestyle and ability changes often caused by dystonia. There is no right or wrong way to grieve. Although loss can feel overwhelming, it is possible to move through the grieving process. Learn healthy ways to help you through difficult times.

To help cope with loss:

- Take care of yourself.
- Share your feelings with a caring friend or family member.
- Try not to make any major changes right away after a painful loss.
- Join a support group.
- Consider professional mental health support.
- Talk to your doctor if you are having trouble with everyday activities.
- Be patient. Grief takes time.

STRENGTHEN SOCIAL CONNECTIONS

Social connections help protect health and lengthen life. Scientists are finding that our links to other people can have powerful effects on our health—both emotionally and physically. Whether with romantic partners, family, friends, neighbors, or others, social connections can influence our biology and well-being.



To build healthy support systems:

- Build strong relationships with your family.
- Share good habits with family and friends, such as exercise or wellness activities.
- If you are a caregiver, ask for help from others—especially if you are also coping with dystonia.
- Join a group focused on a favorite hobby, such as reading, hiking, or painting.
- Take a class to learn something new.
- Volunteer for things you care about in your community, like a community group, school, library, or place of worship.
- Experience different places and meet new people.
- Join a DMRF support group.

Adapted from *Your Healthiest Self: Emotional Wellness Toolkit*. Source: nih.gov/wellnesstoolkits



Research Bits

There have been a record number of dystonia clinical studies published in recent years. Here are just a few interesting examples.

Investigating Remission

There are occasional reports of dystonia symptoms going into remission. Given the research and prognosis implications of this, investigators did a systematic review of reports of remission in the medical literature. They reviewed data from 2,551 cases with reports predominantly from individuals with cervical dystonia or blepharospasm/Meige syndrome. Complete remission was reported in 12% and partial remission for 4% of cases. Remission rates were higher in cervical dystonia (15%) than in blepharospasm/Meige (6%). Remission occurred on average 4.5 years after onset of symptoms. However, the majority of patients (64%) experienced a return of symptoms. The data suggested that patients with remission were significantly younger at symptom onset than patients without remission. The investigators called for additional study into this important phenomenon.

Mainka T, Erro R, Rothwell J, Kühn AA, Bhatia KP, Ganos C. Remission in dystonia - Systematic review of the literature and meta-analysis. Parkinsonism Relat Disord. 2019 Sep;66:9-15.

Deep Dive into Oromandibular Dystonia

In a study partially supported by DMRF, an international team of investigators embarked on the most comprehensive examination of the clinical features of oromandibular dystonia (OMD) with the purpose of reducing misdiagnosis. The symptoms of OMD include varying combinations of abnormal jaw, tongue, or lower face movements. OMD is particularly disabling because it often interferes with eating and speaking and can cause severe discomfort. Isolated OMD is estimated to account for only 3–5% of all dystonias. Of the 2,020 cases of OMD reviewed, typical age at onset was in the 50s, and 70% of patients were female. The muscles of the lower face were most commonly affected, followed by jaw, and sometimes tongue. OMD more commonly appeared as part of segmental dystonia, rather than occurring as a focal dystonia or within generalized dystonia. Social anxiety and depression were prominent. Botulinum neurotoxin injections improved symptom severity by more than 50% in approximately 80% of patients.

Scorr LM, Factor SA, Parra SP, Kaye R, Paniello RC, Norris SA, Perlmutter JS, Bäumer T, Usnich T, Berman BD, Maily M, Roze E, Vidailhet M, Jankovic J, LeDoux MS, Barbano R, Chang FCF, Fung VSC, Pirio Richardson S, Blitzer A, Jinnah HA. Oromandibular Dystonia: A Clinical Examination of 2,020 Cases. Front Neurol. 2021 Sep 16;12:700714.

Survey of Musicians Reveal 2%+ Affected by Dystonia

A team of researchers in Brazil set out to evaluate the clinical characteristics and frequency of task-specific dystonia in musicians and to promote awareness of the condition among musicians across the country. They visited orchestras and music schools to deliver lectures on musician's dystonia and invite musicians to complete a questionnaire. They visited 51 orchestras and music schools in 19 Brazilian cities, collecting over 2,200 questionnaires. Of those, 72 individuals with suspicion of dystonia were video recorded and evaluated for motor impairment. Forty-nine individuals (2%+) were diagnosed with dystonia. This is in range with rates reported in previous studies from various countries. The instruments most associated with task-specific dystonia were acoustic guitar (37%) and brass instruments (31%). They concluded that Brazilian musicians with dystonia tend to be male, classical music professionals, around 30 years of age, with arms, hands, or oromandibular muscles most often affected. The research team stressed the career-altering impact of dystonia in musicians and called for greater awareness among musicians, music instructors, and health professionals.

Moura RC, de Carvalho Aguiar PM, Bortz G, Ferraz HB. Clinical and Epidemiological Correlates of Task-Specific Dystonia in a Large Cohort of Brazilian Music Players. Front Neurol. 2017 Mar 6;8:73.

"Musicians with Dystonia Support Forum" is a private Facebook group available for musicians affected by task-specific focal dystonia. Visit the group at: facebook.com/groups/musiciansdystonia

Worth a Thousand Words: How to be a Respectful Communication Partner to AAC Users

By Caitlyn Connelly

You may have seen someone write in a notebook or text to answer a question. Maybe you have seen people using sign language or other gestures. You may have seen someone push buttons on a computer that speaks for them. These are all forms of augmentative and alternative communication (AAC).

AAC includes all the ways we share ideas and feelings without talking. It encompasses the communication methods used to supplement or replace speech or writing for those with impairments in the production or comprehension of spoken or written language. Some members of the dystonia community may use AAC. The goal of AAC is to achieve the most effective communication for the individual in order to maximize their potential and lead the highest quality of life possible.

**“Be patient. Talk to me.
Listen to me.”**

There are many types of AAC from alphabet boards to

iPads with specialized apps to DynaVoxes or specialized computers. The Picture Exchange Communication System (PECS) allows people with little or no communication abilities to communicate using images. People using PECS are taught to approach another person and show them an image of a desired item in exchange for that item.

I am able to speak, but it is often difficult for outsiders to understand me; you need to focus on what I am saying and not be distracted. I cannot speak clearly or fast enough for strangers to understand and be patient. When I have a lot to say it can become jumbled and you may only get bits and pieces. A lot gets lost in translation. My parents, siblings, and close friends understand me, especially in one-on-one, quieter settings. I use my iPad to communicate with others. People make assumptions that I cannot speak and/or understand. If I only communicated through text people would not realize I had a disability. People, including doctors, are impressed with my writing ability. It changes their view of me.

Be patient. Talk to me. Listen to me. These are key messages for good communication partners. Consider these strategies to put the AAC user first in your conversations.

Good communication partners...

- Respect the effort AAC takes
- Are patient and wait
- Help manage noise
- Help manage physical space
- Watch the person, not the device
- Pay attention to the message—and other cues
- Do not dominate the conversation
- Respect an AAC user’s voice
- Ask questions
- Accept that communication is on the AAC user’s terms

For me, personally, if it is quiet, I prefer to try using my voice first especially if the person I am talking to is patient or familiar with me, because typing takes a lot out of me and takes a really long time. However, sometimes words just get stuck and it’s easier to use my iPad.

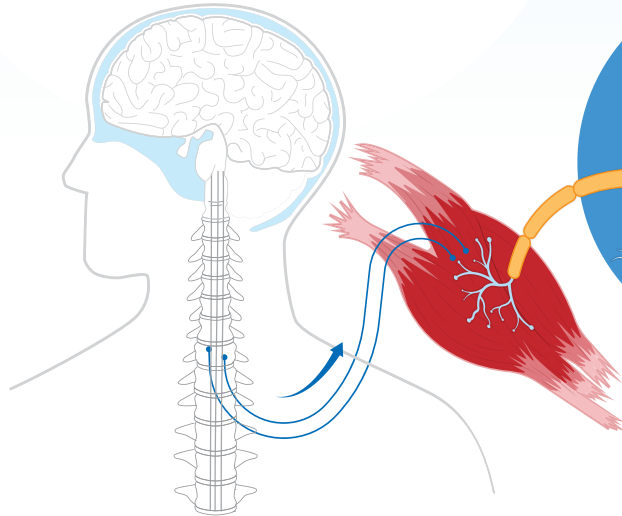
Everyone has their own unique voice, experiences, and desire to connect and be included, no matter their communication style.

It is always important to ask the individual their own personal preferences, to be patient and understanding, to speak to the individual as an equal, to not hover over the individual, to not touch the person’s device unless the person says it’s okay and always, always put the person first.

Caitlyn Connelly is 24 years old, a graduate of Holy Family University, and lives in Pennsylvania. She has cerebral palsy and dystonia. She received a Certificate of Appreciation from DMRF as a high school student for organizing the first-ever Stomp Out Dystonia Event. Caitlyn and her family are dedicated DMRF supporters. This article is adapted from her blog, A Little Imperfect, Entirely Me 2.0!

MAKING THE CONNECTION

Untangling Complex Neural Networks is Central to Understanding Dystonia



Excessive muscle contractions are a hallmark sign of dystonia, but the muscles are not the source of the problem. The muscles are responding to abnormal signals from the brain to flex or relax, which cause the dystonic movements and postures.

Research shows that the origins of dystonia lie in the complex pathways and networks of neurons that carry signals from one part of the brain to another.

There are an estimated 86 billion neurons in the human brain, making roughly 100 trillion connections. Neural pathways convey the information and instructions necessary for the brain to function. If there is a problem anywhere along a pathway, the communication between brain structures in that network breaks down. When areas of the brain responsible for movement cannot communicate properly, movement disorders such as dystonia can occur.

Researchers from around the world are painstakingly working to identify the neural pathways and networks implicated in dystonia and pin-point dysfunction within these complex

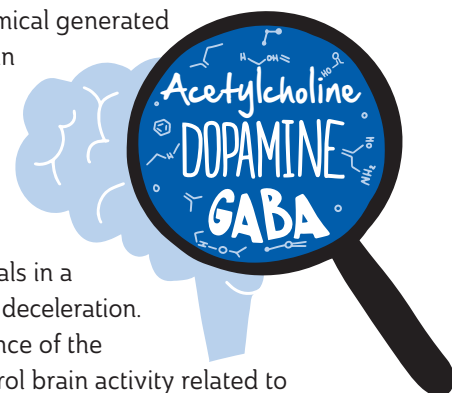
connections. Once the problem areas are identified, researchers can strategize effective treatment approaches.

In late 2021, over three days, dystonia experts from across the world met for a virtual workshop, “Defining the Role of Brain Networks in the Pathophysiology and Treatment of Dystonia.” The meeting’s distinguished Scientific Co-Chairs, Drs. Mark Hallett of National Institute of Neurological Disorders & Stroke, David Peterson of University of California, San Diego, and Kristina Simonyan of Harvard Medical School led an intensive program to review what is known about the neural networks involved in dystonia, discuss emerging research, and identify research gaps. A manuscript from the meeting is planned for publication in the DMRF’s new scientific journal, *Dystonia*.

Although the exact mechanisms underlying the origins of dystonia are not fully understood, several contributing neurological problems have been identified. These help explain what goes wrong in the dystonia brain.

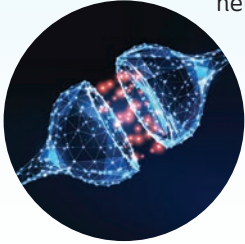
Losing Balance: Neurotransmitters

A neurotransmitter is a chemical generated by one neuron to transmit an electrical signal to another neuron. Some neurotransmitters stimulate neuron activity while others suppress activity, similar to how the gas and brake pedals in a car control acceleration and deceleration. Dystonia creates an imbalance of the neurotransmitters that control brain activity related to moving the body. When neurotransmitter levels are not balanced, movement disorders occur. Many oral medications used to treat dystonia act to restore normal neurotransmitter levels in the brain.



Missing the Connection: Abnormal Neuron Firing

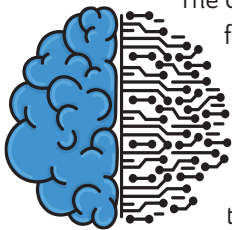
When signals between neurons are compromised by unbalanced neurotransmitters, this disrupts normal firing patterns.



Neurons may have trouble initiating or receiving signals. This results in a loss of connectivity in the brain, which disrupts motor pathways. Researchers are investigating whether this altered connectivity in dystonia is specific to certain brain regions or possibly more widespread beyond brain functions specific to movement.

Forever Learning: Neuroplasticity

Plasticity, or neuroplasticity, is the brain's capacity to change over time.



The developing brain organizes itself and assigns brain functions to various regions. New neurons can be generated, creating new connections. Neuroplasticity explains how we learn, remember, and adapt behavior. Neuroplasticity likely plays a key role in the development of dystonia—the brain's ability to re-organize and adapt is impaired. The brain loses its internal equilibrium. This opens the door for move-

ments that were once mastered to be 're-learned' incorrectly. The more the nervous system 'practices' activating abnormal movements, the more difficult they are to unlearn. This explains why the benefits of therapeutic deep brain stimulation (DBS) occur over weeks and months: the brain is gradually re-learning how to organize and coordinate normal movement patterns.

Overdoing It: Surround Inhibition

A hallmark sign of dystonia is that the brain activates more muscles than needed to complete a movement task. For example,



an individual with focal hand dystonia may pick up a pen to write and experience excessive muscle contractions in the hand and fingers, plus an overflow of involuntary movements in the arm and shoulder. The brain loses the ability to suppress activation of muscles that are not needed to complete a voluntary movement.

By contrast, when the normal brain is planning and coordinating movements, it activates the muscles required for a task while inhibiting the surrounding muscles not needed for the task.



Targeting Brain Pathways in Treatment

A number of existing treatments and investigational therapies act to reduce dystonia symptoms by rebalancing brain activity.

- Deep brain stimulation
- Non-invasive neuromodulation techniques such as repetitive transcranial magnetic stimulation (rTMS) and transcranial direct current stimulation (tDCS)
- Neurorehabilitation/physical therapy
- Medications

Botulinum Helps Balance Brains & Brawn

Even though therapeutic botulinum neurotoxin is injected into muscle and does not enter the brain, this therapy indirectly helps restore balance to neural pathways. While the primary effect of injections is that they reduce muscle contractions, an added benefit is that the relaxed muscles change the sensory information sent back to the brain. This helps balance the motor system in a positive way.

People on the Move

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



Darren H.L. Austin raised \$175 at his lemonade fundraiser last fall to support DMRF. Darren's mom is **Lindsay Hudson-Austin** who led the team "Dystoniadame's Pride" in support of Virtual Dystonia Zoo Day 2021.

The Virtual Indy Hunt for a Dystonia Cure took place in late 2021. Many thanks to organizers **Sarah Ernstberger** and **Sunshine Fox** for creating this unique event which included a virtual scavenger hunt and auction.



Author **Jennifer Senne** donated a portion of profits from sales of her children's book *Wonder Mommy*, a tribute to mothers coping with chronic medical conditions, to DMRF.



Back by popular demand, **"The Singing Chef" Andy LoRusso** partnered with DMRF to host "Cooking for a Cure," a live online cooking event in January. Andy LoRusso is the author of the best-selling cookbook and album combo, *Sing and Cook Italian*, and *Sing & Cook with Andy LoRusso the Singing Chef*. Chef Andy is donating a portion of proceeds from his signature pasta sauces to support the DMRF (singingchef.com). For an interview with Andy LoRusso about his experience with blepharospasm, visit dystonia-foundation.org/singing-chef

Dystonia & Cerebral Palsy

Cerebral palsy is a leading cause of dystonia in children. Adults with cerebral palsy can develop increasing dystonia symptoms with age. Dystonia hosted a webinar, "Dystonia and Cerebral Palsy" with Drs. Bhooma Aravamuthan and John McGuire. Hannah Thompson, longtime disability advocate and DMRF supporter, also provided remarks. The program was made possible by support from Ipsen and Revance. A recording is available for viewing at: dystonia-foundation.org/cp-webinar



Father and son **Tom and Connor Ahern** ran the Newburyport Half Marathon in October in honor of DMRF Board Member **Jon Davis**. Jon pledged to match the first \$1,000 raised, and the campaign has raised over \$3,700.

Many thanks to the expert panel, most of who are past DMRF Clinical Fellows, who participated in the Virtual Dystonia Symposium in November. Speakers included **Drs. Andres Deik, Svyetlana Miocinovic, Scott Norris, Victor Patron Romero, and Harini Sarva**. DMRF Board Member **Carole Rawson** presented on legislative advocacy and Brain Bank Liaison **Martha Murphy** spoke about registering as a brain donor. The pro-

gram was moderated by **Art Kessler**, past DMRF President. The event was made possible by support from Ipsen.



Mark Donovan ran the 2021 TCS New York Marathon in honor of DMRF Board Member **Jon Davis's** 10-year anniversary since deep brain stimulation (DBS) surgery. Jon's response to DBS has been so successful that he and Mark completed a 100-mile bike ride in advance of the marathon. Mark's campaign has raised over \$3,100.

Many thanks to **Robin and Marc Miller** for raising \$11,000 for the DMRF's Cure Dystonia Now Fund through a sold-out SB Squares 4 Dystonia fundraiser in February.

THANK YOU Social Media Fundraisers



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

To collect donations for DMRF through Facebook's fundraiser tools, go to: facebook.com/fundraisers to get started. Learn how to use Instagram donation stickers at: help.instagram.com

Donations from social media fundraisers, up to \$30,000, will be matched this year by a generous anonymous donor, doubling your support!

Please notify DMRF of your social media fundraiser. Tag or DM us on Facebook or Instagram (@dystoniamrf), or send a message to: dystonia-foundation.org/contact

Create a Legacy of Caring



The DMRF Legacy Society was created to recognize those who have made a lifetime commitment to the mission against dystonia. There are a number of planned giving options to build a legacy through the DMRF, either by joining our Legacy Society or making a specific gift. You can make a commitment of support today that is fulfilled in the future.

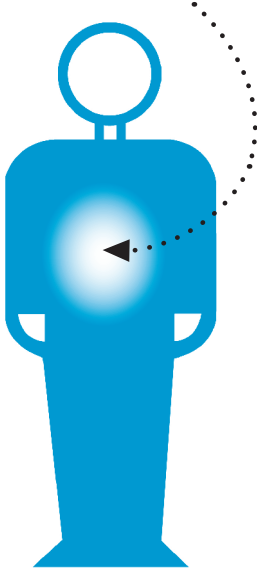
OPTIONS INCLUDE:

- Wills & Bequests
- Life Income Gifts: Charitable Trusts & Gift Annuities
- Qualified Retirement Plans
- Real Estate

For more information, please contact Director of Development Debbie Durrer at ddurrer@dystonia-foundation.org or visit dystonia-foundation.org/legacy

FOCUS ON: Truncal Dystonia

Dystonia of the Chest, Back, & Abdomen



Muscles in the middle trunk of the body, including chest, back, and abdominal muscles, may be affected in both isolated (primary) and acquired (secondary) dystonia. The trunk is often prominently affected in generalized and segmental dystonias. Truncal dystonia, sometimes referred to as axial dystonia, may also occur as an isolated focal dystonia.

Truncal dystonia can occur in a number of neurodegenerative disorders including Parkinson disease, amyotrophic lateral sclerosis, and multiple system atrophy. It can be caused by medications such as antipsychotics and acetylcholinesterase inhibitors in patients with dementia.

The symptoms may be triggered by specific actions or tasks, or may occur across numerous activities. The trunk twists and bends in the direction of overcontracting muscles. This results in forward bending of the torso, back extension or arching, torso tilting to one side, torso twisting, or a combination of these postures.

Symptoms of Truncal Dystonia

- Involuntary contractions of the torso, chest, or abdominal muscles
- Forward bending of the torso
- Back extension or arching
- Torso tilting to one side
- Torso twisting
- Pulling or tightness of back, abdomen, or trunk muscles
- Symptoms triggered by action, often reduced when at rest

Truncal dystonia can alter posture, leading to pain and challenges to daily living. Truncal dystonia may cause changes to the spine and vertebrae, nerve impingement, and structural changes to the chest wall and abdomen.

Truncal dystonia that causes the torso to bend forward may be classified as camptocormia, but it is important to note that not all camptocormia is dystonic. Camptocormia is characterized by forward flexion of the lower spine when standing and walking.

Truncal dystonia that causes the torso to bend to the side, often with twisting of the torso, may be called Pisa syndrome. It is often associated with neurodegenerative disease or the use of antipsychotic medications. In some cases, when associated with antipsychotic medication use, symptoms may be reduced by lowering the dose, stopping, or switching antipsychotic medication.

When truncal dystonia occurs as an isolated focal dystonia, with no apparent underlying cause, the most common symptoms are forward bending of the spine and torso triggered by standing or walking. Symptoms tend to reduce with rest. Sensory tricks that temporarily reduce symptoms may include running, marching, dancing, placing hands in pants pockets, or tucking hands in the waistband behind the body.

Botulinum neurotoxin injections may be used to reduce mild-to-moderate truncal dystonia. Botulinum neurotoxin injections may also reduce pain. Severe truncal dystonias may be treated with oral medications, intrathecal baclofen, and/or deep brain stimulation (DBS).

Special thanks to Debra J. Ehrlich, MD, MS for reviewing the content of this article.

DYSTONIA Q&A

In late 2021, DMRF hosted a Virtual Dystonia Symposium with a panel of experts. A recording is available at: youtube.com/FacesofDystonia

Below are responses to questions from the audience that were not answered during the live program due to time limitations.

How do I find out what kind of dystonia I have?

Dystonia is a neurological disorder that causes excessive muscle contractions. These muscle contractions result in involuntary muscle movements and body postures, making it difficult for individuals to control their movements. The movements and postures may be painful. Dystonic movements are typically patterned, repetitive, and/or tremulous.

There are several types of dystonia, and the disorder may appear quite different from person to person. Types of dystonia are classified by two main factors: 1. The clinical characteristics (for example, what muscles of the body are affected, age at onset, and spread, if any), and 2. Whether there is a known cause (for example, drug exposure, traumatic brain injury, or underlying disease).

A movement disorder neurologist is trained to evaluate an individual and make a diagnosis of dystonia based on a thorough neurological exam and patient history. This is typically the most appropriate approach to help you understand your dystonia diagnosis. You can learn more about types of dystonia at dystonia-foundation.org/types

Are direct-to-consumer genetic testing companies that offer whole genome sequencing a good source of testing?

The genetics of dystonia are complex, and whether an individual is a candidate for genetic testing is largely made on a case-by-case basis. Up until recently, genetic testing has been available only through physicians and genetic counselors. Direct-to-consumer genetic testing is advertised directly to patients by private companies. Patients provide their genetic information via a saliva sample directly to the company without necessarily involving a doctor or insurance provider in the process. Multiple medical and scientific organizations and regulating bodies have expressed concerns

about taking genetic testing out of a healthcare setting. Some of these concerns involve patient education, marketing, lack of regulation, and privacy. Individuals who are considering direct-to-consumer testing should consider speaking with their physician and/or a genetic counselor before they make the decision to test so they can think through the complexities and implications of testing ahead of time.

Whole genome sequencing screens for changes in a person's entire genetic code including parts that do not contain genes but may be important for gene function. This method is used primarily for research purposes and does not yet have a clear clinical application for managing dystonia. For more information about dystonia genetics, visit: dystonia-foundation.org/genetics

Is restless legs syndrome a type of dystonia?

Dystonia and restless legs syndrome (RLS) are distinct diagnoses, though some individuals may experience both. RLS is not considered a type of dystonia. RLS causes unpleasant or uncomfortable sensations in the legs and an irresistible urge to move them. Symptoms commonly occur later in the day and are often most severe at night when a person is resting, for example sitting or lying in bed. Since symptoms can increase in severity during the night, it may become difficult to fall asleep or return to sleep after waking up.

Which is better for guiding botulinum neurotoxin injections, EMG or ultrasound?

Electromyography, or EMG, is a technique that measures muscle activity. During EMG, a small electrode needle is inserted into the muscle to measure the electrical activity of the muscle. Physicians who inject patients with botulinum neurotoxin (BNT) may use EMG to help identify the muscles and precise targets within the muscle to be treated. Ultrasound (sonography) may also be used to visualize muscles targeted for BNT injections, providing a non-invasive alternative to EMG. Whether a doctor uses EMG or ultrasound guidance (or both) when giving BNT injections depends on their training and the muscle groups being treated. One approach is not necessarily superior to the other.

Many thanks to Abhimanyu Mahajan, MD, MHS for reviewing the content of this article.

Dystonia Dialogue

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