

Dystonia Dialogue

NEWSLETTER OF THE DYSTONIA MEDICAL RESEARCH FOUNDATION Winter 2019 | Volume 42 • No.3



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Thank You...

to the DMRF community for all you have done, and continue to do, to increase awareness of dystonia. Dystonia Zoo Walks and community events have cumulatively raised \$1.3 million in support of the DMRF mission.

DMRF supporters have also distributed awareness materials, educated government leaders, spread the word on social media, and supported one another. Your efforts help bring visibility to dystonia and generate the funding investigators need to continue working toward a cure. See page 10 for more in "People on the Move."

Amanda Sleeper organized the 2nd Nashua, New Hampshire Dystance4-Dystonia Walk on May 5.

The **4th Twin Cities Dystonia Zoo Walk** on June 22 was organized by Shanna and Brad Schmitt and Billy McLaughlin.

The 6th Detroit Dystonia Zoo Walk took place July 14. Dr. Ellen Air of Henry Ford Health System was a special guest. The event was organized by DMRF Community Leadership Council Member Rosemary Young.

The **4th Portland Dystonia Zoo Walk** was organized by Dee Linde. The event took place July 20.

Sheila Killham organized the annual **Dogs for Dystonia Dog Walk** in Marion, lowa on August 24.

The **5th St Louis Dystonia Zoo Walk** organized by June Tritley took place August 25. DMRF Scientific Director Dr. Joel Perlmutter of Washington University provided remarks.

Dystonia Community Unites



DMRF Board Member Pamela Sloate organized the **5th Bronx Dystonia Zoo Walk** on September 8. Guest speakers included DMRF Lifetime Honorary Director Dr. Stanley Fahn of Columbia University.

The Flanagan Family, who created the original Dystonia Zoo Walk, hosted the **8th Cleveland Dystonia Zoo Walk** on September 8. Many thanks to the organizing committee: DMRF

Community Leadership Council Member Karen Flanagan, Jane Ann Flanagan, and Gale Flanagan. DMRF Medical & Scientific Advisory Council Member Dr. Aasef Shaikh of University Hospitals, Cleveland VA Medical Center, and Case Western Reserve University provided remarks.

In August, the Pittsburgh Pirates welcomed members of the Western, Pennsylvania Dystonia Support Group

to Raise Awareness & Funds for Research



for Dystonia Awareness Day at PNC Park as a prelude to the **6th Pittsburgh Dystonia Zoo Walk** on September 8. Event organizers were MaryRae Nee, Ed Cwalinski, Tara Sorley, and Chris Mack of The Fan Morning Show on 93.7 Pittsburgh Sports Radio. Dr. Mary Flaherty of Allegheny Health Network provided remarks.

The **4th Providence Dystonia Zoo Walk** took place September 14 at Roger

Williams Zoo. The event organizing committee included longtime DMRF supporters Sue Baron, Beth Paolero, Paula Schneider, and Brian Smuda. Dr. Maria Luisa Moro-de-Casillas of Hartford Healthcare provided remarks.

On September 21, the **6th Cincinnati Dystonia Zoo Walk** took place, organized by DMRF Community Leadership Council Member Melissa Phelps. Special guests included members of the movement disorders teams at University of Cincinnati Medical Center and Cincinnati's Children's Hospital. DMRF Clinical Fellow Dr. Abhimanyu Mahajan provided greetings.

The **3rd Philadelphia Dystonia Zoo Walk** on September 22 was organized by Janice and Len Nachbar, Joanna Manusov, and Raman Patel. Philadelphia

Mayor Jim Kenney provided a Dystonia Awareness Proclamation in recognition of the event.

The **Indy Hunt for a Cure for Dystonia** took place September 29 in Garfield Park, Indianapolis. The organizing committee included Sarah Ernstberger and Sunshine Fox. DMRF Board Member John Downey was a special guest.

The **1st Toledo Dystonia Zoo Walk** took place October 5 thanks to the efforts of Kristin Cinglie. Dr. Ellen Air of Henry Ford Health System was a special guest.

Beth Farber, Steve Laser, and Joel Farber organized the **Chicago Basket Bash** on October 27 in memory of Shari Farber Tritt and Harriett Farber.

The **2nd Phoenix Zoo Day** took place November 2 thanks to the efforts of Mary Stone and her supporters.

The Ist Los Angeles Dystonia Zoo Walk was organized by Jacquelyn Coello and Janis Eiler on November 9. Dr. Michele Tagliati of Cedars-Sinai Medical Center provided remarks.

> The DMRF thanks National Sponsors Allergan, Merz, and Ipsen for their generous event support.

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On the Cover:



There are many types of dystonia, and numerous potential causes. The disorder can present quite differently from person to person. We believe a cure is possible for every dystonia.

For nearly 45 years the DMRF has offered programs and resources to individuals and families impacted by all types of dystonia. The mission from the beginning

has been to advance research for more effective treatments and ultimately a cure, to promote greater dystonia awareness, to provide up-to-date and accurate educational resources, and to support the well-being of dystoniaaffected individuals and their families.

Information about the various types of dystonia as well as DMRF programs is available at www.dystonia-foundation.org

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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Debbie Durrer Director of Development

Jessica Feeley Editor and Special Projects

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Jennifer McNabola Director of Finance

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Chief Scientific Advisor

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Dystonia Medical Research Foundation

One East Wacker Drive Suite 1730 Chicago, Illinois 60601-1980 Phone 312 755 0198 800 377 DYST (3978) Web www.dystonia-foundation.org

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Foundation Update



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Janet L. Hieshetter Executive Director

In this season of gratitude, as the year comes to a close, the DMRF leadership is thankful for our generous supporters and the vibrant community we have built together. DMRF is stronger because of each of our members. You empower us, every day, to work diligently on your behalf. And we will continue to do so for as long as it takes to find a cure.

Collaboration allows us to combine our strengths and continue forward until our mission to cure dystonia is complete. This fall, the Department of Defense (DOD) announced that, once the FY2020 federal budget is approved, dystonia investigators will once again be eligible to apply for funding through its Congressionally Directed Medical Research Program, resulting in millions of new research dollars. This victory was made possible by dedicated dystonia advocates across the country who joined the Dystonia Advocacy Network to educate their Members of Congress about dystonia and the urgent need for federal research funding. Critical new research discoveries are being made because our volunteer advocates came together to give the dystonia community a unified voice to Congress.

Collaboration and cooperation are essential in research. We are grateful for the support of Cure Dystonia Now and the Benign Essential Blepharospasm Research Foundation for partnering with us on investigations this year. In our requests for research proposals, DMRF regularly encourages innovative, collaborative, and cross-disciplinary projects. DMRF is proud to reach across medical and scientific disciplines to invite investigators to get interested in dystonia. These efforts have yielded profoundly exciting results, from dramatic discoveries in dystonia biochemistry to ambitious new contracts to explore novel therapeutics.

The Dystonia Coalition continues to be an outstanding model of collaboration between medical researchers and patient advocacy groups focused on accelerating clinical research. The DMRF is pleased to continue serving as an administrative center. We are also looking forward to the Samuel Belzberg 6th International Dystonia Symposium, June 4–6, 2020, in Dublin, Ireland, organized in partnership with Dystonia Europe. The program is designed for clinicians and investigators to provide a comprehensive overview of important scientific advances in the field and stimulate discussion within and across disciplines. The International Dystonia Symposium is the seminal international dystonia scientific meeting, dating back to 1975.

Amid all the activity and achievements the DMRF shares with our members, we know the dystonia experience at home can at times feel isolating. We hope you will reach out to your nearest DMRF support group, join an online support forum, and consider attending local events and education meetings. If you do not know where to turn for help, contact the DMRF office and we will assist you. No one should have to face dystonia alone. We are here for you.

Thank you for being a part of the DMRF community. Your donations and participation make it possible for the Foundation to continue working toward a cure for every person and family affected by dystonia.

letter Keylen

Art Kessler President

Janer L. Hieshetter

Janet L. Hieshetter Executive Director

Dystonia Community Curious & Cautious about Medical Cannabis



Recent years have seen a dramatic increase in the marketing, availability, and consumption of cannabis (marijuana) products in the United States. While federal laws classify cannabis as having no accepted medical use and a high potential for abuse, legal restrictions on the medical and recreational use of cannabis have loosened in

Dystonia & Medical Cannabis Webinar

For a thorough and thoughtful examination of the role of medical cannabis in treating dystonia, do not miss this presentation with movement disorder specialist Danny Bega, MD at: https://dystonia-foundation.org/ complementary-therapy/

several states. There is renewed interest within segments of the research community to explore its use to treat numerous diseases and conditions. While cannabis products are increasingly easy to obtain, it can be challenging for patients and physicians alike to find credible information about its role in medicine.

650+ people responded to a DMRF survey into perceptions and questions about the use of medical cannabis for dystonia. Below are highlights from the results.

Have you used cannabis to manage your dystonia or dystonia-related symptoms? 48% Yes 52% No Are you currently using cannabis to help manage your symptoms? (Of those who have used.) 74% Yes 26% No Is your doctor aware that you are using cannabis?

70% Yes 30% No

Top Reason for Not Using Medical Cannabis 46% Would like to know more about it before trying it

Did you speak with your doctor about using cannabis prior to using it? 62% Yes 38% No

For what symptoms do you use cannabis?

- 85% Spasms, tremors, movements
- 80% Pain
- 65% Sleep
- 60% Anxiety
- 18% Appetite
- 12% Other: Seizures, mood, nausea

Most Popular Types of Cannabis Used

55% Ingest oils

63% Smoke or vape 44% Topical creams or lotions 30% Gummies or candies

ICYMI: US Centers for Disease Control & Prevention, Food & Drug Administration, state and local health departments, and other health authorities are investigating a multistate outbreak of lung injury associated with the use of vaping products, also called e-cigarettes. Information is available at https://www.cdc.gov/



Research Bits

New dystonia studies are published daily. Below is a tiny sample of new data and discoveries.

CROSSING PATHS.

Advancements in genetics have led to rapid expansion in the number of genes associated with dystonia. These genes encode proteins with very different biological functions. However, many potential shared biological pathways have been proposed, including those related to dopamine signaling, mitochondrial dysfunction and energy maintenance, toxic accumulation of heavy metals in the brain, and calcium channels and abnormal calcium physiology. These shared pathways are being explored for the purpose of better understanding the biological basis for dystonia and for designing new therapies with the broadest potential for multiple types of dystonia.

H.A. Jinnah, Yan V. Sun. Dystonia genes and their biological pathways. Neurobiol Dis. Sept 2019, 129:159-168.

HOW MANY PEOPLE HAVE DYT1 DYSTONIA?

A new study suggests between 54,366–80,891 people in the United State are carriers for the DYTI/TORIA dystonia-causing gene. Because not every gene carrier develops dystonia, this would translate into 16,475–24,513 individuals with DYTI dystonia. DYTI dystonia is among the most common early onset isolated dystonias, yet represents only a fraction of the total patient population.

Joseph Park, Scott M. Damrauer, Aris Baras, Jeffrey G. Reid, John D. Overton, Pedro Gonzalez-Alegre. Epidemiology of DYTI dystonia. Neurol Genet. Oct 2019, 5 (5).

RARE DYSTONIA MISDIAGNOSED AS SPASMODIC DYSPHONIA.

Pharyngeal dystonia is a rare focal dystonia affecting muscles involved in swallowing, which may be misdiagnosed as spasmodic dysphonia/laryngeal dystonia—a focal dystonia affecting the vocal cord muscles. Both can cause voice impairment. A specialized botulinum neurotoxin injection technique targeting the pharyngeal muscles may result in significant improvement in voice with minimal side effects.

Lucy L. Shi, C. Blake Simpson, Edie R. Hapner, H. A. Jinnah, Michael M. Johns. Pharyngeal dystonia mimicking spasmodic dysphonia. Journal of Voice. Mar 2018, 32(2): 234–238.

At the 4th International Congress on Treatment of Dystonia in Hannover, Germany earlier this year, founders of Musicians With Dystonia, Steven Frucht, MD (L) and Glen Estrin (R), met with ophthalmologist Alan Scott, MD, who developed therapeutic botulinum neurotoxin injections for blepharospasm and other conditions of the eye.



In Memoriam: Valerie F. Levitan, PhD



We are deeply saddened to report that DMRF's past Executive Director, Valerie F. Levitan, PhD, passed away on October 15.

She served as Executive Director of the DMRF (1992– 2002) and the National Spasmodic Dysphonia Association (1997–2002). Dr. Levitan joined the DMRF at a critical time for the Foundation, and was integral to the evolution from a private family foundation to a membership-driven organization. Her vision, energy, and wisdom helped build the DMRF into the vibrant community it remains to this day.

She was a tireless advocate on behalf of dystonia patients, their loved ones, and the research community. The DMRF extends our deepest condolences to her children, Dan Levitan and Jeanie Levitan, and family.

Dystonia Diagnosis Times Improving



1500+ people completed a DMRF survey seeking feedback from individuals with dystonia about the diagnosis process and experience with botulinum neurotoxin injections, one of the most common treatments for dystonia.

One of the most significant findings is that individuals diagnosed with dystonia within the last five years were diagnosed faster than those diagnosed more than five years ago, suggesting that diagnosis times continue to improve. Highlights from the survey results are below.

Gender

79% Female21% Male1% Not Listed

48% of individuals diagnosed within the last five years were diagnosed in less than a year

Age

 1%
 Under 18

 1%
 18–24

 4%
 25–34

 8%
 35–44

 17%
 45–54

 28%
 55–64

 42%
 65+

Type of Dystonia

- 76% Cervical dystonia11% Blepharospasm
- 13% Generalized dystonia
- 12% Laryngeal dystonia or spasmodic dysphonia
- 9% Focal foot or leg dystonia

- 10% Oromandibular or facial dystonia
- 8% Other

72% of participants were diagnosed by a neurologist. 40% of participants were diagnosed by a movement disorder neurologist.

How recently were you diagnosed with dystonia?

4% Within the last year
21% 1–5 years ago
22% 5–10 years ago
53% 11+ years ago

96% of participants have been treated with botulinum neurotoxin injections

How long after seeing a doctor were you diagnosed?

- 44% Less than a year
 32% 1–3 years
 9% 4–6 years
 5% 7–9 years
- 10% 10+ years

74% are currently receiving botulinum neurotoxin injections

Top 3 Reported Benefits from Botulinum Neurotoxin Injections

- 57% I have less pain
- 43% I am able to do more in a day
- 33% Regained or increased independence

Do you feel your dystonia symptoms have...

- 49% Improved
- 29% Stayed about the same
- 23% Worsened

44% of participants have received botulinum neurotoxin injections for 10+ years

Investigators Use New Technologies to Understand Origins of Symptoms

Myoclonus-Dystonia Research Projects 2019

The following outstanding research projects were funded with support from the Brown Family Foundation. Each project is applying cutting-edge research techniques to achieve a better understanding and improved treatment options of myoclonus-dystonia (M-D).

Variable Expressivity in SGCE-Associated Myoclonus-Dystonia: Insights from Epigenetics and Transcriptomics Karen Grütz, PhD, University of Lübeck (Germany)

Myoclonus-dystonia (M-D) is a movement disorder presenting with brief, jerking movements (myoclonus) combined with sustained muscle contractions (dystonia). In about 25% of patients, the disorder is caused by mutations in the SGCE gene. M-D is inherited in an autosomal-dominant way, meaning that one mutated version of SGCE, inherited from a parent, is enough to cause the disorder. Typically, only individuals who inherit the mutation from their fathers are affected by the disorder. This is because a mechanism called maternal imprinting silences the information handed down from the mother, leading to incomplete penetrance of clinical symptoms. Of interest, the severity of symptoms and the age of onset varies widely between individuals (related and unrelated) even with the exact same mutation. It is possible that some of these variations can be explained by slight changes in the underlying mechanism of maternal imprinting. The protein product of the SGCE gene is called ε -sarcoglycan and is localized at membranes in various tissues within the body and, importantly, has been identified as component of a membrane-bound complex within the brain.

Dr. Grütz is using highly specialized techniques and cell models to help explain the variation of clinical symptoms and severity among M-D patients. This approach will help clarify the mechanism behind the development of M-D and also contribute to the generation of therapeutics and possibly even personalized treatments.

Deciphering the Sources of Myoclonus in DYT-SGCE Patients *Yulia Worbe, MD, PhD, Salpêtrière Hospital (France)*

Muscle jerks, or myoclonus, often affecting the upper body, represent the most disabling symptom in many patients with myoclonus-dystonia (M-D). Treatment of the jerking movements can be challenging. Understanding the mechanism by which the jerks are generated within the brain may clarify new strategies for treatment. In M-D, two brain circuits have been related to myoclonic jerks: one links the posterior part of the brain called the cerebellum to the upper part of the brain called the cortex. The other potentially involved network links the cortex to the basal ganglia. Recent studies have shown that the cerebellum and basal ganglia are interconnected, and through these connections they can influence how different movements are performed.

Despite some progress in understanding how the brain generates myoclonic jerks, there is so far no direct evidence of brain alteration accounting for myoclonus. This is mainly due to difficulties encountered when studying patients who have jerking movements that interfere with the techniques and technology available to study of their brain activity. Magnetoencephalography (MEG) is a non-invasive technique for investigating human brain activity. Dr. Worbe is using a new and very innovative MEG system that is worn like a helmet, allowing free and natural movement during scanning. This system opens up new possibilities because the myoclonic jerks experienced by patients will no longer interfere with brain activity studies. Investigators will be able to identify the sequence of brain events that leads to the generation of these jerks. Understanding the brain alterations leading to myoclonus could eventually guide the use of non-invasive brain stimulation as a possible treatment.

People on the Move

The DMRF is grateful for the grassroots volunteers across the country working to improve dystonia awareness in their communities. Every volunteer makes a difference!



Mike Delise purchased a #5dollarcure-4dystonia digital billboard in Michigan to promote awareness and encourage people to visit https://5dollarcure.com/ during Dystonia Awareness Month. The campaign was also covered by reporter Deena Centofanti of Fox2Detroit.

The Buffalo Bills hosted a kickball tournament on August 25 and a cornhole tournament on September 7 to raise dystonia awareness and research funds. The events were spearheaded by **Jim Metherell**, member of the DMRF Community Leadership Council.

The Dystonia Support & Advocacy Group of San Diego County led by **Martha Murphy** welcomed **David Peterson, PhD**, Associate Project Scientist in the Institute for Neural Computation at University of California, San Diego and the Computational Neurobiology Laboratory at the Salk Institute for Biological Studies, and **Jeanne Vu**, Master's student at San Diego State University, to present on "Advances in Measuring Motor Outcomes of Dystonia" at the September meeting.



Susan Whaley, creator of Wild Child Paintings by Hippy Susie, partnered with Urban Grind Roasters coffee shop in Mooresville, NC to educate patrons about dystonia and exhibit her artwork, with proceeds from art purchases benefitting the DMRF.

The Denver Dystonia Patient Education Program took place October 5, 2019. **Brian Berman, MD** of the University of Colorado presented on dystonia and treatment options. **Ray Skibitsky** provided a personal perspective on his experience with cervical dystonia and deep brain stimulation. This was the first in a series of meetings scheduled this fall in communities across the country, supported by Allergan.



Beth Paolero, an educator at Saint Philomena School in Portsmouth, RI, and her daughter Maddie Paolero promoted dystonia awareness at back-to-school night. Service dog JJ was a big draw to the table.



Jennifer DeVore ran the Wineglass Marathon in Corning, NY on October 6 to raise awareness and funds for research.

DMRF extends special appreciation to all those who made the extra effort to bring visibility to dystonia during Dystonia Awareness Month in September. Hundreds of volunteers supported DMRF events, distributed awareness materials, and educated friends and neighbors.

Dystonia: Frequently Asked Questions

What is dystonia?

Dystonia is a neurological disorder that causes excessive, involuntary muscle contractions. These muscle contractions result in abnormal 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 and repetitive.

Dystonia can affect any region of the body including the eyelids, face, jaw, neck, vocal cords, torso, limbs, hands, and feet. Depending on the region of the body affected, dystonia may look quite different from person to person.

What causes dystonia?

There are many known causes for dystonia. Dystonia may occur due to an inherited or new genetic mutation. Dystonia may also result from changes in brain activity caused by another health condition such as a traumatic brain injury. Certain drugs are known to cause dystonia. However, for many people who develop dystonia, there is no identifiable cause.

Can dystonia affect the heart and other vital organs?

Dystonia affects muscles that can be controlled voluntarily, mostly the skeletal muscles. Dystonia does not affect smooth muscle, such as the heart.

Is dystonia fatal?

In the overwhelming majority of people with dystonia, it does not shorten life expectancy or result in death. In extreme cases, the development of increasingly frequent or continuous episodes of severe generalized dystonia can cause a medical emergency called status dystonicus. If untreated, this rare condition can cause life-threatening complications. Status dystonicus is a treatable condition and, with prompt medical attention, symptoms typically can be brought under control. Dystonia does occur as a symptom of several degenerative diseases, some of which do impact mortality, but the dystonia itself does not shorten life span.

Dystonia is making it hard to function. Who can help me?

Living well with dystonia is possible. The early stages of symptom onset, diagnosis, and seeking effective treatment are often the most challenging. In some cases, treatment can dramatically reduce or suppress symptoms.

Individuals and families living with dystonia are strongly encouraged to:

- Seek treatment from a neurologist who specializes in movement disorders.
- Learn about dystonia and treatment options.
- Develop a multi-layered support system of support groups, online resources, friends, and family.
- Seek expert mental health professionals to diagnose and treat possible co-existing depression and/or anxiety disorders.
- Investigate complementary therapies that support overall wellness.
- Get active within the dystonia community

Information pertaining to each of these suggestions are available at www.dystonia-foundation.org

Look for additional FAQ in future issues of the Dystonia Dialogue. If you have a question you would like to see addressed, email us a contact@dystonia-foundation.org

DEEP BRAIN STIMULATION FOR DYSTONIA

Neuromodulation is the science of altering brain activity to treat disease, often by using electrical stimulation.

Deep brain stimulation (DBS) is a neuromodulation technique that uses a surgically implanted medical device to treat dystonia and other disorders.

DBS applies electrical stimulation to interrupt the abnormal brain activity that causes dystonia symptoms.

A DBS system includes an electrode placed deep in the brain, an implanted neurostimulator, and a wire that connects the electrode to the neurostimulator. The neurostimulator contains a battery and electronic circuitry that generates the signals delivered to the brain. Once the system is surgically implanted, the electrical stimulation is adjusted by remote control. The neurostimulator batteries must be periodically replaced. Rechargeable and non-rechargeable neurostimulators are available.



Who is a Candidate?

DBS treatment for dystonia is recommended on a case-by-case basis. DBS may be considered if:

- Symptoms cannot be adequately treated with medications and other treatments.
- Surgical risks are justified against quality of life.

Types of Dystonia Most Often Treated with DBS

- Genetic childhood onset dystonias, e.g. DYT1/TOR1A and DYT28/KMT2B
- Cervical dystonia
- Tardive dystonia/dyskinesias
- Myoclonus-dystonia
- X-linked dystonia-parkinsonism

There is encouraging early data about the use of DBS to treat cranial dystonias, including oromandibular dystonia and blepharospasm.

What are the Benefits?

Benefits of DBS include reduced dystonic movements, postures, and pain. It is difficult to predict which patients will benefit and to what degree. Numerous studies have shown that the benefits of DBS are sustained long-term.



Risks

The most common adverse effects from DBS are infection (5%) and lead breakage (4%). There may also be surgical and stimulation-related risks and side effects.

In the USA, Medtronic's DBS device received a special approval for dystonia by the Food & Drug Administration (FDA) called a Humanitarian Device Exemption.

Manufacturer

FDA Approval

Abbott St Jude Boston Scientific Medtronic

Parkinson disease, essential tremor Parkinson disease Parkinson disease, essential tremor, dystonia

Neurostimulator Type

Non-Rechargeable Rechargeable Rechargeable and Non-Rechargeable

DBS Medical Team

DBS is a highly specialized therapy that requires a team of medical professionals to evaluate and treat patients:

- Movement disorder neurologist
- ✓ Neurosurgeon
- ✓ Neuroimaging specialist
- ✓ Neuropsychologist
- Psychiatrist
- ✓ Nurses

Medical centers vary in their DBS procedures and practices. Individuals considering DBS may wish to consult more than one DBS team before making a selection.

Beware

Certain medical MRI imaging, diathermy therapies, security metal detectors, and security wands may interfere with the DBS device. Individuals must clarify with their doctor and device manufacturer what restrictions apply to their implanted DBS system.

Technology Advancements

The neuromodulation field combines biomedical research with cutting-edge engineering design, often inspiring collaboration among academic research programs and private companies.

Multiple efforts are underway to make DBS systems more personalized and more effective. DBS is evolving from a strategy to suppress dystonia symptoms toward the possibility of correcting the underlying brain circuit dysfunction that causes symptoms.

Emerging DBS Advancements

- Creating DBS systems that automatically make stimulation adjustments by sensing and responding to brain activity. This involves improved sensors and increased device memory.
- Deciphering biomarkers in the brain that help predict a positive result from DBS.
- Rechargeable neurostimulator batteries to lengthen the time between battery replacements.
- Smaller devices. Ultimately, neurostimulators may be small enough to implant under the scalp rather than the abdomen.
- Upgradeable devices. Patients may one day have the ability to upgrade their DBS systems as easily as upgrading apps on a smartphone.
- Improving electrode design for more selective targeting and reduction in unwanted stimulation side effects.
- Greater flexibility of stimulation options, including the ability to have different settings for specific brain targets.

Additional information about DBS for dystonia can be found at: www.dystonia-foundation.org/ living-dystonia/treatment/

A brochure is available for free mail order or download at www.dystonia-foundation.org/brochures

Local Dystonia Advocacy Leads to National Recognition

Grassroots dystonia advocacy is a powerful force for public awareness. By engaging civic and government leaders at all levels—federal, state, local—members of the dystonia community are bringing visibility of dystonia to new audiences and influencing policy that impacts lives.



DMRF is proud to provide staff support for the DAN. Member organizations include:

Benign Essential Blepharospasm Research Foundation (BEBRF)

Dystonia Medical Research Foundation (DMRF)

National Spasmodic Dysphonia Association (NSDA)

National Spasmodic Torticollis Association (NSTA) Through the Dystonia Advocacy Network (DAN), volunteer advocates from across the country unite to give the dystonia community a voice to Congress. These volunteers are called to action throughout the year to contact their Members of Congress regarding important dystonia-related issues, often pertaining to pending regulation changes or legislation. Thanks to the efforts of these advocates, dystonia is once again included on the list of eligible conditions for the Department of Defense's Peer Reviewed Medical Research Program for Fiscal Year 2020. This means additional federal dollars available to dystonia researchers. The dystonia community owes a debt of gratitude to Dale Dirks and Phil Goglas II of Health & Medicine



Jason Dunn of Michigan met with Governor Gretchen Whitmer after obtaining a Dystonia Awareness Month Proclamation.

Counsel of Washington who have worked alongside the dystonia community to make this happen.

Congressman Andy Levin of Michigan introduced House Resolution 557 to designate September 2019 as National Dystonia Awareness Month. Congressman Levin authored

the resolution after learning the story of DMRF supporter Jason Dunn, who lives in Warren, Michigan. Jason has lived with dystonia for over 30 years, and despite the challenges of the dystonia, has worked tirelessly to raise awareness. DMRF supporter Mike Delise was integral to making this resolution possible. The bipartisan resolution was co-sponsored by Congresswoman Jan Schakowsky of Illinois, Congressman Chris Smith of New Jersey, and Congressman Brian Fitzpatrick of Pennsylvania. The complete resolution can be found at www.congress.gov/

A number of individuals successfully reached out to local and state leaders to request a dystonia awareness proclamation. In several situations, the individuals were invited to meet with their Governors.

For information about joining the DAN and becoming a dystonia advocate, visit www.dystonia-advocacy.org/contact/



Marcus Bush presented a Dystonia Awareness Proclamation to the Dystonia Support & Advocacy Group of San Diego County on behalf of City Council President Georgette Gomez.



Don LeBlanc obtained a Dystonia Awareness Proclamation from Louisiana Governor John Bel Edwards.



Sheila Kilham and daughter Selena met with Iowa Governor Kim Reynolds to receive a Dystonia Awareness Proclamation.

Linda Davis was presented with a Dystonia Awareness Month proclamation by New York State Assemblyman Joe De Stefano.



Mayor Jim Kenney provided a Dystonia Awareness Proclamation in recognition of the Philadelphia Dystonia Zoo Walk, presented by Councilwoman Jannie Blackwell.



Clinical Researchers Tackle Dystonia Treatment Gaps

Non-Invasive Brain Stimulation Therapies Offer New Therapeutic Opportunities to Explore

"During my fellowship training at Duke, the faces of dystonia patients with painful postures and tremors stayed with me," said Noreen Bukhari-Parlakturk, MD, PhD, a movement disorder neurologist and clinical investigator at Duke Neurological Disorders Clinic. "Their stories of resilience and optimism—despite daily hardship and social misunderstanding—inspired me to ask the hard questions about this disease and affirmed my life mission to develop disease-modifying therapy for them."

Dystonia can be a challenging disorder to treat. Treatment must be customized to the unique needs of each patient. For some patients, available therapies such as botulinum neurotoxin injections or deep brain stimulation can dramatically improve quality of life. For other patients, these treatments provide little to no benefit. "There is absolutely a need for novel therapies," said Dr. Bukhari-Parlakturk. "I see it daily in my dystonia clinic where patients tell me the current combination of treatments helped 'a little' or 'some.' I am not satisfied with that."

Transcranial Magnetic Stimulation Therapy

After years of working to advance therapies for neurological diseases, Dr. Bukhari-Parlakturk received a DMRF Clinical Fellowship in 2017. The award provided the opportunity to pursue specialized training in movement disorders and original research focused on dystonia. "It meant everything!" she explained. "The DMRF clinical fellowship support was the first time I was proposing to study dystonia and enlist the help of volunteers. It validated that my ideas about understanding the disease and developing therapy for dystonia patients will address a critical need in the field."

Dr. Bukhari-Parlakturk is conducting research to investigate the use of transcranial magnetic stimulation therapy (TMS) to treat focal hand dystonia. TMS is a non-invasive, non-surgical technique to stimulate brain activity. Electricity is pulsed into the brain through an electromagnetic coil positioned against the head. The pulses stimulate neurons in the targeted brain region.

TMS has been used since the 1980s as a research tool to study the pathophysiology of neurological disorders, including dystonia. The use of TMS for dystonia for research paved the way for its exploration as a therapy. When used therapeutically, TMS is applied in sessions repeated over a schedule of days or weeks. Adverse effects are rare and may include seizure, fainting, and headache.

"I want to close the gap between understanding the disease and targeting it with therapeutic trials so patients walk away feeling their treatment makes a major change in their quality of life."

Correcting Faulty Brain Circuits

The exact mechanism of action of TMS is not fully understood. Dr. Bukhari-Parlakturk explained: "We know from prior studies that the symptoms of dystonia are due to a problem with the brain's electrical circuit. I am interested in identifying and correcting this electrical circuit in dystonia patients using TMS. My hope is that if I target the underlying problem, I can not only provide prolonged symptom benefit but also affect the dystonia disease pathway."



Dr. Noreen Bukhari-Parlakturk (far right) and research team are studying TMS therapy as a novel treatment for focal hand dystonia.

The potential of TMS for use in dystonia has been shown in a handful of studies. Several trials using low-frequency repeated TMS have shown reduction of focal hand dystonia symptoms. A pilot study demonstrated that low-frequency repeated TMS reduced muscle spasms and pain in three patients with secondary generalized dystonia. A randomized, sham-controlled, blinded study of TMS in eight cervical dystonia patients demonstrated marked benefit in seven patients, with an apparent cumulative benefit over time. In all studies, TMS was safe and well-tolerated by study volunteers.

Benefit from repeated TMS has been shown to last from days to weeks. Dr. Bukhari-Parlakturk is hoping to harness the benefits of TMS into a lasting treatment option: "I am interested in developing this technique to provide stable and prolonged relief in writer's cramp dystonia [focal hand dystonia], so it can be a meaningful treatment option for patients." Hand dystonia is among the most challenging focal dystonias to treat effectively. Symptoms can interfere with holding objects, writing, using utensils, keyboarding virtually any task requiring fine motor control of the hands.

Moving Forward

TMS and additional therapeutic brain stimulation techniques may ultimately provide treatment options to dystonia patients for whom traditional therapies, such as oral medications and injected botulinum neurotoxin are not adequately effective. "There is a high pay off when we get non-invasive brain stimulation techniques to work," said Dr. Bukhari-Parlakturk. "This is why there is such excitement to advance this technique by patients and researchers alike."

TMS might also play a valuable role in helping to better predict which dystonia patients will respond well to deep brain stimulation (DBS).

Advancements in bringing new therapeutic technologies to market require cooperation among basic researchers in the lab, clinical researchers, biotechnology engineers who

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develop medical devices, and—perhaps most importantly—patients willing and able to volunteer for clinical studies. "We are moving in the right direction and our success in developing treatment options is driven by patients—by their hope and participation in the discovery process," said Dr. Bukhari-Parlakturk. "I am grateful to the dystonia participants in my research study who have contributed their time to help advance this work. Their hope and commitment to advancing research is the key to our collective success."



How Does TMS Work?

Transcranial magnetic stimulation (TMS) uses a magnet to activate or suppress brain activity with electricity. An electromagnetic coil is positioned against the head, over an area of the brain involved in coordinating movement.

The coil delivers brief pulses of electricity through the skull, into the brain. The pulses stimulate neurons in the targeted brain region.

A person receiving TMS experiences a tapping sensation on the head as the pulses are administered. The process is loud but painless. No sedation or anesthesia are needed.

TMS has been approved by the US Food & Drug Administration (FDA) as a treatment for depression and obsessivecompulsive disorder.

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 www.clinicaltrials.gov

Register as Brain Donor

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

✓ Join a Patient Registry

Patient registries help researchers better understand dystonia by collecting information directly from volunteers who are diagnosed. The DMRF is proud to support the Global Dystonia Registry www.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 www.dystonia-foundation.org/donate

What Makes You Stronger Mother & Daughter Reflect on Family Legacy of Living Beyond Dystonia

"In my family, we were all aware that my father had a problem with his hands," explained Carole Rawson, "and my parents' generation was one in which, if there was something bad going on, you didn't talk about it. He was very closed-lipped about it." Carole's father went most of his life without a name for his problem until Carole began having problems too.

"I was born a lefty and when I was in the fifth grade, I no longer could write with my left hand. We realized I must have what my father had, so I taught myself to write with my right hand." The solution was temporary. Before the end of fifth grade, Carole could no longer write with either hand.

In 1972, Carole and her father were diagnosed with writer's cramp, a focal hand dystonia. She was 15 years old. He was 53. At the time, treatment options were few. The medication offered did not help. Carole persevered without treatment through high school, college, and a career in retail during a time when writing by hand was pervasive in everyday life. "It was painful," she said. "There were no special accommodations back then. I was always coming up with new, inventive ways to hold my pen. At work, people would ask me *why do you hold the pen so funny?* And as soon as I started telling them, it was like they wanted to crawl under the desk and were sorry they asked."

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Carole and her husband Doug met as undergrads at George Washington University in Washington, DC. Both native New Yorkers, they settled in Santa Monica, California. The couple have four children. "I was always very forthcoming about the dystonia," said Carole. "All the kids knew about it." She joked that between her writer's cramp and Doug's "terrible handwriting," she did not have high expectations for her children's penmanship: "My feeling was as long as they don't have dystonia, I don't care how they write."



Liz and Carole Rawson joined the DMRF Board of Directors in 2018.

When Carole and Doug's third child Liz was around age 11, a familiar pattern began to emerge. "My handwriting was really poor and I remember sitting in class, in either fifth or sixth grade, and my friend would massage my hands because I complained that they hurt so much," said Liz. By the time Liz was 14, it clicked for Carole that Liz's problem was the same as her own. Carole contacted the DMRF for assistance locating a movement disorder specialist, and Liz was diagnosed with dystonia by a leading expert in Los Angeles. She promptly began teaching herself to write with her opposite hand, but, just like her mother, Liz soon lost the ability to write at all.

Continued on page 20

Hand dystonia is among the most challenging types of focal dystonia to treat medically, especially when the symptoms are present only during a specific activity, such as writing. Botulinum neurotoxin injections can be helpful, depending on the muscles affected, but individuals must often permanently adapt to losing fine motor control of the hands.

Carole taught Liz early to advocate for herself, especially regarding accommodations at school mandated by the Americans with Disabilities Act. "At the start of every school year I'd meet with my teachers and explain dystonia," said Liz. "I also made the very wise decision of buddying up with my guidance counselor and people in the office, so I always had them on my side." Carole accompanied Liz to meetings with educators at first, but by sophomore year Liz was handling it on her own. "If it got to a point where I needed reinforcements, she was there in a second," said Liz, "but my mom also gave me the space to attempt it on my own, to go talk to teachers and ask for what I needed. It's something I'm always very grateful for."

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Liz graduated high school and, following in her parents' footsteps, attended George Washington University in Washington, DC. Liz had managed well in high school despite the challenges of dystonia, and there was every expectation that her success would continue.

Carole recalled, "When she left for school, it was like anybody else leaving for school. I mean, she had dystonia in her hands and she was going to deal with it like she always did."

Liz agreed: "This is what we understood dystonia to look like. We didn't understand at that point that there were so many different manifestations of dystonia and that it could spread to my legs. That wasn't a thought in anyone's mind."

When Liz's doctor mentioned deep brain stimulation (DBS) for the first time, the idea seemed extreme relative to her symptoms. DBS is a neurosurgical procedure to treat dystonia that involves implanting electrodes in the brain and a battery-powered stimulator in the chest wall. Liz was not interested: "I've been needle phobic and doctor phobic my whole life and when my doctor offered [DBS] to me as an option, I remember laughing and telling him he was crazy and that I would never get it." (For more information about DBS, see pages 12–13.)

Early-onset isolated dystonia is a disorder characterized by progressive problems with movement, typically beginning in childhood. Symptoms include involuntary muscle contractions, twisting postures in specific body parts such as an arm or a leg, tremors, and other uncontrolled movements. Signs and symptoms of early-onset isolated dystonia typically appear around age 12. Abnormal movements or postures in an arm or leg are a common first sign. The involuntary movements may initially occur while a person is doing a specific action such as writing or walking. In some individuals, dystonia spreads to other parts of the body. The signs and symptoms of early-onset isolated dystonia can vary dramatically, even among family members. Several gene variants have been associated with early-onset isolated dystonia, most notably the DYT1/TOR1A gene discovered in 1997.

While away at college, thousands of miles from home, Liz experienced a slow creep of troubling difficulties. "I was having trouble walking. I ended up fracturing my ankle because I tripped over myself. I would take a cab two blocks because I just couldn't stand it." One day toward the end of her freshman year, she was so exhausted after completing an assignment and walking a short distance from the library, she crashed at her dorm and slept for 22 hours.

Despite a close relationship with her parents, Liz did not readily talk about her deteriorating condition. "It was like pulling teeth to get information from her as to how she was doing," Carole said. "Liz never complained." A check-up with Liz's movement disorder specialist confirmed that the dystonia was spreading to her legs and feet, and could explain the pain and loss of control.

"I wasn't always the most forthright about the state I was in," admitted Liz, "but my parents were always there when I needed them and they were always supportive in a way that allowed me to maintain my independence. I felt like I had a safety net if things went bad." Liz continued: "It can be really hard to establish your independence as a young adult if you are never given the opportunity to fall—and to fall enough until you feel like you need to ask for help. Asking for help is a really important lesson to learn."

At the start of Liz's spring semester of sophomore year, her dystonia symptoms became overwhelming. "It was bad. My body wasn't functioning anymore," she said. She could not walk, and barely left her dorm room for a week—even to attend class. She texted her mom late one night, having reached a breaking point. Liz decided to take a medical leave of absence from school and return to California to have the DBS procedure her doctor had been recommending for years.

Carole immediately suggested she would fly out to help Liz move out of her dorm. Liz refused. "I think that could be hard as a parent," reflected Liz. "I said, no, I don't need help. I think many parents would say, I don't care, I'm coming anyway. But my parents trusted me to know what I needed."

In March of that year, at age 19, Liz underwent DBS surgery. Carole said, "The hardest time for me with Liz was when she went in to have the DBS surgery. I felt so responsible for the whole thing. She would not have been going through any of this if not for me." Fortunately, the surgery went smoothly, without complication. Her recovery was slow at first, but within a few weeks she regained her strength.

Just two months after the DBS device was implanted, Liz returned to Washington, DC for a summer internship with a presidential campaign. That September she continued working full-time for the campaign while enrolled in a full schedule of classes. She was traveling home to California every four or five months for appointments to adjust her DBS settings. Ideally, the appointments would have been scheduled weeks apart, not months, to determine the optimal settings required to control the dystonia, but Liz was determined to keep up with school. "I was about 20% better," recalled Liz. "I was still definitely disabled. But better was better." She was grateful to have had the surgery.

After graduation, Liz spent the summer at home in California, working with her neurologist to fine-tune her DBS settings. In the fall, she relocated to New York City to begin her career. "Within six months of moving, I was basically asymptomatic," she said. She has since taught herself to write again, can walk with little difficulty, and works full-time for a marketing technology company. "I have tried to make every decision in my life based on things that I want and not based on what might be easiest for me. There are so many things in New York that make it hard for a disabled person to exist, but I've always wanted to live in New York and I wasn't going to let any of that stop me."

Liz and Carole joined the DMRF Board of Directors in 2018. Both are registered brain donors. Carole has volunteered for numerous research studies at the National Institutes of Health. The Rawson family has generously supported the DMRF since the 1990s, including recent Dystonia Zoo Walks on both east and west coasts. Carole has yet to find an effective medical therapy to treat her writer's cramp. She recently began experiencing mild dystonia symptoms in her legs and feet.

"Up until Liz [developed dystonia], I looked at my dystonia as a gift because I felt it was a great equalizer," said Carole. "It makes me appreciate life more and has given me the ability to better empathize with others."

As a DMRF Board Member, Liz is especially sensitive to the experiences of young people with dystonia, particularly those who are navigating the transition into adulthood. "Don't rule out jobs or opportunities ahead of yourself apply for the job, apply for the internship," she would suggest. "Give people a chance to welcome you. Give people the chance to make accommodations for you to be able to do the job. Everyone's dystonia presents differently, and for many of us there are jobs that our bodies aren't able to do, but don't rule things out because you make assumptions about the people who are doing the hiring. Go for it."

Carole added: "Liz has so much grit and perseverance. Learning to be an advocate for herself at an early age has been an amazing tool for her throughout her life. Don't let your disability define you—let it make you stronger. It will always be a part of you, but don't let it be who you are."



PERSONAL PROFILE Lauren Bianco

Lauren Bianco was diagnosed with dopa-responsive dystonia about three years ago. Her passion is photographing the wildlife and natural beauty of Long Island, NY.

What is it like to have doparesponsive dystonia due to SPR deficiency? How were you diagnosed?

Growing up was really rough. I had a lot of issues with low tone. I had Special Ed services when I was two and a half, I had speech and occupational therapy. They didn't know what was wrong with me. By third grade, I was very delayed in all my areas such as math, reading, science, all that. I was diagnosed with OCD [obsessive compulsive disorder] when I was eight. I had speech until high school. In my 20s, my mom noticed that my feet would turn in and the orthopedist said, yeah, she definitely needs braces. Still not diagnosed yet. Years later, I started getting migraines and the neurologist asked, what happened to Lauren, why is she the way she is? And we were like we don't know. Because l research everything online one day l thought, hypotonia [low tone] is not a disease by itself, it's triggered by something else. I said, I want to





go for genetic testing and my neurologist said, that's a great idea. After three months of waiting she said you have a condition called dopa-responsive dystonia and one of your genes, the SPR gene, is mutated. This is what causes the hypotonia, the intellectual disability, the learning disability, the OCD, anxiety, all that. Once I read about it, it brought me to tears because I was so happy that I finally had a diagnosis. I was thinking maybe the doctor is thinking that I'm lying or maybe my therapist thought I was a hypochondriac, but I was always in pain growing up, even doing the simplest things like writing on the board, writing my homework. I would get tired easily. I was always the slowest one. I knew I was different but I never knew what was wrong with me.

What treatments have helped you?

Botulinum toxin and trigger point injections for my migraines, occupational therapy and physical therapy, mindfulness for anxiety, and just having a positive outlook in life. I am on Sinemet[®]. I still get tired easily. Walking is a little better. I still have balance issues. I still have a lot of tremor. The biggest challenge is the emotional part of it—the OCD and anxiety and irritability. I'm a

very sensitive person. I have a tough time accepting that I am 34 years old. I was told that I have a mental age of about I2 years old. I may not sound like I'm I2, but mentally and emotionally I am, and I accept this about myself. I'll go to stores and look at toys and be so happy like a little kid, or I go to the movies and watch kid shows. I look at myself and think, wow, 34 years old but I'm stuck in this 12 year old mind. But I accept who I am.

How do you spend your time? Talk about your photography.

I go to a school-based program for people that have intellectual and developmental disabilities. We do things like cooking, reading, science, math. We go on community trips. I also have my one-to-one [mentoring program] and go out three times a week.

I have always loved animals. I said to my mom one day, let's see if I can start taking pictures. My dad got me my first camera. I started posting the pictures on my Facebook and the response I got was unbelievable. It built my confidence up tremendously. My mom is my partner in crime. Since I cannot drive, she drives me around all these places. We get really excited. Photography helps me focus. It takes away my anxieties. Sometimes I don't even realize I'm wearing braces and walking in the sand on the beach or I'm in the freezing cold weather. Once you see these animals in their natural habitat, it is unbelievable.

What advice do you have for others living with disabilities?

Just because you have disabilities, it may take you longer than others to do the same things, but eventually you will get it. I never thought that I would be a photographer. I love being out in nature and exploring Long Island. It's an amazing place.

Dopa-responsive dystonia (DRD) is a group of movement disorders that include symptoms of dystonia and parkinsonism. Mutations in multiple genes can cause of dopa-responsive dystonia, including GCH1, TH, or SPR. Sepiapterin reductase (SPR) deficiency is a rare genetic condition characterized by movement problems including dystonia, muscle stiffness, tremors, difficulty with coordination and balance, and involuntary jerking movements. People with SPR deficiency may also experience intellectual disability, seizures, sleep disturbances, and mood disorders.

Social Media Fundraisers Matched in 2019



Thanks to a generous anonymous donor, all donations raised for DMRF through Instagram and Facebook fundraisers are eligible to be matched dollar for dollar throughout 2019. This generous challenge will help us continue to collect support for dystonia research towards a cure. If you or a family member are raising

donations for DMRF via social media, you must notify us by emailing events@dystonia-foundation.org or inviting us to participate in your fundraiser to qualify for the match. Instagram and Facebook do not notify DMRF of these fundraisers, and we wish to acknowledge the generosity of all our supporters.

During Dystonia Awareness Month in September, DMRF received \$10,000+ in Facebook donations!

Thank you to our social media friends and followers for participating in this exciting challenge.

Creative Giving: Tax-Free Gifts from Retirement Accounts

If you are 70 1/2 or older, you can jumpstart your legacy giving by making a tax-free gift from your IRA (individual retirement account). Your contribution will lay the groundwork for vital research supported by the DMRF now and in the years ahead.

Normally, a distribution from your IRA is taxed as ordinary income. However, if you are 70 ½ years of age or older, you are eligible to make a direct gift from your IRA to a qualified charitable organization during any tax year. The tax-free transfer can count toward your required minimum distribution.

For more information on this giving option, plus additional creative ways to financially support DMRF, visit https://dystonia-foundation.org/donate-other-ways/ or call us at 312-755-0198.

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Thank You are essential to the DMRF mission to find a cure. Donate today at dystonia-foundation.org/donate