



DYSTONIA
MEDICAL
RESEARCH
FOUNDATION

Dystonia Dialogue

NEWSLETTER OF THE DYSTONIA MEDICAL RESEARCH FOUNDATION

Spring 2021 | Volume 44 • No.1

Shining a Light on Dystonia

And a Path Forward for Every Family



4

Dystonia Advocates
Rescue DOD Research
Funding

7

What is a Physical Medicine
& Rehabilitation Physician?

12

Journeys in Parenthood:
Stories from Parents
with Dystonia

Inside this Issue

4 Dystonia Advocates Rescue Critical Source of DOD Research Funding

Volunteer Advocates Needed to Support Legislative Agenda in 2021

5 Dystonia Community Hard Hit by Covid-19 Pandemic

Highlights from a DMRF Survey

7 What is a Physical Medicine & Rehabilitation Physician?

Insights from John McGuire, MD

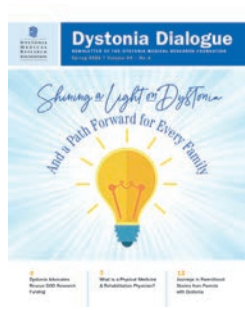
12 Journeys into Parenthood

Stories from Parents with Dystonia

18 Personal Profile

Meet Amber Hall

On the Cover:



Since 1976, the Dystonia Medical Research Foundation (DMRF) has led a global effort to advance dystonia research and empower patients and families. The DMRF brought dystonia out of obscurity and into the spotlight of neuroscience and movement disorders research. Thanks to our generous supporters, we continue to uncover dystonia's mysteries with every new research discovery. For 45

years, the DMRF has helped light a path forward for individuals and families who don't know where to turn after a dystonia diagnosis.

No one has to face dystonia alone. See page 10 for more information on the DMRF's programs and accomplishments.

The Dystonia Dialogue is supported by an educational grant from Revance Therapeutics.

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.

Staff

Janet L. Hieshetter
Executive Director
Veronica Diaz
Manager of Community Engagement
Debbie Durrer
Director of Development
Jessica Feeley
Director of Communications & Education
Jennifer McNabola
Director of Finance
Martha Murphy
Information Coordinator & Brain Bank Liaison
Emma Pinto
Associate Director of Development
Jody Roosevelt
Science and Technology Manager

Chief Scientific Advisor

Jan Teller, MA, PhD

Dystonia Dialogue Editorial Board

Barbara Kessler, *Chair*
Ed Cwalinski
Paul Kavanaugh
Dee Linde
Allison London
Anneliese Ornelis
Diane Rudolph
Paula Schneider

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

Printed in the USA. © Dystonia Medical Research Foundation

Dystonia Medical Research Foundation Board of Directors

Samuel Belzberg
(1928-2018) Co-Founder
Frances Belzberg
Honorary Chairperson & Co-Founder
Mahlon R. DeLong, MD
Scientific Director Emeritus, Lifetime Honorary Director
Stanley Fahn, MD
Lifetime Honorary Director
Art Kessler, *President*
Richard A. Lewis, MD
Vice President of Science
Barbara Kessler
Vice President of Awareness & Education
Karen K. Ross, PhD
Vice President of Support
Carole Rawson
Vice President of Public Policy
Ronald Hersh
Vice President of Development
Mark Rudolph, *Treasurer*
Sandra Weil, *Network Liaison*
Jon Davis, *Director*
John Downey, *Director*
Donna Driscoll, *Director*
Marilynne Herbert, *Director*
Erwin Jackson, PhD, *Director*
Stefanie Jackson, *Director*
Brian Keane, *Director*
Dennis Kessler, *Director*
Rosalie Lewis, *Director*
Allison London, *Director*
Liz Rawson, *Director*
Diane Rudolph, *Director*
Paula Schneider, *Director*
Pamela Sloate, JD, *Director*
Joel S. Perlmutter, MD
Scientific Director, ex officio
Billy McLaughlin
Awareness Ambassador, ex officio
Janet Hieshetter
Executive Director, ex officio

Foundation Update



Art Kessler
President

This year marks the 45th anniversary of the founding of the DMRF. The milestone is an opportunity to reflect on the Foundation's many important accomplishments, but it is not an occasion we celebrate. The DMRF is working hard to close its doors for all the right reasons: because a cure is discovered and no person or family is ever again burdened by dystonia. Thanks to the commitment of generous friends who have supported our service to the community, we know this day is coming.

As long as individuals are diagnosed with dystonia, the DMRF will continue pushing the research forward and attending to the needs of affected individuals and families. We know our community struggles with access to effective treatment, social isolation, and the societal challenges so many with disabilities and chronic health conditions face on a daily basis. The Covid-19 pandemic has exacerbated these challenges for many, and we are redoubling efforts to address them in our programs and activities.

Your support allows DMRF to sustain our research programs while continuing to offer the free support and education programs that people with dystonia and their families need now.

It is more important than ever for the dystonia community to have a voice in federal policy and legislation matters. DMRF is calling on the community to join the Dystonia Advocacy Network (DAN) to lend your stories and experience to help Members of Congress understand what dystonia is and how it changes lives. Learn how you can become a dystonia advocate and participate in efforts throughout the year on page 5.

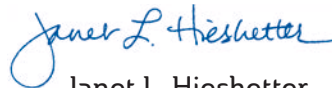
It is a privilege for the DMRF leadership to be of service to the dystonia community, and we are only able to do so because of generous people like you. Thank you for being a part of the DMRF.



Janet L. Hieshetter
Executive Director



Art Kessler
President



Janet L. Hieshetter
Executive Director

Renew Your Membership or Join Today

dystonia-foundation.org/membership

Thank you for your support!

Dystonia Advocates Rescue Critical Source of DOD Research Funding

Volunteer Advocates Needed to Support Legislative Agenda

Dystonia advocates acted swiftly to protect a critical source of research funding for dystonia investigators. In late 2020, the Dystonia Advocacy Network (DAN) was alerted that dystonia was not included in the list of conditions eligible for study through the Department of Defense (DOD) Peer-Reviewed Medical Research Program (PRMRP).

Unlike the National Institutes of Health (NIH), the medical conditions included in the PRMRP must be specifically recommended by lawmakers each year. Because of the advocacy of the DAN, dystonia has been included on this exclusive list of conditions annually since 2010, resulting in more than \$17 million awarded to dystonia investigators.

“As a community, it’s vital that we take time to advocate for dystonia to ensure that our representatives not only understand the disorder but understand the importance of funding research to find a cure,” explained Carole Rawson, Chair of the DMRF Public Policy Committee and newly appointed Chair of the DAN.

In response to learning that dystonia was not on the PRMRP list of conditions, the DAN mobilized dystonia advocates in key districts. Advocates contacted their Members of Congress and requested that dystonia be included in the PRMRP. Thanks to their quick action, dystonia was added as a condition eligible for funding, avoiding a potentially devastating disruption to dystonia research supported by DOD. “As a result of the overwhelming response



Carole Rawson and daughter Liz Rawson serve on DMRF’s Board of Directors.

from our action alert, dystonia was the only condition added to the list!” said Carole.

DAN held Virtual Advocacy Day on March 3, 2021 to launch plans for the coming year. Hundreds of dystonia advocates contacted their US Senators and US Representatives to educate them about dystonia and the needs of the dystonia community. In advance of Advocacy Day, every newly elected Member of Congress received a personalized letter and information about dystonia. The DAN reaches out to

new members following the start of every Congress.

Carole encourages individuals with all types of dystonia, as well as family members, to consider becoming a dystonia advocate: “Through our collective voices, we can unlock much-needed funding for research and new treatments. When you share your story, it helps our legislators put a face to our daily struggles and our dreams for the future. We are a powerful force when we all join together.”

The DAN is a coalition of patient organizations focused on issues relevant to the dystonia patient community. Many of these issues are anchored in advocating for federal research funding and accessible, affordable treatment.

Victories for dystonia advocates include increased dystonia research funding for the NIH, new dystonia research funding through the DOD, the first-ever Congressional Briefing on Dystonia, protecting access to deep brain stimulation and botulinum neurotoxin therapies, and safeguards for new biologic therapies.



Dystonia Advocacy Network

DMRF is proud to provide
staff support for the DAN.

Benign Essential Blepharospasm
Research Foundation (BEBRF)

Dystonia Medical Research
Foundation (DMRF)

National Spasmodic
Dysphonia Association (NSDA)

National Spasmodic
Torticollis Association (NSTA)

How You Can Help

By becoming a dystonia advocate, you can have a powerful effect on the laws and policies that affect your life and the lives of others in the dystonia community. To get started:

- Sign up to receive DAN action alerts via email at: dystonia-foundation.org/advocacy
- When you receive a DAN action alert, respond promptly by calling or sending an email to your legislators. The DAN makes it easy. Every alert includes clear, simple instructions to take action.

For more information on the DAN and becoming a dystonia advocate, visit dystonia-foundation.org/advocacy or contact the DMRF at dystonia@dystonia-foundation.org or 312-755-0198.

Dystonia Community Hard Hit by Covid-19 Pandemic

Nearly 450 people responded to a DMRF survey seeking insights into how the Covid-19 pandemic is impacting the dystonia community.

The following are highlights from the results.

How concerned do you feel for your health and safety during the pandemic?

Very concerned	44%
Somewhat concerned	35%
A little concerned	17%
Not concerned	4%

How concerned do you feel for the health and safety of your family and loved ones during the pandemic?

Very concerned	55%
Somewhat concerned	36%
A little concerned	8%
Not concerned	1%

56% of survey participants experienced canceled or delayed healthcare.

Of these, 70% report canceled or delayed dystonia-related healthcare.

68% of survey participants have had telemedicine appointments.

Of those, only 6% were not satisfied with their telemedicine experience.

How has the pandemic affected your overall physical health?

Improved a lot	3%
Improved a little	8%
Has not changed	40%
A little worse	36%
A lot worse	13%

How has the pandemic affected your dystonia symptoms?

Improved a lot	1%
Improved a little	6%
Has not changed	36%
A little worse	38%
A lot worse	19%

How has the pandemic affected your mental health?

Improved a lot	1%
Improved a little	4%
Has not changed	34%
A little worse	44%
A lot worse	17%

Top five most important things DMRF should be doing for people during the pandemic:

1. Offering dystonia research updates
2. Offering information and resources about emotional and mental health
3. Offering information and resources about self-care
4. Promoting awareness of how the pandemic is impacting dystonia community
5. Offering updates on healthcare legislation and policy

Dystonia, Depression & Anxiety

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.

People who live with dystonia may also experience a range of non-motor symptoms that dramatically impact daily activities and quality of life.

Depression is the #1 cause of disability in the US

Anxiety and depression are among the most common non-motor issues seen in individuals with dystonia.

Studies have shown that as many as 70% of individuals with dystonia will experience depression and/or anxiety over their lifetime.

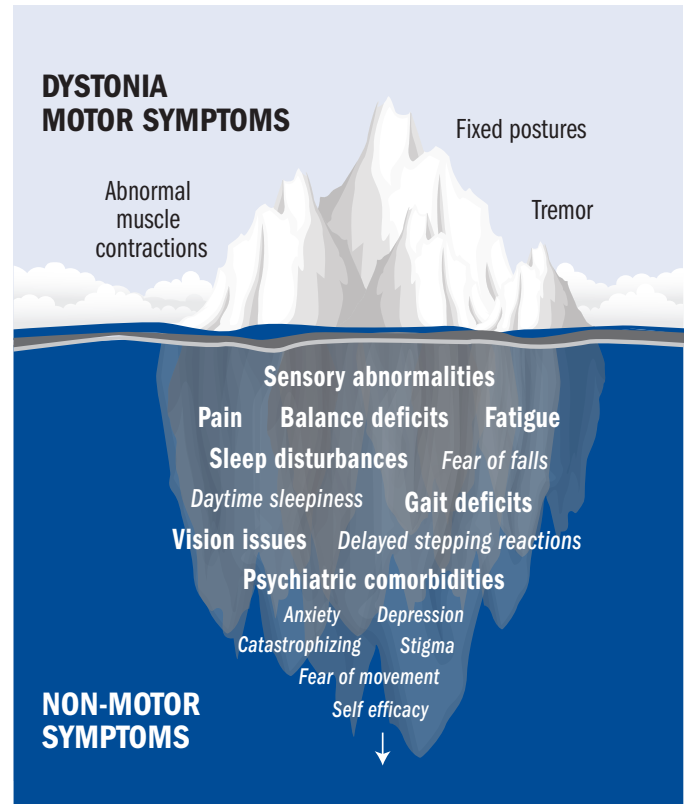
Anxiety and depression significantly worsen disability and quality of life in dystonia.

The relationship between dystonia, depression, and anxiety is complex. Research demonstrates that in some cases, depression and anxiety may develop in response to dystonia. In other cases, they occur before the onset of dystonia and appear to be part of the dystonia syndrome.

Evaluating for and treating co-existing depression and anxiety disorders is an important part of a holistic approach to treating dystonia.

Reducing movement symptoms associated with dystonia may not always alleviate depression or anxiety.

Treatment options for depression and anxiety depend on the individual needs of the patient. Interventions may include lifestyle adjustments, self-help activities, cognitive behavioral therapy, other types of counseling, and/or a range of intensive psychological therapies.



The hallmark symptoms of dystonia are involuntary movements, but individuals often deal with a range of less obvious, non-motor symptoms.

To locate a mental health professional for help, ask your doctor, health insurance provider, or a professional organization such as the Anxiety and Depression Association (adaa.org). Or ask for recommendations from family and friends.

Adapted from the webinar “Dystonia is More than a Movement Disorder: Depression & Anxiety” with Brian Berman, MD, Director of Virginia Commonwealth University’s Parkinson’s and Movement Disorders Center. The webinar is available for viewing at dystonia-foundation.org/living-dystonia/mental-health/

Image adapted from a figure in Bradham, L.V., Meiring, R.M., Boyce, M. et al. Neurorehabilitation in dystonia: a holistic perspective. J Neural Transm (2020).

What is a Physical Medicine & Rehabilitation Physician?

“We’re experts in the diagnosis, pathophysiology, and management of any disabling condition,” explained John McGuire, MD, a physical medicine and rehabilitation (PM&R) specialist and Director of Stroke Rehabilitation & Spasticity Management at Froedtert Medical College of Wisconsin.

PM&R physicians evaluate and treat patients with physical and cognitive impairments resulting from musculoskeletal conditions (neck or back pain, or injury), neurological conditions (stroke, brain injury, or spinal cord injury), and other medical conditions. “We approach all these conditions in the same light,” said Dr. McGuire, “as far as asking, what are your deficits and how can we get you as functional as possible?”

PM&R physicians are also known as physiatrists. Some in the field avoid using this term due to the frequent confusion with psychiatrists and psychiatry.

The field of PM&R emerged after World War I and World War II in response to hundreds of thousands of soldiers returning from service with brain injuries, spinal cord injuries, amputations, and other traumatic injuries. Dr. McGuire explained: “Basically the surgeon would piece them back together and now they have to figure out how are they going to dress themselves, feed themselves, walk, and get back to work and doing the things they need to do. Our goal is to improve function and reduce pain, basically a non-surgical approach to overall well-being that goes beyond the medical model. It’s more of a holistic approach to all the ways that someone’s life is impacted by a condition or injury.”

PM&R physicians practice in a number of clinical settings. Subspecialties range from brain injury and spinal cord injury to sports medicine, pediatric rehabilitation, and more. PM&R physicians typically work in an interdisciplinary fashion with physical therapists, occupational therapists, speech therapists, and other supportive therapies depending on the needs of patients.

When assessing a patient, careful consideration is made to overall wellness. Dr. McGuire explained: “Whatever ails you, if you’re not eating right, sleeping right, if you’re not exercising, no matter what your problem is, that’s going to create more problems. We have to look at everything to get your life back together.”

Dr. McGuire was trained in stroke rehabilitation and electromyography (EMG). He was an early leader in the therapeutic use of botulinum neurotoxin and intrathecal baclofen. This exposed him to patients with dystonia and spasticity resulting from stroke, cerebral palsy, and other conditions. “I love working with my movement disorders colleagues. We all work together,” said Dr. McGuire. “If a patient turns out to be more in the dystonia sphere, I may send them to the neurologist; if the patient is more in the spasticity sphere, they may send them to me. There’s this cross fertilization with all these patients.”

The nature of PM&R requires keen understanding of human anatomy and body mechanics. However, McGuire stressed that not all PM&R physicians can be expected to treat movement disorders. Credentials that lend PM&R physicians to expertise in dystonia and movement disorders include certification in electrodiagnostic medicine (EMG) and advanced training in neurorehabilitation. A small number of medical institutions offer advanced fellowship training for PM&R physicians specifically in movement disorders neurorehabilitation.

The DMRF offers a Find a Doctor directory at dystonia-foundation.org/find-a-doctor

Official Keith Emerson Tribute Concert Release to Support DMRF

Proceeds from *The Official Keith Emerson Tribute Concert* disc set, released March 11, 2021, will benefit The Leon Fleisher Foundation for Musicians with Dystonia, a program of the DMRF. This highly



anticipated concert captures the essence of what made Keith Emerson a pioneer of progressive rock, a treasure among his many fans and friends, and a legendary keyboardist and composer with the iconic supergroup Emerson, Lake & Palmer. Emerson died in 2016.

The Official Keith Emerson Tribute Concert is available in both DVD Digipak format and in a Blu-Ray Hi-Def download, playable on any device, both accompanied by a bonus audio version of the concert. The 3-disc set documents the historical 2016 tribute show in Los Angeles featuring a once-in-a-lifetime line-up of musical luminaries performing music from Emerson, Lake & Palmer, The Nice, Emerson, Lake & Powell, The Keith Emerson Band, and the Three Fates Project.

The 2 1/2-hour concert film features stellar performances by over two dozen prominent artists in rock, jazz, and classical music as well as artist interviews, behind-the-scenes footage, tribute speeches, and a gallery of personal never-before-seen photos from the Emerson estate.

All of the artists, crew, and support personnel kindly donated their skills and time to bring visibility to musician's dystonia and support research toward a cure.

The Official Keith Emerson Tribute Concert disc set is available for purchase at: fanfarefortheuncommonman.com

New Resource for Musicians



The DMRF has created a private peer support group on Facebook for musicians affected by dystonia.

Musicians with Dystonia Support Forum
facebook.com/groups/musiciansdystonia

Samuel Belzberg 6th International Dystonia Symposium Rescheduled

Due to Covid-19, the Samuel Belzberg 6th International Dystonia Symposium in Dublin, Ireland has been rescheduled for June 1–3, 2023. The scientific meeting is named for DMRF Co-Founder Samuel Belzberg who sadly passed away in 2018. Co-Chairs are H. A. “Buz” Jinnah, MD, PhD of Emory University School of Medicine and Antonio Pisani, MD, PhD of University of Pavia. The symposium is a joint effort of Dystonia Europe and the DMRF. 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 meeting for investigators and clinicians, dating back to 1975.

Dystonia Support Available via Video Conference & Online

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

- **Support Groups** – DMRF offers peer support groups in communities across the USA, many of which are meeting virtually by video conference. Search support groups at dystonia-foundation.org/support-groups
- **Online Forums** – Online groups are available for specific types of dystonias, musicians, parents, loved ones, young adults, and more. Find links for online forums at dystonia-foundation.org/online
- **Individual Support** – DMRF is available by phone, email, web, and social media to those seeking information and support.

People on the Move

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

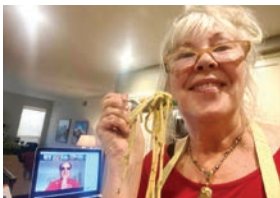
“The Singing Chef” Andy LoRusso partnered with DMRF to host “Cooking for a Cure,” a live interactive online cooking event on January 29, 2021. Participants prepared two authentic Italian dishes, *cacio e pepe* and Sicilian ricotta cheesecake, while crooning classic songs in Italian and English, including *Nel Blu Dipinto di Blu (Volare)*, *That’s Amore*, and *Arriverderci, Roma*.



For two years, Andy has been coping with blepharospasm, a focal dystonia that causes uncontrollable blinking and closure of the eyes. “I wanted to give back in hopes of finding a cure,” he said. “It’s also about fun. Cooking and singing, creating a happy, positive mood, bringing family together in the kitchen—that’s been my passion for 30 years.”



Since the 1990s, “The Singing Chef” Andy LoRusso has performed thousands of shows blending food, song, and dance to bring people together. He is the author of the best-selling cookbook and album combo, *Sing and Cook Italian*, and *Sing & Cook with Andy LoRusso the Singing Chef*.



Andy is donating a portion of proceeds from his signature pasta sauces to support the DMRF. Shop at singingchef.com

THANK YOU to 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 limited information about these fundraisers, and we wish to acknowledge all of our supporters. Plus, social media fundraisers in 2021 will be matched up to \$30,000 by an anonymous donor!

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

Be sure to notify DMRF of your social media fundraiser to be eligible for the match. Tag or DM us on Facebook or Instagram (@dystoniamrf), or send a message via dystonia-foundation.org/contact



Instead of the annual Chicago Basket Bash, longtime DMRF supporters **Beth Farber and Steve Laser** produced and hosted “Dish for Dystonia” a one-episode, online parody cooking competition to benefit DMRF. Chefs **Adrienne Sabora, Dani Fingard, Elena Tubridy, Jill Povol, and Scott Weisman** each prepared a dish to be evaluated by guest judges. The audience voted for their favorite chef by contributing a dollar per vote. After a week of voting, Elena Tubridy earned the most donations and was announced the winner of the challenge.

Why is DMRF Important to You?

2021 marks the DMRF’s 45th anniversary. Please consider sending us a brief video or typed testimonial about why the work of DMRF is important to you.

For details and instructions, visit dystonia-foundation.org/45



Shining a Light on Dystonia

And a Path Forward for Every Family

Since 1976, the Dystonia Medical Research Foundation (DMRF) has led a global effort to advance dystonia research and empower patients and families.

The DMRF brought dystonia out of obscurity and into the spotlight of neuroscience and movement disorders research. Thanks to our generous supporters, we continue to uncover dystonia's mysteries with every new research discovery.

DMRF continues to fight for recognition of dystonia as a life-changing neurological disorder in urgent need of research and resources. We bring awareness to policymakers, government institutions, and the general public.

For 45 years, the DMRF has helped light a path forward for individuals and families who don't know where to turn after a dystonia diagnosis. No one has to face dystonia in the dark. No one has to face dystonia alone.

The DMRF's accomplishments are more than notable bright spots in the organization's history. Every accomplishment shortens the path to a cure and generates new opportunities to improve the lives of individuals impacted by dystonia.

For more information on the programs and activities of the DMRF, visit dystonia-foundation.org

\$18M
National
Institutes of Health
dystonia research
funding FY2021

30+
movement
disorder
fellows
trained

Health
ch
21

60
peer support
& online
groups

6,000
Global Dystonia
Registry
sign-ups

270
investigators
funded

120,000
*Dystonia
Dialogues*
mailed per
year

\$37M
invested
in science

**GENES
DISCOVERED**
DYT1/TOR1A
DYT6/THAP1
DYT16/PRKRA
DYT25/GNAL

\$17M
Department
of Defense
research
funding

\$1.7M
savings
for Dystonia
Coalition

Pharma
pursuing
new drugs

- 💡 Full-time Chief Scientific Advisor
- 💡 Clinical trials
- 💡 Drug compounds tested
- 💡 Molecular pathways mapped
- 💡 Protein structures defined
- 💡 30+ scientific meetings
- 💡 Advocacy to Congress and policy makers
- 💡 63 Dystonia Zoo Days
- 💡 Free educational materials
- 💡 Personal assistance for every office contact

Journeys into Parenthood

The path to becoming a parent for individuals in the dystonia community is as varied as society at large. There are more options than ever for having children: biologically, via reproductive medicine, fostering, and adoption. Parents with dystonia may encounter challenges that parents without health issues might never imagine. Here are two parenthood stories from the dystonia community.

The Downey/Botman Family

When John Downey began showing symptoms of dystonia at age 13, his parents knew exactly what it was. Because of a known family history, John was promptly diagnosed with DYT1 dystonia, an inherited dystonia that typically begins in childhood and progresses throughout the body and limbs. Dystonia caused John's left foot to turn inward, causing difficulty walking that worsened as he got older.

John relocated from Indiana to Chicago to pursue a career as a software developer. His view of the future included conflicted feelings about having children, both because of the physical demands of parenting and the genetics of DYT1 dystonia. Carriers of the gene mutation for DYT1 dystonia can pass the disorder onto their biological children. "I was really kind of against having kids because I struggled so much with dystonia, was in pain a lot of the time, and I did not want to bring another person into that," he said.

Consulting an internationally recognized movement disorder neurologist in 2012 proved "life-changing." For one, she outlined a treatment plan with several new options to try including oral medications, botulinum neurotoxin injections, and, if needed, deep brain stimulation. John was surprised when she asked if he had given any thought to family planning. She explained that some families with DYT1 dystonia were using in vitro fertilization (IVF) and pre-implantation genetic testing (PGT) to essentially eliminate the risk of future generations being born with the dystonia-causing mutation. She encouraged John to contact the Dystonia Medical Research Foundation (DMRF) to connect with couples who had pursued this option, if he was interested. Until this interaction, John explained, "I didn't understand the genetics enough to know that IVF and PGT could be a viable option for me. I didn't even know what PGT was, and I had a loose understanding of IVF."

...

John and Mara Botman started dating in 2013. Mara works in non-profits and had lived in Chicago since college. "I was always very embarrassed about my dystonia, people would look at me funny," said John. "I intentionally tried very hard to hide it from her at first. I would always walk behind her when we were at restaurants or out together because it was a hard conversation to have."

A few dates in, John shared what dystonia was and how it affected his life. Mara was immediately supportive. "My biggest goal for John was always figuring out what he could do to increase mobility and comfort," she said. "I was fortunate to have a very well-educated partner in John. I did go to some of his doctor's appointments with him, and I was always eager to be there. But it was less for an educational experience and more of wanting to advocate for him to consider all his options."

When the couple started talking about the future and the possibility of a family, Mara was unfazed by the idea of consulting a genetic counselor to discuss options for reducing the risk of their children developing dystonia. "I'm from a Jewish family and genetic counseling is very common in the Jewish community given all the Jewish genetic disorders, so it felt like genetic counseling that I would do any way," she said.

Similarly, she was open to learning about IVF and PGT: "I always assumed I would want children and doing in vitro seemed like something that people sometimes did based on genetic counseling. None of this seemed particularly extreme. It seemed much more like a planning hurdle."

...

On Valentine's Day in 2015, John had deep brain stimulation surgery (DBS). "I saw a pretty tremendous response



DMRF Board Member John Downey and Mara Botman welcomed their first child in 2019.

from that,” he said, in terms of reduced dystonia symptoms and pain. “It was great for a while. It was really good. We took a lot of trips, we were a lot more active.” The couple married in 2016.

The success of DBS further convinced John to seriously consider IVF and PGT. “I went on this tear of, yes, everything I can do to leverage science and technology to improve my life, why wouldn’t I try to leverage that? After DBS, I even got Lasik,” he laughed.

John joined the Board of Directors of the DMRF and the couple connected with others in the dystonia community whose families had undergone PGT.

Mara reached out to a friend

for a recommendation for an IVF clinic. The consultation with the IVF clinic set the process toward parenthood in motion.

IVF is not a single treatment but a series of procedures. PGT is a step in the process. An average IVF cycle takes months from the first consultation to—hopefully—a successful pregnancy. The process is complicated and unpredictable. “A logistical nightmare,” said Mara. She elaborated: “I was going to a clinic three days a week and getting shots and giving myself shots and getting blood taken and all that stuff. It was a lot of medication in my body, extending the pregnancy process in a way that I didn’t fully appreciate. I also didn’t realize how long it was going to take. You have to find the right doctor. You have to make sure the clinic has capacity for you. You have to order the medication. You have to get the right timing to start the medication.” The couple also had to figure out what aspects of the process were covered by health insurance and what would be paid out of pocket, very little of which was straightforward or even logical.

• • •

Lewis was born in 2019. He was named after Mara’s father who died when she was 9 years old. “He is a happy, delightful baby,” said Mara. “I was very, very sick during both the in vitro process and pregnancy, and so it was a tough year, and then he came, and he’s this delightful child who loves the world.”

What is In Vitro Fertilization?

In vitro fertilization (IVF) is a method of assisted reproduction that involves combining an egg with sperm in a laboratory. If the egg fertilizes and becomes an embryo, the embryo is transferred to the uterus where it will hopefully implant in the uterine lining and develop into a successful pregnancy.

What is Pre-implantation Genetic Testing?

Pre-implantation genetic testing (PGT) is a group of techniques to examine embryos during in vitro fertilization (IVF) for a range of genetic problems before possible transfer to the uterus.

These genetic defects include single gene disorders (such as DYT1 dystonia), a missing or extra chromosome in the embryo, or the rearrangement of genes that can cause pregnancy loss and birth defects.

Embryos free of the genetic problems are transferred to the uterus for implantation and, hopefully, pregnancy.

When PGT is used to detect single gene disorders, a child has a 99%+ chance of being born without the gene mutation for which the embryos are screened.

The first step for individuals who wish to learn more about PGT is to consult a genetic counselor.

To locate a genetic counselor, consider asking your movement disorder specialist or obstetrician/gynecologist for a recommendation, consult your health insurance provider, or contact a local reproductive health clinic.

Like many new parents, John jokes that he did not entirely anticipate how drastically life would change after becoming a dad. Or how sweet the rewards would be: “Every time he snuggles into you or learns something new, it can take even the worst day you’re having and make it good again.”

Becoming a parent also gave John a deeper appreciation for parents of children with dystonia: “When I go back and talk to my parents, I feel a little bit closer to what they were going through. Until I had a child I never would have been close to understanding how painful it is to have your child struggle with something like dystonia.” A second DBS procedure in 2018 helped control the increasing dystonia symptoms John began noticing in the years before Lewis was born.

At the time of the interview for this article, Lewis was a year and a half old. He was learning lots of new words, loved books, and enjoyed woofing at dogs encountered on walks in the neighborhood.

Mara reflected, “I was sitting in a room with a lot of board games and I think about the Candy Land game and Chutes and Ladders—for us, becoming parents was very much this complicated journey as opposed to a straight line. That’s how I think about it: three steps forward, one step back, but eventually you get there. And it’s all worth it.”

The Dorner Family

“There are unique aspects of parenting with a disability that somebody without a disability isn’t going to have to think about, and that’s okay. We just have to figure it out,” explained Jenelle Dorner. Jenelle is a longtime DMRF supporter and moderator of Parenting with Dystonia, a private Facebook group created by DMRF.

Jenelle is diagnosed with dystonia and gastroparesis, a disorder that prevents food from moving normally through



DMRF Support Leader Jenelle Dorner is pictured with son Leif and service dog Herbie.

the digestive system. The dystonia and gastroparesis are caused by underlying mitochondrial disease, a chronic genetic disorder that occurs when a person’s cells cannot produce enough energy for the body to function properly. For years she has battled dystonia, malnutrition, fatigue, and pain.

She was a 19-year-old college student when the health challenges began with a sudden onset of dystonia triggered by medication. Despite recurrent illness and hospitalizations, the mitochondrial disease would remain undiagnosed until she was in her 30s. In the meantime, she adapted to using a wheelchair for mobility, got a service dog, and married her husband Greg in 1999, shortly after she finished her Bachelor’s degree.

Jenelle obtained a Master’s degree in motor control kinesiology and got to work toward a PhD in neuroscience. The demands of graduate studies were grueling, but ironically some of her chronic symptoms stabilized when she became pregnant. “I was so healthy during my pregnancy that it was kind of crazy,” said Jenelle. “At that time, I didn’t have the mitochondrial disease diagnosis, but we knew I had dystonia and my dystonia was a lot better.”

...

Leif was born in 2005. He was named after one of Jenelle’s mentors, a reproductive physiologist who would push her around his farm in a wheelbarrow when the mud was too thick for her wheelchair.

Given a history of unusual drug reactions, Jenelle said she was “adamant” about not having an epidural or medications during delivery. She prepared for a natural birth, enlisting the help of a labor coach. After 22 hours of labor her son came into the world weighing well over 9 lbs. “Looking back on it, I have no idea how I did it, or really what the heck I was thinking!” laughed Jenelle.

At home, Jenelle and Greg got creative about accessible parenting. They had a crib that swung open sideways and a changing table that could be lowered to the floor in case Jenelle didn't feel she could lift Leif safely. "My wheelchair was an asset because I could put a baby sling on and he was right there in my lap, or eventually he could climb into my lap and I didn't have to worry about picking him up as much," she said. "This is kind of funny, but when he was really tiny, I would put him in the laundry basket and drag the laundry basket around the house." Greg was able to work part-time temporarily after Leif was born. Then they hired a nanny to help occasionally with housework and caring for Leif.

When Leif was two years old, Jenelle was hospitalized with life-threatening complications from mitochondrial disease. She was in intensive care for two months. She recalled, "That's the first time I remember thinking, wow, I don't know if I'm going to be here for Leif, and it was really important to me to be here, on Earth, to parent him."

...

Jenelle remained passionate about a career in neuroscience, but slowed the pace of her research to spend more time with her son. "After I got my PhD," she said, "I was completely IV-fed, plus fluids, and I was having respiratory problems, all kinds of mitochondrial disease symptoms, and I realized it's going to get worse if I keep stressing myself like this. I went on postdoc interviews, and I was thinking I can't do this." Jenelle decided to focus on stabilizing her health and being a mom. "I felt this intense loss, like I lost my intended career, and I was very depressed when that happened. Yet motherhood gave me purpose and helped me find new things in my life that I wouldn't have had if Leif wasn't there."

Multiple prolonged hospitalizations when Leif was a toddler were the most difficult times. Jenelle explained, "When kids are that little, like between two and five, they don't understand what's happening, and they don't know why you left them. They think you left them. And at times I did." She came to recognize those separations as opportunities to strengthen their relationship: "We've been forced to take these little breaks when I've been sick, or there was a time he was in the hospital and I was at home missing him, and it makes you recognize how important you are to each other."

When Leif was old enough to attend school, he enjoyed introducing the other kids to Jenelle's service dog Herbie.

Jenelle and Herbie would do presentations to Leif's class each year. "Service dogs are a big catalyst to social interaction and teaching kids about disabilities," said Jenelle. "I think that made Leif feel okay about his mom, and for the other kids to feel okay about peers that had disabilities—and to learn about service dogs. It was fun."

As Leif grew older, the resources Jenelle counted on for assistance evolved. Her parents and in-laws have been ongoing sources of support. Her mother, an expert in special education, was especially helpful with navigating how to communicate with Leif in an age appropriate way when challenges or issues came up. As needed, Jenelle and Greg involved Leif in play therapy, cognitive behavioral therapy, and other therapeutic activities to build resilience.

...

Leif is now 15 years old. He is a big help to his mom, whether by doing laundry or walking the dogs. They read together in their own little book club and have in-depth philosophical conversations.

Jenelle has weathered numerous feeding tube surgeries and complications, intravenous feeding to her heart, immunoglobulin (IVIG) infusions to fortify her immune system, and years of trial and error with medications and supplements. At the time of this article, she was anticipating yet another surgery to fix a painful stomach tube.

"Parenthood is very challenging, but it's something that can be empowering and provide growth to your soul, to you as an individual and your understanding of other human beings in the world, because you are nurturing someone else," said Jenelle. "Not everyone has that desire, and that's okay. We all have our own journey. But if you do have the desire and the willingness to put your heart into loving a child, and learning through those challenges and hardships that are inherent to life or living with a disability, it can be a very a wonderful journey."



Support for Parents

Connect with other parents in the dystonia community by joining DMRF's private Facebook group:

Parenting with Dystonia

facebook.com/groups/dmrf.parenting

Becoming a Mom

My Experience with Gestational Surrogacy

By Allison London, DMRF Board of Directors

Spoiler alert: This story has a very happy ending.

I am a mom. I. Am. A. Mom. Those four words once seemed like a given, but then dystonia hit. Diagnosed at 30 with truncal dystonia, I was at the time physically unable to care for myself let alone a child, even with lots of assistance. Yet the possibility of not being a mother did not really sink in until a year later, when my movement disorder specialist confirmed what we already knew deep down: I should not (or was it could not?) carry a pregnancy.

The reasons why made so much sense—from my forward flexing posture and spasms posing potentially serious risks to both me and the fetus, to the unknown effects on pregnancy of deep brain stimulation (DBS), which I was hoping to undergo. Still it was the second biggest gut punch of my life, the first being the dystonia diagnosis itself.

Dystonia had taken a lot from my husband Dan and me in the five years since it had crept into our lives. But parenthood was non-negotiable. Two weeks later I was in a fertility doctor's office discussing gestational surrogacy. A gestational surrogate (or carrier) is a woman who carries a pregnancy that is not biologically her own. That is the path we chose to take.

To create embryos, we started the process of in vitro fertilization (IVF). Different than typical IVF, though, the embryo created would ultimately be transferred into the gestational carrier's uterus instead of my own. The embryos we made were frozen, waiting to be used post-DBS when my dystonia would, hopefully, be improved.

Nine months later I underwent DBS surgery and, 10 months after that, we were ready to start trying. We now had to choose a surrogacy agency, essential for many reasons, chief among is that it is the agency's job to find the right carrier. Our fertility doctor had relationships with two highly reputable agencies. After researching, we scheduled an appointment to meet with one. At that first meeting, the



DMRF Board Member Allison London with husband Dan and daughter Daisy.

agency captured us with their incredibly thorough carrier screening process and additional services. We retained them.

While they went to work, we did too, with marching orders given to us by the agency. We spoke with their lawyer about the legal aspects of gestational surrogacy. We spoke with a high-risk obstetrician since IVF pregnancies can cause complications. We spoke with the agency's psychologist. We created a profile about ourselves for potential carriers to review. Then we waited.

On September 11, 2013, one year to the day of my DBS surgery, we met our would-be carrier at the agency's "match meeting." It was surreal. We were there to decide whether this stranger was the right woman to entrust with the biggest responsibility we had ever asked of anyone.

Two days later we decided to move forward. Sadly, after three embryo transfers, she did not become pregnant, and according to stipulations in our contract, the agreement was terminated. We were inconsolable, a feeling that had become all too common, but we were not giving up. We waited. Again.

Two months later we met Iesha, the woman who will forever have our gratitude. I went through additional rounds of IVF egg harvesting to create more embryos. Iesha got pregnant. Dan and I shouted the news from the rooftops, including that our daughter was in another woman's belly. Though I was unable to drive and uncomfortable in long car rides, I did not miss an obstetrician appointment. We texted frequently and got to know her kids, who found out we were having a girl before our own family did. We appeared before a judge in Family Court to obtain a pre-birth order, naming Dan and me our unborn daughter's parents.

Our agency did so much behind the scenes including making the hospital aware this was a gestational surrogacy situation and all that entailed. The hospital was informed that we wanted our last name on our daughter's medical bracelet and bassinet upon birth, and that we would like a room at the hospital of our own, to spend that first night with her. The due date was getting closer and our obstetrician let us know that he thought our daughter would arrive a few days early. We got a hotel room near the hospital and waited. On December 27, 2015, Dan and I were in the delivery room at Iesha's side as our daughter came into the world. Dan cut the umbilical cord. Lots of tears were shed. Our entire family, as well as Iesha's, were gathered in the waiting room.

Choosing to start a family via gestational surrogacy is a decision fraught with sleepless nights. It is expensive and a true test of your determination and relationship. But if like me, you do not want dystonia to define you, this may be a viable option.

After all, I am a mom and Dan is a dad. Our daughter, Daisy, is sunshine personified, our seemingly impossible wish come true.

DMRF Merchandise to Promote Awareness

Have you checked out the DMRF online store lately? You'll find many practical items to promote dystonia awareness: face masks, tote bags, pop sockets, pins, key chains, and more. For details and to order visit: dystonia-foundation.org/merch



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.

- ☒ 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

PERSONAL PROFILE

Amber Hall

Eighteen-year-old Amber Hall is a high school senior in Florida. She was born extremely premature, weighing just 1 lb, 12 ounces, which resulted in cerebral palsy (CP), dystonia, and hearing impairment.

What is a typical day for you? How have you been coping during the pandemic?

I've been doing virtual online classes. At first it was a bit of a struggle because I was used to actual school, my regular classes. I have a job at a small farm. I have a stand where I sell my fruits and vegetables on Saturday mornings, now that the markets are open again. I'm graduating this year and then off to college and probably I'll get a second job.

I've also been doing this other thing, which is amazing, and it's called Taekwondo. I started doing Taekwondo in July. It has done so many things for me. Physically, I've been doing very well. It helps me to be more mindful about my body. Master Lee helped me align my spine, I learned the pressure points of my body, and he helped me walk better. I take stretching very seriously. I never had heel-toe movement in my ankles before Taekwondo. I never had sensation in the back of my calves. I am building muscle. I never really had muscle control in my legs, but now I do, which was a great achievement and it's really awesome.

How I got going with Taekwondo in the pandemic is: we went to four specialists to see about a tendon repositioning procedure in my legs, but they said no because of the dystonia. We decided to go to Taekwondo and see if it could help. We found out that Master Lee's brother has CP, and he helped his brother with rehab when he was young. Every week his understanding about my body gets better. He's amazing in his knowledge, and I work very hard. I have been going three days a week. Jumping was another thing that I started doing. I learned how to jump during the pandemic.

I practice real Taekwondo moves, like kicking, blocking, and punching, about one day a week, and the other days are focused on learning how to place my body in space and keep my balance. Trying to rehabilitate my brain to

Cerebral palsy (CP) is a group of disorders that affect a person's ability to move and maintain balance and posture. CP is caused by changes in brain development or damage to the developing brain that affect a person's ability to control their muscles. CP is the leading cause of dystonia in children.

Complementary & Integrative Therapies

The DMRF encourages individuals with dystonia to consult their doctor about non-traditional therapies they may be interested in pursuing including dietary supplements, products, and mind-body practices. Information about the use of complementary therapies for dystonia is available at: dystonia-foundation.org/complementary-therapy/

take control of my muscles without falling, getting jerking movements, or being startled. It has also helped me emotionally and helps me speak better. I have confidence and motivation. I sleep better. Since I started Taekwondo I don't get these awful cramps in my legs and my toes. My toes used to fold underneath when I walked. Now they are flat and I can control them and wiggle them and paint my toenails. I can keep the polish on the nails because I don't walk on my toes anymore. At first I even struggled with taking off my uniform. I had to do it over and over again until I could get it off finally, but now I don't struggle with it.

What therapies or interventions have helped you?

I used to take medicines, baclofen and Artane® [trihexyphenidyl]. Artane® helped me get balance and stop my jerky movements, but there were side effects. Now the only thing I take is CBD oil [cannabidiol] and



Amber Hall studies Taekwondo with Grandmaster Chu Young Lee. Taekwondo is an ancient Korean martial art.

some teas, naturopathic things that maybe relax muscles. I used a walker until I was six years old. Then I started using the walker like a skateboard, and they took it away. I love skateboards. I wish I could ride one. My physical therapist was very hands-on and she would strap my legs to her legs and we'd ride the skateboard down the street together and go to the park. I wore AFOs with SMO inserts, and twister cables to keep my knees apart. And some nighttime bracing and casting and Botox® and so many things throughout the years. And then conductive education—summer camp for CP and other people who are struggling with mobility. I had an occupational therapist who helped me a lot with my feelings and thinking skills and executive functioning skills.

What impact has CP and dystonia had on your life?

I have had CP and dystonia for my whole life. It affected me more as I became more aware about myself. It also affected

me emotionally and mentally. When I was like nine or eight, I knew I had a disability but I felt I was still like every other kid. As I got older, like middle school and high school, I became more aware of how other people felt around me because I had CP. It's 50/50. Either they bully you or they take it seriously and try to be nice to you. I had very challenging problems with bullies in high school. That's when I started focusing on things that help me cope through these problems. I had arts, music, family, and my mom—she's the best. My mom took me to private school junior year. It's my second year there. Before the pandemic hit, I was independent for the first time. It was amazing. I met a lot of kids there. I have a troubled time trusting people. I made one or two friends. I'm fine with that. Private school was amazing. It made me feel like I'm an average high schooler.

I was mainstreamed with a para [classroom assistant] in sixth grade. Before that I was in the deaf and hard-of-hearing classes. My para was awesome in middle school. But at the same time I was struggling because I wanted to be free. I didn't want anybody helping me. Now I am independent, and I'm happy.

I want to become a digital artist and a comic book illustrator. I like doing portraits and cartoons. I love movie posters, especially old ones. One of my favorite all-time movies is *Halloween*, the classic 1978 one. I also like *Friday the 13th* from the 1980s. As a kid I loved *Looney Tunes* and *The Three Stooges*. I also take cartoon shows seriously. Especially from around the late 1990s and the 2000s. I get inspiration from them, like *Invader Zim* and *SpongeBob SquarePants*. I love my mom's art books. My art is a combination of things. It's either dark humor or comedy, like slapstick comedy. Sometimes it's venting my feelings. It helps me cope with anxiety. I also want to become an animator in movies.

What are you most proud of?

I'm proud of what I am doing with Taekwondo: balancing, walking, thinking, speaking, and having conversations with people, trying to be very aware. I'm proud I'm alive. I'm proud that I am independent and strong and determined. Focus is the number one thing. Number two is determination. The greatest challenge I overcame was bullying. Always speak up for yourself and be an advocate for yourself, and you can help others too. Be kind.

Dystonia Dialogue

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



Non-profit
Organization
U.S. Postage
PAID
Random Lake, WI
Permit No. 399

Renew Your DMRF Membership Today

As a member of the DMRF, you will continue to receive the latest news and information.

Renew at dystonia-foundation.org/membership or use the enclosed envelope.



**DMRF earned the HIGHEST RATINGS from
CHARITY NAVIGATOR and GUIDESTAR.**

***Give with confidence, knowing your contributions
are used effectively and responsibly.***