Dystonia is a Journey

Dystonia MEDICAL RESEARCH FOUNDATION
2018 ANNUAL REPORT
Dystonia is more than a diagnosis in a medical record. It is a life-changing journey, not only for those affected but for the entire family. Your support of the Dystonia Medical Research Foundation (DMRF) ensures that no one has to face this journey alone. For nearly 45 years, DMRF has supported people through the dystonia experience while advocating on their behalf and stimulating medical research to improve lives.

The DMRF’s mission to cure dystonia is also a journey. The organization has evolved dramatically since its founding in 1976. The promise to find a cure has not wavered, and we continue to transcend the many obstacles between our vision and the cure, including limitations in funding and the inherent unpredictability of science. The remarkable expansion of dystonia research and flow of medical advancements prove our approach is working, and we continue to be inspired by the constant progress.

We suffered a difficult loss in 2018 with the death of Co-Founder and Chairman Samuel Belzberg. His wise guidance and tenacious enthusiasm for pursuing our mission are sorely missed. Yet his legacy continues. The strength of the DMRF and the undeniable impact the Foundation has had on dystonia research and patient resources is a testament to Sam’s vision and the unconditional commitment he and Frances Belzberg made to the dystonia community. We are grateful Fran agreed to step into the role of Honorary Chair, and the Board of Directors is as determined as ever to fulfill the promise of a cure.

Our progress in 2018 simply would not be possible without your support. Every accomplishment highlighted in this report was possible because of you. Thank you for being a part of the DMRF’s global effort to find a cure.

Sincerely,

Art Kessler
President

Janet L. Hiesemer
Executive Director

Sam and Fran Belzberg founded the DMRF in 1976. The Foundation has since transformed the field of dystonia research and the resources available to affected individuals and families.
The mission of the Dystonia Medical Research Foundation (DMRF) is to advance research for more effective treatments and ultimately a cure, to promote awareness and education, and to support the well-being of affected individuals and families.

Dystonia is a neurological disorder that causes excessive, involuntary muscle contractions. These involuntary contractions force the body and limbs into abnormal movements and awkward postures. Dystonia may affect a single body area or be generalized throughout multiple muscle groups. Dystonia affects men, women, and children of all ages and backgrounds. Estimates suggest no fewer than 250,000 people in the United States are affected. Dystonia causes varying degrees of disability and pain, from mild to severe. Although treatments exist, dystonia remains a chronic disorder for which there is not yet a cure.

Each of the 20+ members of the DMRF Board of Directors is personally affected by dystonia, either diagnosed themselves or a loved one is diagnosed.

Sam’s impact on dystonia research and those affected by it was immeasurable. He literally kick-started the field of dystonia research. Through his determination to unravel this mystery, he inspired countless others to tackle it. Like so many of us, Sam never accepted defeat to dystonia. Our dystonia family lost a great crusader, but his legacy will live on and we won’t let him down.

—Art Kessler, DMRF President, on the passing of Co-Founder & Chairman Samuel Belzberg
Neuroanatomical Substrates for Disrupted eIF2alpha Signaling in Dystonia
Nicole Calakos, MD, PhD
Duke University (USA)

Myoclonus-Dystonia, a Study of Motor and Non-Motor Symptoms: Is there a Role for Serotonin?
Marina A.J. de Koning-Tijssen, MD, PhD
University of Groningen (The Netherlands)
Supported by the Brown Family Foundation

Machine Learning Guided Deep Brain Stimulation to Cure Neurological Disease
Jesse Goldberg, MD, PhD
Cornell University (USA)
Supported by the Dorothy Feiss Scientific and Medical Research Fund

Striatal Neuron Activity Patterns in Dystonia
Ellen Hess, PhD
Emory University (USA)

Tremor, Oscillations, Synaptic Plasticity, and DBS for Dystonia
William Hutchison, PhD
Toronto Western Hospital (Canada)
Supported by Dystonia Medical Research Foundation Canada

Determining the Role of Torsin in Nuclear Pore Complex Assembly
Patrick Lusk, PhD, Yale University (USA)
Supported in part by Dystonia Medical Research Foundation Canada

Synaptic Plasticity in a Mouse Model of Paroxysmal Dystonia
Alexandra Nelson, MD, PhD
University of California, San Francisco (USA)

An International Study to Investigate the Non-Motor and Psychological Impact of Myoclonus-Dystonia
Kathryn Peall, MD
Cardiff University (United Kingdom)
Supported by the Brown Family Foundation
Dystonia is one of the network disorders, and I’m trying to understand how different nodes of the network talk to each other and how we can modulate the network in a counter-intuitive way. If something is broken, we don’t focus on what is broken. We make up for what is broken with something that is working.

—Aasef Shaikh, MD, PhD, DMRF Grant Recipient & Past Clinical Fellow
4 YOUNG DYSTONIA INVESTIGATORS RECEIVED RESEARCH FELLOWSHIPS.

Dystonia research requires a constant flow of new people and new ideas. DMRF provides funding opportunities for young investigators, at strategic stages in a research career, designed to help foster the next generation of dystonia innovators.

Barbara Oliver Memorial Dystonia Research Award
Using the Nematode Caenorhabditis elegans to Identify Candidate Substrates for OOC-5/TorsinA
Maria Daniela Cirnaru, PhD
University of Minnesota
Mentor: Michelle Ehrlich, MD

CRISPR/Cas9 System Targeting Specific DYT1 Allele Mutation in Patient Induced Pluripotent Stem Cells (iPSCs): A Strategy for Phenotype Reversion in iPSCs-Derived Neurons
Lilian Cruz, PhD
Massachusetts General Hospital (USA)
Mentors: Xandra Breakefield, PhD & Cris Bragg, PhD

Role of the X-linked Dystonia Parkinsonism (XDP) Causative Gene TAF1 in the Striatal Development and Maintenance
Anthony Rampello, PhD, Yale University
Mentor: Christian Schlieker, PhD
Supported by the family of Barbara and Ron Oliver.

A Genetic Approach towards Identifying Torsin Function in Relation to DYT1 Dystonia
Gabriela Huelgas-Morales, PhD
University of Minnesota
Mentor: David Greenstein, PhD

Mahlon DeLong Young Investigator Award recipient Aloysius Domingo, MD was invited to present at the annual Medical & Scientific Advisory Council meeting. Pictured with MSAC Member Kristina Simonyan, MD, PhD, Dr. med.
DMRF LEADERSHIP MET WITH NINDS TO IDENTIFY OPPORTUNITIES FOR DYSTONIA RESEARCH.

“Defining Emergent Opportunities in Dystonia Research” was the title of a meeting organized by the National Institute of Neurological Disorders & Stroke (NINDS), October 29–30, 2018, in Bethesda, Maryland. The meeting convened world renowned dystonia experts and brought increased visibility of dystonia to a critical federal medical research agency. Director of NINDS, Walter J. Koroshetz, MD, opened the meeting which was co-chaired by Laurie Ozelius, PhD of Harvard Medical School and David G. Standaert, MD, PhD of University of Alabama at Birmingham. DMRF Chief Scientific Advisor Jan Teller, MA, PhD was among the presenters.

The program agenda included a detailed review of advancements in dystonia research and discussion of future research priorities.

A NEW SECTION OF THE DMRF WEBSITE CENTRALIZED RESOURCES FOR INVESTIGATORS.

The DMRF’s support of dystonia investigators is beyond financial. The DMRF also prioritizes curating tools and resources to help dystonia investigators do their important work. A new section on the DMRF website added in 2018 includes links to information about animal models, cell lines, biosamples, and additional resources.
Evaluation by a movement disorder specialist can have a dramatic impact on quickening dystonia diagnosis time, increasing treatment options, and maximizing outcomes from treatment. Many patients throughout the country must travel significant distance and/or endure long waits for appointment openings to access a physician with the necessary qualifications to diagnose and treat dystonia. In response to the need for additional clinical experts, the DMRF sponsors one-year clinical fellowships to train second-year fellow physicians in the diagnosis and treatment of movement disorders with special competence in dystonia. The outstanding clinicians who complete training have joined movement disorder programs at prestigious institutions and begun practicing in communities previously without a dystonia expert.

The Clinical Fellowship Program was funded by grants from the Allergan Foundation and Merz Pharmaceuticals.

5 CLINICAL FELLOWS RECEIVED EXPERT TRAINING TO QUICKEN DYSTONIA DIAGNOSIS AND TREATMENT.

CLINICAL FELLOWS AND MENTORS ACTIVE IN 2018 INCLUDED:

  Mentor: Nicole Calakos, MD, PhD
  Research: Using transcranial magnetic stimulation (TMS) in patients with writer’s cramp (focal hand dystonia) to reduce dystonic movements.

- Avram Fraint, MD – Rush University Medical Center (2017–2018)
  Mentor: Cynthia Comella, MD

- Christopher Groth, MD – University of Colorado Denver (2017–2018)
  Mentor: Brian Berman, MD, MS
  Research: Exploring possible correlation between reduced GABA receptor availability in global pallidus and susceptibility to developing cervical dystonia.

  Mentor: Mark Hallett, MD
  Research: Investigating frequency of anxiety and depression in dopa-responsive dystonia.

- Lindsey Neimand – Icahn School of Medicine at Mount Sinai (2017–2018)
  Mentor: Rachel Saunders-Pullman, MD, MPH, MS
  Research: Exploring differences in the pathophysiology of dystonia associated with complex regional pain syndrome compared to organic focal hand dystonia.
INVESTIGATORS HAD ACCESS TO DATA AND SAMPLES FOR NEW STUDIES.

The Global Dystonia Registry is a community-based research initiative that depends on the DMRF for operational and administrative management. The Global Dystonia Registry supports future dystonia studies, including clinical and research trials, through the voluntary collection of patient data. To date, more than 5,300 individuals have joined.

The Dystonia Brain Collective is another DMRF-led collaborative effort among participating dystonia organizations. The purpose is to promote brain donation registration in support of research and provide investigators with access to tissue samples. Three brains were successfully recovered in 2018. DMRF is deeply grateful to the donors’ families for facilitating these invaluable contributions to research.

“I was fortunate to find a movement disorder specialist through DMRF. Thank you so much. If it wasn’t for the Foundation I would be stuck using a pain clinic with very little knowledge of dystonia.

—Jason Padden, DMRF Supporter

Drs. Christian Schlieker, David Peterson, and Roy Sillitoe were among the guest presenters at the 2018 Medical & Scientific Advisory Council meeting.
EXPERTS FROM ACROSS THE GLOBE GATHERED FOR SCIENTIFIC MEETINGS.

With support from the Brown Family Foundation, the 4th DMRF Myoclonus-Dystonia Workshop took place September 27–28, 2018 in Paris. The meeting was chaired by Marina de Koning-Tijssen, MD, PhD of University Medical Center Groningen (The Netherlands) and Marie Vidailhet, MD, PhD of Salpêtrière Hospital (France). An international cohort of experts presented the latest myoclonus-dystonia research and brainstormed next steps to accelerate research progress.

DMRF organized a workshop entitled “Targeted Drug Discovery for Dystonia,” November 8-9, 2019 in Chicago. Meeting participants included seasoned dystonia investigators and non-dystonia experts experienced in protein-based drug discovery. The workshop was co-chaired by Christian Schlieker, PhD of Yale School of Medicine and Thomas Schwartz, PhD of Massachusetts Institute of Technology. The goal of the meeting was to summarize and discuss recent efforts to identify drug targets for dystonia.

The Joan Miller Young Investigator Scholarship Award supported up-and-coming dystonia researchers by providing travel stipends to attend DMRF workshops and meetings. Participation in these workshops and meetings is particularly important for young researchers because they provide opportunities to present their work to expert audiences and develop useful connections with thought-leaders in the field.

“Research grants awarded by the Foundation along with its dedication to raising public awareness make me truly hopeful that finding a cure for all forms of dystonia is possible.
—Ed Gewirtz, DMRF Supporter”
THE DYSTONIA COALITION RECEIVED
FREE STAFF AND ADMINISTRATIVE SUPPORT.

The Dystonia Coalition is a collaboration of medical researchers and patient advocacy groups supported by the Office of Rare Diseases Research in the National Center for Advancing Translational Sciences and The National Institute of Neurological Disorders & Stroke (NINDS) at the National Institutes of Health (NIH). The mission is to advance the pace of clinical and translational research in the dystonias to find better treatments and a cure. The objectives are to develop a fuller understanding of the many features of dystonia and how they change over time, to develop validated diagnostic strategies and rating tools for diagnosis and monitoring patients in clinical trials, to establish a biorepository where blood and other samples can be stored and distributed for research, to stimulate clinical trials for new treatments, and to promote education and awareness. The DMRF plays an integral role by providing free logistical and planning support for the Coalition as an administrative center, for example managing payments to clinical sites and supporting the renewal grant to the NIH. This has saved the Dystonia Coalition $1.5 million in indirect costs, maximizing funds invested in research.
THE DYSTONIA ADVOCACY NETWORK MADE 150 VISITS TO MEMBERS OF CONGRESS TO EDUCATE THEM ABOUT DYSTONIA.

On March 19–20, 2018, the Dystonia Advocacy Network (DAN) hosted Advocacy Day in Washington, DC to encourage Members of Congress to support the DAN’s legislative priorities. Advocates asked that dystonia continue to be recognized as a condition eligible for study through the Department of Defense Congressionally Directed Medical Research Programs and to provide the National Institutes of Health with ample funds for the 2019 fiscal year. The DAN also collaborated with like-minded organizations to ensure pre-existing condition protections are not eliminated from federal healthcare policies. The DMRF continued to provide staff support in 2018 to ensure the DAN implemented and achieved the legislative agenda.

The Douglas Kram er Young Advocate Award recognizes young people who are giving voice to dystonia through advocacy. Recipients in 2018 were Rachael Mathews, Sophia Taylor, and Melissa Rentfrow. Special thanks to the Kramer Family for generously supporting this award.

"The time I spent with the Dystonia Advocacy Network was a tremendous turning point for me. Being with other people who had been through similar experiences made me feel like I wasn’t alone anymore.”
—Mike Cahall, DMRF Supporter & DAN Advocate
DYSTONIA WAS INCLUDED IN THE DOD CONGRESSIONALLY DIRECTED MEDICAL RESEARCH PROGRAMS.

Since 2010, the Department of Defense (DOD) has funded dystonia research because volunteer advocates push every year to make sure dystonia is included in its Congressionally Directed Medical Research Programs (CDMRP). Unlike the National Institutes of Health and other federal medical research initiatives, the CDMRP only funds research into conditions that are deemed eligible for study by Congress. The eligible conditions list is renewed by Congress every year. The hard work of dystonia advocates on this issue has resulted in $14 million from the DOD awarded to outstanding dystonia investigators. The DOD integrates patients and family members into the CDMRP scientific review process by assigning consumer reviewers to participate alongside research experts. The DMRF is invited each year to nominate members of the dystonia community for these important positions.

DAN advocates presented Senator Susan Collins with a Distinguished Public Service Award to recognize her support of the dystonia community.
TEAM DMRF RAISED AWARENESS AT THE TCS NYC MARATHON.

In honor of those struggling with dystonia, Team DMRF has competed in the world famous TCS New York City Marathon since 2013, literally running for those who can’t. In 2018, Team DMRF included Amy Amendola, Heather Barskaya, Ginny Bryan, Jaime Dimitri, Larry Dubill, Stephen Gebeloff, Jim Metherell, Marissa Rozenfeld, Carrie Siu Butt, and Carole Tordi. Dozens of supporters gathered on the sidelines to cheer them on.

"I would never run for any organization except for the DMRF. The research they fund changes lives, including mine and my sister’s. This organization changed my life!"

Carrie Siu Butt, Team DMRF, TCS NYC Marathon
Em poweri ng patie nts with info rm at i on & support

50,000+
PEOPLE RECEIVED INFORMATION & RESOURCES.

The DMRF is here for you when dystonia turns life upside down. Every member of the DMRF’s Board of Directors is navigating the dystonia journey, either themselves or beside a loved one. We are reminded every day of the urgency of our work because we see and feel the impact of dystonia in our own lives. We are all on this journey together.

50+ SUPPORT GROUPS AND MODERATED ONLINE FORUMS REDUCED ISOLATION AND INCREASED SOCIAL CONNECTION.

Feelings of isolation are common among individuals and families impacted by dystonia, and can be devastating to physical and emotional health. Social connection and peer support are vital to living well with dystonia. DMRF support groups provide local peer support, offering the opportunity to gather with others who know the challenges of living with dystonia. Online forums provide connection to thousands around the country, and around the world. The DMRF volunteer network is also made up of individuals devoted to awareness, fundraising, and/or serving as area contacts.

"Support groups provide a space for those of us with dystonia to share our stories and struggles in a safe and accepting environment. We feel less alone and more understood. We become better informed about our treatment choices and walk away with useful information to support us in our journey with dystonia.”

—Alli Feeley, Founder & Leader of Greater Chicagoland Dystonia Support Group
The Central Indiana Dystonia Support Group held its first meeting in 2018.
DMRF PROVIDED THOUSANDS OF FREE EDUCATIONAL MATERIALS TO PEOPLE SEEKING INFORMATION, RESOURCES, AND REASSURANCE.

Access to accurate information about dystonia and treatment options is as vital to living well as access to appropriate medical care. Sharing a brochure about dystonia can be a simple but powerful act to inform family members, educators, colleagues, and even strangers about the disorder and promote greater awareness. More than 25 brochures and pamphlets are available for free mail order or download from the DMRF website. These materials are also distributed through local dystonia support groups, educational meetings, awareness events, and in medical offices.

8 EDUCATIONAL MEETINGS PROVIDED THE LATEST INFORMATION FROM TOP EXPERTS.

Hurried medical appointments in which physicians have limited time with patients are not a conducive space for in-depth learning about dystonia and the latest clinical advancements. DMRF creates opportunities for individuals to interact with expert clinicians and researchers in an educational setting, with ample time for interactive discussion. DMRF provided thousands of people with the latest information on living well with dystonia by organizing free educational meeting across the country. These meetings also provide opportunities for peer support and social connection. Locations included: Albuquerque, Boston, Chicago, Cleveland, Gig Harbor (Washington), Los Angeles, Phoenix, and St Louis. Allergan provided a generous educational grant in support of the meetings.
20+ community events

PATIENTS, FAMILIES, AND HEALTHCARE INSTITUTIONS UNITED IN THE MISSION TO FIND A CURE.

The DMRF partners with volunteers and support groups around the country on creative and fun events that unite the local dystonia community, promote awareness, highlight movement disorder programs, and raise urgently needed funds for research.

Among DMRF’s most popular community programs are Dystonia Zoo Walks, family-friendly events to raise awareness and support DMRF. Thirteen Zoo Walks attracted more than 5,000 participants in 2018. Locations included: Bronx/New York City, Detroit, Cleveland, Cincinnati, Fresno, Philadelphia, Phoenix, Pittsburgh, Portland (Oregon), Providence (Rhode Island), St Louis, San Diego, and Twin Cities.

(L to R) Allison Hersh London and June Hersh led “Ali’s Zoo-Gooders” at the Bronx Zoo Walk. US Congressman Dwight Evans is pictured with organizers of the Philadelphia Zoo Walk. Team BillyMac participated at the Twin Cities Zoo Walk. Scotty Tritley made a new friend in St. Louis. The Young Family traveled to Cleveland to support Karen Flanagan and her event. Ed Cwalinski and MaryRae Nee were recognized at the Pittsburgh Zoo Walk. Mary Stone lead team “Rolling Stones” in Phoenix. The Baron Family is pictured at the Providence Zoo Walk.
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(1928–2018) Co-Founder  
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Barbara Kessler, Chair  
Paul Kavanaugh  
Diane Rudolph  
Elizabeth Schultz  
Ed Cwalinski  
Dee Linde  
Paula Schneider  
Todd Spotti

DMRF distributed 110,000+ copies of the Dystonia Dialogue newsletter.
The DMRF has been a wonderful community of families who have walked the path. We followed those who went before us, and now there are people following us. We pay it forward through the DMRF.
—Diane Rudolph, DMRF Board of Directors

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Federal Liaison

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Ginny Bryan
Karen A. Flanagan
Melissa Phelps
Beth S. Farber
James A. Metherell
Rosemary Young
## Statement of Activities

**Dystonia Medical Research Foundation | Year Ending December 31, 2018**

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<th>Without Donor Restrictions</th>
<th>With Donor Restrictions</th>
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| **Expenses**             |                             |                          |            |
| Program services expense |                             |                          |            |
| Science                  | 1,534,744                   |                          | 1,534,744  |
| Awareness and education  | 419,167                     |                          | 419,167    |
| Membership and support   | 190,386                     |                          | 190,386    |
| Advocacy                 | 122,586                     |                          | 122,586    |
| **Total program services expense** | 2,266,883                   |                          | 2,266,883  |
| Supporting services expense |                             |                          |            |
| Administration           | 174,785                     |                          | 174,785    |
| Fundraising              | 299,679                     |                          | 299,679    |
| **Total Expenses**       | 2,741,347                   |                          | 2,741,347  |
| Change in Net Assets     | (601,031)                   | 57,371                   | (543,660)  |
| Net assets, Beginning of Year | 2,555,579                  | 1,662,807                | 4,218,386  |
| **Net Assets, End of Year** | $1,954,548                  | $1,720,178               | $3,674,726 |

A complete copy of financial statements audited by Barnes, Givens & Barnes, Ltd. is available upon request from the Dystonia Medical Research Foundation, One E. Wacker Drive, Suite 1730, Chicago, Illinois 60601.
Statement of Financial Position

DYSTONIA MEDICAL RESEARCH FOUNDATION | YEAR ENDING DECEMBER 31, 2018

LIABILITIES

Grants and fellowships payable $ 558,947
Accounts payable and accrued expenses 115,691
Deferred rent payable 114,519
Dystonia Coalition agency liability funds 198,578
Total Liabilities $ 987,735

Net assets without donor restrictions:
Undesignated 454,548
Board designated for science 1,500,000
Total net assets without donor restrictions 1,954,548

Net assets with donor restrictions:
Purpose restricted 1,720,178
Total net assets with donor restrictions: 1,720,178
Total Net Assets 3,674,726

Total Liabilities and Net Assets $ 4,662,461
In memoriam

THE DMRF WISHES TO ACKNOWLEDGE THE GENEROUS GIFTS RECEIVED IN MEMORY OF THE FOLLOWING:

Raymond Adolfson
Wanda Ahlquist
Emma and Donald Ajlony
Louis Amato, Sr.
Robert Amerio
John F. Anderson
Suzanne Arnold
Harriet Ball
Basya Rochel Bas
Shmuel Hirsh obm
Eileen Bean
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Shari Farber-Tritt
Tommy Ford
Pat Frost
Irene Galassi
Shirley and Harry Goldberg
Dr. George Goldman
Howard Golub
Robert Goodman
John Jessee Gossett
Joyce Greenberg
Jerry Groner
Alfred Gruman
Robert Haid
Ellen Hansen
Georgia Harrington
Henrichson
Bernard Harris
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Connie Walter
Charlotte Weaver
Susan Pitts (Weiss)
Ida Larine Williams
James L. Williams
George Winston Gilpin
Allan Womac
Joann Wyeth
Thank you 2018 donors

$50,000 or More
Allergan Corporation
Allergan Foundation
Frances and Samuel Belzberg
Wendy Bloom and Arthur Kessler
Brown Family Foundation
Cure Dystonia Now
Dystonia Medical Research Foundation Canada
Dystonia Study Group/Rush University Medical Center
Family Alliance Foundation
June and Ron Hersh
Barbara and Dennis Kessler
Rosalie and Richard Lewis, MD
Merz North America, Inc.
Janet Nast
Ronald Oliver/Barbara Oliver Memorial Research Fund
C. Northrop and A. Marder Pond Foundation
Pamela Sloate/Bronx Dystonia Zoo Walk
Team Ali’s ZooGoods
Bronx Dystonia Zoo Walk
Sandra Weil

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