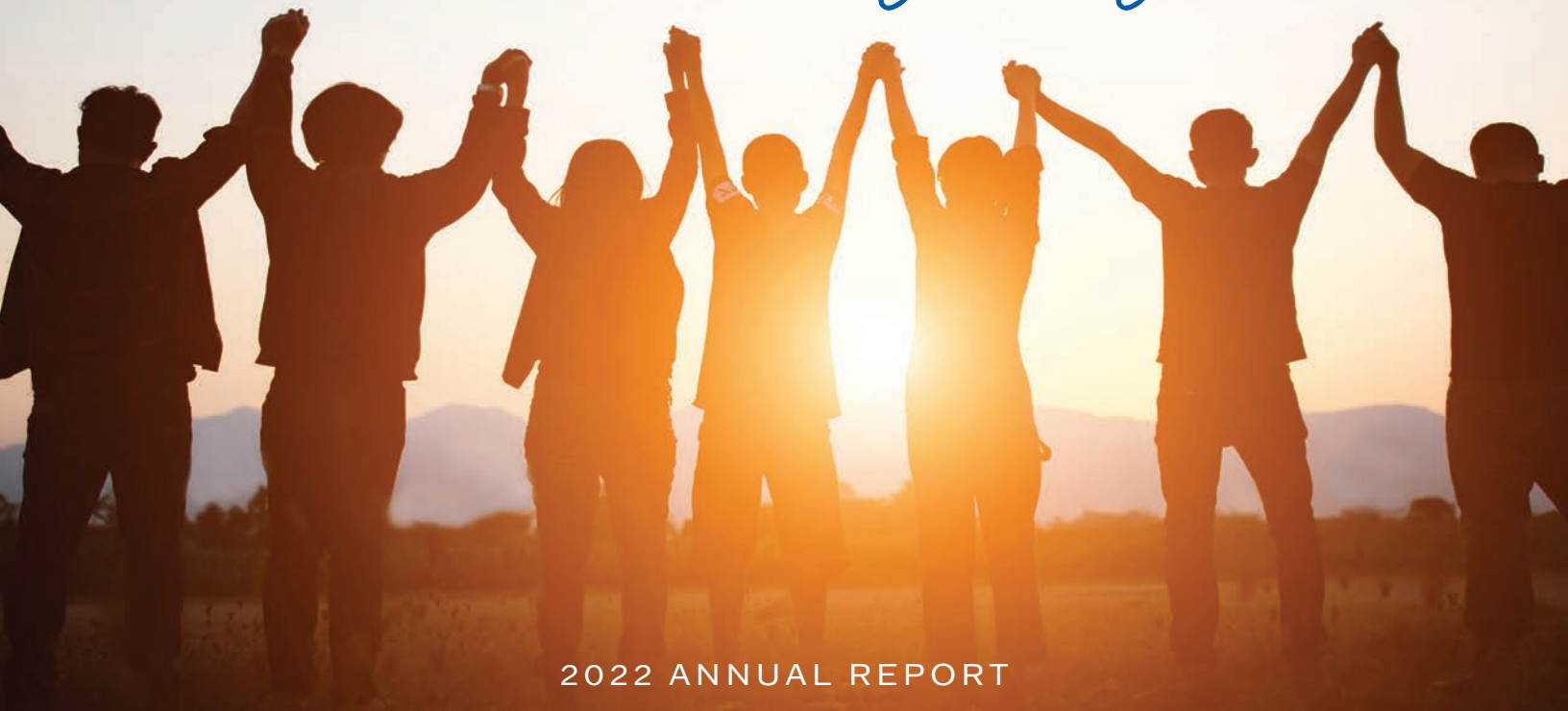




Stronger Together



2022 ANNUAL REPORT

Message From President and Executive Director



MARK RUDOLPH
President

In 2022, the Dystonia Medical Research Foundation (DMRF) experienced a year of transition and reflection.

Mark Rudolph was elected president of the DMRF, taking the reins from Art Kessler, who had been president of the foundation since 2010. "I'm honored to lead this organization," Mark said. "Together with the DMRF Board of Directors, we carry on the DMRF's commitment to solving the mysteries of dystonia, helping everyone impacted by the disorder, and ultimately finding a cure."



JANET L. HIESHETTER
Executive Director

The DMRF is grateful to Art for his many years of dedication and leadership. Under Art's direction, the DMRF's outreach programs grew significantly with the launch of Dystonia Zoo Day events, additional support groups, community education meetings, and regular use of webinars to provide helpful and accurate information to those in need.

Reconnecting in 2022 after enduring the seclusion of the pandemic was a welcome transition back to 'normal.' It reminded us of the strength that comes from being together and supporting one another through personal contact. While the DMRF never stopped connecting our community, we all appreciated being together again. Dystonia Zoo Day events were back in-person in eight different cities, support groups began to meet face-to-face again, and researchers were fully and gladly back in laboratories.

Using the platforms that helped us connect during the pandemic, the DMRF continues to expand our reach to the community. Our webinars and virtual programs kept our community informed and helped battle the isolation people affected by dystonia so often feel. The Dystonia Dialogue, in its 44th year of publication in 2022, continues to be a "lifeline" to many by providing the latest dystonia news, research updates and messages of hope.

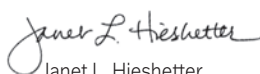
The DMRF has long recognized the mental health needs of those living with dystonia. This year, we formalized our efforts even further by forming the Mental Health Programming Committee to help identify and meet the non-motor symptom needs of the disorder.

We couldn't have done any of this without you. Thank you for all you have done to help the DMRF provide critically important support, information, and hope for families this past year. You organized events, rallied your communities, shared your dystonia experiences, and donated funds for the DMRF's many programs.

We are **STRONGER TOGETHER** because of you.



Mark Rudolph
President



Janet L. Hieshetter
Executive Director

Mission

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.

About Dystonia

Dystonia is a neurological disorder that causes excessive, involuntary muscle contractions. These muscle contractions force the body and limbs into abnormal movements and postures, making it difficult for individuals to control their movements. Dystonia can affect virtually any region of the body including the face, jaw, eyelids, neck, vocal cords, torso, limbs, hands, and feet. 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.

COLLABORATE



Advocates Collaborate For Dystonia Funding

The DMRF provided staff support to the collaborative efforts of the Dystonia Advocacy Network again in 2022. Joining with the Dysphonia International (formerly NSDA), the Benign Essential Blepharospasm Research Foundation and the National Spasmodic Torticollis Association, the DMRF worked to educate elected officials about dystonia and how it changes the lives of those affected.

In 2022, dystonia advocates met virtually with congressional offices about the needs of the dystonia community. Advocates were once again successful in having dystonia included on the list of eligible conditions through the Department of Defense Peer Review Medical Research Program. This program has been an important funding source for dystonia researchers with \$28,155,678 awarded since dystonia was first included on the program in 2009.

David Peterson, PhD, who received funding through this program said, "The DOD PRMRP is an important source of funding for dystonia research. It has allowed me to do work that I might otherwise not have been able to do, and I am grateful to all of the dystonia advocates who work so hard to create these opportunities."

STRONGER TOGETHER

Celebrating 25 Years of Progress



DMRF Past President Art Kessler
and Drs. Xandra Breakefield and
Laurie Ozelius



DMRF Board Member Liz Rawson
and Dr. Susan Bressman

Among the most significant breakthroughs in dystonia research, 2022 marked the 25th anniversary of the identification of the DYT1/TOR1A gene responsible for early onset torsion dystonia. The DMRF supported the research that resulted in this life-changing discovery allowing for faster diagnosis and the development of treatment plans with better expected outcomes. The DMRF also presented the Changing Lives Through Science Award to Drs. Xandra Breakefield and Laurie Ozelius during the Boston Zoo Day and Dr. Susan Bressman during the Pamela Sloate Bronx Zoo Day, recognizing how this seminal research has improved the lives of many in the dystonia community.

Collaboration Drives Dystonia Research

The DMRF continues to serve as the administrative support for initiatives and programs that are resources for dystonia research.



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 **Global Dystonia Registry** supports future dystonia studies through the confidential collection of patient data and distribution to dystonia investigators. To date, more than 6,500 individuals have joined the registry.



The **Dystonia Brain Collective** promotes brain donation registration to further research and provide investigators with access to critically important brain samples.

DMRF and Cure Dystonia Now Join Forces



Robin and Marc Miller with Cris Bragg, PhD, former advisor to CDN and former DMRF MSAC Member

DMRF and Cure Dystonia Now (CDN) began a new chapter of collaboration as the organizations formally joined together in 2022 to further their mutual goal of supporting research for better treatments and ultimately a cure for dystonia and to support those living with dystonia. CDN Founders Robin and Marc Miller were elected to the DMRF Board of Directors and established the DMRF Cure Dystonia Now Fund to continue their commitment to advancing dystonia research. The CDN Fund supported a special request for applications on noninvasive brain modulation and is supporting a three-year project of Leighton Hinkley, PhD (see page 8) at University of California San Francisco.

“This partnership reduces redundant efforts and gets us closer to what we all want—better treatments and ultimately a cure for dystonia,” Marc Miller said. “CDN has worked with the DMRF for years. But, this year as a partner and board member, I have a much greater appreciation for the breadth of all the work that DMRF does to drive dystonia research and support for the community.”

The Millers hit the ground running early in the year by launching the SB Squares program that created dystonia awareness and raised funds for research around the Super Bowl in February. They hosted their annual Rockin for a Cure event in July, raising additional support for research.

“We are extremely grateful to Robin and Marc for all they have done for dystonia, and we are pleased to have them as members of the DMRF Board of Directors. I look forward to continued progress. We are definitely stronger together,” DMRF President Mark Rudolph added.

“Dystonia” Journal Published 12 Articles in 2022

The DMRF's journal, *Dystonia*, published 12 articles in this first full year of operation, bringing more visibility to the growing field of dystonia and highlighting advancements in science and clinical practice. The scientific journal is the only open-access journal dedicated to dystonia research, serving as the central repository for all dystonia research.

“The number of scientific studies on dystonia has grown immensely, and the time has come to create a central place to house and facilitate access to dystonia research,” said Richard Lewis, MD, DMRF Vice President of Science. “There are many general neurology journals and some disease specific journals, but none that focus exclusively on dystonia. Now we have it.”

Dystonia aims to be the leading journal in the field by publishing premier research on all basic, clinical, and translational aspects of dystonia, providing a centralized publication by and for dystonia investigators. Articles published this year covered a wide range of dystonia types and research fields. Some of the most viewed articles covered treatment of pediatric dystonia, a feasibility trial of tele-yoga to manage cervical dystonia, and tremor in writer's cramp, to name a few.

Dystonia is a resource for researchers, clinicians and patients and is partially supported by the Joan Miller Young Investigator Fund and the Tuft Family Foundation.

Access *Dystonia* and all published research articles from 2022 at: frontierspartnerships.org/journals/dystonia

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Infirmary, Harvard Medical School*

Committed to advancing dystonia research, DMRF supported 18 research grants and 5 research fellowships and completed one research contract in 2022. The DMRF has invested more than \$37 million in grants and fellowships since 1976 in an effort to provide investigators with the funding needed to address the most pressing, unresolved questions in dystonia research.

Grants and Contracts

The research grant program supports the advancement of our understanding of the underlying mechanisms of dystonia, supports the exploration of new areas for drug discovery, and allows investigators to pursue new technologies and how these can be applied to improving the lives of those living with dystonia.

Genetic Modifiers of Penetrance in DYT1 Dystonia

David Arkadir, MD, PhD
Hebrew University of Jerusalem

GABA Abnormalities and Stability in Cervical Dystonia

Brian Berman, MD
Virginia Commonwealth University

Normalizing DYT1 Cholinergic Neurons by CRISPR Disruption of Mutant TOR1A Allele

Xandra O. Breakefield, PhD
Massachusetts General Hospital

Octanol Effect on Symptoms and Network Dysfunction in Myoclonus Dystonia: An Intoxication Free Therapy?

Cecile Gallea, PhD
Pitié-Salpêtrière Hospital

Personalized Functional Neuromodulation of Common Deficits in Focal Dystonias

Leighton Hinkley, PhD
University of California San Francisco

A Study to Identify Kinematic and Force Measures Capturing Impairment in Musician's Dystonia Among String Players and Improvement with Retraining Therapy

Christine Kim, MD, *Yale University*

A Next Generation Sensing Neural Interface Study for Adaptive DBS In Dystonia

Simon Little, MBBS, PhD
University of California San Francisco

Protein Kinase R Dysfunction in Dystonia
Stephanie Moon, PhD, *University of Michigan*

The Role of Cholinergic Neurons in Isolated Focal Cervical Dystonia

Scott Norris, MD, *Washington University*

Role of Cerebellar Network Excitability and Plasticity in the Pathophysiology of Dystonia

Antonio Pisani, MD, PhD
IRCCS Mondino Foundation

Investigating Multimodal Neuroimaging for Probing Brain Networks in Cervical Dystonia

Richard Reilly, PhD
Trinity College Dublin

Targeting the cAMP Pathway in the Striatum to Treat Dystonia

Emmanuel Roze, MD, PhD
Paris Brain Institute

Cholinergic Interneuron Dysfunction in a Phenotypic Mouse Model of Dystonia

Mariangela Scarduzio, PhD
University of Alabama at Birmingham

Cerebellar Repetitive Transcranial Magnetic Stimulation in Monogenic Myoclonus-Dystonia

Anne Weissbach, PhD
University of Lubeck

Deciphering the Sources of Myoclonus in DYT-SGCE Patients

Yulia Worbe, MD, PhD
Pitié-Salpêtrière Hospital

Molecular Mechanisms of Dysmyelination in Dystonia

Dan Yellajoshiyula, PhD
Case Western Reserve University

RESEARCH CONTRACT

Machine Learning Guided Deep Brain Stimulation to Cure Neurological Disease

Jesse Goldberg, MD, PhD, *Cornell University*

DYSTONIA COALITION PILOT PROJECTS PROGRAM AWARDS

The goal of the Pilot Projects Program is to foster the most promising clinical and translational studies of direct relevance to dystonia. Another example of working collaboratively to support more research, the DMRF co-funded these grants with the Dystonia Coalition.

The Effect of Multi-Day Continuous Theta-Burst Stimulation on Symptoms of Cervical Dystonia

Daniel Corp, PhD, *Deakin University*

Moodscreen For Cervical Dystonia: A Diagnostic Accuracy Study of Depression and Anxiety

Davide Martino, MD, PhD, *University of Calgary*

POSTDOCTORAL RESEARCH FELLOWSHIPS

Postdoctoral fellowships support outstanding young scientists who have earned a doctoral degree and embarked on a period of mentored research. DMRF supports postdoctoral fellows who are working to fundamentally improve our understanding of brain dysfunction and molecular mechanisms underlying dystonia.

Beyond Theta: Analyzing Oscillations Across the Frequency Spectrum in Patients with Dystonia Implanted with Sensing-Enabled Pulse Generators

Stephanie Cernera, PhD
University of California San Francisco

Investigating Abnormal Neurodevelopment in a Novel in Vivo Model of Inherited Dystonia

Simon Lowe, PhD
UCL Institute of Neurology

Investigating Sensory Dysfunction in a Mouse Model of Early-Onset, Generalized Dystonia

Amanda Pocratsky, PhD
University College London

Neural Signals in the Cerebellar Nuclei Gate the Manifestation of Dystonia-like Symptoms

Meike van der Heijden, PhD
Baylor College of Medicine

Impact of Microglia on Abnormal Synaptic Plasticity and Development of Dystonia in Transgenic DYT1 Mouse Models

Lisa Rauschenberger, MD, PhD
University Hospital of Würzburg

CLINICAL FELLOWSHIPS

The DMRF supports the training of young physician scientists—both to help them be better in the clinic and to support their important clinical research. The DMRF has supported 33 clinical fellows since the program launched in 2012.

Jessica Frey, MD

Mentor: Aparna Wagle Shukla, MD
University of Florida

John Yu, MD

Mentor: Michael Okun, MD
University of Florida

Providing Critical Connections

DMRF support groups across the country hosted **Dystonia Strong**-themed events to help the community re-connect after the social isolation of the Covid-19 pandemic. Members of the community gathered at homes or parks to meet, provide support for one another, and stand strong together in living with dystonia. DMRF also launched a Dystonia Strong awareness campaign to help promote awareness and build community.

This year, DMRF hosted 119 in-person and virtual support group meetings, providing local peer support and connecting the community with others who understand the challenges of living with dystonia. DMRF support groups play a critical role in battling the isolation that dystonia often brings.

Along with local support groups, DMRF's moderated online forums provide dystonia support and information around the clock to people all over the world. Various DMRF online forum groups are available for specific areas of interest, such as dystonia and deep brain stimulation, parenting with dystonia, and dystonia spouses and loved ones.

CONNECT





DMRF produced and distributed three issues of the *Dystonia Dialogue* to 55,000 people. The DMRF's magazine is considered a 'lifeline' to many by connecting them to the community and keeping them informed of the latest dystonia news and research.



Furthering our reach to the DMRF community, the foundation hosted 8 webinars and participated in multiple

podcasts this year. Our most popular webinar, *How to Get the Most From Your Botulinum Neurotoxin Injections*, drew more than 1,800 registrants.

DMRF Mental Health Programming Committee Gets to Work

DMRF formalized its commitment to addressing mental health in the dystonia community by creating a Mental Health Programming Committee in early 2022 to identify needs in the dystonia community related to the non-motor symptoms of the disorder and to assemble experts and resources to address those needs.

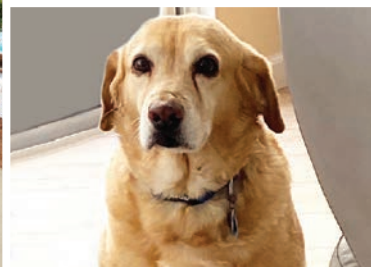
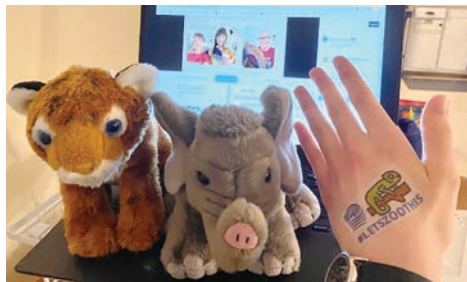
"While the DMRF has long recognized the need for emotional and mental support for individuals and families affected by dystonia, we've been wanting to do a more formalized mental health program for some time," said Karen Ross, PhD, Vice President of Mental Health Programming. "I'm thrilled the DMRF is organizationally supporting this important program need. We're in the process of discovering what the dystonia community's greatest mental health needs are and finding ways to help them however we can as an organization."

To better direct their efforts, the committee developed a web-based survey on mental health and dystonia, which was open to the DMRF community for five weeks in the fall. More than 800 individuals over the age of 18 with a dystonia diagnosis responded to the informal survey. The overwhelming response reinforced the high need for mental health programming, and data will be used to help DMRF develop a range of mental health resources.

DMRF Events Unite Community

The DMRF community came together in person and virtually in 2022 to promote dystonia awareness, raise funds for the foundation... and have some fun. The foundation's network events **raised nearly \$500,000 in support of our mission** to advance dystonia research, support those living with dystonia, and ultimately find a cure. Here's a sampling of some of the many events held in 2022:

- **Dystonia Pet Palooza** brought animal lovers together to raise awareness while celebrating the special role pets have in the lives of those affected by dystonia.
- **The Nashua Distance4Dystonia 5K Walk/Run** drew more than 100 people and raised more than \$5,000 after the event had been paused in recent years due to the pandemic.
- **DMRF's Virtual Zoo Days** kicked off with an online live stream event, and participants shared #letszoothis on social media to spread awareness.
- Friends and family gathered for the **Cooking For a Cure** event held virtually.



Terry Chapman's dog, Sydney, (pictured above right) was the top-earning pet fundraiser for DMRF. Sadly, Sydney died the day Terry registered for Pet Palooza, and all gifts received were made in her memory.



Dystonia Zoo Days Resume In-Person Gatherings



Dystonia Zoo Days were back in-person in 2023 and, along with Virtual Dystonia Zoo Days events, brought together members of the dystonia community from all 50 states to raise awareness and funds for research. After a two-year hiatus during the pandemic, eight in-person events were held at zoos in select cities across the U.S.

"It was so uplifting to finally be back together again with my DMRF friends and meet face to face to support the dystonia community," said Paula Schneider, DMRF Board Member and organizer of the Boston event.

Virtual celebrations kicked off and closed Zoo Day season with appearances by Lolly Lardpop and Leslie Carrera Rudolph, other special guests, a zookeeper chat, inspiring stories, and research Q&A. Continuing to host the Virtual Zoo Day provides the opportunity for people from coast to coast to come together on the same day at the same time to support one another and create awareness.



GRATITUDE

Statement of Activities

Dystonia Medical Research Foundation | Year Ending December 31, 2022

	WITHOUT DONOR RESTRICTIONS	WITH DONOR RESTRICTIONS	TOTAL
REVENUES, GAINS & SUPPORT			
Donations and grants	\$ 1,585,532	\$ 302,468	\$ 1,888,000
Bequests	111,583		111,583
Special events, net of direct benefits to donors of \$65,821	428,416		428,416
Investment income	(609,969)	-	(609,969)
Other	44,707		44,707
Net assets released from restrictions	384,375	(384,375)	-
Total Revenues, Gains and Support	<u>1,944,644</u>	<u>(81,907)</u>	<u>1,862,737</u>
EXPENSES			
Program services -			
Science	1,346,775		1,346,775
Awareness and education	457,154		457,154
Membership and support	146,924		146,924
Advocacy	76,067		76,067
Total program services expense	<u>2,026,920</u>		<u>2,026,920</u>
Supporting services -			
Administration	235,304		235,304
Fundraising	244,606		244,606
Total Expenses	<u>2,506,830</u>		<u>2,506,830</u>
Change in Net Assets	(562,186)	(81,907)	(644,093)
Net assets, Beginning of Year	<u>3,204,477</u>	<u>2,235,769</u>	<u>5,440,246</u>
Net Assets, End of Year	<u>\$ 2,642,291</u>	<u>\$ 2,153,862</u>	<u>\$ 4,796,153</u>

Statement of Financial Position

Dystonia Medical Research Foundation | Year Ending December 31, 2022

CURRENT ASSETS

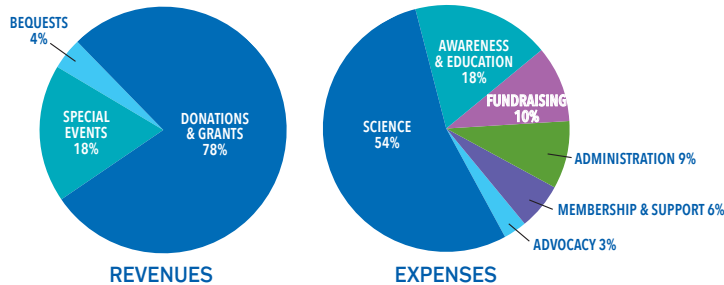
	2022
Cash and cash equivalents including segregated	
Dystonia Coalition cash of \$378,854 in 2022	\$ 657,300
Unconditional promises to give	73,439
Other current assets	57,191
Finance lease right-of-use asset, current	4,721
Operating lease right-of-use asset, current	41,719
Total Current Assets	<u>834,370</u>

NONCURRENT ASSETS

Investments	5,195,634
Equipment and leasehold improvements,	
less \$32,817 of accumulated depreciation	9,322
Finance lease right-of-use asset, noncurrent	8,575
Operating lease right-of-use asset, noncurrent	208,137
Total Noncurrent Assets	<u>5,421,668</u>

Total Assets

\$ 6,256,038



CURRENT LIABILITIES

	2022
Grants and fellowships payable	\$ 527,975
Accounts payable and accrued expenses	74,578
Deferred revenue	168,573
Dystonia Coalition agency liability funds	378,855
Financing lease liability, current	4,721
Operating lease liability, current	47,095
Total Current Liabilities	<u>1,201,797</u>

NONCURRENT LIABILITIES

Financing lease liability, noncurrent	8,575
Operating lease liability, noncurrent	249,513
Total Noncurrent Liabilities	<u>258,088</u>
Total Liabilities	<u><u>\$ 1,459,885</u></u>

NET ASSETS WITHOUT DONOR RESTRICTIONS

Undesignated	1,142,291
Board designated for science	1,500,000
Total net assets without donor restrictions	<u>2,642,291</u>

NET ASSETS WITH DONOR RESTRICTIONS

Purpose restricted	2,153,862
Total net assets with donor restrictions	<u>2,153,862</u>
Total Net Assets	<u>4,796,153</u>
Total Liabilities and Net Assets	<u><u>\$ 6,256,038</u></u>

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.

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Dystonia Coalition

Sophie Cho, MD
Federal Liaison

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Donna Russow

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Paula Schneider

Virtual Zoo Day Teams 2022

\$25,000 or More

Team AbbVie
Team Ipsen Biopharmaceuticals
Team Merz Therapeutics

\$10,000 to \$24,999

Team Dystonia Muse/Pamela Sloate Bronx
Team Flanagan/Cleveland

\$5,000 to \$9,999

Team Caleb's Crew/Virtual
Team Kristin/Toledo
Team Revance Therapeutics/Virtual
Team Rudolph/Los Angeles

\$2,500 to \$4,999

Team Barrow Neurological Institute/Virtual
Team Braden's Brigade/Pittsburgh
Team Gerald/Cleveland
Team Jester/Los Angeles
Team Medtronic/Twin Cities
Team Mount Sinai/Pamela Sloate Bronx
Team Paige's Peeps/Pittsburgh
Team The E Team/Portland

Team Tina/Portland
Team Twisted Troops/Pittsburgh

\$1,000 to \$2,499

Team A Twist of Ed/Pittsburgh
Team Allegheny Health Network (AHN)/Pittsburgh
Team Bayou Pelicans/Virtual
Team Cedars Sinai/Los Angeles
Team Columbia University/Pamela Sloate Bronx
Team Dee/Portland
Team Destiny's Squad/Boston
Team Dystoniadame's Pride/Virtual
Team Eisel/Toledo
Team Feinstein/Pamela Sloate Bronx
Team Goldman Strong/Boston
Team Hartford Healthcare for Dystonia/
Pamela Sloate Bronx
Team Henry Ford Movement Disorders/Toledo
Team Impact Networking/Virtual
Team Interpretations Dance Academy/Cleveland
Team Ipsen/Boston
Team Liam's Unshakeable Ninjas/Pittsburgh
Team Maddie & JJ/Boston
Team Metro Detroit Dystonia Support Group/Toledo

Team MGB Dystonia Clinic/Boston
Team Movers & Shakers 2022/Twin Cities
Team N.A.Q./Twin Cities
Team Pacific Neuroscience Institute (PNI)
Foundation/Los Angeles
Team RAWRsons/Los Angeles
Team Rawson/Pamela Sloate Bronx
Team Revance Therapeutics/Los Angeles
Team Robin's Nest/Pamela Sloate Bronx
Team San Diego Strong!/Virtual
Team SEP Dystonia Support Group/Virtual
Team The Wild Rumpus!/Virtual
Team Theisenators/Twin Cities
Team University of Pittsburgh Medical Center
(UPMC)/Pittsburgh
Team Val's Crew/Toledo

\$500 to \$999

Team Beth Israel Deaconess Medical Center
(BIDMC)/Boston
Team Cali Walkers/Los Angeles
Team Carson Strong/Toledo
Team Daniel's PoZOOTivity/Virtual
Team East River Energy/Pamela Sloate Bronx

Team Ellen's Warriors/Virtual
 Team Hakuna Matata Sweeney/Virtual
 Team Lafuente/Boston
 Team Marie/Boston
 Team Metro Detroit Support Group/Virtual
 Team MGB Dystonia Clinic/Boston
 Team Montefiore Neurology Dept/
 Pamela Sloate Bronx
 Team Paula S/Boston
 Team Rudolph/Pamela Sloate Bronx
 Team Ryan's Buddies/Virtual
 Team Shaken Not Stirred/Twin Cities
 Team Sullivan/Pamela Sloate Bronx
 Team The Fuzz/Pittsburgh

\$250 to \$499

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