5 Report from Musician’s Dystonia Summit

8 2012 Research Funding

14 Donna Hensley Shaves Head for Research
inside this issue

5  Report from Musician’s Summit
Scientists and Musicians Come Together at First-Ever Event

6  Report from Dystonia Advocacy Day
Volunteers Descend on Capitol Hill

8  2012 Research Funding
Check Out the Investigations the DMRF is Funding this Year

11 The Next Generation of Dystonia Experts
DMRF Announces Clinical Fellowship Recipients

14 Donna Hensley Shaves Head for Research
Teacher Launches Unique Campaign

22 Personal Profiles
Meet Nima Patel, MD and Scottie Roberts

What is Dystonia?
Dystonia is a disorder that affects the nervous system. Improper signaling from the brain causes muscles to contract and twist involuntarily. Dystonia can affect a single body area or multiple muscle groups. There are several forms of dystonia, and dozens of diseases and conditions include dystonia as a significant symptom. For more information visit: http://www.dystonia-foundation.org

On the Cover:
This year, the DMRF and Musicians With Dystonia hosted the first ever Musicians’ Summit which included lectures, discussion sessions, a poster gallery, and a special concert. Ryan Thomson, whose career was interrupted by the diagnosis of focal dystonia, performed with his son Brennish as The Fiddling Thomsons. Read more about this event on page 5.

Partial support of the Dystonia Dialogue is provided by educational grants from Allergan, Inc., The Medtronic Foundation, and Merz Pharmaceuticals.
Foundation Update

Dear Friends,

The DMRF believes the best service it can provide the dystonia community is to work every day toward improved therapies and a cure. The field of dystonia research has a lot of moving parts. Many people, medical centers, agencies, and organizations must work together to keep the science progressing. The DMRF aspires to serve as a leader in the field, putting in as much time, effort, and resources as needed to get results that make a difference in people’s lives. The DMRF is committed to serving individuals and families living with all forms of dystonia.

We firmly believe in collaborating with partners to meet mutual goals. For example, as you read about the research investigations the DMRF is funding this year, you may notice we are partnering with other dystonia organizations on specific projects to advance science. The DMRF is proud to join forces with Tyler’s Hope for a Dystonia Cure to fund Phase 2 of the research contract with BioFocus, DPI to identify new drug targets for DYT1 dystonia. The BioFocus project is a step toward a whole new generation of dystonia therapies that interrupt, alleviate, or prevent symptoms at the cellular level. While research into all forms of dystonia is ongoing in centers around the world, each form of the disorder demonstrates something about dystonia as a whole, and DYT1 dystonia is teaching us lessons that apply to all dystonias.

Similarly, the DMRF is partnering with the National Spasmodic Dysphonia Association to fund a project led by Dr. Tatiana Fuchs to identify gene mutations associated with dystonia of the vocal cord muscles. As new techniques become available and the genetics of dystonia are increasingly understood, the number of genes known to be associated with certain forms of dystonia continues to grow. Discovering genes and gene mutations associated with dystonia are critical to the development of new treatments because they provide new therapeutic targets. If scientists learn that a mutation in a specific gene triggers the development of dystonia, it is only rational to devise a therapy that attempts to correct or compensate for that mutation.

The dystonia community is working together for the common good. The DMRF is cooperating with partners to maximize results. We remain extremely grateful to our organizational partners, to the scientists who devote their time and expertise to dystonia, and—especially—to all of you, our supporters without whom none of this would be possible.

Thank you, always, for your support.

Art Kessler
President

Janet L. Hieshetter
Executive Director
Sign Up for Updates to Your Phone

The DMRF is going mobile! Through mGive, a national mobile communications provider for non-profits, we are now able to send dystonia updates directly to your cell phone. To sign up to receive news alerts, event information, and cutting edge research updates, scan the code above with your smartphone or text DYSTONIA to 90999.

Meeting Lays Groundwork for Dystonia Clinical Trials

One of the cornerstones of the DMRF mission is to advance research for improved therapies and a cure. Continuous advances in our understanding of dystonia have led to identification of various therapeutic targets that may result in the development of new treatments. Consequently, several pharmaceutical companies have begun to develop new medications for dystonia and related disorders. The gold standard for testing the effectiveness of a new therapy is the random, blinded, placebo-controlled clinical study. To lay the groundwork for dystonia clinical trials to test new compounds and therapies in human volunteers, the DMRF hosted in May a meeting of clinical researchers and biostatisticians, in collaboration with the National Institute of Neurological Disorders & Stroke (NINDS) and the Dystonia Coalition.

The major outcome of the meeting was a set of clinical trial proposals specifically designed for dystonia. Proceedings from the meeting will be published and are designed to help prepare the dystonia community for clinical trials. These preparations will expedite the process of bringing new treatment options to market.

The workshop was organized by Christopher Coffey, PhD (Co-Chair), H. A. Jinnah, MD, PhD (Co-Chair), Wendy Galpern, MD, PhD, and Jan Teller, MA, PhD. Funding for the meeting was provided by DMRF, a grant from NINDS, and additional support provided by the Dystonia Coalition (which is funded through the Office of Rare Disease Research and NINDS).

Upcoming Events

August 20, 2012
4th Annual Minnesota Dystonia Golf Classic
Hastings, MN

September 2012
Dogs4Dystonia Virtual Dog Walk Month

September 8, 2012
Dystance4Dystonia Capitol Hill Walk
Washington, DC

September 23, 2012
7th Annual Dogs for Dystonia
Freehold, NJ

October 13, 2012
Dogs4Dystonia Dog Walk
Arlington, TX

October 21, 2012
Mid-Atlantic Regional Dystonia Symposium
Rockville, MD

October 28, 2012
7th Annual Basket Bash
Chicago, IL

Go to http://www.dystonia-foundation.org and click on "Calendar of Events" for a complete list of events as dates are confirmed.
DMRF Hosts First-ever Musicians’ Summit

*Scientists and Artists Gather in NY to Discuss Dystonia*

The Musicians’ Dystonia Summit that took place March 9–10, 2012 in New York City was the very first event of its kind. The Summit was made possible by sponsorships from Allergan, The Medtronic Foundation, Merz, and Jack and Nancy Britts. The DMRF has been funding research on musician’s dystonia for many years, and this event provided an historic occasion to stop and review what the research is telling us and what we need to do next to accelerate improved treatments. The Summit also provided an important opportunity for affected musicians to meet and exchange information. Some of the discussion became very emotional as participants shared the depth of how dystonia shattered their careers and livelihoods—not an uncommon experience for people with all forms of the disorder. The DMRF extends our deep appreciation to the musicians who shared their stories and the researchers and physicians who are working tirelessly to meet the needs of this community.

Sessions included Natural History and Phenomenology/Epidemiology; Imaging; Physiology; Genetics; Treatment and Management for the Healthcare Professional and Patient. The distinguished roster of speakers included Eckart Altenmüller, MD, Mark Hallett, MD, Gottfried Schlaug, MD, and David M. Simpson, MD.

The DMRF was proud to honor Musicians With Dystonia founders Glen Estrin and Steven Frucht, MD for their years of service to the musician community. They each played an essential role in bringing the Musician’s Summit to fruition. Glen and Dr. Frucht served on the Summit planning committee along with Mark Hallett, MD, Billy McLaughlin, and Jan Teller, MA, PhD.

Many thanks, also, to the performers who shared their talents for a spectacular concert: David Leisner, The Fiddling Thomsons, and Billy McLaughlin joined by a quartet including Steven Leung, Dr. Frucht, and Dr. Frucht’s talented daughters.

*New York Times* reporter James R. Oestreich covered the Musicians’ Summit and a story was published in the March 13 issue. The summit also led to articles authored by Glen Estrin and Dr. Frucht in spring issues of *International Musician* and *Making Music*.

Research on musician’s dystonia will help scientists better understand all dystonias. Scientists are challenged to connect the dots between a musician’s intense, repetitive practice of specific fine motor movements and the onset of symptoms—this will reveal fundamental information about how and why dystonia occurs.
Volunteers Make Dystonia Advocacy Day 2012 a Big Success

Nearly 120 people from across the country, representing diverse forms of dystonia, traveled to Washington, DC to participate in Dystonia Advocacy Day, May 8–9, 2012. Dystonia Advocacy Day is organized by the Dystonia Advocacy Network (DAN), a grassroots coalition of dystonia organizations that brings the community together to speak out with a single, powerful voice on legislative and public policy issues. The DMRF is proud to serve as the administrative center for the DAN.

DAN volunteers visited 125 Congressional offices to discuss the following issues that impact the dystonia community:

• Allocating $32 billion in funding for the National Institutes of Health (NIH) so that highly scored science can be funded. Volunteers informed legislators about the Dystonia Coalition, and urged them to request an update on the Coalition from Francis Collins, MD, PhD, Director of NIH.

• Requesting that dystonia continue to be included as a condition eligible for study in the Department of Defense Peer-Reviewed Medical Research Program. This is significant because it provides additional federal funding of dystonia research in addition to the NIH.

• Patient access to treatments and care continues to be a concern, and advocates asked for a long-term solution to the problem of determining the appropriate reimbursement for Medicare for physician services. This is significant because many physicians are no longer accepting Medicare/Medicaid patients because of inadequate reimbursement rates. Also, private insurance companies follow the lead the Centers for Medicare and Medicaid Services set for procedures, and private coverage may soon be problematic.

• Advocates asked for Congress to lift the profit restrictions on Humanitarian Use Devices (such as the implants used in deep brain stimulation for dystonia), because this restriction limits companies from entering the field and contributing to the development of new devices. The DMRF later learned that Congress approved the request to lift the profit restriction on Humanitarian Use Devices and it was sent to President Obama for his signature.

Dystonia Advocacy Day attracts a mix of first time attendees and regular participants. Laurie Ozelius, PhD of Mount Sinai Hospital, a dedicated researcher whose acclaimed work has contributed to the discovery of multiple gene mutations associated with dystonia, was among the first-timers. Dr. Ozelius joined members of the dystonia community to represent the state of New York and meet with their Senators and U.S. Representatives.

“We had people with all types of dystonia in our group,” explained Dr. Ozelius. “They showed great empathy to one another and shared some very personal things in order to get their messages across. It was very heartfelt and courageous. The group really worked together. We
were a well-oiled machine, presenting very compelling arguments!"

Dr. Ozelius explains why, as a researcher, it is so important for legislators to hear from their constituents with dystonia: “I am personally grateful to all the advocates because it’s very tough to get research funding these days. Each different source of federal funding and every dollar committed by Congress to medical research can really make a difference to dystonia research. I enjoyed meeting and working with the other advocates. I would urge people to think about participating. This grassroots type of advocacy really makes a difference on many different levels.”

Many thanks to the DAN organizations and all the DAN advocates who made the effort to attend Dystonia Advocacy Day on behalf of the entire dystonia community.

DMRF Offers New Dogs4Dystonia Collars and Leashes

Your dog can help promote dystonia awareness! Brand new collars and leashes feature the Dogs4Dystonia logo, the DMRF website, and have a unique seatbelt-style buckle. Your four-legged friend will spread the word about dystonia and the DMRF wherever he goes. A great conversation piece, these collars are the perfect segue to tell your fellow dogs lovers about dystonia, whether it’s the veterinarian, groomer, or (in only rare cases, we hope) the neighborhood animal control officer. Collars are available in sizes small and medium. Small collar expands to approximately 13”. Medium collar expands to approximately 25”. Leash is 4’ long. To order, go to: https://dystonia.donorshops.com
The DMRF is proud to announce this year’s outstanding research investigations, some of which are continuing from 2011. Congratulations to this year’s award recipients, and infinite thanks to our supporters for making this research funding possible.

STANLEY FAHN AWARDS
Regulatory RNA Networks in Inherited Dystonia
Pedro Gonzalez-Alegre, MD
University of Iowa

Dr. Gonzalez-Alegre is a pioneer in applying RNA interference techniques to genetic forms of dystonia, which typically affect children and are profoundly disabling. RNA interference is the process of deactivating disease-causing genes. Dr. Gonzalez-Alegre’s latest investigations use molecular methods to advance our understanding of the disease mechanism underlying dystonia.

D2 Dopamine Receptor Signaling Alteration in a Mouse Model of DYT1 Dystonia: A Novel Rescue Approach
Antonio Pisani, MD, Fondazione Santa Lucia, University of Rome

Dr. Pisani previously identified a link between a dystonia-causing gene mutation and changes in specific receptor proteins in the brain. His latest work further explores the effects of this mutation on neurotransmitters in the brain.

FELLOWSHIPS
Neuroanatomical Abnormalities in DYT1 Dystonia
Chang-hyun Song, DVM
Emory University

Exactly which region of the brain is responsible for the abnormal signals associated with dystonia symptoms is a matter of intense recent debate in the neuroscience community. To better understand how symptoms originate, Dr. Song is exploring the brains of mice with the DYT1 dystonia mutation for suspected anatomical abnormalities in three main brain regions currently believed to be involved in dystonia.

Defining the Parameters of TorsinA Function in Neural Development
Lauren Tanabe, PhD
University of Michigan

Dr. Tanabe is using a new mouse model to pinpoint when and how the dystonia protein torsinA is functioning during the formation of the nervous system in embryonic development. She is also exploring the role of a related protein, torsinB, as possibly protecting the nervous system from the effects of mutant torsinA.

Targeted and Genome Wide Analyses of Factors that Modify TorsinA
Lucia Zacchi, PhD
University of Pittsburgh

One approach for developing new dystonia therapies is to identify cellular processes or genetic factors that selectively affect the protein torsinA, which is known to be abnormal in individuals with specific dystonias. Dr. Zacchi’s work will identify new genetic factors that affect normal and abnormal torsinA, which could eventually lead to the development of therapies specifically designed to reduce the effects of abnormal torsinA while enhancing the effects of normal torsinA.

GRANTS & CONTRACTS
Identification of Novel Drug Targets for DYT1 Dystonia
BioFocus, DPI, The Netherlands

This continuing project is the first step toward the rational design of dystonia therapeutics by identifying new potential drug targets. Phase two is underway, which involves screening for compounds that modify effects of the DYT1 dystonia-causing mutation. The DMRF is proud to partner with Tyler’s Hope for a Dystonia Cure on this project.

Comprehensive Examination of Motor Circuit Activity in Awake Behaving DYT1 Knockin Mice
Nicole Calakos, MD, PhD
Duke University

Dystonia can be caused by many factors, including lesions to a variety of different regions in the central nervous system, peripheral trauma, drug exposure, and defined genetic mutations. One hypothesis is that what all of these diverse insults to the nervous system have in common is that they interfere with the brain networks responsible for movement. In this study, Dr. Calakos is seeking a defined connection between the DYT1 dystonia mutation and these brain networks.
Characterization of Neuronal and Non-Neuronal Forms of THAP1 Protein
Michelle Ehrlich, MD
Mount Sinai School of Medicine
Mutations in the DYT6 gene are associated with a form of early onset dystonia, and may be related to the more common focal dystonias as well. Dr. Ehrlich plans to learn more about the protein associated with this gene (called THAP1) and the relationship between the mutated protein and symptoms of dystonia.

Identification of a Spasmodic Dysphonia Gene Using Exome Sequencing
Tatiana Fuchs, PhD
Mount Sinai School of Medicine
This grant is focused on finding a genetic cause of spasmodic dysphonia (SD), focal dystonia of the vocal cords. Dr. Fuchs is using an innovative, powerful technique to screen all genes in a family affected by SD to identify a common gene mutation. To determine whether mutations in this gene contribute to additional SD and other focal/segmental primary dystonia cases, Dr. Fuchs will also screen for this gene in an additional group of volunteers with focal dystonia. The DMRF is proud to partner with the National Spasmodic Dysphonia Association to support this project.

Abundance and Cellular Localization of Wildtype and Mutant SGCE in Human Skin Fibroblasts and Induced Pluripotent Stem Cell-derived Neurons
Anne Grünewald, PhD
University of Lübeck, Germany
Myoclonus dystonia is caused by mutations in the epsilon-sarcoglycan (SGCE) gene in about 25% of cases. This study seeks to clarify why only a percentage of individuals who inherit the gene manifest symptoms and the differences between the proteins produced by normal and mutant SGCE. This may help clarify penetrance patterns for additional dystonias.

Morphological-Functional Analysis of the Endoplasmic Reticulum in a Mouse Model of DYT1 Dystonia
N. Charles Harata, MD, PhD
University of Iowa
The proposed research is designed to identify how dystonia changes the way neurons function. Dr. Harata is also taking a close look at a possible link between dystonia and calcium ions, which play a critical role in the function of cells.

Dystonia Gene Discovery Through Autozygosity Mapping and Massively Parallel Sequencing
Michael Kruer, MD
Oregon Health & Science University
This project seeks to identify previously unknown genetic causes of dystonia using novel comprehensive genetic screening. The findings from this work have the potential to significantly advance our understanding of dystonia at the molecular level and identify new mechanisms of dystonia. This project is a collaboration with Dr. Henry Houlden.

Neurophysiological Study of Myoclonus Dystonia
Sabine Meunier, MD, PhD,
National Institute of Health and Medical Research, France (INSERM)
John Rothwell, PhD, University College London, United Kingdom
Although the protein associated with the gene responsible for myoclonus dystonia is very common, it is not clear why mutations in this gene cause the jerks and spasms that continue on next page

“I would like my research to take us to new frontiers of a cure for dystonia. The hope is that a better understanding of the molecular pathways affected by mutant forms of dystonia-causing genes will bring us a few steps closer to finding that cure. My goal is to figure out how one particular gene—DYT16—may lead to early onset dystonia when mutated.”
– Rekha Patel, PhD

In 2008, dystonia researchers discovered that mutations in the DYT16 gene are associated with early onset dystonia. Little did they know that Rekha Patel, PhD of University of South Carolina had spent over 10 years researching the same gene (a.k.a PRKRA) and its role in regulating cell proliferation and programmed cell death—concepts especially relevant to cancer research. Dr. Patel has since brought her expertise to the dystonia field. Her work is providing a significant leap in progress toward possible therapeutic targets and novel treatments.
Continued from page 9

are so apparent in patients. This study uses new methods of brain stimulation to learn more about how symptoms arise. The findings will provide clues toward new treatments.

The C. elegans OOC-5 Protein as a Model for Understanding the Role of Torsins at the Nuclear Envelope
Lesilee Rose, PhD, University of California, Davis
Recent studies suggest that torsinA has an important role in the membrane that surrounds the nucleus of the cell, the nuclear envelope. Dr. Rose and her team are using a worm model to clarify what this role is and how torsinA functions in cells.

Structural Characterization of Torsin1A with its Interactors at the Nuclear Envelope
Thomas Schwartz, PhD, Massachusetts Institute of Technology
Dr. Schwartz’s project seeks to learn more about the function of the dystonia protein torsinA by defining its three-dimensional atomic structure. Within the atomic structure lie the clues to fully understand its function and the ability to influence this function by developing specific drugs.

Patient-specific Induced Pluripotent Stem (iPS) Cells as a Model for DYT1 Dystonia
Nutan Sharma, MD, PhD and Christopher Bragg, PhD, Massachusetts General Hospital/Harvard University
The objective of this research is to establish a collection of dystonia cell models that will serve as a public resource for the research community. These models enable multiple dystonia research groups to study defects in neurons associated with the DYT1 gene mutation.

Clinical and Genetic Studies of African American Patients with Dystonia
Zbigniew Wszolek, MD, Mayo Clinic Jacksonville
Dr. Wszolek, in partnership with Dr. Mark LeDoux, is studying the genetics of several African American families impacted by dystonia, hoping to identify new gene(s) associated with adult onset, focal dystonia. This study will expand what is known about the genetics of focal dystonias and is providing important outreach to African American dystonia-affected families in Southern Georgia and Northern Florida.

Definitions & Concepts

Brain networks/neural network – A circuit of neurons in the brain.

Disease mechanism – The origins and development of a disease or disorder. Ideally, treatments target the disease mechanism.

Drug target – A protein involved in the disease mechanism that can be manipulated by a medication or therapy.

DYT – A designation given to genes associated with a specific type of dystonia. There are over 20 genes known to be associated with dystonia.

DYT1 – Gene associated with early onset generalized dystonia. TorsinA is the protein associated with this gene.

DYT6 – Gene associated with early onset dystonia-parkinsonism. THAP1 is the protein associated with this gene.

DYT11 – Gene associated with myoclonus dystonia. SGCE is the protein associated with this gene.

Genes & Proteins – Genes are the building blocks of living things. Each gene contains a code of DNA that corresponds to a protein. Proteins participate in virtually every process with the human body. When a gene becomes mutated, this changes the protein associated with that gene and how that protein functions. When a protein cannot properly fulfill its function, this may disrupt normal biological processes and lead to a disease or disorder.

Neurons & Receptors – A neuron is a nerve cell or brain cell. Signals between neurons must cross a small gap called the synapse. A neurotransmitter is released from one neuron and crosses the synapse. The neurotransmitter then may be accepted by the next neuron at its receptor.

Neurotransmitter – A chemical in the brain that transmits signals from one neuron to another.

Penetrance – The percentage of people with a disease-causing gene that actually develop symptoms of the disease. For example, approximately 30% of people who have the DYT1 mutation develop symptoms. The remaining 70% of people with the mutation never develop dystonia.
Training the Next Generation of Dystonia Experts

DMRF Announces New Clinical Fellowship Program

Few things are more critical to living well with dystonia than a knowledgeable doctor who is trained in the diagnosis and treatment of movement disorders. Creating opportunities to help young physicians establish careers in movement disorders is also vital to the advancement of dystonia research.

The DMRF is fostering the next generation of dystonia leaders by offering one-year fellowships to support clinical training of physicians in preparation for a clinical and/or research career in movement disorders with special focus on dystonia. The training is patient-oriented and includes hands-on experience in clinics as well as participation in professional meetings. The fellowship encourages participants to be involved in the activities of the Dystonia Coalition and participate in courses related to dystonia at major neurological meetings. The Clinical Fellowship Program is made possible by a grant from Merz Pharmaceuticals.

Congratulations to this year's Clinical Fellowship recipients:

**Florence Chang, MD**
Fellowship institution: Mount Sinai Hospital, New York, New York
Mentor: Steven Frucht, MD

**Andres Deik, MD**
Fellowship institution: Beth Israel Medical Center, New York, New York
Mentors: Susan Bressman, MD and Rachel Saunders-Pullman, MD, MPH

**Scott Norris, MD**
Fellowship institution: Washington University, St. Louis, Missouri
Mentor: Joel Perlmutter, MD

**Neepa Patel, MD**
Fellowship institution: Baylor College of Medicine, Houston, Texas
Mentor: Joseph Jankovic, MD

**Vesper Ramos, MD**
Fellowship institution: National Institute of Neurological Disorders & Stroke, Bethesda, Maryland
Mentor: Mark Hallett, MD

**Jeri Williams, MD**
Fellowship institution: University of Alabama at Birmingham, Birmingham, Alabama
Mentor: David Standaert, MD, PhD

To learn more about these impressive individuals and to read, in their own words, why they chose movement disorders as their medical specialty, go to: http://www.dystonia-foundation.org/dialogue

**Participate in Dystonia Research From Your Computer**

Volunteers Needed for Global Dystonia Registry

The Global Dystonia Registry is a survey designed to collect data submitted from persons affected with dystonia to assist in clinical research efforts including clinical and research trials.

As a supporter of this registry, the DMRF encourages you to consider participating. Although the focal dystonias have many different manifestations, most experts believe they share a common pathogenesis or mechanism that causes the disorder. The common causes may be a similar gene defect, similar lifetime experiences, or both. Collecting information from different patient populations may help identify the common features they may share. This Registry compliments the current scope of research for the Dystonia Coalition, a clinical research effort supported by the National Institutes of Health.

Your participation is essential for this effort to be successful. To learn more or to register with the Global Dystonia Registry please go to: http://www.globaldystoniaregistry.org/
The DMRF is deeply grateful for our grassroots volunteers who work all year to promote dystonia awareness and fundraise for medical research. Every effort and every volunteer makes a difference! We appreciate your support.

**Stefanie and Erwin Jackson** organized the Tallahassee Valentine’s Day Dinner & Show in support of the DMRF in February. The Jacksons work year round to promote dystonia awareness and support research in honor of their adult son **Brian Jackson**, who is diagnosed. Over 300 people supported this event, raising $22,000.

**April Bradbury** hosted a booth at the Mohegan Strawberry Social in Uncasville, Connecticut to promote greater dystonia awareness. Nearly 675 people attended the event, and April distributed 200 pieces of literature.

In April, **Alex Rudolph** crossed the finish line of the Illinois Half-Marathon, celebrating not just a successful run on behalf of the DMRF, but also raising twice as much as she had set for her goal, bringing in $1,000 for dystonia research. Alex is the cousin of DMRF board members, Mark and Diane Rudolph.

Cheers to **Mandi Sleeper** for hosting the first-ever Fitchburg, MA Dystance4Dystonia Walk for a Cure in May. The event raised over $5,000! Mandi developed dystonia as a child and has twice undergone deep brain stimulation surgery.

**Jodi Logman**, a.k.a. Miss Wheelchair Indiana 2011, was among the models featured in the fashion show at this year’s disabilities Expo at Memorial Coliseum in Indiana earlier this year. Jodi is diagnosed with generalized dystonia and is a vocal disabilities advocate.

The **35th Rochester Institute of Technology Spring Juggle-In** took place in April, bigger and better than ever. For the last 12 years the Juggle-In has focused on raising awareness and funds for research to cure dystonia, inspired by **Rosalie and Richard Lewis** and their four sons, all of who are diagnosed. Special thanks to RIT Juggling Club Co-Presidents **Kyla Ciranni** and **Ross Lancaster** for their many efforts.

**April Bradbury**

**Jodi Logman**

Honor Society students at West Essex High School in North Caldwell New Jersey raised $400 on behalf of the DMRF this spring by selling lollipops and dystonia awareness bracelets. Pictured are **Lydia Nathans**, **Sara Jo Notte**, **Nicole Del Mauro**, **Stephen O’Neill**, **Natalia Zak**, and **Hannah Pappas**. Lydia and her brother Jack are diagnosed with dystonia, and her parents **Sandra Calvert Nathans** and **Robert Nathans** lead the New Jersey Support Group. Special thanks to Journalism/English teacher **Cassandra Lo**.

Dystonia awareness duo **Mike Delise** and **Jason Dunn** partnered with Michigan Senator **Steven Bieda** to pass Michigan Senate Resolution 0130 (2012) to recognize the need for improved dystonia awareness.

This spring, leader of the Spokane, Washington Support Group, **Denise Gibson** ran the Lilac Bloomsday Run, an annual event with over 50,000 runners, walkers, and participants in wheelchairs. This year, Denise raised pledges to help fund dystonia research. Go Denise!
Patricia Bergeron and the Verville family organized the 7th Hands for Movement Freedom benefit piano recital at the Richmond Library in Vermont. Alexandre Verville, whose diagnosis inspired this annual concert, played piano at the recital along with 19 pianists. The pianists are all students of Patricia Bergeron. The event raised $300!

The Dystonia Support Group of Alabama hosted the Southeast Regional Dystonia Symposium on May 12 in Birmingham. Medical speakers included DMRF Scientific Director Mahlon DeLong, MD of Emory University; Erwin Montgomery, MD of University of Alabama, Birmingham; and Carlos Arce, MD of University of Florida, Jacksonville. Eighty people traveled from through the surrounding states to attend. Many thanks to Ken Williams for his leadership in organizing this symposium.

Congrats to Meryl (Ohlhausen) Kasdan for being recognized for her volunteerism in the May 25 issue of the Jewish Community of Louisville’s “Community” newspaper. Meryl was diagnosed with generalized dystonia in 1970 at age 12, after three years of misdiagnosis.

In May, Passion Cut Diamonds hosted a special event at E. E. Robbins jewelers in Bellevue, Washington to bring visibility to the remarkable story of newly married DMRF member Meghan Fell and her husband Jeff Fell. Passion Cut Diamond presented Meghan with a specially designed ring in acknowledgement of the challenges dystonia has caused.

In June, Stephen Lichty and Neil Marcus performed their original dance composition “Special Effects” at Oberlin Dance Collective in San Francisco. Stephen and Neil incorporated their dystonia symptoms into the performance, renegotiating dystonic movements to become sculpture.

Jean Kaluza of Orlando, Florida has reached over 100,000 people and raised hundreds of dollars through her “Free the Brains” campaign.” Jean was a professional percussionist when she developed focal hand dystonia, and she retrained herself to perform with opposite hands. Jean built a website and posts weekly online videos demonstrating how she has adapted her performance to incorporate dystonia and discuss the importance for research. Jean has taken advantage of every opportunity to amplify her message via drumming contests, an appearance on the TV show Daytime, and courting the support of radio stations.

Many thanks to Len and Janice Nachbar and Joanna Manusov, leaders of the Central Jersey Dystonia Support & Action Group, for hosting “Dine for Dystonia” in partnership with Solo Trattoria in Freehold, NJ on June 4.

In June, Hunter Webster organized the 3rd Annual Dogs4Dystonia Dog Walk in Reston, Virginia. The event attracted a great crowd and raised much-needed support for DMRF programs.

“Sparks of Imagination” flew at the third annual Women Who Weld exhibit in Omaha, Nebraska. The exhibit, which was held over two weekends in April, was organized by metal sculpture artist Melinda Eames. The event not only raised money and awareness for the DMRF, but was also a tribute to the 10 talented women sculpture artists in the show. One of the artists featured in the exhibit, Antonella Gosselin, is diagnosed with dystonia.

In June, Stephen Lichty and Neil Marcus performed their original dance composition “Special Effects” at Oberlin Dance Collective in San Francisco. Stephen and Neil incorporated their dystonia symptoms into the performance, renegotiating dystonic movements to become sculpture.

Many thanks to Ernie and Val Inman, leaders of the Tampa Bay, FL Dystonia Support Group for creating a dystonia awareness QR code in time for Dystonia Awareness Week. Special dystonia outreach to the technologically savvy.

In June, Hunter Webster organized the 3rd Annual Dogs4Dystonia Dog Walk in Reston, Virginia. The event attracted a great crowd and raised much-needed support for DMRF programs.

Jean Kaluza of Orlando, Florida has reached over 100,000 people and raised hundreds of dollars through her “Free the Brains” campaign.” Jean was a professional percussionist when she developed focal hand dystonia, and she retrained herself to perform with opposite hands. Jean built a website and posts weekly online videos demonstrating how she has adapted her performance to incorporate dystonia and discuss the importance for research. Jean has taken advantage of every opportunity to amplify her message via drumming contests, an appearance on the TV show Daytime, and courting the support of radio stations.

Many thanks to Len and Janice Nachbar and Joanna Manusov, leaders of the Central Jersey Dystonia Support & Action Group, for hosting “Dine for Dystonia” in partnership with Solo Trattoria in Freehold, NJ on June 4.

In June, Hunter Webster organized the 3rd Annual Dogs4Dystonia Dog Walk in Reston, Virginia. The event attracted a great crowd and raised much-needed support for DMRF programs.

“Sparks of Imagination” flew at the third annual Women Who Weld exhibit in Omaha, Nebraska. The exhibit, which was held over two weekends in April, was organized by metal sculpture artist Melinda Eames. The event not only raised money and awareness for the DMRF, but was also a tribute to the 10 talented women sculpture artists in the show. One of the artists featured in the exhibit, Antonella Gosselin, is diagnosed with dystonia.

Continued on next page
**Donna’s Good Hair Day**  
*Teacher Declares She Will Shave Head for Research*

When Donna Hensley of Sandusky, Ohio announced that she would do a fundraising event for the DMRF, she decided to do it in a way that would capture national attention and raise funds for research. Donna, a high school art teacher whose younger sister, Nancy Dennison, is diagnosed with dystonia, decided to challenge her students and the entire community by promising to shave her head if $10,000 was raised by May 16, the day before her sister’s birthday.

“My sister is only 48 and has always been active and very smart,” says Donna. “It is so hard to see what she is going through. I wanted to raise money for research. I thought if I told the students that I would shave my head if I could raise $10,000 that we could probably raise quite a bit.”

Although her event was focused in Sandusky, along the way toward her goal, she captured the hearts of people all across the country. Donna and her students flooded the Twitter universe with tweets about the campaign, posted online videos, secured articles in local newspapers, and reached out to celebrities for support.

Donna and her supporters were successful in meeting—and exceeding—their goal. On May 16, a gymnasium full of excited high school students cheered as a hair stylist cut off Donna’s hair, which was donated to Locks of Love. To date, Donna’s event has generated nearly $12,000 for dystonia research. Although she won’t be able to exactly recreate the event in 2013, she is already considering how she can continue raising money to help her sister and others with dystonia.

The DMRF is immensely grateful and uniquely inspired by Donna’s efforts and the enthusiastic participation of her students and supporters. To view a video Donna created about the campaign, go to: http://www.hooplaha.com/a-sisters-wish-comes-true

---

Despite sizzling temperatures, a team of volunteers led by MaryRae Nee represented the DMRF at the Pittsburgh Pirates Disability Awareness Celebration on July 6. The purpose of the event was to educate the public on the spectrum of disabilities and, given the Pirates record so far this season, fans packed the park. Eric Jansen traveled back to his hometown of Pittsburgh from Florida to attend.

Earlier this year, Juice Newton headlined a concert to benefit DMRF and the National Spasmodic Dysphonia Association in El Cajon, CA. Billy McLaughlin and D.C. Hathaway also performed. The concert was hosted by the Dystonia Support and Advocacy Group of San Diego County, and live-streamed to the DMRF website courtesy of San Diego State University. Donations continue to come in. Special thanks to Martha Murphy and Paul Fowler for their efforts in making this event possible, and to Ms. Newton for once again lending her time and incredible talents in support of the dystonia community.
Volunteers Needed for New Cervical Dystonia Study

The DMRF wants to inform the community of a new opportunity to participate in a clinical study. Quest Research Institute is coordinating a study to compare Xeomin®, a botulinum neurotoxin treatment, in shorter treatment intervals (“short flex dosing”) to the standard interval dosing (“long flex dosing”) to determine if more frequent dosing leads to development of botulinum neurotoxin resistance, making the treatment less effective. Xeomin® is approved by the FDA for the treatment of cervical dystonia. The use of Short Flex Xeomin® is investigational, which means it is not approved by the FDA.

To qualify for this study, volunteers must:
• Be between the ages of 18 and 81.
• Have a documented clinical diagnosis of primary cervical dystonia.
• Be on a stable dose of other medications (if any) for dystonia treatment for 30 days prior to baseline.
• Have been previously treated with onabotulinumtoxin A (Botox®).
• Have had at least 12 weeks elapsed since the most recent onabotulinumtoxinA injection.

Additional inclusion or exclusion criteria must be met for eligible volunteers. For more information about participating in this study, please contact Quest Research Institute at 248-957-8940 or visit http://www.questri.com

DID YOU KNOW?
You may be eligible for financial assistance for your prescription medications. Visit the following websites to learn more:

Partnership for Prescription Assistance
http://www.pparx.org/

Needy Meds
http://www.needymeds.org/

Face2Face
Promoting Dystonia Awareness

You can improve dystonia awareness, one person at a time, as part of the DMRF’s “Face2Face” campaign. The DMRF encourages individuals impacted by dystonia to share their stories with the people they encounter in their daily lives: your neighbors, members of your religious congregation, the wait staff at your favorite restaurant, your mail carrier — anyone and everyone you find yourself in contact with.

Your story is a powerful instrument for improving awareness, and you can help put a face to this disorder simply by telling others about your diagnosis.

For example, Amy Behar and Silas Courson took advantage of a backstage visit at a Flaming Lips concert in June to share Amy’s experience with dystonia with frontman Wayne Coyne. Talk about seizing an opportunity for awareness!

How many people can you impact, Face2Face?
At http://www.dystonia-foundation.org/face2face you will find the following tools to help you in your efforts:
• A fact sheet to download, print, and share with the people you educate.
• An order form to request a supply of wallet-sized information cards by mail.
• A “Dystonia is Not a Country” image to post on Facebook, Twitter, and other social media networks.

Stay in touch and let us know about your adventures in dystonia awareness. Tell us how opening up to someone about dystonia has made a difference by posting in the DMRF’s Face2Face Facebook group: https://www.facebook.com/groups/face2facedmrf/

We appreciate your support.
Take Me Out to the Ball Game
Colin Returns to the Baseball Field

Seven-year-old Colin loves sports, but dystonia was making it hard for him to walk and run. It got harder to run the bases when he played baseball. At school, he sometimes fell while trying to keep up with the other kids at recess. So, his teachers created a special room for him. Every day at recess, Colin got to choose one friend to play with him in the room while the class went outside. Pretty soon, all the kids wanted to have a turn and spend recess inside with Colin.

Colin had deep brain stimulation (DBS), which is a type of surgery to treat dystonia. When asked what advice he would give to other kids with dystonia who are having DBS, Colin said, “I would tell other kids it’s not that bad. It’s not scary.”

A few months have passed since Colin’s DBS. His dystonia is slowly going away. He no longer uses a wheelchair, and it is now easier for him to play baseball again.

Read a special interview with Colin’s parents Kevin and Angela Baldwin on page 19.

“Never Give Up!”
Nathan Shares his Story

This is how Nathan Simmons describes living with dystonia: “It was really rough. Every time I took a step I would fall down. I couldn’t write. Sometimes the pills make me nauseous and dizzy.”

In February, Nathan and his parents, Steve and Jeannie Simmons, appeared on TV on News 12 WRDW in Augusta, Georgia to help educate other people about dystonia.

Shortly before appearing on TV, Nathan had a type of surgery called deep brain stimulation (DBS) to treat his dystonia. He was nine years old. Because Nathan had to shave his head to have the surgery, several of his friends and family members also shaved their heads so he wasn’t the only one.

After the surgery, Nathan’s dystonia began to lessen. He can now run and move easily.

Nathan’s advice to other kids with dystonia is: “Never give up! If you walk weird, keep walking and trying. Maybe you can have DBS.”

Parents can read more about dystonia treatments and DBS at http://www.dystoniafoundation.org/treatment

The Little Girl Who Could
Gigi Runs for her Mom

Seven-year-old Gigi Kenney ran her second 5K race to raise money for research in honor of her mother Yanett Kenney, who has dystonia. Gigi ran in the Oakland, CA Running Festival with her father, John Kenney. Gigi’s goal was to raise $500, and she did it! Way to go, Gigi! And many thanks to the many people who supported her.
Dystonia: A Debilitating Disorder

A Whole Lot of Shaking Going On

By Robbie Nabors, Leader, Mobile, Alabama Dystonia Support Group

In the early stages of this illness, I had the disconcerting feeling that something was wrong, but there were no clearly definable symptoms. It took me years to discover the name of my illness.

At first there was a whole lot of shaking going on—barely noticeable in the beginning but gradually affecting everyday routines such as putting on make-up. I would find it difficult to apply mascara or lipstick. Inexplicable smudges and smears began to irritate me because I felt like a child who couldn’t color inside the lines. Then my head began shaking noticeably, and others would comment. I went to a neurologist, but he assured me that it was only a jerk in my neck.

After about seven years, my eyes began to blink and feel gritty, as though they had sand in them. Occasionally, when I walked or drove a car, they would involuntarily close and I would have to stop and use my fingers to pry them open again. These were frightening symptoms so again I sought medical advice only to be told by my eye doctor, “You have a chronic dry eye problem.” Somehow, that did not ring true. “I cannot accept that!” I thought.

I saw an article in the paper about a woman with blinking eyes, and I wrote down the name of the doctor she used. I called and got an appointment, and the doctor said I had the beginnings of dystonia. Now, at least I knew the name of this monstrous malady that was slowly stealing my dignity. I saw a neurologist, who diagnosed me with generalized dystonia because my body was affected from the waist up.

The shaking of my head and neck and uncontrollable jerking of muscles in my arms became a painful reality of the disease. I had to walk with my arms tightly clasped behind my back to keep from flailing about. Sometimes the muscles in my neck would swell as if there were a balloon being inflated inside, making it difficult to swallow or speak or breathe. Sometimes when I talk on the phone, my neck pulls down to the left, making my voice change, my words slur, and my tongue feel like a plank. It is hard to understand me sometimes. Eating has become an ordeal. When the muscles contract, it is hard to chew. Sometimes my jaws will just clamp down, and it takes a while to release. Going out to eat in fine restaurants used to be a great pleasure, but now it has become a source of embarrassment. I know people are staring. The violent shaking has traveled to my arms and my legs. My muscles are in perpetual motion, and it is truly exhausting. When I go to bed, it all goes away after a while and I can get a good night’s sleep. My husband says I still move in my sleep.

Most of these things happen inside my house, so only my husband can see the difficulty and daily struggle. Dystonia can be more active when you are excited or under stress, and sometimes my muscles are more active when I am at church, the grocery store, or a department store.

I take medication for this disorder, which helps some. My husband is a wonderful, patient caretaker and my family gives me encouragement and love. I refuse to give up church, and I try to stay busy by volunteering. I stay out doing things with people because this takes my mind off the constant motion, and it makes me feel better to help others. My friends and acquaintances sustain my spirit and bring me great joy. I ask God to give me strength, courage, and wisdom every day to do all the things I do. I am hopeful research will find a cure for this debilitating disease. Above all, I pray for those who suffer with this illness.
Be a Social (Media) Butterfly
Check Out DMRF Online Support Forums

If you do not have access to a local support group, you can still connect with others in the dystonia community. Check out the following online social forums.

**YouTube**  http://www.youtube.com/facesofdystonia

**Twitter**  http://twitter.com/dmrf

**Facebook**  http://www.facebook.com
Search “Dystonia Medical Research Foundation” and “Dystonia Friend”

**NEW! Facebook Group:** “Cervical Dystonia Support Forum”
Special thanks to volunteer administrator Denise Gaskell.  https://www.facebook.com/groups/dmrf.cervical/

**NEW! Facebook Group:** “20/30 Dystonia Group – A Forum for People in Their 20s and 30s”
Special thanks to volunteer administrators Marcie Povitsky and Ginny Bryan.  https://www.facebook.com/groups/2030dmrf/

**NEW! Facebook Group:** “Support4Parents of Children with Dystonia”
https://www.facebook.com/groups/support4parents.dmrf/

**NEW! Facebook Group:** “Caring4Parents with Movement Disorders”
Email contact@dystonia-foundation.org for more information.

**Online Dystonia Bulletin Boards**
http://www.dystonia-bb.org/
Many thanks to volunteer moderators Bob Campbell, Jeff Harris, Linda Walking Woman, and to Mary Beth Chan who recently retired from her moderator role.

**DBSforDystonia Yahoo Group**
http://health.groups.yahoo.com/group/DBSforDystonia/
Much appreciation to founder and moderator Dee Linde.

For a complete list of DMRF’s online social forums, visit:  http://www.dystonia-foundation.org/online

To search for a live DMRF support group in your area, go to http://www.dystonia-foundation.org/supportgroups

---

**Priceless Gift to Research**
**Costs Nothing At All**

**Consider Registering as a Brain Donor**

Brain donation is a priceless gift that dystonia-affected persons and their family members can give to the dystonia community. Providing investigators with brain tissue samples for research is invaluable because it supports the development of new therapies and ultimately a cure for dystonia.

The DMRF partners with the Harvard Brain Tissue Resource Center (HBTRC) at McLean Hospital in Belmont, Massachusetts as part of the Brain Bank Collective. The DMRF is partnering with McLean and other dystonia patient groups to build a private dystonia tissue sample collection.

The brain recovery process does not interfere with funeral or memorial services, or affect the outward appearance of the donor. All expenses related to the recovery or transport of the tissue to the HBTRC are covered by the DMRF. There is no cost to the donor or the donor’s family.

If you chose to enroll, there is a short registration form to fill out and return. Once enrolled, you will receive a laminated donor card with notification instructions and contact information for the HBTRC.

If you are interested in learning more, please contact Martha Murphy, Brain Bank Liaison, at mmurphy@dystonia-foundation.org or call DMRF headquarters at 800-377-DYST (3978). Additional information on brain donation can be found at http://www.dystonia-foundation.org/brain

Thank you for considering this very special gift in support of dystonia research.
Getting Colin’s Smile Back

*Parents Kevin and Angela Baldwin Talk about the Decision to Try DBS*

By the time Colin Baldwin was five years old, he was fascinated by sports. “Whatever sport was on TV, he would stand there in front of the TV and act it out,” recalls his father Kevin. “He didn’t want to watch cartoons. He wanted to watch NFL News, MLB News.”

By age six, Colin excelled athletically. He especially loved baseball.

A slight limp was the earliest sign that something wasn’t quite right. “The next week, it got worse,” said Kevin. “It was in his gait. Then we noticed the twisting.”

The orthopedist gave Colin a clean bill of health, and said let’s wait and see. The doctor also made a vague reference to a neurologist referral if things didn’t improve. Kevin seized on this subtle inference, and it began to dawn on him that his six-year-old boy might have a brain disorder. It was an eight month wait to see the neurologist.

“It felt like everything stopped,” says Angela. “Of course we tried to keep everything moving and normal, but it becomes a little mechanical.”

Kevin threw himself into researching online for a possible explanation for Colin’s symptoms. “All I did was research,” he said. “I said to Angela, I think he has dystonia. I had no denial. I was the first one to hit reality, and I hit it hard.”

Angela was not immediately convinced: “I said, let a doctor tell us what’s going on. The last thing I wanted to do was put a label on it, if we didn’t know for sure.”

Kevin was ready for the diagnosis. “I already knew DBS was a possibility. I did the research and realized the risks are actually low—of course there is risk with every surgery. I knew the results could be mixed.”

Kevin and Angela took Colin to a major movement disorder center known for its expertise in deep brain stimulation for dystonia. The movement disorder team determined that Colin was an excellent candidate for the procedure.

In March and April of 2012, one year after Colin’s symptoms had first surfaced, had the procedure in two separate surgeries. In most cases, individuals with dystonia must wait a number of weeks to begin seeing improvements, and full benefit may take months or years to achieve. Colin’s symptoms began to diminish almost immediately. Within four weeks, the dystonia was barely noticeable.

“I’m holding my breath because the results have been so good,” says Angela.

Kevin adds, “I had forgotten what his smile truly looked like. Now it’s back.”

Angela and Kevin are cautiously delighted with Colin’s experience. “DBS is working for Colin,” says Angela. “If your child is a candidate, talk to patients who’ve had the DBS by the same medical team. Talk to as many people as you can.”

Kevin echoes Angela’s encouragement to parents to research their options carefully: “Get a diagnosis, research the best doctors. Learn everything you can. Don’t be afraid.”

Colin has returned to school full-time and is back on the baseball field. In May, the 30th Annual Ramona Pony Baseball Invitational baseball tournament honored Colin and his family by raising over $2,000 for dystonia research in his name. At the tournament, just five weeks after needing a wheelchair to walk, he threw out the first pitch and ran the bases. To learn more about Colin’s story, visit the family’s blog at http://www.colinrbaldwin.com/
Kindness of Strangers
Unexpected DMRF Supporters & Volunteers

Most of the champions of the dystonia community volunteer because of a very direct, personal connection to the disorder: dystonia impacts them, a child, or a spouse. But sometimes people on the periphery are so taken by the cause that they too go above and beyond to help support the dystonia community. Here we highlight three such volunteers, who came to be involved with the DMRF in a more round-a-bout way.

Susan Strawgate Code
In 2004, Susan Strawgate Code’s nephew David Rudolph was diagnosed with DYT1 dystonia at age eight. “I never heard of dystonia before my nephew’s diagnosis,” she recalls. “As soon as we learned all about it, I realized that if people affected by it don’t take action, who will? Of course, wanting to do anything I could for my sister and nephew is a driving force.”

At the time of David’s diagnosis, Susan’s son Ethan was preparing for his Bar Mitzvah. Ethan chose to raise money for dystonia research for his required community service. The family decided to have a garage sale and donate the proceeds to the DMRF. By this time, David’s parents, Mark and Diane Rudolph, had joined the DMRF Board of Directors. The entire extended family was rallying around the cause. The Code family enlisted the help of friends and neighbors as well, and Ethan’s garage sale was a huge success. “He felt so good, and it was so successful that we didn’t hesitate to make it an annual thing,” said Susan. “It has grown so that neighbors and friends contribute items to sell with pleasure, local shops donate raffle items to make extra funds, and buyers return year after year to buy or just make a donation!”

In addition to the annual garage sale (for which Susan collects items year-round), Susan traveled from her home in New York to Washington, DC for Dystonia Advocacy Day earlier this year. “My only experience with someone with dystonia was with David, and what I read in the DMRF newsletter. But at Advocacy Day I met all kinds of people, all ages and types of dystonia, all brave and wonderful. I felt part of something big and important,” she explains.

Susan learned that she was a carrier for the gene mutation associated with DYT1 dystonia, which only fueled her to work harder to support research. “A cure is attainable if we put our money and time and effort towards it,” she says. “David was an energetic little boy who excelled at karate and basketball, and wanted to be a pilot. Dystonia has changed all that. He has to fight to do the things he loves to do, and some things he has to let go of. If we all keep raising awareness and funds, change can happen.”

Mike Delise
Mike Delise of Warren, Michigan recalls meeting Jason Dunn for the first time: “It was that grin of his. That stuck out. And then, the condition he was in, and I had never heard of dystonia. I just knew we had to do something.”

Jason attended high school with Mike’s daughters. Mike met Jason at his younger daughter’s graduation party. Jason developed dystonia as a young child, and the disorder had taken a heavy toll on his posture, gait, and voice. He uses a typing device to communicate, cannot sit in a chair, and walking is extremely difficult.
In addition to becoming fast friends, Mike and Jason became a powerful dystonia awareness duo. Because Jason is unable to speak on the phone or drive, they joke that Mike is Jason’s agent. “I believe in Jason so much,” says Mike. “I just want to get him in front of people and let him take over. He has more charm in one finger than I do in my whole body.” Mike and Jason have gotten stories about dystonia in all the major Detroit metropolitan news outlets. Jason has reached millions of viewers around the world by appearing in a TV program Extraordinary Humans: Muscles. They traveled to Washington, DC to advocate on behalf of the greater dystonia community, and they secured a Dystonia Awareness Proclamation in the Michigan Senate. They regularly attend public relations events associated hosted by Detroit sports teams to help get the word out. Mike worked for two years to convince community organizations and agencies to build Jason an accessible house when it became clear that the childhood home he was living in on his own was not safe. Mike has accompanied Jason to medical appointments, and overall been his right hand man.

“I feel blessed to have this opportunity. I feel like dystonia awareness is a purpose for me. I wish everybody could spend one day with Jason and see how difficult it is for him to get through a day. To have to type every letter of every word, when he has so much to say. And he does it all with a smile. If a celebrity could spend one day, one hour, with him, I think they would want to help find a cure. We just have to keep working at it. I can’t wait for the day that Jay can grab his keys off the table and jump in his car and take off without depending on someone else.”

Melinda Eames
Melinda Eames of Omaha, Nebraska is an interior design professional with a passion for metal sculpture and welding. Since beginning to work in metal, she was inspired by the talented group of women metal sculpture artists she has met. These women inspired her to organize an annual art show called “Women Who Weld” to showcase female metal artists.

One of the artists who inspired Melinda with her work, a woman named Antonella Gosselin, also inspired her in other ways. Antonella began developing multifocal dystonia at the age of 35. Melinda was so taken by Antonella’s story and spirit that she added a silent auction at the annual Women Who Weld show to the benefit the DMRF. Each participating artist contributes a sculptural piece that becomes a welded “quilt” and is auctioned off to support dystonia research. “Those who find a way to live with this disorder, like Antonella, inspire us to create art that tells a story of passion and strength. Working in this medium is empowering. This year’s theme, ‘Sparks of Imagination,’ was not only a reference to welding and creativity, but also a reference to the hope that comes from extensive research and treatments that can ‘spark’ the brain activity in a person suffering from dystonia.”

Melinda and Women Who Weld secured multiple stories in Omaha metropolitan news outlets and have raised thousands of dollars to support the DMRF over three years.
PERSONAL PROFILE

Meet Nima Patel, MD

Dr. Nima Patel is a pediatrician who owned her own practice for 30 years and is an Associate Professor in the Department of Pediatrics at the University of Connecticut. Dr. Patel was diagnosed with oromandibular dystonia and antecollis causing symptoms affecting her jaw, neck, and scalp muscles.

How did your symptoms begin and how were you diagnosed?
I am a doctor, I didn't know what I had for five years. I had a molar extracted and, with complications, healing took three months. Suddenly, I felt like I forgot how to chew. My muscles just wouldn’t move. I talked to my dentist and dental specialists resulting in having 22 teeth capped, braces for two years, and nine root canal procedures made necessary by the excessive work in capping. Things didn’t improve. I consulted an oral surgeon, then a dental school, and no one provided answers. I was depressed because I didn’t know what was wrong with me. I was prescribed an antidepressant that left me suicidal. I went on to a neurologist who said it was all in my head.

At the time, my son was a medical resident at Yale. He came home for a holiday visit and saw that I couldn’t eat anything. I started to cry. He said, “Let me watch you eat.” He said, “Mom, you have a movement disorder.” I saw a doctor at Yale who was very good, very kind. He identified oromandibular dystonia (OMD) and antecollis. With muscle spasms, my jaw would not close and my head tipped down so I couldn’t keep food in my mouth. I followed treatment options and consulted several dystonia experts.

What has helped you cope?
A psychologist I was seeing suggested I attend a course on hypnosis where I learned how important it is for a body to relax and how to make the body relax. The problem was that hypnosis takes time to learn and you must practice every day. I said to the lecturer “I don’t have this kind of time.” I am 4’10” and was down to 75 pounds and needed something. He said to me, “It’s your body, your problem. Find the time!” And he left. I thought, Oh my God, he is so right. I decided to change my whole lifestyle. I now take breaks at work to meditate. I take walks. In between patients, I’m telling my body to relax. At home, I have meditation tapes, which I had listened to six times a day initially. This was my routine for a year. Now the hypnosis is inside me, I don’t even have to do the tapes, I know how to self hypnotize. I do yoga. I get botulinum neurotoxin injections every three months. When I work, I do not take medications. I take medication at night and over-the-counter pain meds here and there. I’m fortunate to have a good husband. If I do get down, I just need to talk to someone.

What is your life like today?
Over the years, I was embarrassed by my poor eating, voice difficulties, odd motions, and inability to commit to anything because I could not predict whether I would be well enough to function. Yet, I wanted to socialize, to be normal. Finally, I explained my condition to my friends. When we get together, I bring my own food or eat beforehand. When I can’t function due to pain or side effects, I explain. Three years ago, I reached a stage where I couldn’t count on my voice being available 24/7 due to spasms and botulinum neurotoxin side effects. I had to sell my practice after 30 years. I now work two days a week. I try to think positively, it makes all the difference. Positive thinking, botulinum neurotoxin treatments, medications, and distractions don’t always work but in combination they make life manageable and whole.

For more information about oromandibular dystonia and available treatments, visit: http://www.dystonia-foundation.org
When and how did your symptoms begin?
In 1992, I was 11 years old. I had a twitch in my head. I was diagnosed with Tourette's syndrome. I thought I would grow out of it. As I got older, other people noticed the progression, but I really didn’t. In 2007, it changed from a twitch to constant turning. It was creeping into my trunk and back. By 2009, something wasn’t right. I was working in Booking at the main jail in Sacramento. Co-workers complained I was looking over my shoulder at them. I started running into lockers, walls. Crossing the street was a struggle. I had people telling me over and over something was not right. But I’m stubborn. You have to drag me into the doctor’s office. I didn’t want to believe it was that bad. That was the hardest to accept, that I couldn’t control this. It had control of me.

How has dystonia affected your life?
My last day of work was in January 2011. Not working took a lot out of me. For two or three months, it was really hard. I was feeling useless. I wasn’t providing for my family. I would hide in the garage. I would worry about what I was going to do, what is going to happen. My wife almost had to drag me into the doctor’s office. I didn’t want to believe it was that bad. That was the hardest to accept, that I couldn’t control this. It had control of me.

How has dystonia impacted your family?
This affects my whole family. We have to accept that we don’t know what’s going to happen. It’s mentally hard on me and on my wife. My co-workers made donations, donated leave time at work when I had exhausted mine, and got us through some really tough financial times. I’ve been married to my wife for seven years, together for 10 years. We made a vow for better or for worse, and right now we say we’re in the “worse.” But we’re going through it together. I have to keep active, keep the right perspective, have responsibilities. Coaching my son’s t-ball team was a stepping stone to help me open up. That first year I coached from my wheelchair. I pulled the parents aside to explain, and found out one of the parents has a mother with dystonia. Right now the best thing I can do is to get the word out, and focus on getting back to work. I will never stop wanting to teach people. I think a lot about all the kids who are misdiagnosed.

How are you feeling about DBS?
There is not a single thing I would do differently. I did my research, and if this is what needed to be done to get me back to work, I was going to do it. I knew the risks. I looked at the pros and cons. I talked to people on the DBSforDystonia bulletin board. The struggle I’m having now with the infection, it’s uncertain. I have to roll with the punches and make the best of it. With the bulletin board, I didn’t expect to create relationships, all over the country. I have people across the nation that I’m able to listen to, learn from. It’s been awesome to have these people in my life.

Since this interview, Scottie’s infection required removal of all DBS hardware, including the leads. His symptoms began to return and he is waiting to be cleared to re-do the procedure. For more information about cervical dystonia and available treatments, visit: http://www.dystonia-foundation.org

PERSONAL PROFILE
Meet Scottie Roberts

Scottie Roberts is a 31-year-old corrections professional who was diagnosed with cervical dystonia in 2009. In 2011, he had deep brain stimulation (DBS). Persistent infection forced his physicians to remove the stimulator and wires in December. He is now waiting for clearance to have the hardware replaced. Scottie resides in California with his wife Whitney, sons Darien and Caiden, and young brother-in-law Andrew.
Planned Giving Provides Important Support for DMRF Programs

A number of bequests have provided essential support for DMRF programs, especially in years when fundraising proved challenging. This is the lovely irony of planned giving: gifts made possible by an individual’s death are lifesavers for the Foundation. These generous contributions help the DMRF move forward in its mission to find a cure and improve quality of life for those affected by this debilitating disorder.

For more information on how to make a bequest to the DMRF, consult your financial advisor or contact the DMRF at 312-755-0198 or dystonia@dystonia-foundation.org.