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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:
The DMRF is a leader in dystonia research, investing the time, effort, and resources needed to get results that improve lives. Recent discoveries such as the GNAL gene demonstrate that our efforts are making a difference; progress continues to accelerate toward a better understanding of this complicated, challenging disorder. On page 4, read how the newly uncovered GNAL gene is yet another piece researchers have added to the dystonia puzzle. Be sure to check out Science Officer Jan Teller’s new column, “Research Reality Check” on page 7. (Word cloud design courtesy of http://www.tagxedo.com)

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Foundation Update

Dear Friends,

Thank you, always, for your support. The Dystonia Medical Research Foundation (DMRF) remains absolute in our commitment to achieve our mission, and we would not be able to continue our important work without you.

Dystonia research has never been more active and productive, with new discoveries and data published regularly in the medical literature. The DMRF received grant and fellowship applications for the 2013 cycle from investigators all over the world. Our Medical and Scientific Advisory Council members are reviewing each application and will gather to discuss and rate them at the annual February meeting. In the meantime, the DMRF faces the challenge to fund as much outstanding research as possible with the limited funds at our disposal, and your support in the coming year is critical to our ability to do so.

Individuals and families impacted by dystonia have many opportunities to participate in research: clinical studies, registering as a brain donor, and patient registries. On page 4 you’ll read how readers of the Dystonia Dialogue played a critical role in one of the field’s latest discoveries, the identification of the GNAL gene for primary torsion dystonia. We are indebted to our readers for helping researchers by submitting medical histories and blood samples toward the ongoing effort to discover dystonia genes. Your commitment to research has once again led to another piece of the dystonia puzzle sliding into place. You are part of the global effort to find a cure.

Greater public awareness of dystonia is critical to everything we do. We thank those of you who use your stories to help others understand dystonia and convey the urgent need for medical research. Legislative advocacy through the Dystonia Advocacy Network is a targeted effort to educate our federal legislators and policymakers on the needs of the dystonia community. Our efforts in 2013 are underway. On page 6, read how you can join fellow advocates across the country and give dystonia a voice in Washington.

The dystonia diagnosis is a life-changing experience, and no one should have to navigate it alone. A network of support groups and online forums are available to help connect you with people who know what you are going through. We expect a number of new support groups to be up and running in 2013. Meetings and local symposia provide an outlet for accurate information from experts, and we will announce those events as dates are confirmed. Contact the DMRF with questions or for individual assistance to make sure you are aware of all the resources available to you.

We love to hear from our community, so please do not hesitate to share your thoughts on how we can serve you better—or what we are doing well. You can contact us by phone, email, or social media. We look forward to hearing from you.

Sincerely,

Art Kessler
President

Janet L. Hieshetter
Executive Director
Scientists from Mount Sinai School of Medicine and Beth Israel Medical Center have announced the latest discovery in dystonia research: a novel gene for primary torsion dystonia, which can include adult onset focal dystonia and early onset dystonia. The DMRF is proud to have partially supported this work.

The discovery was made through the collaboration of the molecular genetic laboratory of Laurie Ozelius, PhD at Mount Sinai and the clinical research team led by Susan Bressman, MD at Beth Israel as well as movement disorder specialists in the US and Canada. Ultimately, this work depended on the generosity of patients and families who volunteered in studies over a 25 year period. Many responded through advertisements placed in the Dystonia Dialogue to participate.

Published in Nature Genetics in December, the findings describe the GNAL gene, the first primary torsion dystonia gene that directly points to signal transduction pathways in the dopamine system as the origin of pathophysiology. Genetic testing in volunteers from two dystonia families revealed mutations in GNAL. Further screening of 39 additional families identified another six mutations in the gene.

This discovery will lead to genetic tests to confirm diagnosis and identify carriers. The research also unveils a new potential therapeutic target and the opportunity for developing new treatments.

“The technique used for the identification of the GNAL gene—called exome sequencing—is a powerful and efficient tool that will accelerate the pace of dystonia gene discovery and, consequently, our understanding of the pathways involved in primary torsion dystonia.” says Dr. Ozelius, who led the research team. “Any new gene offers the potential to develop new therapeutics, but because GNAL belongs to a well-studied pathway, other components in this pathway may also be targets for drug development,” adds Tania Fuchs, PhD, fellow member of the research team from Mount Sinai.

Primary torsion dystonia is characterized by repetitive twisting muscle contractions and postures that can affect the face, neck, arms, legs, or torso. Common symptoms include tremors, voice problems, or a dragging foot. Primary torsion dystonia may be adult onset or childhood onset. Symptoms can be focal, segmental or generalized. The disorder is dominantly inherited with reduced penetrance, making it difficult to predict which family members may be at risk without genetic screening.

Three additional genes associated with primary torsion dystonia have been identified: DYT1, THAP1, and CIZ1.

Art Kessler, President of the DMRF says, “As someone who has lived essentially my whole life with dystonia, I’m thrilled that we were able to help support this research to find GNAL. It’s especially exciting that this gene appears relevant to families impacted by adult onset and childhood onset dystonia.”

Susan Bressman, MD, Chair of Neurology at Beth Israel Medical Center, also noted “this study emphasizes the great utility of a truly long-term collaborative study of patients and families, including families spread out throughout North America. And the importance of investing in the clinical and laboratory infrastructure so the study of these patients and families can continue.”

Study authors also include scientists from Scripps Research Institute; Toronto Western Hospital; University of California, San Francisco; Emory University School of Medicine; Jefferson Hospital for Neuroscience; Massachusetts General Hospital; and Institute National de la Santé et la Recherche Médicale (INSERM).

Go to http://www.dystonia-foundation.orgresearch to learn more about the DMRF’s research efforts.
Difference4Dystonia Donors Earn DMRF $10,000 Bonus

For the second year in a row, hundreds of supporters joined together to help the DMRF raise funds in response to the Difference4Dystonia Challenge issued by a generous anonymous donor. Last fall, the donor offered to match, dollar for dollar, all new and increased donations up to $100,000. And if the goal was met before December 1, the donor promised a $10,000 bonus. The DMRF community dug deep and met the fundraising deadline, earning a total of $210,000 in support of the DMRF’s mission.

“We couldn’t be where are today without the amazing support of our community,” explains DMRF President Art Kessler. “We appreciate every single donor who participated in this campaign.” The funds raised are critically important to the DMRF’s ability to advance research and serve the community.

See “People on the Move” on pages 14–15 to read how volunteers across the country raised funds in support of the Difference4Dystonia Challenge.

Clinical Fellowship Training Program

Physician Education in Dystonia Advanced through Contributions by Ipsen Biopharmaceuticals, Inc. and Merz Pharmaceuticals

The DMRF’s clinical fellowship program helps train emerging physicians in the evaluation and treatment of dystonia through one-year fellowship grants, at $75,000 each. The fellowship grants have been funded by Ipsen Biopharmaceuticals, Inc. and Merz Pharmaceuticals.

“Despite improvements in research and treatment, dystonia patients may still have difficulty finding a doctor who knows how to diagnosis and appropriately treat their symptoms,” says Art Kessler, President of the DMRF. “Too often diagnosis is delayed or missed. Physician education is critically important.”

The DMRF is proud to announce our 2013 fellows:

**Aasef Shaikh, MD**
Fellowship Institution: Emory University
Mentor: H.A. Jinnah, MD, PhD

Now in its second year, the DMRF Clinical Fellowship Training Program focuses on clinical diagnosis and evaluation, ongoing patient care and management, pharmacotherapy with emphasis on neurotoxin therapy, and neurosurgical interventions. Training is patient-oriented and includes hands-on experience in clinics and professional workshops.

Andres Deik, MD, who pursued his 2012 clinical fellowship at Beth Israel Medical Center under mentors Susan Bressman, MD and Rachel Saunders-Pullman, MD, MPH, says this about his training: “Thanks to the DMRF’s clinical fellowship program, I have far expanded my expertise in the nature and treatment of the different dystonias. It has also provided me with insight on the existing gaps in the management of these conditions, giving me new focus for future research.”

This year’s fellowship grants will be awarded by July 1, 2013. For more information about the Clinical Fellowship Training Program, visit [http://www.dystonia-foundation.org/research](http://www.dystonia-foundation.org/research)
By becoming a legislative advocate, you can have a powerful effect on the laws and policies that affect your life and the lives of countless others in the dystonia community. Taking action is not as difficult or time-consuming as you may think.

The Dystonia Advocacy Network (DAN) is a collaborative, grassroots organization that brings people together to speak out with a single, powerful voice on legislative and public policy issues that impact individuals and families affected by dystonia.

The DAN develops and advances a legislative agenda that raises awareness of dystonia, educates policymakers, addresses patient care issues, and moves research forward. Your voice is needed to help get the message out. Dystonia advocates develop relationships with their legislative leaders to help them understand the challenges of living with dystonia; sharing your story is the power behind DAN’s success.

You can start getting involved today! Sign up to receive legislative alerts and be part of the network of volunteers reaching out to their legislators. From time to time, the DAN will issue a legislative alert via email to enlist your help on an important issue that affects the dystonia community.

This simple process will help you get started:

• Visit http://dystonia-advocacy.org/agenda to read a simple summary of the dystonia community’s legislative agenda.

• Know who your legislators are and establish communication with them. The DMRF can help you draft correspondence to your Members of Congress to articulate your dystonia story and ask them to support the dystonia community. This simple activity will introduce you and dystonia to your congressional offices, and make them more likely to act when you reach out to them about specific legislative issues.

• Sign up to receive alerts. Contact the DMRF at dystonia@dystonia-foundation.org or 312-755-0198.

• When you receive a legislative alert, promptly follow the simple instructions to take action.

• Finally, consider attending Dystonia Advocacy Day in Washington, DC, April 30–May 1, 2013. This empowering event provides the opportunity to meet with your legislators and share your story firsthand.

The DAN is also working to develop a program for volunteers to advocate at the state level to protect treatment and healthcare access. Please consider becoming a dystonia advocate. For more information, contact the DMRF at dystonia@dystonia-foundation.org or 312-755-0198.
“From Genes to Mechanisms”

You have heard this theory many times: It’s all in your genes. The key to solving the mysteries of human behavior and disease resides in the depths of our DNA (deoxyribonucleic acid). So, naturally you might conclude that research strategies that strive to link genes with diseases, including dystonia, will easily provide answers and gradually lead to cures.

Members of the dystonia community occasionally ask, “So, now that we have all these dystonia genes, finding the cure should be easy, right?”

The truth is, gene discovery is only the beginning of a long and arduous process of deciphering disease mechanisms. True, it all starts with genes, but single genes rarely, if ever, are responsible for complex diseases. The dystonias are no different. Clinicians distinguish many forms of dystonia that are now linked to known mutated genes. Over the years, the DMRF has funded genetic research that has resulted in breakthrough discoveries, for example the DYT1 or DYT6 genes. Each gene discovery is a great achievement that gives more insight into the inner workings of the dystonia brain. But all these genes are very different, and we don’t yet know what they do or how they cause dystonia. In most cases, many years pass before researchers can connect the dots and understand the real role of a faulty gene.

Should we be skeptical about the value of genetic research then? Certainly not. We just have to understand that finding a gene provides a target for research; it anchors our efforts in the right place. The rest is hard work and sheer opportunity. That’s why we all got so excited in late 2012 when an international team from New York led by Laurie Ozelius identified another gene for primary torsion dystonia. (Read more about this discovery on page 4.) The gene, esoterically called GNAL, is a blueprint for a protein that is strategically linked to other known proteins. GNAL might just deliver what we have been waiting for: a protein with relatively known function placed in an area of the cell where we can start cracking the mechanism of dystonia. What is even more fascinating and promising is that the proteins that hang around GNAL have already been implicated in dystonia and can potentially be easily “manipulated” with drugs—drugs that already exist but have never been tested for dystonia. Somehow, the GNAL discovery may prove once again that it’s not all in our genes, but that answers may be near after all: this single gene may help us understand the complex cellular processes that malfunction in dystonia, perhaps pointing us to additional proteins whose functions we do not yet understand. So, the plot thickens. Exciting times are ahead.
The latest physical therapy (PT) techniques used to treat dystonia are based on an advanced understanding of the neuromuscular system and how it is affected by dystonia. When provided by a well-trained, experienced physical therapist, these newer techniques can be vitally helpful to patients.

While PT cannot cure or eliminate dystonia, it may improve function by reducing involuntary muscle contractions, postures, and pain. This is important because the long-term stress on muscle, tendons, and ligaments associated with dystonia have implications for the entire body including increased risk for premature spinal disc degeneration, damage to connective tissues and joints, and diminished exercise capacity. As a treatment option, PT can help limit some of the long-term complications of dystonia.

PT is most effective when combined with other medical interventions such as botulinum neurotoxin injections, deep brain stimulation, or intrathecal baclofen therapy. PT is not a "quick fix" and requires significant commitment from the patient. Dystonia symptoms can change over time, and ongoing re-evaluation and modification of a PT program may be needed.

**How to Select a Physical Therapist**

Not every physical therapist possesses the necessary experience or skills to treat dystonia. As a first step to finding someone qualified, ask your movement disorder specialist for a referral or check with another dystonia patient, a local dystonia support group, or physiatrist (Doctor of Physical Medicine/Rehabilitation). It may be helpful to seek out a physical therapist with an advanced degree such as a Doctorate (DPT) or board certification as an Orthopedic or Neurologic Specialist.

That said, there are gifted, experienced physical therapists without special designations who provide excellent care. Ideally seek out a physical therapist who has experience with dystonia, possesses advanced manual skills, and can offer one-on-one sessions.

The American Physical Therapy Association (www.apta.org) offers a “Find a PT” link on their website that allows users to search by geography as well as by specialty.

**Treatment Options**

Similar to other medical consultations, a PT appointment starts with an evaluation. The goal is to learn the patient’s chief complaints, identify needs and goals, and understand pertinent background information. Based on this, a treatment plan is developed that may include any combination of the following:

**Therapeutic Exercise:** A physical therapist may recommend aerobic conditioning, strengthening, and stretching, modified appropriately, to improve fitness and reduces stress. Aerobic exercise may include walking, stepping or elliptical machines, swimming or aquatic exercise, and bicycling. An experienced physical therapist may offer suggestions for overcoming some of the limitations associated with dystonia, such as swimming with a snorkel or using an adult tricycle as a safer alternative to a bicycle. To improve strength (especially of the core muscles) weight bearing and non-weight bearing exercises may be recommended. Tools such as foam rollers, therapy balls, light resistance bands, and cable systems may be used to customize exercises to the individual patient. Like other modalities, stretching can be beneficial when performed appropriately but can easily aggravate dystonia symptoms if used inappropriately.
**Manual Techniques:** Manual therapy is a clinical approach using skilled, specific hands-on techniques to lessen pain, improve range of motion, reduce inflammation, induce relaxation, and improve function. Advances in manual therapy represent some of the newest, most promising approaches for dystonia patients. One technique known as *functional mobilization* uses a combination of passive and active techniques. First, the therapist uses his/her hands to identify an area of tightness, then applies sustained pressure to calm it (the passive component). After the muscle releases, the patient is cued to move in a certain pattern (the active component). With repeated sessions and follow-up exercises, the patient can achieve more controlled movements. Other techniques might include strain-counterstrain, pressure bio-feedback, muscle energy, active release, and gentle manual traction.

**Neural Muscular Re-education:** This involves retraining the muscles to contract and relax properly through improved posture, gait, balance, and body mechanics. By making patients aware of optimal posture and form, it can help them use their muscles more efficiently with less fatigue and pain. For example, swinging the arms while walking can relax overactive muscles in the upper back, thereby reducing tension in the neck. Kinesiotaping is a rehabilitative taping technique that provides support and stability to muscles and joints without restricting range of motion. For dystonia, it is used to inhibit overactive muscles while helping to support and retrain those that are underactive.

**Supportive Modalities:** These include the familiar use of heat, ice, and ultrasound as well as transcutaneous electrical nerve stimulation or electrical stimulation. A newer device is *cold laser therapy* or *low level laser therapy*. This treatment uses specific wavelengths of light to accelerate the tissue healing process. All of these are used to complement other treatments and help eliminate pain, reduce swelling, reduce spasms, and increase functionality.

**Aquatic Therapy:** Aquatic therapy is a gentle way to maintain or regain range of motion, build strength, and perform aerobic exercise. People with dystonia may find it easier to exercise in water because the added buoyancy supports the body, helping to compensate for balance, posture, mobility, strength, and flexibility issues. The water also provides gentle resistance to increase strength and endurance.

**Activity, Environment, and Ergonomic Considerations:** A physical therapist may identify conditions in the patient’s environment that reduce or aggravate the dystonia and make recommendations accordingly. In addition, sensory tricks are simple gestures that help some dystonia patients temporarily decrease or eliminate muscles spasms or postures, and the use of physical devices (orthotics) may be explored.

**Future of PT for Dystonia**
PT is not a cure for dystonia, but it may help lessen symptoms. Physical therapists with the experience and skills necessary to treat dystonia are limited, but PT programs now require students to obtain doctoral degrees and this bodes well for a future in which a greater number of physical therapists have the training to help movement disorder patients. PT and rehabilitation are areas of great interest in dystonia research because of the potential to improve function and quality of life.

*For the full-length version of this article, go to: http://www.dystonia-foundation.org/dialogue*
There are forms of dystonia that are known to be genetically inherited and forms that may or may not have a genetic component—researchers simply cannot confirm or rule it out at this time. To date, several gene mutations have been linked to specific forms of dystonia. Below are answers to common questions about the genetics of dystonia.

What Forms of Dystonia are Inherited?
Individuals who have a form of dystonia that is known to be genetic may pass the disorder on to their biological children. These include primary torsion dystonia (which may include childhood and adult onset, generalized and focal symptoms), dopa-responsive dystonias, myoclonus dystonia, paroxysmal dystonias/dyskinesias, rapid-onset dystonia-parkinsonism, X-linked dystonia-parkinsonism, and others.

What about Secondary Dystonias?
For individuals whose dystonia is secondary to an injury to the brain or nervous system (including exposure to certain medications or stroke), their children are in all likelihood not at increased risk of inheriting a known dystonia-causing mutation. Secondary dystonia can also occur as a symptom of various genetically-caused diseases, in which case the inheritance patterns will vary depending on those diseases.

Who is at Risk of Developing a Genetic Dystonia?
The degree of risk varies with the form of dystonia and what is known about inheritance patterns of those specific forms. Most dystonia-causing genes are inherited in a dominant pattern, which means that only one parent needs to have the mutation for a child to inherit the mutation and develop dystonia. However, most people who inherit dystonia-causing mutations never develop symptoms. This phenomenon is known as reduced penetrance. A genetic counselor can help families understand who may be at risk of inheriting a dystonia-causing gene mutation, but at present there is no way to predict who will develop dystonia.

Some people develop dystonia due to a gene mutation without any apparent family history. Furthermore, there are families in which many members are diagnosed with dystonia but no specific genetic mutation has yet been identified.

Who is a Candidate for Genetic Testing for Dystonia?
Genetic testing is available to determine if an individual has a specific gene mutation known to cause dystonia. The first step is to consult a genetic counselor trained to educate families on the likelihood and risks of inherited diseases.

Genetic testing is only available for the specific forms of dystonia for which gene mutations have been identified. An individual can still have dystonia without testing positive for a dystonia mutation. Testing negative for known dystonia mutations does not necessarily negate a clinical diagnosis. As additional dystonia gene mutations are discovered, genetic testing will become informative for a wider spectrum of dystonia patients.

A genetic counselor can help individuals and families understand what testing may be appropriate for them. Testing may be recommended for adults and children who exhibit clinical symptoms that resemble the forms of dystonia for which gene tests are available. This includes, but is not limited to, some primary torsion dystonias, early onset dystonias, dopa-responsive dystonias, myoclonus dystonia, rapid-onset dystonia parkinsonism, and X-linked dystonia parkinsonism.

Adult biological relatives of individuals who have a genetic form of dystonia may also be eligible for testing. Genetic testing is not recommended for children who do not exhibit symptoms, even if a biological relative tests positive for a dystonia mutation. Options exist for individuals with specific genetic forms of dystonia who wish to decrease the likelihood of passing the mutation.
Dystonia 101

Dystonia can be a confusing disorder to understand. It never hurts to brush up on the basics:

- Dystonia is a neurological disorder. It affects the nervous system’s ability to control voluntary muscle movements.
- Dystonia does not affect smooth muscles, such as the heart.
- There are many forms of dystonia. It can affect a single body area or multiple muscle groups.
- Dystonia can exist on its own, or be a symptom of another neurological or metabolic disorder.
- In primary dystonia, the affected person has no other neurological symptoms and the dystonia is known or suspected to be genetic.
- In secondary dystonia, the symptoms can be attributed to an insult to the brain such as physical trauma, drug-exposure, or other diseases/conditions.
- People with secondary dystonia often have other neurological symptoms, some of which may affect more than just muscle movement.
- Each case of dystonia is classified by: the age symptoms started, whether it can be classified as primary or secondary, the body parts affected, and the presence or absence of other disorders.
- Treatment options include oral medications, botulinum neurotoxin injections, surgery, and less invasive methods such as physical and occupational therapy, and relaxation practices.
- Stress does not cause dystonia, but symptoms may worsen in stressful situations.

For more information, visit http://www.dystonia-foundation.org

How Do I Find a Genetic Counselor?
Individuals may locate a genetic counselor by consulting their movement disorder specialist or health insurance provider. Several professional organizations can also provide contact information for genetic counselors such as GeneTests: National Center for Biotechnology Information and the National Society of Genetic Counselors.

For more information on dystonia, including genetics, visit http://www.dystonia-foundation.org
Double Trouble
*siblings cope with dystonia*

Although they vigorously disagree over who was first to coin the analogy, teen siblings Jack and Lydia Nathans use a common childhood game to describe how dystonia affects them. Jack explains: “My body plays telephone with itself. If my brain tells my foot to move, by the time the signal gets down there, the message gets misinterpreted and it’s telling my toes to curl instead.”

Jack, age 17, and Lydia, age 15, are both diagnosed with dystonia, yet their stories and symptoms are quite different.

**Jack**

Jack was the first to develop symptoms, though at first it wasn’t clear anything was seriously wrong. His handwriting began to suffer in second grade. An occupational therapist suggested he simply switch hands to write, and the issue seemed to be resolved. During the third grade, blistered feet and a slight limp quickly deteriorated into a complete inability to walk. He was eventually diagnosed with dystonia. Lydia recalls watching her brother struggle: “It was pretty terrifying. He would be in so much pain he would be screaming and we didn’t know what to do.” Jack’s symptoms became severe, affecting his arm, back, legs, and feet.

“I learned about DBS [deep brain stimulation surgery] by watching the movie *Twisted,*” Jack says. “We were at a screening and I turned to my dad and said: *I want that.* I had my first surgery on my parents’ wedding anniversary in 2007. What did I have to lose?”

“Just your hair,” quips Lydia, as if on cue.

The surgery greatly reduced Jack’s dystonia, but mechanical setbacks—a broken wire, a faulty stimulator—delayed his full recovery. Now that those issues have been resolved, he is doing well.

**Lydia**

Meanwhile, just as Jack’s dystonia began to settle down, Lydia’s flared up. “I don’t want to say I’m happy Jack got dystonia first,” she says, “but I know how it looks from the outside, when it looks like someone is getting special attention. And I also know what it’s like to be that person who needs that kind of help. Dystonia has helped me grow and be more mature.”

Lydia’s symptoms affect her hands and cause pain in her lower back. So far the dystonia is reasonably controlled with medication. “I love writing, making lists. Sometimes I can’t do that—I’ll have a bad day, and it’s frustrating. So, when that happens, I think about what I can do, not what I can’t.”

**Coping**

Jack also credits having a positive attitude as a way he copes with dystonia: “I’ve always been able to stay optimistic. If you tell yourself everything is going to be ok, you’ll feel better. If you keep telling yourself this is terrible, you’re setting yourself up for failure.”
Jack is cautious about how much he shares with friends about his dystonia experience, carefully gauging what he thinks each friend can handle. “People may not always get what you are going through,” he explains, “but as long as you know what’s going on and understand it, it’s ok.”

Lydia struggles for balance in how she deals with dystonia in social situations: “I talk a lot. Sometimes I look back and realize I went into more detail than I had to. Think about how much you want to tell people about dystonia, and only tell what you are comfortable with.” She offers further advice for coping at school: “If you need something—if you can’t keep up with the class or someone is giving you a hard time—talk to your teachers one-on-one. I email my teachers so I don’t have to bring things up in the middle of class. Guidance counselors can help too.”

Taking Action
The Nathans family is active in raising funds in support of the DMRF and promoting dystonia awareness. Parents Sandra and Robert Nathans lead the New Jersey Dystonia Support Group and the family has hosted numerous fundraisers. Lydia uses social media to promote awareness and encourage support for the DMRF. She has also led several fundraisers for the DMRF in partnership with her school.

Lydia sums up her family’s dystonia experience like this: “Even though dystonia can be the worst thing, it’s also been one of the best things. I learned to value what I can do. I also feel an obligation to help.”

Ideas for Promoting Dystonia Awareness
• Share a link to the DMRF website on your Twitter or Facebook page.
• Talk to a favorite teacher or your guidance counselor about doing a fundraiser for dystonia research.
• Offer to do a presentation about dystonia for your science class.
• Come up with a two-sentence answer to educate people who ask about your symptoms.
• Invent your own creative way to get the word out.

How Can I Participate in Dystonia Research?
You can support dystonia research discoveries in more ways than one. Consider these opportunities to make an impact:

Ask your Doctor: Your movement disorder specialist may be participating in a clinical trial that needs volunteers.

Search Online for NIH Studies: Search for dystonia clinical studies supported by the National Institutes of Health at http://www.nih.gov/health/clinicaltrials/

Register as Brain Donor: Individuals with dystonia as well as unaffected family members are urgently needed. See page 17 for more information.

Join a Patient Registry: Patient registries help researchers better understand dystonia by collecting information directly from volunteers who are diagnosed. The DMRF is proud to support the following patient registries:
• Global Dystonia Registry https://globaldystoniaregistry.org/
• Dystonia International Patient Registry http://www.dipregistry.com/

Donate: Financial contributions to the DMRF support cutting-edge research by the world’s foremost thought leaders and up-and-coming experts. Use the envelope enclosed in this newsletter to make a gift or donate online at http://www.dystonia-foundation.org/donate
PEOPLE ON THE MOVE

The DMRF is deeply grateful for our grassroots volunteers who work year round to promote dystonia awareness and fundraise for medical research. Every effort and every volunteer makes a difference! We appreciate your support.

Roberta Senzel of Maryland and family held a jewelry fundraiser and raised nearly $700 in support of dystonia research and DMRF programs.

Patricia Evangelista of Florida created beautiful watercolor greeting cards to promote dystonia awareness and raise funds for research. She raised just under $300!

Kayla Pounds of Ohio partnered with Dance Bag of Westchester and sold hand-knitted scarves to benefit the DMRF, raising over $1,750. Special thanks to Debbie Meiners for her essential role in developing this unique fundraiser. Kayla is diagnosed with stiff person syndrome (Moersch-Woltmann syndrome) with dystonic symptoms.

Denise Gibson and Donna Suiter of the Dystonia Support Group of Spokane, Washington spoke before classes of graduate students from the Eastern Washington University Masters of Science in Communication Disorders program and the Washington State University Master of Arts in Speech & Hearing Sciences program. The pair shared their experiences with dystonia to inform the students about the early signs of dystonia and need for increased awareness within medical communities.

In November, DMRF Treasurer Mark Rudolph and daughter Rachel Rudolph participated in the Calabasas Classic 5K to raise funds for dystonia research. This is Mark’s ninth year as a runner and Rachel’s fifth. Rachel’s friend Lily Braunstein joined in and together they raised hundreds of dollars for dystonia research and raised much-needed awareness.

Following 16 weeks of training, Rodrigo Gomez ran an astounding 50 miles in 9 hours 48 minutes to finish the Nashville Ultra Marathon in November. Rodrigo ran in honor of his mother Goya Rubio de Gomez, a resident of Mexico who for two decades traveled to the US for dystonia treatment.

Leader of the Dystonia Support and Advocacy Group of San Diego County Martha Murphy and member Bette George represented the dystonia community at Jobtoberfest, an annual job fair for individuals with disabilities. In January, Martha and group member D.C. Hathaway appeared on Drew Schlosberg’s Union-Tribune Community Spotlight online radio show to inform listeners about dystonia.

MaryRae Nee of Pittsburgh partnered with Three Rivers Confections to raise funds in support of the DMRF through online sales of their Fudgie Wudgie treats. To order, visit http://www.fudgiwudgie.com/#!_dystonia and use promo code CUREDYSTONIA.

In September, Timmy Samec competed in the Pocono Ironman 70.3 in honor of his friend and fellow triathlete Pat Brogan. He raised over $1,200!

Kevin Oetjens ran the 2012 Bank of America Chicago Marathon, raising over $1,000 in honor of his brother Mark and cousin Zach. This was Kevin’s third marathon, and the first he ran in his hometown of Chicago.
A supportive network of family and friends cheered on **Misty Mullenbach** in the Lady Speed Stick Half Marathon in Phoenix. Misty raised nearly $1,000 in honor of her brother Ryan and her niece Sarah.

**Lisa Barbadora** ran the Philadelphia Marathon in honor of her lifelong friend Richard who has dystonia. She successfully met her goal to raise $200 per mile for more than $5,000!

**Karen Rodriguez** considered herself a half-marathon runner, but this year decided to take it to the next level at the Disney Space Coast Marathon in Florida. She raised over $1,700 in honor of sisters **Carrie Siu Butt** and **Suzie Siu Butt**.

**April and Dave Bradbury** hosted a pasta dinner fundraiser at The North Italian Club in Meriden, Connecticut which raised over $4,000. Guests were treated to gourmet food prepared by local chefs and live entertainment.

**The Farber family** hosted the 2012 Chicago Basket Bash in October. More than 120 friends and supporters came together to raise over $19,000 in honor of the late **Shari Farber-Tritt**, whose experience with dystonia was featured in the 2006 documentary, *Twisted*.

**Pat Brogan** and his committee of family and friends have hosted the Help Find a Cure 4 Dystonia pub event in Hazleton, Pennsylvania each holiday season. The event featured an extensive raffle, special guests, and lots of fun. This year’s event raised over $10,000. Pat shared his story with secondary dystonia and deep brain stimulation in the documentary film *Twisted*.

**Kay Cooksey** raised $2,575 in support of the DMRF through her annual writing campaign to family and friends. Kay’s teenage granddaughter, **Maddie Paolero** is diagnosed with dystonia.

**Nancy and Larry Present** of St. Louis, Missouri hosted the Annual “Moving Forward” Dystonia Walk-a-thon, raising over $5,500 for research and boosting local awareness.

**Rebecca Sharp** raised over $1,000 through a writing campaign in honor of her father **Timothy Hornsby**, who is diagnosed. The Hornsby family founded the Dothan, Alabama Dystonia Support Group and has hosted numerous Dystance4Dystonia events.

**Sheila Williams** organized the Arlington, Texas Dogs4Dystonia Walk in October at River Legacy Park. Over 30 individuals attended the event, which generated more than $10,000 in support of the DMRF.
Dystance4Dystonia Runners Make Strides Toward the Cure

Since 2010, runners participating in Dystance4Dystonia have competed in races across the country to raise funds for research and promote awareness. Each runner who embarks on a 5K or marathon is part of the national Dystance4Dystonia team, which is growing in members each year. Whether a runner raises $5,000 or $500, every dollar is multiplied by the efforts of fellow team members. Cumulatively, Dystance4Dystonia runners have raised tens of thousands of dollars in support of urgently needed research and DMRF programs.

Dystance4Dystonia runners often compete in honor of family members and friends who are diagnosed. For example, six-year-old Gigi Kenney has run two 5K races in honor of her mother who has dystonia. Denise Gibson competed despite being diagnosed herself with dystonia affecting her neck and vocal cords muscles.

To learn more about Dystance4Dystonia runners and what inspires them to train and compete on behalf of the DMRF, go to http://www.dystonia-foundation.org/dystance

You can be part of the Dystance4Dystonia team! If you are a runner and can lend your talents to the fight against dystonia, contact the DMRF at 312-755-0198 or dystonia@dystonia-foundation.org

Dystance4Dystonia is a program for volunteers interested in participating in local marathons, runs, and/or walks in support of the DMRF.

Dogs4Dystonia Dog Walks are a fun way to bring family, friends, and the community together in support of a good cause.

For more information about Dystance4Dystonia or Dogs4Dystonia, contact the DMRF at 312-755-0198 or dystonia@dystonia-foundation.org

Expert Speakers Present Diverse Topics at Regional Symposium

The Mid-Atlantic Regional Dystonia Symposium took place October 21, 2012 in Rockville, Maryland. Topics addressed include an overview of dystonia and treatments, deep brain stimulation, genetic counseling, traditional Asian medicine techniques, research, and others. Attendees from throughout the region gathered to participate.

General session speakers included Stephen Grill, MD, PhD, Parkinisons & Movement Disorders Center of Maryland; Zachary Levine, MD, FAANS, Holy Cross Hospital; Jan Teller, MA, PhD, Dystonia Medical Research Foundation; and Carol Smith, LPC.

Many thanks to all the stellar speakers who volunteered their time, talents, and expertise.

Sincere thanks also to the planning committee for their essential assistance with this important event: Virginia Foster, Art & Bernice Hindle, Elena Nacamuli, Raman Patel, Marcie Povitsky, Sally Presti, Paula Schneider, Roberta Senzel, and Hunter Webster.
Teams of Researchers Discover Gene for Spasmodic Dysphonia

Christine Klein, MD, Professor of Neurology and the Schilling Professor of Clinical and Molecular Neurogenetics at the University of Lubeck, Germany, was the principal investigator of an international study group that has identified the DYT4 gene associated with a dominantly inherited form of dystonia called whispering dysphonia. Dr. Klein is a past member of the DMRF’s Medical & Scientific Advisory Council and guest author for the Dystonia Dialogue.

Whispering dysphonia caused by the DYT4 gene causes uncontrollable muscles spasms beginning in the voice and also affecting the neck muscles.

Expanding on previous work on DYT4, Dr. Klein and her team conducted genome-wide linkage analysis in 14 members of a large Australian family followed by genome sequencing in two individuals. The findings were published online in December in the Annals of Neurology along with a second article describing another research team’s work on this gene, led by Henry Houlden MD, MRCP, PhD, Professor of Neurology and Neurogenetics, The National Hospital for Neurology and Neurosurgery Institute of Neurology Queen Square, London.

Symptoms of spasmodic dysphonia (sometimes referred to as laryngeal dystonia) include involuntary contractions of the vocal cords muscles, causing interruptions in speech and affecting voice quality. One of the most characteristic features of spasmodic dysphonia is the patterned, repeated “breaks” in speech, causing a strangled-sounding or breathy voice. Treatment may include botulinum neurotoxin injections, voice therapy, and breathing techniques.

The DMRF congratulates Dr. Klein, Professor Houlden, and the outstanding investigators who contributed to this exciting discovery. Each new gene associated with dystonia provides a potential new target for therapeutics and adds to science’s growing understanding of all the dystonias.

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
Look for new interviews with Zachary Weinstein and Alyssa Dver, authors of Never Look Down.

Twitter: http://twitter.com/dmrf

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

Facebook: “Generalized Dystonia Support Forum”
Special thanks to volunteer administrator Paula Schneider.
https://www.facebook.com/groups/dmrf.gen/

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

Facebook: “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/

Facebook: “Support4Parents of Children with Dystonia”
Many thanks to volunteer administrators Carol-Ann Peralta and Dena Sherry.
https://www.facebook.com/groups/support4parents.dmrf/

Facebook: “Caring4Parents with Movement Disorders”
Email contact@dystonia-foundation.org to join.

Online Dystonia Bulletin Boards
http://www.dystonia-bb.org/
Many thanks to volunteer moderators Bob Campbell, Jeff Harris, and Linda Walking Woman.

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 DMRF support group in your area, go to http://www.dystonia-foundation.org/supportgroups
Meet D.C. Hathaway

D.C. Hathaway was a successful singer, musician, and radio DJ before developing spasmodic dysphonia in 1996. In 2012, D.C. performed with recording artist Juice Newton and DMRF Awareness Ambassador Billy McLaughlin in a benefit concert to support the DMRF. He resides in California.

What were your earliest symptoms and how were you diagnosed?
I was hospitalized for four weeks after a near-fatal car accident in 1996. Three weeks after I was sent home, I went to my band rehearsal and my voice just wasn’t the same. I was living in Florida at the time. I can’t tell you how many doctors I saw. They all said there is nothing wrong; it’s all in your head. All I knew was I wasn’t getting better. My voice was my livelihood. I was always out doing shows—I was singing almost every night. I lost my career, my wife of 20 years. It was an awful time. I moved to California for a fresh start. Nine years after the symptoms started, I was diagnosed with spasmodic dysphonia by an otolaryngologist [ear, nose, and throat specialist]. The doctor explained it’s a form of dystonia. Dystonia was a foreign language I had to learn.

What treatments have helped you?
Botulinum neurotoxin injections are the only thing that has somewhat worked for me. Without treatment, every word I speak sounds like I am choking. With stress, my voice gets really bad—almost to a whisper. I’m not sure how my voice will be from one day to the next.

How have you adapted to the effects of dystonia on your career and music?
Spasmodic dysphonia turned my life upside down. I couldn’t make a living any more. I was working as a DJ, and the station was forced to let me go. I don’t blame them. Main street radio won’t touch me with this voice. My passion for music is even more now, because I know what I have lost. I can’t sing anymore, but I can write songs. I just need other people to sing them for me.

Or I write instrumentals now. I started a radio show. Every day I take a moment to talk about dystonia. I’m having fun with it. I’m killing two birds with one stone with the radio show: I get to do music and promote dystonia awareness. Listeners ask about my voice all the time, and I take advantage of every opportunity to explain.

What helps you cope?
My wife Gloria Mucci gives me so much support. Billy McLaughlin has been the biggest source of inspiration for me. Our stories are so similar, just different parts of the body are affected. He said to me once, at a DMRF symposium in Pasadena, “Quitting is not an option.” It was a big wake up call. Another huge source of support has been the San Diego Support Group. After that first support group meeting, I was hooked. Martha Murphy [leader of the support group] has been my guardian angel. She put me in touch with her attorney, who worked with me for four years to get disability benefits.

Why is promoting dystonia awareness so important to you?
I am on this crusade until we find a cure. I have no idea what else to do. I hope there is a cure before I die. There is no other way for me to look at it than, “quitting is not an option.”

“D.C. Coast to Coast” airs live Monday through Saturday, 3:00–9:00 ET at http://www.livestream.com/ddcoasttocoast

To learn about spasmodic dysphonia and other focal dystonias, visit http://www.dystonia-foundation.org
What were your earliest symptoms and how were you diagnosed?

My first symptoms were so subtle I brushed them off. I sought help in 2002 when my eyes started blinking so rapidly it was making me blind. I was diagnosed with benign essential blepharospasm by an ophthalmologist. I was a nurse and I had never heard of this. That day the ophthalmologist made me an appointment at a university eye institute. Meanwhile the symptoms changed to forceful spasms. I would have to physically pry my eyes open. The eye institute confirmed it was blepharospasm. I had botulinum neurotoxin injections that day. Even though they told me it wasn’t a cure, I thought I was cured. After three months, the blinking started again. I saw several neurologists over the years, but never had such excellent results since that first time.

In 2009 I started feeling like my head was shaking internally. My right arm started shaking; I would drop whatever I was holding. I saw a movement disorder specialist. Right away she suggested I have spasmodic dysphonia—my voice was hoarse. It had been like that for years—I didn’t even think of it. The doctor also noticed I had a head tilt. My neck did ache a lot and I had a frozen shoulder. Now I know I have segmental dystonia: my eye muscles are affected the most severely, and neck and vocal cord symptoms are mild so far.

What helps you cope?

I was always so independent. I was always there for other people. It was hard for me to ask anyone for help—even my husband. I thought my life was over, but it’s far from it. I have a routine I have to do every morning for my eyes and voice. It takes 30 minutes before I can assess how my eyes are going to be that day. For my neck and shoulders, I do gentle stretching, relaxation, massage, heating pad and, over-the-counter pain medicines. In the beginning I remember feeling profound sadness. I was grieving the old “normal.” I can see that in hindsight, but when you’re in it you can’t see it. The physical changes were hard. I felt like people looked at me differently. I was lucky that my worst symptoms were in my eyes—I could wear big dark sunglasses and people couldn’t see my eyes spasming and judge me for it.

What guidance do you have for others coping with dystonia?

Help and support are all around you. You are stronger than you realize. You will adapt, you will learn. A big part of my acceptance is knowing I’m not alone. If I’m having a bad day, I can go online and see that someone is having a worse time than me. I can give support to others. Getting and giving support at the same time. Seeing other people in person at a local support group meeting validated me. It validated that we’re not insane, this is for real.

What do you wish the public understood about dystonia?

We need a cure. Dystonia is profoundly life-changing, whether it’s focal, segmental, or generalized. Those of us who develop dystonia later in life know what it’s like to not have it. Those who have had dystonia since they were young kids may not know any different. I would love to see a cure. I would love for people who haven’t experienced life without dystonia to get that chance.
Mother and Son Shine Light on Invisible Symptoms

New Book Empowers Children with Dystonia

When meeting 13-year-old Zachary Weinstein, it’s not obvious that he has dystonia. He doesn’t have overt postures or use a wheelchair. However, the moment he extends his arm to shake hands it becomes clear he struggles to control the movements of his body. This simple gesture triggers exaggerated, flailing movements in his arm that he cannot hold back. Zachary is now accustomed to the shocked looks—and careless comments—from strangers who are caught off guard. His mother, Alyssa Dver, feels a pang of mixed emotion each time a routine social situation requires yet another explanation for why Zachary doesn’t move like everyone else. Suddenly, the symptoms that were invisible a few moments ago become as conspicuous and awkward as the proverbial elephant in the room. Dystonia? What is that?

It’s not uncommon for dystonia to remain somewhat subtle. Especially in children, the symptoms may be mischaracterized as clumsiness, not paying attention, or bad manners. Zachary and Alyssa have written a book entitled *Never Look Down* in partnership with the DMRF so that other children like Zachary feel empowered, not diminished, by this often misunderstood illness.

*Never Look Down* provides newly diagnosed children and their families a simple way to understand dystonia: what the symptoms may be, how it can affect everyday life, and resources available to help them cope.

If a child in your family is impacted by dystonia, you may be eligible to receive a copy of *Never Look Down* at no charge. Supplies are limited so please contact the DMRF at 312-755-0198 or dystonia@dystonia-foundation.org

*Look for interviews with Zachary and Alyssa about living with dystonia and Never Look Down on the DMRF’s YouTube Channel: http://www.youtube.com/facesofdystonia*

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Become a Brain Donor and Make a Difference

You can make a significant contribution to dystonia research by registering as a brain donor. The study of brain tissue provides researchers with unique opportunities to develop improved treatments, gain a more thorough understanding of the disorder, and, ultimately, advance closer to finding a cure.

The DMRF is part of the Dystonia Brain Collective, a collaborative program among several dystonia patient organizations, in partnership with the Harvard Brain Tissue Resource Center (HBTRC) at McLean Hospital in Belmont, Massachusetts. Individuals residing in the United States who are diagnosed with dystonia as well as unaffected family members are encouraged to register.

The registration process is easy. There is no cost involved in being a brain donor. The recovery process does not interfere with funeral or memorial services or affect the outward appearance of the donor.

Individuals considering brain donation must discuss their intentions with family members because the next-of-kin or legal representative will be responsible for notifying the HBTRC when the donor passes away as well as granting permission for recovery to take place and for the donor’s medical records to be sent to the HBTRC. The recovery process must be completed within 24 hours from the time of death, so time is of the essence.

If you wish to learn more about brain donation, the DMRF is happy to mail you information about the program and answer any questions you may have. Contact DMRF headquarters at 800-377-3978 or email Martha Murphy, Brain Bank Liaison, at brainbank@dystonia-foundation.org. You may also visit the DMRF website to download an informational brochure and registration form: http://www.dystonia-foundation.org/brain