



MYASTHENIA GRAVIS
FOUNDATION OF AMERICA, INC.

Foundation Focus

News about myasthenia gravis for patients, family and friends

Spring 2014

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Follow the MG Walk Campaign:

[Facebook.com/MGWalks](https://www.facebook.com/MGWalks)

[Twitter.com/MG_Walk](https://twitter.com/MG_Walk)

Upcoming Myasthenia Gravis Spring Walks

- Georgia - March 15
- Tampa Bay - March 16
- Tallahassee - March 22
- Jacksonville - March 23
- South Florida - March 30
- New Orleans - April 5
- Orlando - April 27
- New England - May 3
- TriState (NYC) - May 31
- Minnesota - June 21



355 Lexington Avenue, 15th Floor
New York, NY 10017-6603
(800) 541-5454 | (212) 370-9047 fax
mgfa@myasthenia.org | www.myasthenia.org

This publication is intended to provide the reader with general information to be used solely for educational purposes. As such, it does not address individual patient needs and should not be used as a basis for decision making concerning diagnosis, care, or treatment of any condition. Instead, such decisions should be based upon the advice of a physician or health care professional who is directly familiar with the patient.

MGFA

Message from Chairman Sam Schulhof

It's hard to believe we are already three months into the New Year with the 2014 National Conference almost upon us! Time does fly when you are busy and we at MGFA have been, as you will read about in this issue of the Focus.

The MGFA Board of Directors has been looking at governance and organizational design and staffing issues with an eye on how we might more effectively identify, develop and deliver services to the MG community in a cost effective and efficient manner while providing the Foundation with the flexibility to meet the changing needs of the MG community. With that in mind, I am pleased to announce the first step in that direction with the hiring of Kathleen Brown, MPA, CAE, as MGFA's National Program Director. As Director, Kathy will focus on support group services to include the establishment of new support groups across the country and the development and delivery of programs for both those with MG and their caregivers. You can read more about Kathy on page 3.

The MG Patient Registry™ launched last spring continues to grow with over 1,400 individuals registered to date. My MG™ phone app has been downloaded by over 2,700 individuals, has been translated into Japanese and is being translated in to several other languages making it truly international in its scope. You can learn more about both the registry and phone app in this issue of the Foundation Focus. I strongly encourage you to take advantage of these two initiatives.

We are both excited and proud to have received The Child Neurology Foundation's Advocacy Award for 2013. See related article on page 3.

We congratulate Dr. Ricardo Roda for being selected to receive the MGFA/ABF (American Brain Foundation) three years fellowship. See related article on page 3.

This issue of the Focus includes several articles highlighting MG related research including a summary of the research results

from our first MGFA/ABF Fellow, Dr. Jeffery Guptil, and the Foundation's new brochure on the "Effects of MG on Voice, Speech and Swallowing".

MGFA funded an international Consensus Conference on MG Treatment Guidelines. The conference was organized by Dr. Donald Sanders, and was held earlier this month in Durham, NC. Watch for the report out from the conference in the next issue of the Foundation Focus.

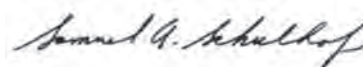
The MGFA Board has appointed Jonathan Bitting to the National Board of Directors to fill the vacancy created by the resignation of Carl Hansson. Please read about Jonathan on page 14.

The MG Walk™, a key component of MGFA's awareness and fundraising programs, continues to grow each year and is generating greater awareness about MG on the part of the general public while raising money to fund research initiatives, develop educational materials and programs. The Walks have raised over \$1.75 million. I personally want to thank all of you who have helped to make this program a success, as it is helping fund a number of the initiatives you will read about in this issue of the Foundation Focus including the hiring of a National Program Director.

Before closing I want to call your attention to and thank two chapters that have made significant gifts in support of MGFA's research initiatives. The Garden State New Chapter, \$100,000. and the Georgia Chapter, \$10,000.

We would love to hear from you! Send us your questions, comments and suggestions about these programs and others you think we should be considering.

Hope to see many of you in Philadelphia, PA, for the MGFA National Conference, April 17-18.



2014 MGFA/ABF Clinician Scientist Development Award in Myasthenia Gravis

We are excited to announce that The Myasthenia Gravis Foundation of America in conjunction with the American Brain Foundation (formally AANF) sponsors another three-year fellowship award. These fellowships are designed to train the next generation of clinical researchers. Dr. Ricardo Roda is this year's winner of the award, which will grant him \$240,000 over the course of three years to conduct research and provide new insights into new therapies for Myasthenia Gravis. His study is "Clinical Significance and Pathogenic Mechanisms of LRP4 Antibodies in Myasthenia Gravis."

Dr. Roda completed a neuromuscular fellowship at John Hopkins Hospital and is currently a neurogenetics fellow at the National Institutes of Health, NINDS/Neurogenetics Branch. To conduct his research, Dr. Roda will be using the resources of The Clinical and Translational Science Institute (CTSI) which is a partnership between New York University and the New York City Health and Hospitals Corporation. The CTSI provides services for research scientists to enable them to create a research protocol from the inception of the core idea, through to the development of a sound research plan, the clinical research approval process, deployment as an active protocol and the analysis of the results garnered. The MGFA will post updates on the findings of his research.

A New National Program Director Joins the MGFA

We are pleased to announce that Kathleen Brown, MPA, CAE, has joined MGFA as National Program Director. Ms. Brown comes to MGFA from the Lymphoma Research Foundation (LRF), where she was Director of Research. Her background also includes serving as Director of Programs for the National Kidney Foundation of New York/New Jersey among other not-for-profit organizations.

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As the National Program Director of MGFA, Ms. Brown will focus on support group services and developing new programs on issues essential to patients and caregivers. With over 25 years of experience, Ms. Brown has worked closely with boards and committees to create resources and activities aimed at successfully achieving a mission. With a Master

degree in Public Administration, Ms. Brown was inducted into Pi Alpha Alpha of the National Honor Society for Public Affairs and Administration, and is an American Society of Association Executives' Certified Association Executive.



MGFA Receives Advocacy Award from the Child Neurology Foundation

It is with pleasure that we announce the Myasthenia Gravis Foundation of America (MGFA) was a recipient of The Child Neurology Foundation Advocacy Award of Merit, created in 2002 through the efforts of its Board of Directors and the Foundation's Advocacy Committee to recognize patient support organizations that make outstanding achievements on behalf of patients and families with neurologic and developmental disorders.

This award was received on behalf of the MGFA by former Board member, Nancy Kuntz, MD of the Ann & Robert H. Lurie Children's Hospital of Chicago.

The MGFA was nominated by Eric Kossoff, assistant professor of neurology and pediatrics at Johns Hopkins University. Eligible patient support organizations must be a not-for-profit organization. Submitted nominations are reviewed by the membership of the Advocacy Committee of the Child Neurology Foundation.

The 2014 MGFA National Conference is Fast Approaching



MGFA's 2014 National Conference is being held in Philadelphia, PA on April 16-18 at the Loews Hotel. The National Conference combines the Foundation's annual business meeting with the opportunity for patients and their families to learn more about the disease and its treatment, current research, and tips to learn 'how to live' with MG from experienced medical professionals and fellow patients. For registration information and hotel details, please visit www.myasthenia.org.

MGFA National Conference Agenda

Wednesday April 16th

5:00PM – 6:30P
Welcome Reception

6:30PM
Nurses CE Dinner

Thursday April 17th

7:30 AM
Relaxation/Mind Body Exercises

8:00 AM – 9:00 AM
Continental Breakfast

9:00 AM – 9:45 AM
State of the Foundation
Mr. Samuel Schulhof, Chair, Myasthenia
Gravis Foundation of America, Inc.

9:45 AM – 10:15 AM
Morning Break

10:15 AM – Noon
General Assembly

(voting members only) Runs concurrent
with MG101 and Tell Your Stories
Basic Track

10:15 AM – 11:30 AM
MG101-The Basics

Neil C. Porter, MD, Assistant Professor, Department of
Neurology, University of Maryland
All Others Track

10:15 AM- 11:00 AM
Tell Your MG Stories - Patients

Carolyn Davis – MG Patient

11:00 AM - Noon
Tell Your MG Stories - Caregivers

Noon – 1:00 PM
Lunch

1:00 PM – 1:45 PM
Keynote Address

Fernando Ferrer , MG Patient, MTA Board of
Directors, Mercury Partners LLC – Co-Chair, Former
Member NYC Council, Former Bronx Borough
President, Former NYC Mayoral Candidate

1:45 PM – 2:00 PM

Break

2:00 PM – 4:00 PM

Three Concurrent Tracks –

Interactive Round Table Discussions

- Mature MG Patients – Robert Ruff and Charlene Hafer Macko
- Young Adults – Jennifer Faucett and Mike Ursic
- Parenting Child with MG – Carlana Hoffman

4:00PM – 4:45PM

GWAS Update

Daniel B Drachman, MD, Johns Hopkins School of Medicine, Professor of Neurology & Neuroscience, WW Smith Charitable Trust Professor of Neuroimmunology

4:45PM – 5:15PM

MG Patient Registry Update and Data

Ted M Burns, MD, University of Virginia, Department of Neurology

6:30 PM

Awards Dinner (also MG Walk awards)

MG Jeopardy, Neil C. Porter, MD, University of Maryland, Assistant Professor, Department of Neurology

Friday April 18th

7:30 AM – 8:00 AM

Exercise

Charlene Hafer-Macko, MD & Richard Macko, MD

8:00 AM – 9:00 AM

Continental Breakfast

9:00 AM – 9:30 AM

Research Update - General

Ted M Burns, MD, University of Virginia, Department of Neurology

9:30AM – 9:45AM

Rituximab Clinical Trial – Getting Involved

Richard Nowak, MD, Department of Neurology, Yale University

9:45AM – 10:00AM

Research update: Understanding How MG Treatment Effects Autoimmunity

Kevin C. O'Connor, Ph.D., Assistant Professor of Neurology, Department of Neurology, Human and Translational Immunology Program, Yale School of Medicine

10:00AM- 10:15AM

Break

10:15AM- 11:15AM

Family Communication

11:15 AM – 12:00 PM

Roundtable Discussions

1. Workplace Issues and MG - Christina Forster
2. Work to Retirement Transition - Does MG Make a Difference – Janet Myder

12:00 PM– 1:00 PM

Lunch

1:00PM – 1:30PM

Robotic Thymectomy

Abbas E. Abbas, MD, MS, FACS, Chief, Division of Thoracic Surgery, Director of Thoracic and Foregut Surgery, Associate Professor of Surgery, Temple University School of Medicine

1:30 PM – 2:30 PM

Ask the Professionals

2:30 PM

Closing Remarks

Mr. Samuel Schulhof, Chair, Myasthenia Gravis Foundation of America, Inc.

New MGFA Brochure: Effects of MG on Voice, Speech, and Swallowing



For persons who have myasthenia gravis (MG), the disturbance of voice, speech and swallowing is more than an inconvenience. It is a disabling and often embarrassing interruption or deterioration of daily functions that are vital to survival and quality of life. It also affects the way people view us and interpret our attitude and possibly our intentions.

Understanding how MG may influence voice, speech and swallowing is important for persons who have MG, as well as those who care for them on a professional or personal level. Understanding helps in coping with and treating MG.

The Myasthenia Gravis Foundation of America has published a new brochure “Effects of Myasthenia Gravis on Voice, Speech and Swallowing.” Written by two experts at the Medical University of South Carolina Evelyn Trammel Institute for Voice and Swallowing, the brochure is a significant addition to the foundation’s collection of educational materials. This article provides highlights of the new brochure.

Dysphonia is a broad term used to describe a voice disturbance that causes a change in vocal quality that may sound like hoarseness. It affects about two percent of persons with MG.

Dysarthria is a problem with articulation, or a problem with how speech sounds are pronounced. It affects about ten percent of persons who have MG. Dysarthria is often characterized as “slurred speech” and can be caused by incoordination or muscle weakness.

Dysphagia is the medical term for the swallowing difficulty that is common in individuals with MG. It can have serious implications that range from drooling, food residue in the mouth or throat, gagging, coughing when eating or drinking, to life threatening complications such as aspiration pneumonia, which is a lung infection caused by food material (saliva, food, liquid) in the lung.

Because care for MG is individualized, treating voice, speech and swallowing difficulties varies according to the nature and extent of other symptoms. The effect of interventions is often related to the effectiveness of treatment of the overall MG. In the case of speech and voice disorders, speech-language therapy may be used

in addition to the pharmacological therapies used for treating the MG. This therapy may involve an active therapeutic program and/or compensatory strategies to increase understandability.

The mechanism of swallowing is a complex process which involves approximately 50 pairs of muscles that must work in a coordinated manner in order to prevent saliva, food or liquid from entering the airway (aspiration). MG may cause muscles to fatigue as a meal progresses or when certain foods require a lot of chewing. Persons with thymomas (tumors of the thymus gland) may have dysphagia, particularly if the tumor is compressing the nerves and muscles involved in swallowing.

The assessment of a person’s swallowing problems may be referred to a speech-language pathologist to determine how the muscles of the face and throat are working. An initial evaluation may look at how a person drinks and eat foods of various consistencies. If further studies are needed, a modified barium swallow or fiberoptic endoscopic evaluation of the swallowing may be performed.

Similarly to treatment of voice and speech disorders in MG, treatment for dysphagia is based on the individual, the underlying cause and severity of the swallowing problem. In addition to pharmacological therapy for the MG, fatigue of the swallowing muscles can be reduced by smaller and more frequent meals, resting prior to eating, and avoiding talking while eating. Many other strategies may be employed such as consuming cold foods and liquids, alternating a small bite of solid food with a small sip of a liquid, and drinking thicker liquids. It also may be helpful to plan meals around the peak of medication, e.g. one hour after taking Mestinon/pyridostigmine. Individuals who have trouble swallowing pills crush them and mix them with applesauce or pudding. Additionally, liquid or dissolvable gel cap forms may be available. Talking with a pharmacist can help with choosing the right manner for taking medication.

For the full text of the brochure, go to <http://myasthenia.org/LivingwithMG/InformationalMaterials.aspx>
To obtain hard copies call 800-541-5454.

Authors:

Kendrea L. Focht, CScD, CCC-SLP, CBIS, Medical University of South Carolina Evelyn Trammel Institute for Voice and Swallowing

Janet A. Myder, MPA, Member of MGFA Communications; Editor, E-Update; Co-Chair, Low Country South Carolina Myasthenia Gravis Support Group



The First MGFA/ABF Fellowship Was a Huge Success

In 2010, the MGFA in conjunction with the American Academy of Neurology awarded Dr. Jeffrey T. Guptill MD, MA, MHS the American Brain Foundation /MGFA Clinician Scientist Development Award (CSDA), which supported a two year fellowship in MG. Dr. Guptill trained under the careful direction of Dr. Donald Sanders and his colleagues at Duke University. Dr. Guptill worked simultaneously on three important areas of MG, 1) studies of the overall cost of MG care, 2) clinical trials of biomarkers of different forms of MG and 3) clinical trials of different treatments for MG. Biomarkers refers to measurable things, such as concentrations of specific antibodies in the blood of person suspected of having MG that can be used to support the diagnosis that an individual has MG or that an individual has a specific type of MG (example, the presence of antibodies to the protein MuSK or to the acetylcholine receptor (AChR) can be used to distinguish different forms of MG).

Duke University was an excellent facility for Dr. Guptill's training because it is a Center for MG care in the US, it has a rich educational environment of clinicians and basic researchers devoted to MG care and Dr. Sanders has a strong track record of training clinicians and researchers who have made great contributions to the field of MG diagnosis and treatment.

Dr. Guptill's two year fellowship was extremely successful. He produced two papers and several presentations on the cost of MG care in the United States. The information in these papers is important for healthcare planners to appreciate how costly MG care is.^{1,2}

Dr. Guptill gave several scientific presentations and wrote papers on the types of antibodies and the composition of the immune cells in people with different forms of MG.^{3,4,5} He also became involved in several multi-site clinical trials of treatments for MG.^{6,7,8}

Additionally, Dr. Guptill has become a member of the MG treatment community. He is now a member of the Medical Scientific Advisory Board of the MGFA. He was promoted to the academic rank of Assistant Professor at Duke University. The work from Dr. Guptill's CSDA led to one funded research grant program in MG and he has 5 other research grants that are in review or undergoing revision prior to funding.

In summary, the CSDA program was very fruitful for Dr. Guptill and enriched the MG treatment community by the addition of a well-trained, funded researcher who is dedicated to improving the lives of people with MG.

References

1. Guptill JT, Marano A, Krueger A, Sanders DB (2011). Cost Analysis of Myasthenia Gravis From a Large U.S. Insurance Database. *Muscle & Nerve*, 44: 907-911.
2. Guptill JT, Sharma B, Marano A, Soucy A, Krueger A, Sanders DB (2012). The Estimated Cost of Treating Myasthenia Gravis in an Insured US Population. *Muscle & Nerve*, 45:363-366.
3. Yi JS, Guidon AC, Sparks S, Medinas R, Massey JM, Juel VC, Hobson-Webb LD, Sanders DB, Weinhold KJ, Guptill JT. Characterization of CD4 and CD8 T cell responses in MuSK myasthenia gravis. *Muscle Nerve* 2013;48(4):691.
4. Yi JS, DeCrosos EC, Sanders DB, Weinhold KJ, Guptill JT. Prolonged B cell depletion in MuSK myasthenia gravis following rituximab treatment. *Muscle Nerve*, ePub Sep 4, 2013. PMID: 24006142
5. Guptill JT, Yi JS, Sanders DB, Guidon A, Sparks S, Osborne R, Massey JM, Juel VC, Hobson-Webb LD, Weinhold KJ. Characterization of B-Cell Subsets in MuSK Myasthenia Gravis. To be presented at the 2014 American Academy of Neurology Meeting.
6. DAPPER: a trial of 3,4-diaminopyridine in the Lambert-Eaton myasthenic syndrome sponsored by Jacobus. Dr. Guptill is a subinvestigator on this study
7. EPITOME: a trial of prednisone versus pyridostigmine for the treatment of ocular MG. Dr. Guptill is a blinded evaluator.
8. Study of eculizumab (A monoclonal antibody treatment that targets a specific class of immune cells) in refractory MG.



2013 Delaware Valley MG Walk



2013 Northern California MG Walk



2013 Delaware Valley MG Walk



2014 Southern California MG Walk

2013 MG Walks Raise More Than \$700K; Fundraising, Awareness Kicks Off in New Year for 2014 Spring Walks

15-Year-Old Victor Mendevil to Represent the Walks as National MG Walk Hero



The National MG Walk Campaign's success is unprecedented for the Myasthenia Gravis Foundation of America as the combined efforts of MG Walkers, since 2010, have raised more than \$1.75 million to

fund critical research and programs of patient support and also raise awareness nationwide of this rarely publicized neuromuscular disease.

In 2013, nearly 5,000 participants raised more than \$700,000, from media attention, to walkers in their T-shirts, to Walk materials in public locations, like in Walgreens pharmacies and doctor's offices, to literally thousands of participants raising funds by talking to everyone about MG and its effects on people's lives, the MG Walk Campaign was a nationwide, awareness-raising machine.

In 2013, the Walks also raised awareness in 32 cities nationwide, from California and the Pacific Northwest across the map to the Tri-State Walk in New York and as far south as Miami, Florida.

This year, the National MG Walk Campaign will honor Victor Mendevil, a 15-year-old living with MG. Victor experienced some strange symptoms around the age of five: droopy eyelids, severe double vision, swallowing difficulty, breathing problems, and loss of muscle strength. A number of physicians tested for asthma, a brain tumor, MS or other illnesses, but nothing seemed to fit completely.

At 8 years old while practicing violin, Victor collapsed to the floor unable to move his arms or legs and having great difficulty breathing. It was this visit to the Emergency Room where the on-call physician happened to specialize in myasthenia gravis (MG), which led to his diagnosis.

The family was stunned to hear of this disease for the first time. "I didn't understand how something so debilitating could be so easily overlooked, misunderstood and ignored. I soon learned that it is a complicated disease that is not widely recognized," says Victor.

Victor's family and friends joined the MG Walk Campaign last year with the team name "Victor's Victory" and walked in recognition of his journey with this rare disease, and to raise awareness of the illness in their community. His enthusiasm caught on, and after giving a passionate speech about the importance of raising funds and awareness, Victor was asked to be the National MG Walk Hero in 2014 and act as spokesperson for the fundraiser.

With new support groups growing, local businesses stepping up to sponsor the walk and new relationships with pharmaceutical providers being established, the momentum of the MG Walk Campaign raises not only individual awareness, but also corporate awareness in communities nationwide.

The MG Walk has raised awareness, renewed hope and worked to build a connected and caring community of those affected by MG. Through the MG Walk, patients along with families and friends unite their voices to say; "MG must be stopped. We need your help to find a cure!"

If your local community would benefit from an MG Walk event, contact the Walk Office.

For information on the Walk, to find a Walk near you, and to register, visit MGWalk.org or call 855-MG-Walks (855-649-2557).

Follow the MG Walk Campaign:

[Facebook.com/MGWalks](https://www.facebook.com/MGWalks)

[Twitter.com/MG_Walk](https://twitter.com/MG_Walk)

Upcoming Myasthenia Gravis Spring Walks

Georgia - March 15

Tampa Bay - March 16

Tallahassee - March 22

Jacksonville - March 23

South Florida - March 30

New Orleans - April 5

Orlando - April 27

New England - May 3

TriState (NYC) - May 31

Minnesota - June 21



2013 DC Metro Virginia MG Walk

Fundraising Tips

Set a goal. Be realistic, but challenge yourself! Once you get your first donations, you'll be pleasantly surprised to see how easy it is. You will also find it more rewarding than you first thought possible!

Consider how many people you'd like to join your team, and how much money you'd like to raise. Every person helps, and every dollar gets us closer to a cure.

Register - Sign up and get your free online fundraising page. It only takes a couple of minutes whether you do it online or by phone. As you recruit people to your team, they do the same.

Personalize - By signing up in advance, you gain a Personal Fundraising Page. Once you register, you will be sent an email with log in instructions. This page, especially once customized, becomes a great tool to ask your family, friends and colleagues to donate and join your team. While the page comes pre-set with information about the Walk, try to invest a few minutes here to share your story on why you are involved with the MG Walk. Add a picture or two. It's easy.

Share - After your page reflects your passion for ending MG, you're ready to tell everyone you know. Send it via e-mail, Facebook, and word of mouth. Ask friends to share it with their friends: the signal boost will reach a multitude of people, and even if a small percentage of them give, you'll watch your goal get nearer. See our MG Walk tutorials for more information.

Ask - It might feel funny the first time or two, but watching your fundraising thermometer inch towards 100% is thrilling. Focus on the idea that you are inviting others to invest along with you in a better tomorrow for all with MG.

Don't be afraid to share your goal! People like to invest in your success.

Once you get rolling, you'll start seeing opportunities everywhere. The number one reason people don't give is that they were not asked. So just ask everyone - friends, colleagues, neighbors. Ask at a book club meeting. Ask at bingo night. Ask businesses you frequent. (They can become local sponsors!) Ask people to ask. Just ask. You'll be happy to see how many people are willing to help when they see your dedication to our cause.

Remind - Even the best intentions need a helping hand. Check in with people if you don't have their donation in hand or see their contribution on your Personal Fundraising Page. Studies show that people need to hear your message more than once to really 'hear' it and respond.

You CAN do this! We know you can; we see people do it every day. Maybe you'll be our next top fundraiser?! And never get discouraged.



2013 Delaware Valley MG Walk

Raise \$200 in 5 Days

Day 1 Sponsor yourself = \$25

Day 2 Ask 5 Family Members/Friends for \$10 each = \$50

Day 3 Ask 10 Co-workers for \$5 each = \$50

Day 4 Secure \$25 from 2 businesses you frequent = \$50

Day 5 Ask your Boss to support your efforts = \$25

***Your Grand Total = \$200

Don't stop there! Continue to ask others in order to help you reach a larger goal!



2014 Houston MG Walk



2013 DC Metro Virginia MG Walk



2014 North Texas MG Walk



2013 Northern California MG Walk



2013 Northern California MG Walk



Raising Awareness

Orlando Support Group Kicks Off MG Walk Fundraising Efforts

The Orlando Myasthenia Gravis Support Group met on Saturday, February 8, 2014, to kick off its upcoming MG Walk fundraising and awareness efforts. The group, which meets every second Saturday of every even month at 10 a.m., got together at the Orlando Regional Medical Center - Classroom 102 (1414 South Orange Avenue). The Orlando Walk will be held on Sunday, April 27, 2014, at Baldwin Park in Orlando, with the walk beginning at Harbor Park at 10 a.m. Registration opens at 9 a.m.. Leah Nash, Chief Innovator at Trusted Source and Amber McKeon of Origami Owl, both MG patients, are committed to reaching Orlando's goal of \$20,000. For more information on the Orlando MG Walk, please contact Leah Nash at leah@trustedsourcefl.com.



New Orleans Group Holds Valentine's Day Comedy, Musical to Raise Awareness

The Myasthenia Gravis New Orleans Resource Group held a special Valentine's Day/Night Comedy and Musical Fundraiser to kick off the group's fundraising efforts for the New Orleans

MG Walk. Local New Orleans theatre talent "Uncle" Wayne Daigrepoint performed a one-man show, "Valentine-Y Show ... with a Mardi Grawdy Look," on Friday, February 14, 2014, at The Jefferson Orleans North in Metairie. The event raised more than \$1,000. Tickets were \$25 each, and all ticket sales benefitted the upcoming New Orleans Myasthenia Gravis Walk, which in its third year is scheduled for Saturday, April 5, 2014, at Lafreniere Park in Metairie. The special "Valentine's Day" night Performance by "Uncle" Wayne Daigrepoint featured over 70 love songs, dancing, comedy acts, sing-a-long, Mardi Gras beads, second lines and much more!!!

Tampa MG Patient Launches T-Shirt Drive to Raise Money for Walk

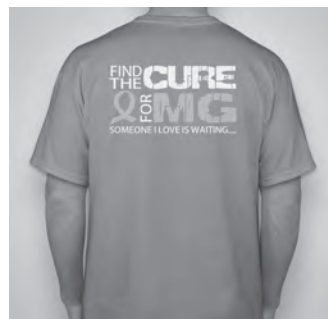


Melissa Choquette, of the Tampa MG Walk team Myssi's Snowflakes and who was diagnosed with MG in 2007, launched a T-shirt fundraising drive through booster.com to raise awareness and money for the upcoming MG Tampa Walk. The shirt is labeled with "Myasthenia Gravis Awareness"

surrounded by a snowflake and MG ribbon, and went on sale for \$20 each. Missy sold close to 40 shirts and raised \$300 for her team for the Walk. Missy's fundraising drive can be found at:

<https://www.booster.com/mgwalk>

New Orleans Walker Raises Money through T-Shirt Drive



Melchor Pavon, the uncle of MG patient Tommy Santora, launched a T-shirt drive, also through booster.com, to raise money for the upcoming New Orleans MG Walk. The shirt is labeled with "Find The Cure for MG, Someone I Love is Waiting." The drive has thus far sold close to 80 shirts and raised

about \$1,000 for Mel's team, Team Santora. Mel's fundraising drive can be found at: <https://www.booster.com/nolamgwalk>

What's Hot off the Press in Neuromuscular Junction Disorders?

Nicholas J. Silvestri, MD, Gil I. Wolfe, MD
Members of the M/SAB

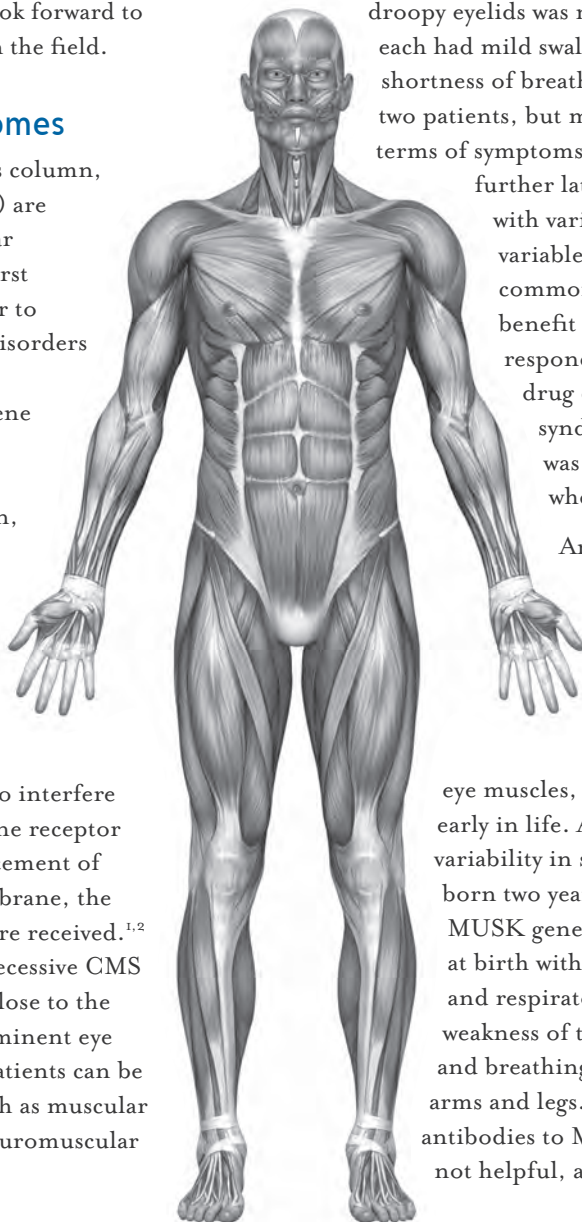
Department of Neurology, Neuromuscular Division, University at Buffalo School of Medicine and Biomedical Sciences, The State University of New York, Buffalo, NY

Introduction

Welcome back to our quarterly column highlighting some of the latest developments in research in neuromuscular junction disorders. We hope you enjoyed the first edition which appeared in the Fall 2013 Foundation Focus and look forward to keeping you updated on breakthroughs in the field.

Congenital Myasthenic Syndromes

As you recall from the last edition of this column, congenital myasthenic syndromes (CMS) are inherited disorders of the neuromuscular junction. Folks typically present in the first two decades of life with symptoms similar to myasthenia gravis (MG). Most of these disorders are inherited in an autosomal recessive fashion. This means that an abnormal gene must be inherited from both a person's mother and father in order to have the disease. In the last edition of this column, we noted that the spectrum of CMS has recently expanded beyond mutations in genes responsible for encoding specific proteins which make up the neuromuscular junction to include mutations in genes encoding for two enzymes that normally modify these proteins. These mutations are believed to interfere with modifications of muscle acetylcholine receptor subunits, affecting the assembly and placement of the receptor into the post-synaptic membrane, the region of muscle where nerve impulses are received.^{1,2} Patients with these forms of autosomal recessive CMS present with weakness of the proximal (close to the trunk) arm and leg muscles without prominent eye muscle involvement. As a result, these patients can be confused for having a muscle disease such as muscular dystrophy as opposed to a disorder of neuromuscular transmission.¹



Five more patients with dolichyl-phosphate N-acetylglucosaminophosphotransferase I (DPAGT1) mutations have now been described in a report led by a group of researchers in Oxford, England.³ The report included patients in the United Kingdom, Zimbabwe, and Argentina, all of whom demonstrated symptoms by age 7 years. Low muscle tone (hypotonia), falls, and delayed walking were early features of the disease. Weakness of the proximal arm and leg muscles was also typical, and weakness of neck muscles was seen in four of the patients. As previously observed, weakness of eye muscles leading to double vision or droopy eyelids was mild if present at all. Two of the patients each had mild swallowing difficulty, facial weakness, and shortness of breath. Mild learning difficulties also affected two patients, but may be incidental. Patients may stabilize in terms of symptoms in early adulthood, but tend to weaken further later in life. The response to treatment with various medications in these patients was variable. Pyridostigmine (Mestinon)- a drug commonly used in myasthenia gravis- was of benefit in all to varying degrees.³ Two of three responded favorably to^{3,4} -diaminopyridine (a drug often used in Lambert-Eaton myasthenic syndrome). Another medication, salbutamol, was dramatically helpful in a single patient when started at age 42.

Another uncommon form of CMS results from mutations in muscle-specific tyrosine kinase (MuSK), a protein that can also be involved in a subset of patients with myasthenia gravis. This rare form of CMS is characterized by droopy eyelids, severe weakness of eye muscles, and breathing difficulties at birth or early in life. A brief report highlighted the striking variability in symptoms between two Italian siblings born two years apart with the same mutations in the MUSK gene.⁴ The sister had a severe presentation at birth with weakness of the muscles of the throat and respiratory failure. She also had fluctuating weakness of the eyelids and eye muscles, swallowing and breathing difficulty, and severe weakness of her arms and legs. As is typical for myasthenia caused by antibodies to MuSK (MuSK MG), pyridostigmine was not helpful, and in fact made her symptoms worse.

Meanwhile, her brother only had fluctuating droopiness of his eyelids since birth and minimal difficulty with running. This brief report highlights the marked variability in symptoms that can be seen in this and other forms of CMS.

Myasthenia Gravis

Turning to MG, several prior studies have confirmed that fatigue is a common symptom.^{5,6} Fatigue— the subjective sense of tiredness or lack of energy— is distinguished from fatigability which is a classic feature seen in disorders of neuromuscular transmission and refers to an inability to sustain muscle contraction, leading to weakness. Symptoms attributable to dysfunction of the autonomic nervous system in patients with MG, such as constipation, blurred vision, and lightheadedness, have been described but appear to be rare.⁷ A recent paper from Norway investigated how often patients with MG experience fatigue and whether its presence is associated with symptoms of autonomic dysfunction.⁸ In the study of 82 MG patients and 410 healthy controls, the researchers administered three questionnaires: one assessing fatigue, one assessing signs and symptoms attributable to MG, and one assessing autonomic symptoms. The investigators found that patients with MG had higher fatigue scores than controls, especially on measures of physical fatigue. In addition, high fatigue levels were found to be more common in the presence of autonomic symptoms, especially as they related to sleep and sweating disturbances. The authors of this paper admit that even though the findings demonstrate an association between the presence of autonomic disturbance and fatigue in patients with MG, there is no proof of causality.

Thanks for reading and see you next time! We will plan on a longer research update section for MG, itself, in the next column.

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MGFA Appoints New Board Member Jonathan Bitting



Jonathan Bitting, a financial executive for more than 35 years with companies such as Citibank, Bloomberg, Chase Manhattan and Morgan Stanley Smith Barney, has joined the Board of the Myasthenia Gravis Foundation of America.

Jonathan is retired, and he said he wants to give back with his time and help the MGFA as much as he can. Jonathan also brings with him an extensive background in volunteer work with numerous not-for-profit organizations.

Jonathan is a Certified Financial Planner since 1986, and he is a financial executive who is adept at driving sales of new high net-worth investment accounts, providing presentations on

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investment products and strategy to brokers and high net-worth individual clients and managing portfolios.

Jonathan was most recently the Marketing Director and Investment Committee Member of The Solaris Group, in Bedford Hills,

New York. For eleven years prior, Jonathan was the Executive Director and Portfolio Manager for Morgan Stanley Smith Barney and its predecessor companies. While at Morgan Stanley, Jonathan helped grow assets under management to more than \$2 billion, traveling nationally with a 12-person wholesaling team, and he personally managed \$500MM in custom, MDA's utilizing proprietary trading platform, which facilitated using multiple investment styles and portfolio managers in a single account. He also served as Executive Vice President for Putnam Capital Alliance; Vice President of CitiBank; Regional Business Development Officer for Bloomberg; and Vice President, Portfolio Manager for Chase Manhattan Bank.

The MGFA is honored to have Jonathan join the Board.

Chapter's Corner

Arizona Chapter Elects Board Members for 2014

The Jim L. Walker, Arizona Chapter of the Myasthenia Gravis Foundation of America, held its annual meeting and elected Sally and Ron Luna, Meg and Lexi Ganx, Anna Lopez, and Dr. Suraj Muley to the board. The Board accepted Luann Green's request to step down as treasurer after many years of service and we thank her for her tenure and hard work with the Board. Luann is the daughter of the namesake Jim L. Walker.

The new executive committee consists of Chair Jim Lovecchio; Vice Chair Steve Borski; Secretary Anna Lopez; and Treasurer Meg Ganz. Doctors Scherer and Muley are our medical advisors, and Marilyn Ricci is our nurse advisor. New board member, Ron Luna, is helping to redo the web site and IT issues.

The Jim L. Walker, Arizona Chapter of the Myasthenia Gravis Foundation of America, was originally founded in 1972 by Peggy Marquard. It has grown from 16 members at that first meeting to just over 900 members.

For more information on the Arizona chapter, contact Jim Lovecchio at 520-889-6910 or e-mail lovecchio@dakotacom.net

Have Chapter or Support Group news to share? Contact Foundation Focus editor Tommy Santora at tommy.santora@gmail.com or call 504-376-7474

In Less Than A Year More Than 1,100 MG Patients Are Registered

A total of 1,140 Myasthenia Gravis patients have so far registered in the MG Registry, a confidential and patient-driven research project, funded by the Myasthenia Gravis Foundation of America (MGFA), managed by the University of Alabama at Birmingham (UAB) with oversight by the MGFA Patient Registry Committee. Patients can sign up at: <https://mgregistry.soph.uab.edu/MGRegistry/SignUp.aspx>

The registry is a confidential means for patients to provide information to an active database of persons with MG and was developed for the purpose of research, treatment and patient information.

It provides a system for researchers to gauge the potential for recruiting patients for clinical trials and communicating with them in a manner that respects their privacy. The registry also can be used to provide education about MG to patients, care givers, non-expert health care providers and funding sources.

To enroll, follow these steps:

- Click on the following link to go to the "Participant Sign Up" page: <https://mgregistry.soph.uab.edu/MGRegistry/SignUp.aspx>
- Read and agree to the Terms and Conditions of participation and complete all of the fields under Register. (For future reference, record the username, password, and email address that you use to register, as well as your security questions and answers.)
- Click the button "Sign me up".
- Complete and submit the enrollment survey.
- If you have questions or need assistance contact the MG Patient Registry Coordinating Center at: Email: MGR@MGregistry.org; Phone: 205-975-8633 or Toll-free 855-337-8633; Web address: <http://www.soph.uab.edu/mgregistry/>

More Than 2,700 MG Patients Take New App "MyMG" For Test Run

More than 2,700 Myasthenia Gravis patients have tried out the new "MyMG" software application designed to run on smartphones, tablet computers and other mobile devices. With "MyMG", the MG community records symptoms and their impact on daily activities. The app allows you to record notes with each survey. The notes can collect information about medicines and life events that can impact your MG symptoms. You can then print your notes and survey results from your PC to share with your doctor.

Approximately 1,700 people have downloaded the app through Apple, while more than 1,000 people have downloaded the app through Google Play.

The new app is available free by downloading from the Google app store or iTunes. For patients without access to a smartphone, tablet computers or other mobile device, "MyMG" is available online at the MGFA website <http://www.myasthenia.org>.

Go to your app store today and see what is new in the conversation between MG patients and their doctors: "MyMG."



MYASTHENIA GRAVIS
FOUNDATION OF AMERICA, INC.

355 Lexington Avenue, 15th Floor
New York, NY 10017-6603

(800) 541-5454
(212) 370-9047 fax
mgfa@myasthenia.org
www.myasthenia.org

Myasthenia gravis is an autoimmune neuromuscular disorder. Symptoms may include double vision, drooping eyelids, slurred speech, difficulty chewing and swallowing, weakness in arms and/or legs.

MGFA is committed to finding a cure for myasthenia gravis and closely related disorders, improving treatment options, and providing information and support to people with myasthenia gravis through research, education, community programs, and advocacy.

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If you would like to receive Foundation Focus by email only, please email mgfa@myasthenia.org.

Thank you

**...to all of our
wonderful
donors!**

*Your generosity
brings us closer to a
world without MG.*

The goal of the MG Walk Campaign is to expand into new markets where we can bring together patients, create a community of active/engaged MG families and raise vital awareness & funding for myasthenia gravis! It is crucial that we go where we know we can garner the support needed to ensure success. If you are interested in seeing the MG Walk come to your area and you are excited to play an active part in its planning, promotion and production, we want to hear from you! Please contact the MG Walk Office at 1-855-MG-WALKS or Info@MGWalk.org or fill out our interest form found online at www.MGWalk.org. Thanks so much!

