



Raising MG Awareness One Month at a Time

June is a special month for a lot of reasons, weddings and young love, blue skies, birds calling. But for the MG Community June is special for another reason — June is MG Awareness Month. June brings us all together to promote awareness and educate our larger community about MG. June is a time when everyone in the MG Community

needs to proactively bring MG forward to others. Ask yourself –

- Do my relatives, friends, and others important to me, know that I have MG and what MG is and does to sufferers?
- Have I shared information and stories with them?
- Do I take actions that can help others become aware of and understand MG?

Maybe that seems like a really daunting task. But like every journey of 1,000 miles, it begins with one step. Remember you are not alone and you don't have to do it all, but if you can do your bit, and someone else does theirs, they add together and make a difference.

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2017 MG Walks

NEW ENGLAND: May 6

NORTHERN WISCONSIN: May 6

SOUTHERN WISCONSIN: May 6

TRISTATE: June 10

CONNECTICUT: June 11

INDIANAPOLIS: September 10

CLEVELAND: September 23

CHICAGOLAND: September 24

and more!

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MG STRONG

Lauren Jarman 2017 South Carolina MG Walk Hero

Lauren has been living with MG for over 6 years now, and it has certainly been a challenging diagnosis. She was first diagnosed with myasthenia gravis in April 2011, right before her wedding. She had a droopy eyelid that was causing concern, and with her wedding day approaching, she got it checked out right away. She went to multiple urgent care centers to figure out what was going on, but was ultimately diagnosed by a neurologist working on the same floor that she was



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CEO'S LETTER



Dear MG Community Friends and Colleagues,



2016. It was a year of progress, a year of change. A time for action, and unification. A time to listen and learn from each other. Our theme for the year was “Together We Are Stronger.” Together, we celebrated progress in research, advocacy and awareness. Together, we grew the MG Patient Registry by more than 50%—creating not only the opportunity for better understanding of MG, but also new pathways for clinical trials. And together, we mourned the loss of too many MG warriors—reminding us that MG can still alter and even take lives, and that there is much yet to be done. To quote Dickens, “It was the best of times. It was the worst of times.” I feel so fortunate and thankful to have been able to be a part of this strong, caring and determined MG Community—through the best times, and the worst ones, too.

The best of times? It was an unprecedented year in research in MG. The publication of the results of the Randomized Trial of Thymectomy in Myasthenia Gravis was the culmination of TEN years of work from scientists who sought proof of the effectiveness of this procedure in MG. The publication of the International Consensus Guidance for Management of Myasthenia Gravis has given us a framework that can and will be used to educate those treating MG on options and protocols, and also a platform for advocacy when faced with denials for reimbursement for treatments. And following the completion of the phase three trial, the Alexion Pharmaceuticals Company filed for regulatory approval of eculizimab (Soliris) for people with refractory forms of generalized myasthenia gravis. Not only does this announcement provide hope for those who don't respond well to standard therapies for MG, it has also created new interest in myasthenia gravis within the pharmaceutical industry. Oh, yes—the BEST of times!

The MG Community united around the June is MG Awareness Month campaign, promoting the “**#IHaveHeardofMG**” hashtag, coined by MG Awareness warrior, Sandra Hardin. More than

5000 people affixed the Twibbon to their Facebook profiles, and through social media reached as many as 260,000 individuals. Through 11 news media stories, and 35 calendar listings, we reached approximately 6 million people! Just think how many more people can now say, “I have heard of MG! And we also raised the bar on Advocacy, uniting in lobbying efforts with partners such as the National Health Council, National Organization for Rare Disorders, and Research!America around issues important to the MG Community. And our network of community and support groups continues to grow, as new groups are being formed nearly every month. And we are excited to have new and stronger partnerships with other organizations serving people with MG, including social media groups.

We made progress in fund raising, too. The MGWalk drew more than 4900 participants and brought in nearly \$795,000,

while raising visibility and awareness in 38 communities nationwide. MGFA also saw a 50% increase in individual giving—and an additional gift of \$50,000 for research from Philanthropists of the Year, Mona Roth, of Long Island. And almost unbelievably, MGFA was the recipient of several large and unexpected bequests, totally more than \$1 million. Best of times.

We move into 2017 with commitments to use the funds so generously given by our donors to expand research, programs and services, awareness and advocacy—and to also launch new initiatives to raise more funds for all that we want to accomplish. The focus for this year is “Living Your Best Life with MG” and that will be the theme of the MGFA national conference—where representatives from all parts of the MG Community will come together to learn, support each other, and set direction for the future.

“I feel so fortunate and thankful to have been able to be a part of this strong, caring and determined MG Community—through the best times, and the worst ones, too.”

We will execute the first year of an ambitious 3 year research plan that in 2017 alone includes: support for the MG Patient Registry, funding for new pilot grants and continuation of our 2015 grants, continuation funding of our post-doctoral fellowship and transformative research grants, a new RFA for a consensus conference on diagnostic criteria, the initiation of the MGFA Research Opportunity Fund, the MG Scientific Session at the AANEM meeting, and perhaps most exciting, May 15-17, The International Conference on MG and Related Disorders—held once every 5 years and considered to be the premier meeting on myasthenia gravis in the world!

You can expect to see the launch of our “MG Friends” telephone peer support program—engaging trained volunteers to help ensure that no one has to face MG alone. We will initiate our MG Partners in Care Program, designed to ensure that people can find and connect with the top clinicians in the country. And the rebranding of MGFA will continue, with the launch of a new website with an updated infrastructure that will be mobile friendly, and will ensure that people can easily find the information that they need. You will be hearing about new fund raising campaigns and initiatives—because all of what we want to accomplish takes money. We are going to ASK BOLDLY—because that is what is needed to change the world for people with MG.

The worst of times? We need to be vigilant about access to health care and treatments. MGFA will continue to reach out to you to support important legislative issues as they arise. With uncertainty about changes in the health care landscape, we will be ready to raise our voices to ensure people with MG and other chronic and/or pre-existing conditions have access to the health care and treatments that they need.

And there is so much to do. While a disease that was once fatal for 30-40% of us, is now fatal for only 3-4%, that is NOT good enough. Not even close to good enough. No one should die from MG. It will take all of us to move forward towards a world in which every person affected by MG can achieve the hopes and dream of their “best life”—without MG getting in the way. And for those with refractory MG to not live in fear. There is a place in our work for every single member of the MG Community: whether you volunteer, donate money, tell your story and spread awareness, form a Walk team, write a letter to your legislators, and/or ask others to donate or volunteer, YOU can make a difference. What will you do? I look forward to working with you for another exciting and productive year for the MG Community.

Thank you,

Nancy Law

MGFA Chief Executive

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Catch up with the MG Community, learn more and share your story...on our website, Facebook and Twitter.

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.

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As of this writing, MGFA is planning for June is MG Awareness Month, so stay tuned to MGFA e-blasts, our Facebook site, and website for more details about the 2017 Program. On our list is a new MG Twibbon to be used on Facebook/social media to help you declare your participation. Watch the MGFA website and MGFA Facebook for announcements and links to resources. Meanwhile, make your own plans for June is MG Awareness Month. Here are some ideas:

1.

APPROACH YOUR MEDICAL CAREGIVERS AND ASK IF YOU CAN PLACE INFORMATION ABOUT MG IN THEIR OFFICES OR ON THEIR BULLETIN BOARD.

You can order brochures for distribution at doctors' offices for newly diagnosed patients. Reach out to MGFA at 800-551-5454 to order. Or, go to the website and print out resources you can find at Living with MG/Informational Materials <http://www.myasthenia.org/LivingwithMG/InformationalMaterials.aspx>



2.

CONNECT WITH YOUR EMERGENCY MEDICAL SERVICE AND HOSPITAL EMERGENCY ROOM STAFF.

Make them aware of Myasthenia Gravis and why it can be a challenge for emergency professionals to treat. For instance, it's rare, it responds differently/adversely to many common medications and standard test results may be misleading. You can find the Emergency Management I brochure for professionals on the website and by calling for a copy/copies at 800-551-5454. At the time of this writing, MGFA is working on a slide presentation for emergency medical professionals – consider working with your doctor, support group or other MG friends to show this slide presentation for your local emergency medical professionals this coming summer.



3.

USE THE JUNE IS MG AWARENESS MONTH POSTER

– go to public places where bulletin boards are available – libraries, supermarkets, pharmacies, senior centers, houses of worship. Ask if you can post the MG Awareness Poster as a public service. You may want to be ready to explain that MG is a rare disease and people often aren't aware they have it or find it hard to get a good diagnosis. So you are trying to raise awareness. Posters will be available in Spring 2017.

4.

CREATE YOUR PERSONAL MG VIDEO FOR YOU TUBE

– tell your story. Be compelling, succinct, and relatable.



5.



BE A SOCIAL MEDIA BUG

– as we all know, huge numbers of people use social media these days: Facebook, Instagram, You Tube, Twitter, Pinterest, Tumblr, Reddit and more. Find ways to use these resources. Ideas: A Day in the Life of an MG Patient – post throughout your day with your ups and downs. Or, post daily, helping others follow along through your June. Post links to resources on the MGFA website such as those listed at the end of this article.

6.

PARTICIPATE IN AN MG WALK.

Visit the mgwalk.org website and learn more about walks near you. If you aren't physically close to a walk, you can still participate remotely. Some feel a walk isn't for them – maybe you aren't sure about raising money, for instance. Even though raising money is important to our cause, you can also go to a walk to meet others in the MG community. Or, you can volunteer to help out. Find out more from the mgwalk.org website or call the Walk Department at 855-649-2557. MGAers and friends are always welcome.



WEAR AN MG T-SHIRT. Use it as an opportunity to speak to others about MG. Have your “speech” in mind for when the opportunity arises. For instance,

- **WHAT’S MG?** MG, myasthenia gravis, is an auto-immune neuromuscular disease that causes fluctuating weakness in the voluntary muscles.
- **WHAT ARE THE SYMPTOMS?** Typical symptoms are droopy eyelids, crossed eyes, loss of limb strength, trouble swallowing or breathing.
- **CAN IT BE CURED?** No. But most patients can get treatment that helps to control the symptoms to a degree. Some find that current medications aren’t very effective.



7.

For more thoughts on what you can say or share see the Spring 2016 Foundation Focus, June is MG Awareness Month starting on the cover page at <http://myasthenia.org/AboutMGFA/FoundationFocusNewsletter.aspx>

8.



ALERT THE MEDIA IN YOUR COMMUNITY. Make them aware of MG and of June is MG Awareness Month. This is an ideal activity for a support group who can share a compelling patient story, while also helping the reporter to understand MG from both a medical and personal point of view. Be ready with the essential information – who, what, when, where and why.

9.

TAKE A MOMENT TO THANK YOUR CAREGIVERS, FAMILY AND FRIENDS FOR THEIR SUPPORT AND UNDERSTANDING. Do something special whether a kiss or a card or a cake to say how much you appreciate their love, time and effort.



10.



MAKE OR ENCOURAGE A DONATION. Support a Walker through the www.mgwalk.org website. Lead a virtual team. Encourage friends and family to participate with donations. Visit the www.myasthenia.org website and donate go to <http://www.myasthenia.org/HowcanIhelp/Donations.aspx>.

11.

RUN YOUR OWN FUNDRAISER. Host a party or event and ask for donations or hold a raffle. Ask local businesses to contribute prizes. Contribute proceeds to MGFA. Remember to check your state and localities regulations regarding activities such as raffles before you proceed.

12.



HOST A LITERATURE TABLE. Check for health fairs and similar events in your area. Order literature from MGFA. Remember to leave ample fulfillment and shipping time. Work with your friends and family or support group to staff the table.

13.

REMEMBER MG AWARENESS ALL YEAR LONG. June may be our special month, but MG Awareness can be pursued all year long.



RESOURCES TO USE

A. LITERATURE – <http://myasthenia.org/LivingwithMG/InformationalMaterials.aspx>

B. WEBCASTS – <http://myasthenia.org/LivingwithMG/MGFAWebinarSeries.aspx>

C. AND PODCASTS – <http://myasthenia.org/LivingwithMG/PodcastEducationalSeries.aspx>

D. ANNUAL CONFERENCE VIDEOS – <http://myasthenia.org/CommunitySupport/MGFAConferenceVideos.aspx>

14.





Circle of Strength

We have been fortunate to be blessed by the generosity of many individuals and families who have written checks and made bequests to help in the fight against MG. This is a list of all those who have made contributions over \$1,000 in 2016. Thank you to these generous supporters, our Circle of Strength. Thank you too, to all of you who have given to the Foundation at whatever level, and to all those who give their time and energy to our cause. Thank you!



\$25,000+

Mona Roth

\$10,000 – \$24,999

Mark Aitken-Cade

Steven Berline

Brian Gladden

\$5,000 – \$9,999

Nancy Law

Darrell and Linda Webb

Edward Walsh

\$2,500 – \$4,999

Jack Ambrosio

Judith Craver

Paul Goldstein

Steven P. Grant

Thomas Larsen

Nelson Machado

Tatsuji Namba

Suzanne & Robert Ruff

Mark Selden

Gerald Smith

David Wind

\$1,000 – \$2,499

Michael Alderman

Leonard & Margaret Aldridge

David Bernstein

Jim Blankenship

Joubin Bral

\$1,000 – \$2,499

Beverly Bremer

Donald Brooks

Ted Burns

Stephanie Cawein

Tobi Cawthra

Steven Chittenden

Virginia Cunningham

Frank J. D'Amico Jr. APLC

Charles Daniels III

Rocco Dano

Charles L. DeFanti

Thomas Defanti

Anthony Della Salla

Gary Eder

Robert Elliot

Judy Elmore

Richard Elmore

Richard & Dana England

Robert & Margaret Espinosa

Debra Fienberg

Sylvia Fuhrman

Leni Fuhrman

Sheldon Gartenstein

Roberta Greenberg

Harriet Griesinger

Faith DeLeon and Paster Norman Harding

Brian and Gretchen Heller

Derek Howard

Robert Johns

Julia Johnson

Mike Kaplan

Susan Klinger

Linda Kusner

Marcia & William Lorimer

Helen Machado

Charlene Macko

Kaitlin Masters

\$1,000 – \$2,499

TJ McConnell

Paula McGinnis

Teresa McHugh

Mitchell Modell

Joan Monk

James Nolt

Josephine Pereira

Tony Pieprzycza

Jeffrey Pilgrim

Monica Poff

Anne Rubin

Donald Sanders, MD

Samuel Schulhof

Lucy Ann Sciandra

Terry Shaver

Betty Shine

Frederick Simon

Margot Slater

Christina Smith

Dr. Donald Somers

Jackie Spencer

David Steinberg

Arthur Strauss

Gary Strauss

Jason Strauss

Mark Swift

Elizabeth Swize

Don Taylor

Noah Tepperberg

James & Sandi Thompson

Carol Tome

Bradley Tusk

Thomas Ursic

Katherine Vennetti

Allan Weiss

Joan Wood

Anonymous

Anonymous





continued from cover



Lauren, Dave and Carleigh Grace

working as a nurse. At this time, she had also gone for a CT scan that showed something that was alarming, and went into surgery to have it removed. But because of this

surgery, and its effect on her MG, Lauren went into crisis and was in the hospital on a ventilator for 3 days. This was the first of four very major MG crises she would endure, the second happening a year later and landing her on a ventilator for 5 days, and another while pregnant with her daughter. In the 6 years she's had MG, she has also been intubated about 7 times. 2 years ago, she and her husband welcomed their first child, and it was after the birth of her daughter that Lauren finally went into remission.

One of the things that has shocked her most since her diagnosis is how little doctors know about MG. During her first crisis, the ER didn't seem to know what to do and was overwhelmed, though luckily the pulmonologist knew what to do. Just recently she underwent Lasik eye surgery and was given a prescription for a medication she knows would react negatively with her MG. The knowledge seems to be very limited, and as a member of the medical community, and the MG community, Lauren hopes to bridge that gap.

Last year was the first year Lauren was able to participate in the MG Walk...and became one of the most successful fundraisers of the entire campaign. Her team, Vent Stoppers, raised over \$18,000 for the South Carolina MG Walk, thanks to the generosity and support of her family and friends. This year, she's been an integral part in moving the Walk to Charleston and working to engage the local support groups in the area and the large medical community for the Walk's third year. Her incredible team and support system are back to walk with her again this year. As the MG Walk Hero, she hopes to inspire others to keep their positivity through their new reality with MG. She has been through so much since her diagnosis, but there is a light at the end of the tunnel. She is working hard to raise the funds and awareness needed for MG patients so there can be better treatments, no complications, and ultimately a cure for MG. ✨



Do You Want to Wake up to a World without MG?

YES?

Then help by joining the MG Patient Registry

The MGFA Patient Registry is helping to expand our knowledge of MG and move us closer to improved treatments and a cure. By making a patient community more accessible and understandable, a patient registry and its bounty of information can encourage pharmaceutical developers to pursue drug discovery in a disease.

To learn more about the registry please visit www.myasthenia.org home page banner and click on the banner when it turns to MG Patient Registry. Or, call the MGFA office at (800) 541-5454 and request the *MG Patient Registry* brochure.





Registration for the 2017 MG Walk Season is now OPEN!

With the completion of 2016, the MGFA celebrated its most successful MG Walk Campaign to date. In 38 locations all across the country, 5,000 participants and over 530 teams, came together to raise awareness and vital funds for the MG community, and helped the campaign raise more than \$800,000!

2017 will mark the 7th year of the MG Walk Campaign, and registration is open at www.MGWalk.org. Now is the perfect time to reactivate your teams (or start one for the first time!) and help us to bring even more resources to the MG community by helping us reach our 2017 goal of \$900,000!

The MG Walk Campaign is the flagship national awareness and fundraising campaign for the MGFA, and the perfect opportunity to become active in the amazing MG community, and help us get one step closer to our ultimate goal...a world without myasthenia gravis.



2017 MG Walks

- NEW ENGLAND:** May 6
- NORTHERN WISCONSIN:** May 6
- SOUTHERN WISCONSIN:** May 6
- TRISTATE:** June 10
- CONNECTICUT:** June 11
- INDIANAPOLIS:** September 10
- CLEVELAND:** September 23
- CHICAGOLAND:** September 24
- LONG ISLAND:** TBD Fall
- DC METRO:** September 30
- PORTLAND:** September 30
- GREATER DELAWARE:** October 1
- SEATTLE:** October 1
- GETTYSBURG:** October 8
- SO ILLINOIS:** October 15
- COLORADO:** TBD Fall
- UTAH:** TBD Fall
- KENTUCKY:** TBD Fall
- AUSTIN:** TBD Fall
- HOUSTON:** TBD Fall
- GREATER LA:** November 18
- SACRAMENTO:** November 18
- INLAND EMPIRE:** November 19
- BAY AREA:** November 19
- ARKANSAS:** December 2
- ARIZONA:** December 3

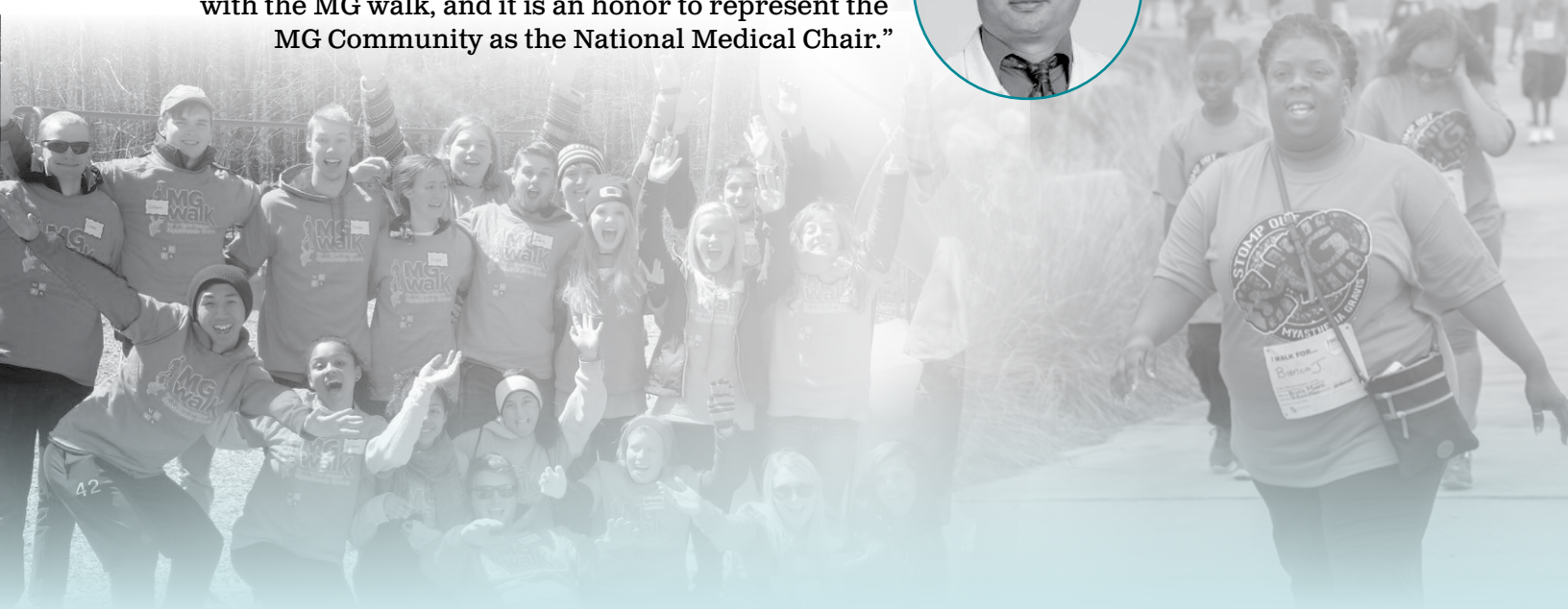


The MG Walk Team is here to make sure you have the best possible experience. We're also here to assist you with setting your goals, increase team recruitment, and tailoring a personal fundraising plan to insure your fundraising is a success! Contact us anytime at 1-855-MG-Walks (1-855-649-2557) or at Info@MGWalk.org. ✨

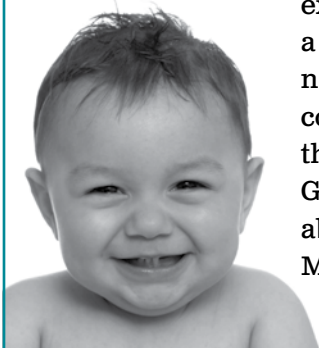


MEET OUR 2017 NATIONAL MG WALK HERO, ALEXIS RODRIGUEZ! "Spreading MG awareness is not something you just do once a year...it's something you do every single day."

MEET OUR 2017 NATIONAL MG WALK MEDICAL CHAIR, DR. YUEBING LI "This will be my third year with the MG walk, and it is an honor to represent the MG Community as the National Medical Chair."



SMILE FOR MGFA



Did you know you could donate to the Myasthenia Gravis Foundation of America while shopping on amazon? It's simple to set-up on an existing account or by creating a new one. You can shop as you normally do, there's no change in cost or convenience to you. Tens of thousands of products are covered. Go to <http://smile.amazon.com/> about to learn more and make MGFA your charity!

Join the fun and use MGFA's new Twibbon to promote awareness of MG.



Go to <https://twibbon.com/support/mgfa-june-awareness> to use the new Twibbon on your social media sites.



New MGFA Board Members

KATHERINE RUZHANSKY, MD, MS



The MGFA is thrilled to be welcoming 2 new board members in this issue of MG in Focus. First, meet Dr. Katherine Ruzhansky, MD, MS, Assistant Professor of Neurology, MUSC, Neuromuscular Division, Director of the EMG Lab. Katherine shared her story with MG in Focus:

I am currently working as a clinical neurologist, Assistant Professor of Neurology and Director of the EMG lab at the Medical University of South Carolina. Prior to relocating to Charleston, SC, I trained at Yale-New Haven hospital and Columbia University Medical Center in neurology and clinical neurophysiology. My clinical focus has been the diagnosis and treatment of neuromuscular disorders.

Since coming to Charleston, I have met many wonderful people and patients who are active on the local and national level in helping bring awareness to Myasthenia Gravis. Treating patients with MG is both challenging and rewarding. Although it is a treatable disease, living and dealing with chronic illness poses daily challenges not only for patients, but also families and caregivers. By getting involved with the local support group and by actively supporting the MG community, I hope to grow as a physician and a person.

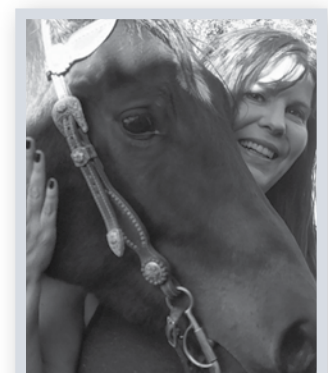
I am excited and humbled to serve on the National Board of Directors. I am hoping to bring my experiences from the provider perspective, and learn from patients, caregivers and leaders.

CELIA MEYER, RN

MGFA is also proud to welcome patient Celia Meyer. Many of you may know Celia from social media focused on MG. Here's Celia's story:

My name is Celia Meyer and I am a retired Navy veteran, with a degree in Information Science. I also have a nursing degree and am an RN and worked in the area of anesthesiology. As well, I am experienced with web design, software development, testing and installation. I have worked as a patient advocate while in the US Navy and coordinated and managed the care for patients in a multi-state area, ensuring minimized cost while establishing continuity of care. I have experience as an instructor and educator and served as a hospital corpsman and EMT, also while serving in the US Navy. I have tutored in math and computer labs during college. I ran my own successful horse business for 5 years, specializing in sales of custom horse barns, as well as raising and showing American Quarter Horses in Reining and Reined Cow Horse events.

Currently I am the administrator of three Facebook Support and Awareness Groups for Myasthenia Gravis. My goals are to make available to every new patient diagnosed with MG, information sources on the disease, information on MGFA and the MG Patient Registry. It is my desire that every individual and their caregivers be educated about MG, the treatments and side effects of treatment, and have these resources readily accessible. Education is key in assisting new patients in getting the care they need. I believe we need to raise awareness not only to the general public, but to the



Celia Meyer and friend, "All About Chrome" aka Marley

medical community that serves these individuals.

As a new board member I hope to increase awareness and assist patients in every aspect of their journey. I am currently retired and have the time to spend, working together with the rest of the Board of Directors to help facilitate new programs to help patients become better advocates for their own care.

I am a resident of Beaufort, SC where I reside on a 3 acre Plantation with my husband and numerous horses, donkeys, dogs and soon, some laying hens! I love being outdoors and active. I promote several dog rescues and assist when I can as a Foster Mom.

In October 2013 I was diagnosed with Myasthenia Gravis. I had had numerous symptoms over the course of many years but no one had put all the pieces together. After an incidental finding of a thymoma on a chest CT, a friend, who is a nurse practitioner, suggested I rule out Myasthenia Gravis, as her cousin had recently been diagnosed with the disease. After confirmation I went on a mission to raise awareness so that others will find answers and can begin a treatment regimen as soon as possible. Subsequently I was diagnosed with Refractory Myasthenia Gravis. I am currently receiving IVIG every three weeks for treatment. I have recently changed my diet and feel it is making a difference in my overall health. I have started to ride again and enjoy spending time with my equine partner, Marley.

My name is Celia Meyer and I have MG. It is what I have, but not who I am. I want to help others and feel I can serve the Board of Directors in that mission. ✨

WELCOME KATHERINE AND CELIA TO THE MGFA BOARD!

Programs and Resources

THE MG FRIENDS PROGRAM

The MGFA offers a one-on-one phone support service, the MG Friends program, which is uniquely designed to assist newly diagnosed patients, family members and caregivers within the MG Community.



Our MG Friends have faced similar challenges and are a valuable resource for their fellow MG community members. As highly trained and experienced volunteers, MG Friends provide patients and family members with practical advice, shared experiences, an understanding ear, as well as emotional and confidential support – no matter where you are located in the United States.

Connect with an MG Friend today by dialing 1-800-541-5454 or by visiting the MGFA website and completing an online referral form on the Support Group Page.

Interested in becoming an MG Friend volunteer? We are currently accepting applications online [How Can I Help](http://www.myasthenia.org/HowcanIhelp.aspx) [<http://www.myasthenia.org/HowcanIhelp.aspx>]. ✨

In remembrance

CLAUDIO MAZIA, MD

We regret to announce that Claudio Mazia, MD, one of the world's leading neurologists dedicated to the study of myasthenia gravis, died recently. Claudio was Head of the Neurological Division in the Institute of Medical Research "A Lanari", of the University of Buenos Aires, Argentina and President of the Argentine Association of Myasthenia Gravis. His dedication during the performance of the MGTX trial, which demonstrated the value of thymectomy for MG patients, was key to the trial's success. Claudio recently completed a textbook, Myasthenia Gravis and Related Problems, along with his many colleagues and friends from across the world. He is survived by his wife and two daughters. – Henry Kaminski, MD. ✨



2016 Scientific Session

By Robert Ruff, MD, PhD



The 2016 Scientific Session of the MGFA was held on September 14, 2016 in New Orleans at the Hilton Riverside Hotel. The meeting was chaired by Drs. Jeff Guptill and Mike Hehir, both of whom were awarded MGFA/AAN Fellowships. There was an audience of about 200 people. Arthur Strauss was in attendance and was recognized for his work as a pioneer in MG research in the mid-1950s to early 1960s. An interesting change in the format this year was to have a break in the middle for selected posters to be presented by

the authors. This was a good opportunity for people to present their work without the pressure of standing in front of 250 people. I liked this touch. The talking posters were an excellent vehicle for young investigators to present their work and get feedback. I thought that this feature should be continued. In the text below I will provide brief discussions of the platform presentations.

DR. SANDERS (DUKE UNIV., NORTH CAROLINA) – International Consensus Guidance for the Management of Myasthenia Gravis

The study was published in the professional journal *Neurology*. An Executive Summary of the report is on the MGFA website. Why do we need guidelines? 1) There have been till now, no accepted standards of care for MG. 2) MG has variable manifestations (remember it is a snowflake disease) and no one treatment is appropriate for all patients. 3) There are few clinicians who are expert in managing people with MG. 4) Guidelines will help justify treatment strategies to insurance companies. How did the guidelines develop? MGFA MSAB appointed a task force in 2013, Drs. Sanders and Gil Wolfe (Buffalo NY) were co-Chairs and Dr. Pushpa Narayanaswami (Boston, MA) organized the task force. The task force used the RAND-UCLA appropriateness method for evaluating each treatment or diagnostic criteria associated with MG. The evaluation process involved panel discussion over several rounds until an intervention achieved consensus or was rejected. The Task Force established the goal for interventions of achieving minimal disease

manifestations with an intervention incurring only acceptable side effects. The Executive Summary details specific interventions and diagnostic criteria. The Task Force work was supported by the MGFA.

DR. SENGUPTA (WASHINGTON DC, MENTORS ARE MSAB MEMBERS DR. HENRY KAMINSKI AND LINDA KUSNER) – Micro RNA profiles associated with ectopic germinal centers formation in thymus samples of patients with Myasthenia Gravis.

The Thymus glands of MG patients differ from healthy Thymus glands in terms of having ectopic germinal centers where sensitized B-cell type of lymphocytes (type of white blood cell) develop and mature. These B-cells produce disease causing antibodies. In patients without MG, the thymus basically atrophies and turns to fat as people age. The researchers studied thymus gland samples from the Thymectomy Trial. They found that gene regulation was changed in the ectopic centers. About 100 genes were either up or down-regulated in ectopic centers compared to control thymus glands. The largest effect was for genes associated with cell death or cell survival, and

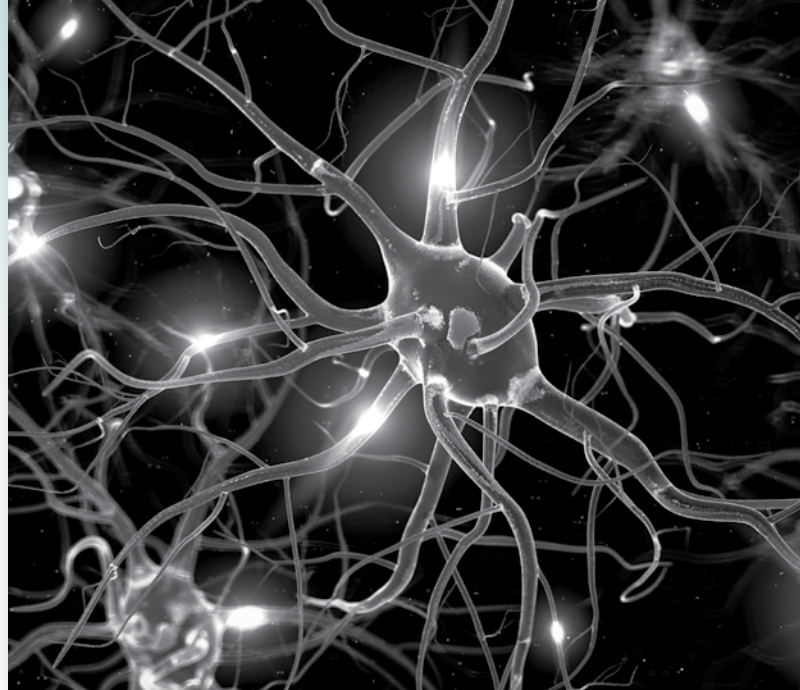
inflammation. Micro-RNAs can alter expression of genes. They identified 34 micro-RNAs that were present in excess. The micro-RNAs in the ectopic centers blocked expression of proteins that would normally inhibit the genes that were up-regulated in association with ectopic centers. Therefore the genetic activity suggests enhanced survival of antibody producing B-cells. The observations explained how germinal centers generate disease causing B-cells. The data also may partially explain why people with MG may manifest other antibody-mediated autoimmune disease and suggest that if the cell proliferation in a germinal center runs amuck that a germinal center could progress to a tumor.

DR. RUSSO (DUKE UNIV., DURHAM NC, MENTOR IS DR. JEFF GUPTILL) - B10 deficiency in MG is not associated with defective T-Cell Suppression.

B10 cells are B lymphocytes that produce interleukin IL-10, a protein that suppresses inflammation. B10 cells are known to be deficient in MuSK MG patients. This study examined B10 cells in AChR + MG patients. They reported decreased IL-10 production in patients with MG, with greater IL-10 reduction in severe compared to mild MG. The T-cells in patients with MG could be suppressed by IL-10 and B10 cells in patients with MG produced normal amounts of IL-10. Therefore, patients with MG due to AChR antibody have decreased anti-inflammatory IL-10 production caused by a paucity of B10 cells. Treatments to enhance B10 cell production may be a new strategy for treating MG.

DR. O'CONNOR (YALE UNIV., NEW HAVEN CT) – B Cell tolerance and abnormal B cell repertoire formation in MG.

This first part of the presentation focused on immune tolerance. Tolerance is a process whereby B-Cells do not attack a person's own proteins. Autoimmune disorders develop when tolerance is disrupted. Under normal circumstances, tolerance increases in B-cells as they mature. In people with autoimmune disorders the process of self-tolerance does not increase during maturation of B cells. Dr. O'Connor's group studied MG patients who had antibodies to AChR or Musk (Musk is a protein at the neuromuscular junction associated with AChR presence on muscle at the



neuromuscular junction where the nerve to the muscle and muscle interact). In patients with AChR and MuSK MG, B cells have impaired self-tolerance and about half make antibodies that attack the self. Therefore, MG is associated with altered B cell maturation resulting in B-cells that make antibodies that attack the body. This part of the study explained why people with MG are also prone to other autoimmune disorders.

The second part of the presentation examined types of antibodies produced by B-cells. The initial result is that the patterns of the antibodies produced in both forms of MG were different compared to antibodies produced by control subjects. The researchers are now evaluating how and why B-cells produce antibodies to MUSK and AChR. The altered activity patterns of B-cells in MG may provide another way to diagnose antibody-negative (seronegative) MG.

DR. BORODOVSKY (Alnylam Pharmaceuticals is a novel pharmaceutical company that is focusing on the clinical application of small pieces of RNA that interfere with the expression of specific genes. Drs. Kaminski and Kusner collaborate with Alnylam to determine if interfering RNA strategies could work for people with MG) – ALN-CC5 is an investigational RNA Therapeutic for the treatment of MG: there is interim phase 1 data in healthy volunteers and the drug has shown efficacy in pre-clinical animal models of MG.

This was a two part presentation of a new agent, ALN-CC5. ALN-CC5 inhibits the production of an essential component in the complement

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pathway, C5. Complement is a chemical system that attacks proteins that are identified by antibodies. In MG, antibodies to AChRs identify the AChRs as targets to be destroyed by complement. Using ALN-CC5 to silence C5 in rat models of MG, reduced disease severity. Silencing C5 early can inhibit disease onset. Treatment with ALN-CC5 after rats are symptomatic reduced symptom severity. A phase 1/2 study of ALN-CC5 is underway. The Phase 1/2 study evaluates how well people tolerate ALN-CC5, what dose of ALN-CC5 suppresses complement in people and how long a dosing suppresses complement. ALN-CC5 is given by injection. The initial findings of the Phase 1/2 study are that ALN-CC5 was tolerated without acute side effects (this means that the side effects for people receiving ALN-CC5 were comparable to the side effects in people getting placebo and none of the side effects were serious). Alnylam found the dose that suppressed complement in people and suppression of complement lasted for many weeks. The next step is to test ALN-CC5 in people with MG to determine if it is tolerated and if it reduces the severity of MG. Note that ALN-CC5 blocks inflammation by acting on a similar pathway to antibodies against C5 (as Eculizumab does).

DR. HEHIR (UNIV. OF VERMONT, RECIPIENT OF MGFA/AAN FELLOWSHIP) – Blinded retrospective Analysis of Rituximab Treatment in Anti-Musk MG.

MG associated with antibodies directed against the neuromuscular protein called Musk accounts for about 1/3 of MG patients who do not have antibodies against the AChR and corresponds to about 7% of all MG patients. About 4-5000 people in US have Musk MG. Among people with Musk MG, about 90% have moderate or worse disease severity. Many people with Musk MG require immune-suppression treatment. Rituximab is a monoclonal antibody treatment that targets B-Cell lymphocytes that is recommended for treating Musk MG. However, people with Musk MG incur problems with insurance companies refusing to cover Rituximab. In order to more strongly justify to insurance companies and others, Dr. Hehir organized a large international multicenter trial of Rituximab vs. placebo in people with Musk MG.

This was an initial report of 68 patients based upon the results from some of the contributing centers. The trial looked at decreasing disease severity, reduction in steroid dose and number of hospitalizations. Rituximab significantly reduced disease severity, but the reduction in steroid dosing was not significant due to the small sample size of this initial partial report. When the study is completed, it will hopefully provide strong support for people with Musk MG to receive Rituximab.

DR. NOWAK (YALE UNIV., NEW HAVEN, CT) – Phase 2 Rituximab trial for MG

This trial used the NIH Clinical Trials Network of participating 26 US sites that are equipped to perform clinical trials. This trial is evaluating Rituximab therapy in people with AChR MG. The target population was 50 patients who had a stable course on mestinon plus prednisone or prednisone plus immunosuppressive treatment. Subjects received weekly infusions for 4 weeks then a second course of infusions after a delay. There were 25 subjects who received Rituximab and 25 received placebo. Target was steroid sparing potential (want to see 75% reduction in steroid dosing). The study reviewed 4030 charts from which 1833 subjects were selected for pre-screening. The study enrolled 68 subjects and after further evaluation 52 subjects were selected and randomized. So far 56% of subjects completed the study, 10% (5 subjects) terminated and the rest are in progress. The reasons for termination included emergence of other conditions that contraindicated using Rituximab. Dr. Nowak expects completion in late 2017.

DR. HOWARD (UNIV. OF NORTH CAROLINA) – A phase 3 trial for Eculizumab in patients with refractory MG.

This was an International multi-site study in which subjects with AChR MG who had inadequate responses to immunosuppressive treatments other than monoclonal antibodies. Eculizumab is a preparation of antibodies directed against C5 a key element in the complement cascade (also look at the study above on ALN-CC5). The study contained 62 subjects who received Eculizumab and 63 who received placebo. The primary



is a good target disease. The pathophysiology of MG is well understood, which makes it easier to determine how an agent works.

DRS. SIDDIQI AND BLACKMORE (UNIV. OF ALBERTA) – Beyond the Antibodies: Sera metabolomic biomarkers signatures discriminate myasthenic and healthy cohorts.

Proteomics looks at the range of proteins produced by a person. Metabolomics examines the spectrum of metabolites present in the blood of an individual. Metabolites include the small molecules that are involved with or produced by ongoing cellular activity. Blood serum is processed through a mass spectrometer to evaluate the presence and contribution of thousands of these metabolite chemicals. The analysis process is complex. The researchers studied 150 patients - 50 with MG, 50 with rheumatoid arthritis (RA) and 50 controls. The subjects were age and gender matched because both age and gender influence metabolomic profiles. The metabolomic profiles of blood from MG patients differed distinctly from patients with RA and controls. The analysis evaluated levels of 5711 metabolites in the blood samples. This research is in a very early stage. In the future the metabolomic profile may be a “biomarker” for MG. Such a biomarker would provide a way to confirm for health care payers that an individual has MG even if that person does not have detectable antibodies to AChR, Musk or any other antibody associated with MG that may be disclosed in the future. ✨

endpoint was change in MG-ADL score at 26 weeks of treatment. Secondary endpoints were other measures of MG severity. The primary endpoint was barely missed $p=0.0698$ (needed to be less than 0.05). Dr. Howard felt that the method used to determine the probability of the primary endpoint being different in those taking Eculizumab vs. control subjects resulted in the p value being too large. Several of the secondary endpoints were achieved. The company is going to approach the FDA to have Eculizumab for use in MG based upon the positive results for the secondary outcome measures. The problem is Rituximab and Eculizumab, which are approved for conditions other than MG, cost about \$400,000 per year for treatment. The out of pocket costs for these agents for people who have insurance coverage are probably lower than the “sticker price” for the two monoclonal antibody treatments (mab indicates monoclonal antibody). Because these treatments are expensive, insurance companies have to pay large amounts if an individual is approved to receive one of these treatments. People with MG need help for third party payers to approve covering expensive treatments. Clinical trials are powerful instruments to get agents covered. By supporting and publicizing clinical trials, the MGFA continues to work for people with MG.

DR. KUSNER (GEORGE WASHINGTON UNIV., WASHINGTON DC) – Survivin in MG

Survivin is a protein that inhibits programmed cell death (apoptosis). T-cell survival and B cell proliferation is enhanced by Survivin. Survivin plays roles in multiple sclerosis and rheumatoid arthritis. For both diseases increased Survivin levels are associated with worse disease course. Dr. Kusner used a rat model of MG in which animals made AChR antibodies. Treating rats with Survivin vaccine induced Survivin Abs, reduced Survivin levels and Survivin-producing B cells. Vaccine treated rats had less severe disease and lower AChR antibody and Survivin levels compared to control rats. I suspect that Survivin enhances inflammation by prolonging the life of inflammatory cells. I spoke with Dr. Kusner after her presentation. She told me that the person who developed the Survivin vaccine is planning a phase 1 clinical trial (to determine the toxicity of Survivin vaccine in people), which is a good thing. Reducing Survivin levels may be useful for some autoimmune disorders and not others. MG

MGFA Support Groups

SUPPORT GROUPS ARE CATERING TO PATIENT NEEDS ACROSS THE COUNTRY

“After corresponding with a mother of a newly diagnosed 17 year old for over a year, I knew that a meeting near her neighborhood would be beneficial,” says one leader of the Connecticut MG Support Group, Toni Brown, as she describes the essence of the group. “Essentially, we bring the Support Group to our patients.”

With keeping every patient in mind, the Connecticut MG Support Group has developed a long standing relationship with its members in the Connecticut Community. Terri Adams, Toni Brown and Ed Czackes are the current leaders who uphold the group’s continued success.

This past winter, leaders of the Connecticut MG Support Group planned a sponsored “Meet and Greet” at Salute Restaurant in Hartford, CT. Welcoming Neuro-Ophthalmologist, Dr. Danielle Rudich and Irene Scanlon, LCSW as guest speakers, members received educational information and information on state services, programs and entitlements. Following the presentation Dr. Rudich and Irene Scanlon gave patients and their caregivers or families the opportunity to speak with them one on one.

Over the years, the Connecticut MG Support Group has partnered with HomeIGRx’s Jami Williams Corbett, to receive sponsorship for their meeting locations and meals. “Our Support Group meetings

are casual and intended to be a day out,” says Terri. “Jami has been an asset to the CT Support Group Team, as her efforts have allowed us to cater to our patients.”

LOOKING FOR A SUPPORT GROUP NEAR YOU?

Support Groups are critical to service delivery for the MGFA. Our groups provide support, education, and mutual aid. The Support Group Calendar can assist you in finding a local support group near you. Go to www.myasthenia.org to find these pages.

Interested in becoming a Support Group Leader? We are always looking for new leaders in the MG Community and currently accepting applications. Visit [How Can I Help](#).

PATIENT RESOURCES GUIDE

The MGFA patient resources guide is here to help those who may be experiencing job loss, seeking prescription assistance, Medicare assistance, social security information and medical equipment. Here’s a sampling of some of the listings, for more go to Patient Resources.

Job Accommodation Network (JAN):

JAN’s mission is to facilitate the employment and retention of workers with disabilities by providing employers, employment providers, people with disabilities, their family members and other interested parties with information on job accommodations, self-employment and small business opportunities. <http://www.jan.wvu.edu/media/MG.html>

Medicare Rights Center: For people with Medicare. The Medicare Rights Center helps older adults and people with disabilities get good, affordable health care. <http://www.medicarerights.org>

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Can MG Cause Urinary incontinence?

URINARY incontinence itself is not rare, but is only rarely seen as a manifestation of myasthenia gravis.¹⁻³ When it does occur in someone with MG, other causes of the incontinence should be investigated. To demonstrate that MG is the cause of incontinence, it should be shown that it improves or worsens with change in MG severity.

DON SANDERS, MD, PROFESSOR OF NEUROLOGY, DUKE UNIVERSITY SCHOOL OF MEDICINE

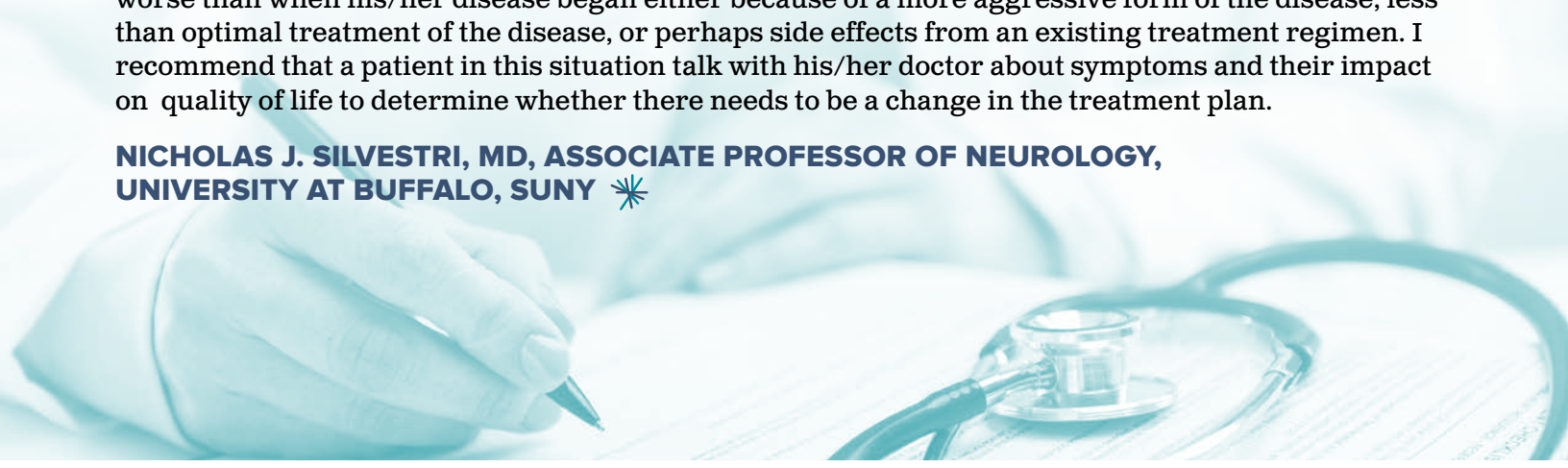
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3. Howard JF, Jr., Donovan MK, Tucker MS. Urinary incontinence in myasthenia gravis: a single-fiber electromyographic study. *Ann Neurol* 1992;32:254.

Is MG a progressive disease?

To answer your question succinctly, MG is usually not a progressive disease. The course of MG is extremely variable from patient to patient. According to several studies that have looked at how MG behaves in relatively large groups of patients, most patients with MG can expect to have the most severe symptoms in the first 2 to 3 years after they begin. After that, the disease is usually less active in the majority of patients which could be due to the characteristics of the disease itself (for example, the antibody attack may be less potent), or due to treatment of the disease with medications or perhaps removal of the thymus gland in selected patients. That being said, MG can be very difficult to remedy in up to 15% of patients despite adequate treatment, and an even larger percentage of patients continue to have milder but at times disabling symptoms despite treatment. At other times, the side effects of certain medications used to treat MG, such as corticosteroids like prednisone, can also hinder a person's health and their quality of life. It is possible that a patient's symptoms seem worse than when his/her disease began either because of a more aggressive form of the disease, less than optimal treatment of the disease, or perhaps side effects from an existing treatment regimen. I recommend that a patient in this situation talk with his/her doctor about symptoms and their impact on quality of life to determine whether there needs to be a change in the treatment plan.

NICHOLAS J. SILVESTRI, MD, ASSOCIATE PROFESSOR OF NEUROLOGY, UNIVERSITY AT BUFFALO, SUNY ✨



THE STATE HAS A PLAN FOR YOUR LEGACY. DO YOU?

Fewer than half of Americans have a will. Without a will, the state will decide how to distribute your hard-earned money. Don't lose control of your legacy. Visit an attorney and prepare a will and when you do, remember the MGFA. Make part of your legacy "A world without Myasthenia Gravis"





Pitfalls and lessons learned: BiPAP for myasthenia gravis



BiPAP (Bi-level positive airway pressure) is a common intervention to help avoid intubation for MG patients who are in myasthenic crisis. BiPAP functions like a ventilator but uses a tight-fitting mask on the face rather than a tube down into the trachea. BiPAP provides breathing assistance with air pressure to push air into the lungs, reducing the work of weakened respiratory muscles. The air pressure drops immediately after each inhalation to allow patients to exhale easily.

I have had myasthenia gravis for the past 6 years and last year I noticed that it was hard for me to breathe when I was laying down flat in bed. I realized that my respiratory function was declining so I went to see a local pulmonologist in the hope of getting a home BiPAP machine to help me breathe. This should have been a relatively simple endeavor. It was not. I am sharing my story to hopefully prevent other MG patients from having to go through the same problems that I went through.

PITFALL #1 – THE DOCTOR

I chose a local pulmonologist that I had worked with in the past. He was knowledgeable and communicated well with nurses so I figured he would be able to meet my needs. Right away I should have realized that he was not a good choice when he said “I hope you have sleep apnea so you can get a CPAP machine.” He was knowledgeable

about sleep apnea and COPD but had no understanding of neuromuscular respiratory management. He ordered pulmonary function testing but I had to ask the technician to perform the MIP and MEP tests to check respiratory muscle strength. He did not understand that CPAP is a poor choice for MG as the weak respiratory muscles cannot exhale against the high pressure and BiPAP is almost always needed instead. After many frustrating appointments, I transferred my care to a pulmonologist who specializes in neuromuscular disease and ventilator management.

Lesson learned

Find a pulmonologist who is familiar with neuromuscular disease, usually connected with a major university hospital or MDA clinic.

PITFALL #2 – INSURANCE CRITERIA

The insurance coverage protocols are very different for sleep apnea than for neuromuscular disease. Patients with sleep apnea are required to start with CPAP machine before insurance will pay for a BiPAP machine. The criteria for patients with neuromuscular disease are much different. Patients with MG automatically qualify for BiPAP if their MIP is < 60 mm H₂O under the qualifying guidelines used by most insurance companies. My MIP was 30 so I definitely met the criteria for BiPAP. Unfortunately neither the medical supply company nor the doctor understood this so I had to try a CPAP machine and they could not understand why it was not working for me. Many stressful phone calls were required to get the CPAP switched to the BiPAP machine that I now use.

Lesson learned

Take the time to learn about insurance guidelines for BiPAP for patients with neuromuscular diseases, and provide copies for doctors as needed. A good starting point can be found at http://www.resmed.com/us/dam/documents/articles/1010293_RAD_Guidelines.pdf.

PITFALL #3 — BIPAP SETTINGS

I wound up in the hospital with an acute MG exacerbation just two days after finally getting my BiPAP machine. IV steroids and rest made my breathing easier but unfortunately my improved respiratory function led to new problems with my BiPAP machine. My BiPAP settings needed to be adjusted but my doctor was only familiar with sleep apnea management, not neuromuscular disease. My BiPAP settings were soon so incorrect that my blood gases were unstable and my respiratory status began to deteriorate again. The doctor did not understand what was going on and managed to make things even worse. In the end, I insisted on being discharged home where I figured out the correct settings and my breathing improved again.

Each morning my BiPAP machine records how my breathing was during the night and if there was air leakage around my mask. I check my results each morning and contact the vent clinic when settings need to be changed. My BiPAP also saves the information onto an SD card inserted into the side of the machine. I take the SD card with me to appointments so that the respiratory therapist and doctor can review my information while my machine stays safely at home.

Lesson learned

BiPAP settings need to be adjusted carefully to reflect the current respiratory status of the patient. BiPAP settings that are too low lead to the patient feeling short of breath and retaining too much CO₂ in the blood. BiPAP settings that are too high lead to too much oxygen in the brain that can cause dizziness, confusion, heart palpitations, and periods of apnea (times when the patient stops breathing for a time).

BOTTOM LINE

I now use my BiPAP machine every night and also during the day as needed when I feel short of breath. I have noticed that I sleep better and wake up less frequently during the night. When my breathing gets worse during a flare, my BiPAP machine can be adjusted as needed to help keep me from being hospitalized.

Patients with MG respiratory muscle weakness should not wait until breathing gets really difficult to consider getting a home BiPAP machine. Signs that BiPAP is indicated include:

- **Pulmonary function testing shows MIP is < 60 mm H2O**
- **Shortness of breath when laying down**
- **Waking up during the night feeling short of breath**
- **Restlessness when trying to sleep**
- **Difficulty waking up or feeling sleepy during the day**
- **Waking up with a headache**

It is best to get a BiPAP machine before breathing becomes severely compromised. This provides patients the opportunity to become comfortable with the machine, mask, features and settings. A wide variety of BiPAP machines and masks are available and sometimes it takes a few tries to figure out what works best. Regular follow-up with respiratory therapy and pulmonology doctors is also essential. ✨

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Novartis Patient Assistance Program:

Novartis offers medications to people who meet certain income requirements. Novartis manufactures Sandimmune® (cyclosporine) and Neoral® (cyclosporine). A medical professional must call this program to request the application. 1-800-277-2254

Patient Advocacy Foundation: A national non-profit organization that serves as an active liaison between the patient and their insurer, employer and/or creditors to resolve insurance, job retention and/or debt crisis matters relative

to their diagnosis through case managers, doctors and attorneys. Patient Advocate Foundation seeks to safeguard patients through effective mediation assuring access to care, maintenance of employment and preservation of their financial stability.

www.patientadvocate.org/help.php

The organization's services are provided strictly for informational purposes and are presented only as possible resources for patients. Please note that MGFA is not in any way associated with the companies or organizations referenced above, and we do not endorse their services. ✨



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

NICHOLAS J. SILVESTRI, MD, Associate Professor of Neurology, University at Buffalo, SUNY

**GIL I. WOLFE, MD, Professor and Chairman;
IRVIN AND ROSEMARY SMITH, Chair of Neurology, University at Buffalo, SUNY**

In men, myasthenia gravis (MG) often presents later in life, raising the possibility that age-related declines in the male hormone testosterone levels may play a role. A recent study performed at the University of Oxford of 5,031 men with known low testicular function calculated an adjusted rate ratio or likelihood of developing MG of 4.25 times higher than in men with normal testicular function.¹ Four MG cases were observed, all between ages of 45-74, when less than one would have been expected. The authors note that low testicular function in general has been associated with increased risk of autoimmune diseases such as MG. The number of MG cases was small, however, and the authors expressed caution and the need for further study.

In data extracted from the Duke University MG Patient Database, Andersen et al.² analyzed factors impacting outcome in the disease, including patient demographics, antibody levels, thymus histology, and clinical severity. Outcomes were investigated after 2, 5, and 10 years of treatment. All patients had been treated for at least 2 years by a single MG expert at Duke. Treatment was individualized with the goal of achieving an optimal outcome of minimal manifestation status (a term implying low disease activity) or better, while simplifying the treatment regimen and tapering

medications, but maintaining optimal clinical status. Thymectomy was recommended in all generalized patients with onset prior to 50 years, including MuSK-antibody positive MG patients. Azathioprine, mycophenolate, or cyclosporine were the typical steroid-sparing medications used. Data was available on 268 patients, 74% of whom had acetylcholine receptor (AChR) and 5% MuSK antibodies. Interestingly, 57% were male. Thymectomy was performed in 40% with 22 patients having thymoma, all acetylcholine receptor-antibody positive. Seven MuSK MG patients underwent thymectomy, with 3 showing hyperplasia and 4 a normal thymus. The study includes a large amount of interesting clinical information, but cutting to the treatment outcomes, the main point is that age at onset was the only consistent independent predictor. Patients with onset after age 50 years were more likely to reach an optimal outcome. Although clinical severity based on MGFA classifications tended to be worse for MuSK MG patients early in the disease course, optimal outcomes did not differ between antibody populations at 5 and 10 years. The improved prognosis for onset after age 50 runs counter to other studies, but could be explained by the center's aggressive treatment approach in this age group, with a higher proportion receiving

immunosuppression compared to other reports. Finally, outcomes did not correlate with a particular treatment modality. By 10 years, 75% of patients had an optimal outcome, only 3% had a poor outcome, and these findings were not impacted by age, maximum disease severity, or antibody status. Although only 7% of patients reached complete stable remission off all medications, none of whom were MuSK-positive, the study clearly reflects the favorable prognosis over time in a large majority of MG patients.

Fatigue is a commonly reported symptom in MG. Prior studies have reported fatigue in 75-89% of patients, with a significant impact on quality of life.^{3,4} A recent study by Hoffmann et al⁵ aimed to again assess the prevalence of fatigue in MG and evaluate its impact on both quality of life and activities of daily living. The authors also examined the factors associated with fatigue, including both MG-specific factors and the presence of other disorders such as sleep or mood disorders such as depression. They performed a study of 200 patients with MG, most of whom had generalized disease and acetylcholine receptor antibodies. The prevalence of fatigue in this cohort as measured by the Chalder Fatigue Scale was 56.1%. The presence of fatigue was associated with higher quantitative myasthenia gravis (QMG), MG quality of life (MG QoL-15), and MG activities of daily living (MG-ADL) scores, all highlighting the negative impact of this symptom in patients with MG. The presence of depression and higher MG disease severity were both associated with higher rates of fatigue. The authors conclude that future trials in MG should include fatigue as an endpoint to determine whether its prevalence changes with improvement in disease status over time.

Autoimmune thyroid disorders are frequently seen in conjunction with MG. A recent study out of Poland aimed to determine the prevalence of thyroid disorders in early-onset (diagnosis before 50 years of age) and late-onset MG, and MG associated with thymoma.⁶ In their study of 343 patients aged 4 to 89 years, the authors discovered that autoimmune thyroid disorders were diagnosed in 92 (26.8%). Of these, 4.4% had Graves' disease, 9% had Hashimoto's thyroiditis, and 13.4% had elevated levels of anti-thyroid antibodies without symptoms of thyroid disease. These rates are all higher than in the general population. There was no difference in the presence of autoimmune thyroid disorders in patients with either early or

late-onset MG; patients with thymoma were found to have a higher incidence of non-autoimmune thyroid disorders. The authors noted that the patients with both MG and autoimmune thyroid disorders in their study were less frequently treated with immunosuppressants for MG, possibly indicating a milder disease course.

On the congenital myasthenic syndrome (CMS) front, although defects in the muscle side of the neuromuscular junction (known as the postsynaptic side) are most common, there have been recent advances in identification of causes on the nerve side (known as the presynaptic terminal), for instance mutations in the genes SYT220 and SNAP25.⁷ Since implicated presynaptic genes are present beyond the neuromuscular junction, symptoms and signs beyond muscle weakness have been observed in patients including memory and language difficulties. The gene that contains a single portion of the choline acetyltransferase gene (which leads to the creation of the enzyme responsible for the formation of acetylcholine--the chemical used in nerve and muscle communication or neuromuscular transmission) and also encodes the vesicular acetylcholine transporter (VACHT), SLC18A3, has now been implicated in CMS as well. Two patients were described presenting with typical features long associated with presynaptic defects: droopy eyelids (ptosis), weak eye muscles, feeding difficulty, fatigable weakness, and episodes of breathing difficulty presenting early in the neonatal period.⁸ Relatively high doses of pyridostigmine partially improved not only muscle strength but also the mild heart muscle dysfunction seen in one of the patients. One child also received ephedrine with further improvement. Mutations of the SLC18A3 gene were found in both. Of note, this form of CMS is the first to be associated with heart dysfunction, a feature that has been observed in the VACHT mouse model. Although heart involvement in the one child could have been incidental, the authors do recommend cardiac surveillance in this form of CMS until the disorder is better defined. As for treatment with pyridostigmine, the authors do remind us that this intervention can worsen several forms of postsynaptic CMS, including those arising from mutations that encode DOK7, MuSK and LRP4.

Since most forms of CMS arise from autosomal recessive mutations (where 2 abnormal copies of a gene are needed to manifest disease) - including

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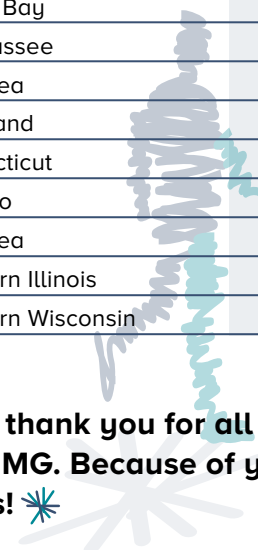
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Jamee Emens	Team Emens	Arizona	\$5,165
Debora Buzinkai	Team Zippy	TriState	\$5,100
Elyse Hausner	Team Hausner	TriState	\$4,064
Michael Daley	Scott's Squad	Connecticut	\$4,045
Shannon Williams	Shannon's Supporters	Austin	\$3,795
Dawn Warner	The Travel Cure	Georgia	\$3,615
Lori Lappe	The Crusaders	Bay Area	\$3,615
Cameron Emens	Team Emens	Arizona	\$3,613
Tiffany Onorato	Team Tiffany	TriState	\$3,558
Joni Kendrick	MG Central Texas	Austin	\$3,516
Stephanie Mikulski		Indiana	\$3,500
Alexis Rodriguez	Rockin' Rodriguez	Georgia	\$3,380
Kelly Smith	Kelly's Heroes	Tennessee	\$3,335
Peyton Emens	Team Emens	Arizona	\$3,250
Nelson Machado	Team Mario	TriState	\$3,150
Terri Adams	Team Ronnie	Connecticut	\$3,110
Shannon Whitfield	Team Lori Whitfield	Georgia	\$3,100
Jesika Hilton	Jilly Bean	Utah	\$3,050
Elizabeth Frenna Roque	I Swear I'm Not Drunk	TriState	\$3,001
Bergen Daley	Scott's Squad	Connecticut	\$2,720
Joan McConnell	Joan's Jaywalkers	Arizona	\$2,675
Diane Boss	Team Boss	Delaware Valley	\$2,638
Kristina Voskes	Team Voskes	Colorado	\$2,630

WALKER	TEAM	WALK	AMOUNT
Roger Morse	Duck Tape Fixes Anything	New England	\$2,620
Steven P. Grant	Team Grant	TriState	\$2,600
Al Weigele	Al's team	Cleveland	\$2,560
Karen K	The Weiss Guys	Tampa Bay	\$2,554
Carlana and Ken Hoffman	Knockout MG for Nicolette	Tallahassee	\$2,550
Angie Espinosa	The Crusaders	Bay Area	\$2,545
Yuebing Li	Team CC Fighting Against MG	Cleveland	\$2,495
Ronnie Adams	Team Ronnie	Connecticut	\$2,490
Richard Elmore		Orlando	\$2,479
Abby Kovalsky		Bay Area	\$2,472
Sue Sumner	The Golden Oldies	Southern Illinois	\$2,440
Maureen Willms	We Willms Find a Cure	Southern Wisconsin	\$2,275

On behalf of everyone in the MG community throughout the country, thank you for all you do, and continue to do to show love and support to all those affected by MG. Because of you, we get closer and closer every day to a world without myasthenia gravis! ✨



Hot off the press continued from page 21

the SLC18A3-linked form just described – a positive family history is often absent. This raises the possibility that patients diagnosed with MG without detectable antibodies in their blood (termed “seronegative”) may actually have CMS, especially if they present in late childhood to early adulthood. Australian and United Kingdom investigators⁹ found that of 25 patients presenting after early childhood with an MG-like syndrome, who were “double seronegative” for antibodies to the acetylcholine receptor and MuSK, seven had a definitely or possibly affected sibling. Mutations in either the RAPSN or CHRNA1 genes were found in each of the seven. Of note, only two of the patients developed symptoms prior to age 10 years, with two developing symptoms after age 25. The authors’ main point is to emphasize that CMS is likely underdiagnosed in seronegative myasthenic disorders. And given the autosomal recessive nature of most forms, an affected relative may not even be present as a tip to the clinician. The misdiagnosis of seronegative MG potentially subjects patients to harmful immunotherapies, corticosteroids, plasma exchange, and thymectomy. All were used in the reported population. Clinical features that should raise suspicion that a patient may have CMS instead of autoimmune MG include slow onset, symmetrical ocular presentation, relative stability

over time, and failure of a predicted response to immunotherapies.

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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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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!