

the transverse myelitis association

# newsletter

summer 2017



*advocating for those with ADEM, NMOSD, ON & TM (including AFM)*

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Find The Transverse Myelitis Association on Facebook! It is a great way to support the TMA and is a wonderful way to network with people in our community. Please take the time to become a fan of our page by clicking "Like," and tell your friends and family about our community's page. Facebook is a great way for us to raise awareness about these disorders and your experiences. Our link is <https://facebook.com/myelitis>.

# FROM THE PRESIDENT OF THE TMA

Change is a constant in my life. Change is certainly one of the more prominent aspects of my life with The Transverse Myelitis Association. When we were first getting the organization off the ground with the Gilmurs, our membership was less than 200 people. When we mailed our first newsletter in 1997, it was sent to our 187 members from the United States and Canada. For years, our mailings were done manually with the help of my parents who meticulously went through my bulk mailing instructions to place the labels on each of the envelopes. Pauline and I then assembled a group of friends, and then later the Ohio TMA Support Group, to stuff envelopes with the next newsletter or journal. Those days are long gone. The TMA has grown to over 12,000 members from more than 100 countries around the world. In part due to the numbers of members we serve, and also because our mission and goals have evolved, our work has become so much more complex.

We were an entirely volunteer organization for the first 17 years of our existence. That the TMA survived that long with only volunteers doing the work was a minor miracle and speaks volumes about the dedication of people such as Jim Lubin, Debbie Capen, and all of our support group leaders who have been serving their countries and communities for many years. Professionalizing the organization gave us a future and it also allowed the organization to offer more sophisticated services to our members, to expand and improve our education programs, to develop an exceptional clinical and research training fellowship for physicians to specialize in our rare neuro-immune disorders, and improve our ability to grow support net-

works around the United States and around the world. We began with one employee in 2012 when we hired Chitra Krishnan as our Executive Director. Our ability to professionalize was made possible by the generous legacy that was provided by our support group leader in the UK, Geoff Treglown ([tma.org/geoff-treglown](http://tma.org/geoff-treglown)). Geoff is always in our memory and the incredible opportunity he provided to our community. In the past five years, we have grown to six staff members who are focused on the mission and goals of the TMA.

Our newest staff member is Kristina Dilger. Based in Columbus, OH, Krissy serves as our Program Associate, and will be involved in every aspect of the day-to-day work of the TMA. Her efforts will support our education programs, from the symposia, to the regular blogs and podcasts. She will be a regular contributor to our newsletters and other publications. Krissy will also support our camp planning and recruitment. She will work with GG, to both expand the research that the TMA performs, as well as to develop the research we are able to support through our fellowships. Krissy is a graduate of The Ohio State University with degrees in both psychology and criminology. She has extensive experience working as a volunteer for several important not-for-profits that do critical work in our community. She's been engaged in this work beginning as a student. As my 14 years of college were filled with having as much fun as I possibly could, Krissy's maturity, devotion to causes in her community, and her selflessness are particularly remarkable and inspiring. We know that Krissy is going to make a difference for the people in our community and for the



TMA. She will be attending camp this summer and also will be at our symposium in October. We hope that you get the opportunity to meet her.

Gabrielle (GG) deFiebre was recently promoted to Associate Director of Research and Education. GG got TM during her senior year in college. She has since earned a master's degree in public health and came to work for the TMA as a volunteer. GG has exceptional research skills and is also a wonderful ambassador to our community, as she has the most intimate understanding of what it means to have one of these disorders. All of us who work for the TMA, both staff and volunteers, were so impressed with her quality of work and her motivation that when a position became available with the TMA, we asked her to come work for us as an employee. GG started part-time because she was also working for a different not-for-profit. She finally came on full time with the TMA about a year ago. At the same time that GG has been working, she began a doctoral program in public health. She is close to completing her coursework for her degree. GG is involved in almost everything we do, and makes an enormous contribution to the day-to-day efforts of the organization. GG has also been able to take our research efforts to a new level. GG has initiated efforts for the TMA to do our own research, and she has

also intensified and expanded our research collaborations with other organizations and with the NIH. GG gives so much to the TMA and we are thrilled and grateful for her contributions.

Dr. Lana Harder's addition to the TMA Board of Directors represents another significant and very positive change for our organization. Dr. Harder is a pediatric clinical psychologist who specializes in caring for children who have neuro-immune disorders, such as ADEM, AFM/TM, NMOSD and MS. Her practice is at the University of Texas Southwestern in Dallas. Dr. Harder is also a Co-Director of the TM Center at UT Southwestern. Lana has been involved in our community and has been making significant contributions to our organization for a long time. She has conducted ground-breaking research trying to better understand cognitive dysfunction in children with these disorders, and was recently awarded a grant from the TMA to study brain imaging in children in an attempt to better understand how the brain might be impacted in transverse myelitis. By using different imaging techniques, Dr. Harder is trying to determine if TM could possibly impact the brain, and not just the spinal cord, as is currently believed.



Dr. Harder is also a regular and critical participant at our TMA Family Camp. She is a presenter in our education program and she makes herself available to the children and the parents throughout the week at camp. Our children and our families love Lana. She offers so much to these families. While at camp, Dr. Harder also offers a group discussion opportunity for the children who come to camp who have ADEM, NMOSD and TM/AFM, and she conducts a similar session for the siblings.

With Dr. Harder's addition to our Board of Directors, we are thrilled that she will have the opportunity to make contributions to our organization in the same way that she has done so for our community. Dr. Harder also serves on our Scientific Advisory Council. We know that her intellect, her creativity and her insights will help to make us a better organization.

Another important change for our organization has been that for the first time in our 23-year existence, the TMA will hold its major education program in Columbus, Ohio. Our education program has been held either in Baltimore or Dallas to take advantage of the TM and NMOSD Centers at Johns Hopkins and UT Southwestern. Those physicians have graciously supported our moving the program to Columbus this year. It is our hope that by offering the program in the Midwest for the first time, that we will make this opportunity available to people who have been unable to attend in the past. We understand the

difficulties involved in traveling for many of you. We are hoping that by making the program more accessible, that many of you will be able to drive into Columbus for the symposium and for the walk. The Ohio Walk-Run-N-Roll will be held the day after the symposium.

Finally, we have added an address for the mailing of donations and for correspondence. The address is a post office at a bank where we make our deposits. This process will greatly improve our accounting practices and our accountability. This new address is:

The Transverse Myelitis Association  
PO Box 826962  
Philadelphia, PA 19182-6962

The international headquarters of The Transverse Myelitis Association remains the address in Powell, Ohio, and you will see that address identified in almost all of our publications. We know that's complicated, but using both of these addresses does actually make us more efficient and accountable. We appreciate your understanding.

Pauline and I are looking forward to seeing many of you at camp this summer and also at the symposium in Columbus this October. It is a great education program.

Please take care of yourselves and each other,  
*Sandy*

# WELCOME TMA SUMMER INTERNS!

We are pleased to introduce Maria Cerio and Maggie Malecky, who will be joining the TMA team as summer interns focusing on communications and development projects. We are excited to work with them and are confident that they will be a great asset to the TMA.



## MAGGIE MALECKY

Maggie is going to be a junior at Ithaca College in the Fall, studying finance and accounting, and is looking to go into corporate finance upon graduation. She contracted transverse myelitis at two and a half years old. Although she is ambulatory, she still experiences the lasting effects of TM. The Transverse Myelitis Association has provided her and her family consistent and compassionate support throughout the years, and various programs that helped her through her recovery. She hopes that as an intern this summer, she can give back to the TMA community some of what they have given her. At school, Maggie has participated in community service projects and business honor societies.



## MARIA CERIO

Maria was diagnosed with transverse myelitis when she was three years old. Initially, she was paralyzed from the neck down. After more than a decade of doctor appointments, assistive devices, and physical therapy, she walks, but with difficulty. Her personal experience has fueled her mission to increase awareness and inclusion of people with disabilities. In Maria's words, "As a TM patient, there is no place I would rather work on this than at the Transverse Myelitis Association." Here is a blog Maria recently wrote: [tma.org/2qCin4r](http://tma.org/2qCin4r)

# BUILDING A FUTURE

I was not familiar with The TMA prior to applying for the Program Associate position in early March of this year, but my interest in public health and advocacy led me to the organization. Since my first interview, my understanding of the TMA has grown with each interaction I have had with the community, and I have witnessed the profound relationship between the TMA and its members. I am humbled and thankful to be welcomed into this community as the newest member of the TMA staff.

The TMA's connection with its members first attracted me to the organization. Fostering community relationships is a passion of mine, and one that I pursue on the neighborhood level by serving as the VP of Community Projects for a local service group. The TMA's community is unique in that it has an international scope, and at the same time, a tight-knit, personal dynamic. I am excited to join that dynamic and will work to cultivate these relationships which allow the TMA to have such an immense impact.

In addition to the community aspect of the organization, the TMA's impressive research and education initiatives appealed to me. The TMA's ability to support, educate, and empower its members is amazing, and I am excited to be a part of its effort to advance understanding and treatment of rare neuro-immune disorders. Health advocacy is another interest of mine, so I will work with the TMA to bring better awareness of these disorders. I will be attending the 2017 RND Symposium and the Ohio Walk-Run-and-Roll in October, which will allow me to learn first-hand the most current research and what it means for the TMA community.

I am incredibly impressed by how much the TMA has grown and accomplished since its inception, and I am excited to join the organization as it continues to grow and establish new projects. I hope to be an asset to both the TMA staff and its members as we navigate the stress and reality of rare neuro-immune disorders together. I believe that the future for the TMA community is hopeful, and that research and education programs will provide answers to the many questions still surrounding rare neuro-immune disorders. I am excited to help build that future.

***Kristina (Krissy) Dilger** recently joined the TMA as a Program Associate. Based in Columbus, OH, Krissy will be focused on supporting and building on our education, research, community advocacy and outreach to help advance our mission.*







# 2017 RARE NEURO-IMMUNE DISORDERS SYMPOSIUM

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# ACUTE DISSEMINATED ENCEPHALOMYELITIS (ADEM): PAST, PRESENT, AND FUTURE

*By Cynthia Wang, MD / James T. Lubin Fellow at the University of Texas Southwestern Medical Center, Dallas*

**Acute disseminated encephalomyelitis (ADEM) is a rare, autoimmune neurological condition that often occurs in young children following an infection. Initial symptoms of ADEM may be quite variable and non-specific, including headache, fever, irritability, confusion, sleepiness, and weakness. Over the years, our understanding of ADEM has matured, though many aspects of the condition remain mysterious. What leads some individuals to develop the condition in the first place? Could it happen again? Why do some individuals make a full recovery while others have lasting impairment?**

ADEM is thought to develop when the body mounts a response to an infection, but rather than solely attacking the invading organism, it mistakenly targets parts of the nervous system as well. ADEM is primarily a clinical diagnosis, meaning that we do not have a perfect test that can tell us definitively whether a child or adult has ADEM. Laboratory studies of blood and spinal fluid, and MRIs of the brain and spine can help rule out other conditions and support the diagnosis of ADEM. If ADEM is suspected, children and adults often receive steroid medications during their hospitalization to help reduce inflammation and suppress the body's confused immune system.

For the vast majority of those affected by ADEM, it is a one-time illness and does not recur. Many children make excellent recoveries and largely seem to return to normal. However, episodes of new neurological symptoms, especially those that develop 3 months or more after the initial event, raise concern for an alternate diagnosis, such as multiple sclerosis or neuromyelitis optica spectrum disorder. In our clinic at UTSW Dallas, our practice has been to repeat MRI brain scans at 3-4 months after ADEM to establish a

baseline for future comparison. Any new clinical events or MRI brain lesions occurring after this period of time would lead to scrutiny of the ADEM diagnosis.

Another practice in our clinic is to perform neuropsychological testing on our patients who have had ADEM. Increasing research highlights that children with ADEM may have subtle cognitive deficits involving processing speed, visual-motor integration, executive functioning, language processing, and behavioral regulation that may not be evident until they get older and are faced with increasing academic and social demands. Neuropsychological testing is geared toward uncovering areas of weakness and may be helpful in guiding school accommodations for children affected by ADEM.

We are just beginning to study the long-term outcomes of children with ADEM, particularly their cognitive

potential, academic achievement, social functioning, and quality of life. In the past couple of years, we have also learned more about a subset of ADEM patients who make antibodies toward a protein on the outer surface of certain cells called myelin oligodendrocyte glycoprotein (MOG). Over time, our ability to test for this antibody has improved, along with increasing understanding that this group of MOG positive ADEM patients may have similarities in their clinical presentation, neuroimaging, and prognoses that may impact their care.

**Introducing APERTURE: Assessment of Pediatric and Adult Encephalomyelitis Related Outcomes: Understand, Reveal, Educate**

To help address gaps in our understanding about ADEM, our group at University of Texas Southwestern Medical Center will be conducting a

study examining the long-term outcomes of individuals with ADEM. We aim to collect data on a large group of children and adults diagnosed with ADEM and review their presenting history, laboratory studies, neuroimaging, treatment course, and recovery from ADEM. Individuals between ages 0-64 with the initial diagnosis of ADEM are welcome to participate. People who cannot travel to our center in Dallas can participate remotely by providing medical records, MRI studies, completing questionnaires, and in some cases, neuropsychological testing. Enrollment will begin in June 2017.

We are eager to learn more about this rare neurological condition, and with your help, work toward improving our ability to better care for those affected by ADEM.

Please email [pediconquer@ut-southwestern.edu](mailto:pediconquer@ut-southwestern.edu) for more details.

# ACUTE DEMYELINATING EVENTS FOLLOWING VACCINES: A CASE-CENTERED ANALYSIS

*This a summary of an article that was published in 2016 by Roger Baxter et al. in the journal Clinical Infectious Diseases.*

**A study was conducted to determine whether vaccines may have a causal effect on Transverse Myelitis (TM) and Acute Disseminated Encephalomyelitis (ADEM). Previous studies consist of mostly anecdotal case reports which do not look at population-level data and do not allow us to see if vaccinations were the cause of the TM or ADEM. However, concern remains among the TM and ADEM community that vaccines could be responsible for triggering autoimmune demyelinating events like TM or ADEM.**

The researchers in this study used the Vaccine Safety Datalink (VSD) to analyze 64 million vaccine doses over the period of January 1, 2007 to December 31, 2012. The VSD is a collection of data from participating healthcare systems across the United States in collaboration with the Centers for Disease Control and Prevention's Immunization Safety Office. This automated database includes immunization registries, and collects information on more than 9 million patients per year. The study population for this research consisted of all children and adults of any age who received one or more vaccines during the study period, and were enrolled in health plans at six VSD sites.

First-ever TM and ADEM diagnoses were identified using each VSD site's internal electronic medical record, and the cases required at least one diagnosis by a neurologist within 3 months of initial diagnosis. The researchers did not include individuals with a history of Multiple Sclerosis or Neuromyelitis Optica Spectrum Disorders. The cases were first reviewed by trained medical records analysts and then reviewed by a neurologist with expertise in demyelinating illnesses to ensure the cases met research criteria. The study utilized the database to match a comparison group to each case based on type of vaccination, age, sex, VSD site, and whether their vaccination occurred during the exposure intervals, which were chosen by the researchers based on prior studies and expert opinion. The two exposure periods analyzed were: (1) 5-28 days from vaccination as the most likely interval to result in a demyelinating event; and (2) 2-48 days from vaccination to the demyelinating event to ensure an increased risk was not missed. The comparison interval that the researchers chose was 43 days through nine months prior to the demyelinating event to avoid duplicate influenza vaccines over two seasons.

**Original research:** Baxter R, Lewis E, Goddard K et al. Acute demyelinating events following vaccines: A case-centered analysis. *Clin Infect Dis*. 2016 Dec 1;63(11):1456-1462. Epub 2016 Sep 1.

The researchers looked at whether or not each case received a vaccine during the exposure interval or the comparison interval before the demyelinating event occurred. They then compared this to the proportion of the entire study population that received a vaccination during the exposure interval before the demyelinating event date. Eighty-one cases of TM and fifty-six cases of ADEM were included. The researchers found that for TM, there was not a statistically significant increased risk of a demyelinating event in either the 5-28 day exposure interval or the 2-48 day exposure interval after receiving any vaccination. The researchers also found that for ADEM, only the Tdap vaccine had a statistically significant association with an increased risk of a demyelinating event. This was only found in the 5-28 day exposure interval. The Tdap vaccine is used for adolescent and adult tetanus, reduced diphtheria, and acellular pertussis. However, the authors noted that there are some concerns with the validity of this outcome. For example, there were only two cases of ADEM that occurred during this time period, which is a very small number, and if there had only been one case, this finding would not have been statistically significant. Also, the authors wrote that they did many statistical tests and did not adjust for this, so they stated that the results could be due to chance. Furthermore, the authors noted that even if we accept an increased risk for ADEM after the Tdap vaccine, the extra risk is very small and not likely to be more than 1.16 cases of ADEM per million doses of the Tdap vaccine. Dr. Benjamin Greenberg from the University of Texas Southwestern Medical Center also noted the study does not reveal the six clinical centers used to collect cases. If these centers were referral centers they may have an overrepresentation of TM and ADEM cases – thus overestimating the risk.

Overall, the authors stated that the results of this study conclude that vaccinations mostly do not have a statistically significant association with demyelinating events that occur in TM and ADEM. They also noted that one limitation of the study was that they chose the exposure interval, so if the interval was chosen incorrectly, an increased risk might have been missed. Also, they stated they did not review combinations of vaccines but rather analyzed each vaccine's risk separately. Finally, the researchers stated that they purposefully did not adjust for multiple observations so that they could achieve higher sensitivity in their results; nonetheless, they noted that they only identified one statistically significant result (the Tdap vaccine). Despite its limitations, the authors noted the study's strength was its review of all TM and ADEM cases by medical records analysts and a neurologist, which likely limited misclassification of cases with these disorders, which reduces bias and makes results more likely to be accurate. According to the authors "In conclusion, TM and ADEM are rarely, if ever, associated with vaccines."

# WHAT IS THE TMA REGISTRY?

**The TMA is excited to announce our latest research initiative, the TMA registry. Through a partnership with the NIH/NCATS Global Rare Diseases Patient Registry Data Repository/GRDR® program, the TMA has developed a survey that collects information from individuals diagnosed with rare neuro-immune disorders.**

The questions in the survey cover many aspects of the participant's experience with his or her disorder, including symptoms, diagnosis, treatment, and complications. The goal of the registry is to develop a deeper understanding of rare neuro-immune disorders and to better advocate for those experiencing these conditions.

The TMA began work on the registry in the summer of 2015. For a year and a half, we collaborated with medical professionals specializing in neuro-immune disorders, including Dr. Benjamin Greenberg from the University of Texas Southwestern, to develop questions. We also drew from other research studies on TM (including AFM), ADEM, ON, and NMOSD to ensure we were asking questions that will be useful for researchers in future studies. We then submitted our research proposal to the Institutional Review Board (IRB) at the NIH for approval, which we received in January of this year. We publicly launched the registry at the end of March and 47 members of our community have participated in the registry to date.

The TMA registry can be completed by individuals diagnosed with a rare neuro-immune disorder from their home at no cost to them. After signing up, we send participants a link to the survey, which they can complete on their own time. To ensure the most accurate data are collected, participants are asked to reference their medical records while answering questions regarding their diagnosis and treatment. Once the survey is completed, we will collect

the responses and follow up with participants if any information needs clarification. We will then de-identify the data before providing it to the NIH/NCATS/GRDR program. By de-identifying the data, participants will not be linked to any of the personal information they provided in the survey. This ensures the confidentiality of participants while their medical information is shared with researchers, healthcare providers, and medical professionals.

The possibilities for the use of the registry are far-reaching. First, we can **conduct analyses** using the results of the registry, share them with you, and publish our findings. We can use the results to **find trends and patterns in different aspects of the disorders**. For example, we will be able to analyze how often neuropathic pain is reported with each disorder. We will distribute our findings to our community, healthcare providers, and medical professionals so that **best practices can be established and individuals experiencing demyelinating events can receive effective and informed care from their doctors**. We are able to use the registry to generate interest in rare neuro-immune disorders within the medical field so that the disorders are better known and more likely to be researched. Additionally, the **data from the registry can be used by researchers studying rare neuro-immune disorders for other research projects**. Our hope is that the information gathered from the registry will eventually be used to help **improve diagnostics, therapies, and**



**“Within an hour  
I couldn't walk  
and was so scared.  
There was so much  
to learn about TM  
and NMO and so  
much hope and  
encouragement  
needed! The TMA  
has been a huge  
help, so when they  
started the registry  
to improve the  
knowledge about  
these diseases  
and help others - I  
absolutely wanted  
to be a part of it!”  
– Kate Krietor,  
(member of the  
TMA)**

**possibly even preventive practices for these rare neuro-immune disorders.**

In order to accomplish the goals of the registry, the TMA needs members of the community to participate in the survey. Eligible participants include adults age 18 years and older who are diagnosed with TM (including AFM), ADEM, ON, or NMOSD. Also,

legal guardians of minors under the age of 18 with these disorders may participate in the survey on the minor's behalf (with the minor's assent if they are age 7 or older). We can also accept information from parents or legally-authorized representatives to complete the survey on behalf of a deceased individual who was diagnosed with one of these disorders. Together, the TMA community can shape the

future for all individuals experiencing rare neuro-immune disorders by contributing to research such as the TMA registry. We believe that there are answers to the many questions surrounding these disorders, and the registry is a powerful tool that can be used to find those answers.

If you are interested in participating, please visit: [tma.org/tma-registry](http://tma.org/tma-registry)

# GENETIC MUTATION FOUND IN FAMILIAL TRANSVERSE MYELITIS

*By Dr. Michael Levy, Associate Professor of Neurology at the Johns Hopkins Hospital and Medical Director of General Neurology at the Johns Hopkins Hospital in Baltimore, MD*

We found a rare genetic mutation in some families of patients with acute transverse myelitis (TM) that may provide new insights into the disease process. Two sisters with TM came to our attention through the TMA who agreed to donate their DNA for a thorough genetic analysis called exome sequencing. Only one meaningful difference was found in both sisters, which their two healthy brothers did not have: a mutation in a gene called VPS37A. As it turns out, a genetic mutation like theirs is extraordinarily rare in nature. No other known organism in the animal kingdom has this mutation so we thought this could be biologically important. But we didn't know how common this was. With the help of the TMA, the Accelerated Cure Project and collaborators at Johns Hopkins, we screened an additional 86 TM patients and found another patient with the same rare mutation! It is statistically beyond coincidence to find three human beings with this same rare genetic mutation unless it has something to do with the rare disease they all share, TM. We do not yet understand how this gene, or the genetic mutation, is associated with TM. But with the continued support of the TMA, funding agencies and of course, our TM patients, we are beginning to piece this puzzle together.

**The abstract below was presented at American Academy of Neurology 2017 in Boston, MA.**

Maureen Mealy<sup>1</sup>, RN, Tai-Seung Nam<sup>2</sup>, MD, PhD, Santiago Pardo<sup>1</sup>, Carlos A. Pardo<sup>1</sup>, MD, PhD, David Valle<sup>3</sup>, MD, Kathleen Burns<sup>3,4</sup>, MD, PhD, Michael Levy<sup>1</sup>, MD, PhD

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**Background.** Idiopathic transverse myelitis is an acute inflammatory attack of the spinal cord leading to weakness, sensory loss and bowel/bladder dysfunction. The prevalence is 0.1-0.2/100,000 and there are no known risk factors.

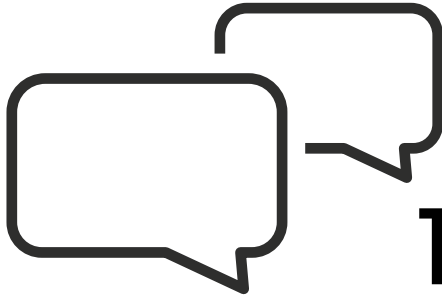
**Objective.** To identify a genetic risk for development of idiopathic transverse myelitis.

**Methods.** We identified two sisters who both have idiopathic transverse myelitis, and compared exome sequencing on DNA samples from them with two healthy siblings. We also sequenced 200 additional samples from patients with idiopathic transverse myelitis, multiple sclerosis, neuromyelitis optica, other neurological conditions and healthy controls.

**Results.** The two sisters with idiopathic transverse myelitis both had acute onset of sensory loss in the legs, followed by weakness and bowel/bladder dysfunction. The first sister developed myelitis at age 15 with clinical nadir of complete paralysis. Over the next few years, she recovered her ability to walk without assistance.

Recent MRI demonstrated persistent T2 lesion in the lower thoracic cord. The second sister developed myelitis at age 50 with nadir of complete sensory loss from T6 down and paraparesis in the legs, associated with an MRI lesion at T6. She also made a partial recovery with treatment. Both sisters share a non-synonymous homozygous mutation in only one gene, VPS37A (c.700C>A, p.Leu234Ile) in the whole genome analyses. One healthy sibling was heterozygous for this mutation. We screened an additional 261 samples from patients with ITM and neuroimmunological diseases by Sanger sequencing of this portion of VPS37A and identified another idiopathic TM patient with this same rare homozygous mutation. No patients with multiple sclerosis, neuromyelitis optica, other neurological conditions or healthy controls contained a homozygous mutation in VPS37A.

**Conclusions.** A mutation in VPS37A may predispose to development of idiopathic transverse myelitis. Further studies are necessary to determine the frequency of this mutation in this patient population and how this genetic mutation might contribute to risk of disease.



# THE MYELITIS HELPLINE

Over the last 22 years, we have worked with leading medical professionals and experienced providers to share resources, information, and up-to-date knowledge with our community of individuals diagnosed with ADEM, NMOSD, ON, and TM, including AFM, caregivers, and medical professionals. Based on the questions and feedback from our community, we recently launched a new online tool, the Myelitis Helpline, a collection of frequently asked questions that covers topics from diagnosis to treatments to research to applying for social security disability.

The goal of this online tool is to provide resources, knowledge and help to our community, whether one has been recently diagnosed or has questions several years after onset of a rare neuro-immune disorder. The information provided is for general information purposes and is not a substitute for professional medical advice, care, treatment or for diagnosis.

Please send an email to GG deFiebre at [gdefiebre@myelitis.org](mailto:gdefiebre@myelitis.org) with additional questions and ideas you would like us to include in the Myelitis Helpline.

**[myelitis.org/mhl](http://myelitis.org/mhl)**

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## VISIT OUR RESOURCE LIBRARY

to access over 250 articles, newsletters, videos and podcasts, filtered by disorder and specific topic.

**[myelitis.org/living-with-myelitis/resources/resource-library](http://myelitis.org/living-with-myelitis/resources/resource-library)**

# 2017 TMA GALA DINNER & AUCTION

in honor of Sarah Robbins

The 2017 TMA Dinner Gala and Auction was held on June 2nd in Cape Canaveral, FL, marking the fourth year that Tina and Jason Robbins have held this event in honor of their daughter, Sarah, and in support of the TMA. This awareness event was a celebration of families and loved ones who triumph every day facing the many challenges and complications from ADEM, NMOSD, ON and TM, including AFM, and brought to light the work we do for those impacted by rare neuro-immune disorders. Additionally, it provided the opportunity to raise crucial funds that will support our education and research programs, such as the TMA Family Camp. We are proud to say over 300 people attended the event.

We want to thank all the event sponsors and extend a special heartfelt thank you to the entire Robbins family for their hard work and devotion to our cause.



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# TMA VOLUNTEER SPOTLIGHT

**TMA volunteers are some of the most powerful members of our community. These individuals dedicate their time, energy, and resources towards advancing our mission of supporting people diagnosed with a rare neuro-immune disorder and their families. Were it not for our volunteers, the TMA would not exist. For almost 20 years, the TMA was an organization operating solely on the hard work of volunteers! We honor those who work so hard for us to support our mission.**

**Through our “Volunteer Spotlight” column, we honor and share our gratitude to some of the amazing people in our community who are the fabric of the TMA.**

**We are pleased to honor Colleen Spaeth. Colleen and her family led the first ever Walk-Run-N-Roll event for the TMA in 2012 and have inspired six Walk-Run-N-Roll events for the TMA over the last five years. Colleen continues to lead the annual New Jersey Walk-Run-N-Roll along with Patty Holston and her friends and family.**







**The 2017 New Jersey Walk-Run-N-Roll took place on Saturday, April 29th at Cooper River Park. We want to thank Colleen Spaeth and Patty Holston for leading the awareness event, their friends and families, and the community for supporting our mission and increasing awareness about rare neuro-immune disorders!**

*In Colleen's words...*

In May 2007, as I was packing to enjoy a weekend at a spa with my sister and daughter, I felt a charley horse in my left leg. I thought a warm shower would alleviate any discomfort I had. During my shower I lost feeling in my right leg, and by the time I got to the bed I lost all ability to walk. Then, by the time I got to the hospital, I was in excruciating pain, and terrifying anyone within earshot with my screaming.

After 12 days in the ICU, and diagnoses of Guillain-Barré syndrome and herniated discs, my second round of radiology results caught the eye of a young radiologist who advised the experts to “consider transverse myelitis.” And so my journey began. A journey that would involve 56 days of intense rehab at Magee Rehab in Philadelphia, and months of outpatient therapy. I went from a wheelchair to a walker, and I was grateful.

When I arrived home I did some research on TM and found out about the Transverse Myelitis Association, and began my search for answers. The following year my family attended a symposium in Seattle. I was anxious to meet people who joined me on this road. I wanted to learn all I could about this awful disease. I was searching for answers and I wanted to get better. It was a wonderful opportunity for me, and I was in awe of the people who worked hard for the TMA, including Sandy Siegel and Jim Lubin. The room was full of people who also wanted to do SOMETHING, and my family joined me in my intention to get involved.

Before I was “hit” with TM, I was a very active person. Tennis, hiking, swimming, biking, and camping; I was an outside person. During the first year I was home and rehabbing, I had bouts of depression, too many bouts. I was determined to turn it all around, but wasn't sure how that would look.

I continued at my job, because I could. As a court reporter all I needed to do my job was the ability to work in the courtroom using my fingers on a court reporting machine, and my job accommodated me. I returned to work using a walker. On “bad” days when the weather affected me, and my pain levels were 8 out of 10, I stayed home.

**I strive to remember I am just  
human, and today I will be the best  
I can be. That's how I deal with each  
day, with the neuropathic pain and  
spasms, and most days it works.  
When it doesn't, well, it doesn't.  
Tomorrow I'll try again anyway!**

I am also an ordained deacon in the Episcopal Church and was surrounded by a fantastic group of people who supported me, and raised \$6,000 to build a ramp for me to make my home more accessible. I was determined to make my “new normal” doable. Some days it worked, others not so much.

In 2012, my daughter Amanda and my son Rob announced that they wanted to do a Walk in my honor. I was amazed at their love for me, and suggested that we raise funds for the TMA at the same time. And so the first Walk was born. It has been an unbelievable experience for me and many others.

It is important to me that I remember that TM doesn't just impact one person, but a larger circle of people who love me. I am a person who believes there are no accidents in life, that all things have a purpose, and so this Walk gives that purpose light. During the last six Walks I have met

some amazing people, and I wouldn't have experienced any of it were it not for two kids who thought they had a great mom. The act of giving back helps me more than I can ever say. Every year my family comes out, my friends come out, strangers who are now my friends come out, and in a few hours we laugh, smile, and some of us walk around the Cooper River, many in wheelchairs, forgetting for just a few hours that we have this crappy disease, and remembering that we are stronger together.

Yes, my life has drastically changed, my outdoor person has been limited to finding other ways to be active, such as chair-yoga, and walking back and forth in the pool at the gym, and I get lots of looks from people as I go through my weight program on the gym floor while using my walker. I can't play tennis any more, and hiking is not happening, but I remember to be grateful for what I can do.

To anyone who is sitting home wondering what's next now that their “new normal” has surfaced, I would say be kind to yourself, allow time to cry and process what has happened, allow others to cry with you, to help you when they want to, take time to be kind to yourself, and then get out of your comfort zone and say **“Now what do I do?”** If you don't, then you are missing out on opportunities you would never have dreamed of for yourself. If you go on YouTube, you can see a video that Patty Holston and I created of the 5th Walk, and at the end of the video we say that while we have TM, TM does not have us. I refuse to be defined by my limitations, I strive to remember I am just human, and today I will be the best I can be. That's how I deal with each day, with the neuropathic pain and spasms, and most days it works. When it doesn't, well, it doesn't. Tomorrow I'll try again anyway!

# IN MEMORY OF GABBY INSCHO

We recently lost a beautiful child. Gabby Inscho has been and will always be a shining star in our community. Gabby and her family regularly attended our family camp and were great supporters of our awareness events. Everyone who knew Gabby loved her – and we were enriched through her positive spirit, her hopeful and outgoing personality, her kindness and her beautiful smile. She will be deeply missed by everyone she touched. She was such a special child. Losing Gabby reminds us all of the fragility of life and the devastating effects of these horrible disorders. We can honor Gabby's memory by remembering the kind of person she was and by trying to be more like Gabby; by being kind to each other, by finding the positive in each other, by being hopeful and optimistic, and by making the very best and the most of every single day we are blessed with. Our memories and our love for Gabby will continue to drive us to fight for a better quality of life for everyone who has these rare disorders. Our hearts break for the Inscho family... and for all of Gabby's friends who are suffering this loss.

*You will be in our thoughts, Gabby, and always in our hearts.*





# DIFFERENT JOURNEYS, ONE HOPE

*By Timi Schrumpf, Community  
Partnerships Manager at The TMA*

**People in our community have so many different perspectives, experiences, and feelings about their journey with a rare neuro-immune disorder, but there is one thing that most will agree on; they have few people in their lives who really understand. This is why sharing our experiences is so important. Support groups are one of the most powerful ways to provide this critical need and to serve our community.**

As our community continues to grow, the need for strong support groups is becoming increasingly evident, and a support group can only be as strong as its leader. Early this year, the TMA implemented a new support group leader program. This program, which includes a training component, was designed to create the best opportunity for a safe, trusted and successful group.

Several of our support group leaders have completed the training and have mentioned the positive impact the tools and skills presented in the training have had on their support group.



Recently, I had the opportunity to attend the Central Texas support group, led by Heather Reynolds in Round Rock, TX. Participating in this meeting allowed me to not only witness how amazing our leaders are, but also to experience the importance of support groups to our community. We had a small group that day, about eight people, but the community members present represented different age groups, backgrounds, and diagnoses. We were a diverse group, just like our community! The small number of people present allowed for there to be an informal, roundtable-style discussion where honesty and vulnerability were welcomed and appreciated. At one point during our discussion, a woman shared something that had been bothering her emotionally in relation to her journey with a rare neuro-immune disorder. At first, many would have assumed this concern was unique to her and not relatable to the other people at the support group meeting. However, as soon as she finished sharing her thoughts, another person immediately spoke up and

shared how she had been experiencing the same concerns and worries. The fact that two different people, decades apart in age, and each living with a different diagnosis could share support and encouragement during what is such a difficult and lonely journey, was the ultimate example of the significance support groups have to those in our community.

It is crucial that everyone who reaches out to the TMA, whether they are seeking information for themselves or help for a loved one, has access to the same level of support that these two people found that day. As we work to build our support group network, we are really working to build a network of hope, encouragement, and support that will have a lasting impact on each of us.

If you are interested in starting a support group in your area, please contact Timi at [tschrumpf@myelitis.org](mailto:tschrumpf@myelitis.org)



# CLINICAL STUDIES & TRIALS

For more information, please visit [bitly.com/tma-clinical-trials](http://bitly.com/tma-clinical-trials)

## 1 **CAPTURE: Collaborative Assessment of Pediatric Transverse Myelitis; Understand, Reveal, Educate**

Principal Investigator: Benjamin Greenberg, MD, MHS  
Lead Study Site: University of Texas Southwestern  
Study includes online and multiple study sites

## 2 **Efficacy and Safety Study as Monotherapy of SA237 to Treat NMO and NMOSD**

Study Sponsor: Chugai Pharmaceuticals

## 3 **Safety and Efficacy of Sustained release Dalfampridine in Transverse Myelitis**

Principal Investigator: Michael Levy, MD, PhD  
Study Site: Johns Hopkins University

*This study is currently not open for recruitment. Participants currently enrolled are being followed.*

## 4 **A Double-masked, Placebo-controlled Study With Open Label Period to Evaluate MEDI-551 in NMO and NMOSD**

Study Sponsor: Astrazeneca

## 5 **Spinal Cord MRI Research Study for Children, Adolescents, and Young Adults with Myelitis**

Principal Investigator: Nadia Barakat, PhD  
Study Site: Boston Children's Hospital

## 6 **A Longitudinal Study of Neuromyelitis Optica and Transverse Myelitis**

Principal Investigator: Benjamin Greenberg, MD, MHS  
Study Site: University of Texas Southwestern

**7      The PREVENT Study**

Study Sponsor: Alexion Pharmaceuticals

**8      The Effect of Pregnancy on Neuromyelitis Optica**

Principal Investigator: Eric Klawiter, MD  
Study Site: Massachusetts General Hospital

**9      Neuroimaging and Neurobehavioral Outcomes of Pediatric Neuromyelitis Optica: A Pilot Study**

Principal Investigator: Ana Arenivas, PhD  
Study Site: Johns Hopkins Medicine

**10      Utilizing Brain Imaging to Understand Cognitive Dysfunction in Transverse Myelitis**

Principal Investigator: Lana Harder, PhD  
Study Site: University of Texas Southwestern

**11      Patient Reported Outcomes for Bladder management in Spinal Cord Injury**

Study Sponsor: Neurogenic Bladder Research Group (NBRG)

**12      The TMA Registry**

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**For detailed information about clinical studies and trials please visit  
[myelitis.org/shaping-the-future/research/clinical-studies-trials](http://myelitis.org/shaping-the-future/research/clinical-studies-trials)**



# 2017 WALK-RUN-N-ROLLS

**We are excited to announce that the TMA is making great progress towards our goal of having ten Walk-Run-N-Roll campaigns across the country in 2017!**

A Walk-Run-N-Roll serves as a great community-building event to raise awareness for rare neuro-immune disorders. These events also help the TMA reach our goal of increasing awareness, developing and strengthening our community, and raising funds for research, education and support. There are other walks being planned, and we will be releasing details as they are finalized. Please visit our website to join a walk near you!

If you would like to help organize a Walk-Run-N-Roll in your area, please contact Timi Schrupf at [tschrumpf@myelitis.org](mailto:tschrumpf@myelitis.org).

## Upcoming Walk-Run-N-Rolls

### 2017 MT Walk Run-N-Roll

Sunday, August 6, 2017

#### North Park, Billings

6th Ave N & North 19th St.  
Billings, MT 59101

**More info:** [tma.org/2017-mt-walk](http://tma.org/2017-mt-walk)

### 2017 TX Walk-Run-N-Roll

Saturday, October 7, 2017

#### Cottonwood Creek Park

4051 N Story Rd  
Irving, TX 75038

**More info:** [tma.org/2017-tx-walk](http://tma.org/2017-tx-walk)

### 2017 OH Walk Run-N-Roll

Sunday, October 22, 2017

#### Coffman Park Pavilion

5200 Emerald Parkway  
Dublin, OH 43017

**More info:** [tma.org/2017-oh-walk](http://tma.org/2017-oh-walk)

### 2017 FL Walk Run-N-Roll

Saturday, September 23, 2017

#### Lake Eola Park

512 E Washington St  
Orlando, FL 32801

**More info:** [tma.org/2017-fl-walk](http://tma.org/2017-fl-walk)

### 2017 MA Walk Run-N-Roll

Saturday, October 21, 2017

#### Endicott Park

57 Forest St  
Danvers, MA 01923

**More info:** [tma.org/2017-ma-walk](http://tma.org/2017-ma-walk)

### Four-States Area Walk-Run-N-Roll

Saturday, September 16, 2017

#### 2017 PA Walk-Run-N-Roll

Date and location TBD

Please visit [myelitis.org/events](http://myelitis.org/events) for more details and updates!



## ASK THE EXPERT PODCAST SERIES

Every month the TMA holds a podcast on a topic that affects those with rare neuro-immune disorders. Experts in the field share the latest knowledge and answer questions from our community. Podcasts have covered topics, such as vaccinations and rare neuro-immune disorders, rehabilitation, and clinical trials.

So far this year our Experts have answered questions about building multidisciplinary teams for complex care, vaccinations, and managing care with a rare neuro-immune disorder. We are very excited about our upcoming podcasts on understanding social security and disability benefits, related auto-immune conditions and myelopathies, and the role of rehabilitation and exercise. We hope you can join us!

Listen and subscribe to our podcast via iTunes today. You can find all past recordings in our resource library: <https://bit.ly/tma-podcasts>.

### Upcoming Podcasts

#### Open Forum on ADEM

Thursday, July 13, 2017 | 1:00 pm ET

**More info:** [tma.org/2017-july-podcast](https://tma.org/2017-july-podcast)

#### Gaining Functional Recovery through OT

Tuesday, August 22, 12:00 pm ET

**More info:** [tma.org/2017-aug-podcast](https://tma.org/2017-aug-podcast)

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## OUR ADDRESS HAS CHANGED!

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## Announcements

2017 MT Walk-Run-N-Roll: August 6, 2017

Four-States Area Walk-Run-N-Roll: September 16, 2017

2017 FL Walk-Run-N-Roll: September 23, 2017

2017 TX Walk-Run-N-Roll: October 7, 2017

2017 Rare Neuro-Immune Disorders Symposium: October 20-21, 2017

2017 MA Walk-Run-N-Roll: October 21, 2017

2017 OH Walk-Run-N-Roll: October 22, 2017

## Contact us

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