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Charcot-Marie-Tooth Association  
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## Boston Charcot-Marie-Tooth Patient/Family Conference

August 29, 2015

Seaport World Trade Center  
Plaza Level  
200 Seaport Boulevard  
Boston, MA

 **CMTA**  
*Charcot-Marie-Tooth Association*



## Boston Charcot-Marie-Tooth Patient/Family Conference

August 29, 2015

- 8:00-9:00am: **REGISTRATION**
- 9:00-9:05am: **WELCOME: INTRODUCTION OF SPEAKERS**  
Elizabeth Ouellette, Vice-Chair  
CMTA Board of Directors
- 9:05-9:15am: **NEUROMUSCULAR DIVISION AND SERVICES AT MGH**  
William S. David, MD, PhD  
Associate Professor of Neurology  
Harvard Medical School
- 9:15-9:45am: **THE CLINICAL SPECTRUM OF CMT**  
David A. Chad, MD  
Associate Professor of Neurology  
Harvard Medical School
- 9:45-10:15am: **OVERCOMING OBSTACLES IN CLINICAL TRIALS OF CMT**  
Vera Fridman, MD  
Co-Director, CMT Center of Excellence  
Massachusetts General Hospital
- 10:15-10:35am: **BREAK**

- 10:35-11:05am: **TUMBLING FORWARD**  
Bethany Meloche  
Director of Social Media  
Charcot-Marie-Tooth Association
- 11:05-11:35am: **INSIGHTS FROM BENCH TO BEDSIDE: PAVING THE WAY FOR TRIALS IN CMT**  
Florian S. Eichler, MD  
Associate Professor of Neurology  
Massachusetts General Hospital
- 11:35-11:55am: **STAR – THE FUTURE IS OURS**  
Patrick A. Livney  
Chief Executive Officer  
Charcot-Marie-Tooth Association
- 11:55am-1:10pm: **LUNCH WITH THE EXPERTS**
- 1:10-1:30pm: **CMTA/SANOFI-GENZYME COLLABORATION: CMT1A**  
Paul R. August, Senior Director  
Genzyme, A Sanofi Company
- 1:30-2:00pm: **LIVING WITH CMT: REHABILITATION ISSUES**  
Sabrina Paganoni, MD, PhD  
Associate Professor  
Harvard Medical School
- 2:00-2:30pm: **BRACING OPTIONS, ADAPTIVE DEVICES**  
Ken Cornell, CO, FAAOP  
Cornell Orthotics & Prosthetics
- 2:30-2:45 pm: **THE TIME IS NOW - GET INVOLVED**  
Jeana Sweeney  
Director of Community Services  
Charcot-Marie-Tooth Association
- 2:45-3:00pm: **BREAK**
- 3:00-3:50pm: **EXPERT ROUND TABLE**
- 3:50-4:00pm: **CLOSING**  
Herb Beron, Chair  
CMTA Board of Directors

## Presenters and Experts



**Paul R. August, PhD, Senior Director**  
**Genzyme, A Sanofi Company**

Paul R. August is a senior director and department head for the discovery biology department of Sanofi in Tucson, Arizona. He has more than 25 years of experience in the discovery of pharmaceuticals and the management of global, collaborative research projects.

He is dedicated to the discovery of pharmaceuticals and the identification of external partnering opportunities, particularly for rare neuromuscular diseases.

Paul received his MS degree in genetics from the University of Connecticut and his PhD from the University of Minnesota, Department of Microbiology, Immunology and Cancer Biology. He followed his passion in pharmaceuticals by performing an NIH Post-Doctoral fellowship in the Department of Organic Chemistry at the University of Washington, Seattle.



**Jonah Berger**  
**CMTA Advisory Board Member**

Jonah Berger is 42 years old and lives in colorful Colorado. He runs a business called The Rhythm Within, working with kids and adults with special needs. He has been diagnosed with CMTX and has enjoyed an interesting journey of challenge and wisdom. He is

the author of *He Walks Like A Cowboy*, a book detailing the life and lessons of the CMT path. He is a proud member of the CMTA Advisory Board, and believes in the challenge and the wisdom that come from CMT!



**Herb Beron, Chair**  
**CMTA Board of Directors**

When his daughter Julia was diagnosed with CMT2E, Herb and his wife Rachael decided that they needed to become actively involved with the CMTA. In 2007, Rachael participated in a one-mile swim to raise funds for the CMTA. Eight years later, "TeamJulia & the Swim for the Cure" has raised in excess of \$640,000 in total

donations for the STAR program. Herb has been on the Board of Directors for the past seven years, and he welcomes the opportunity and challenge to lead the Board in taking the CMTA to even greater heights.

Herb received a BBA in Finance from Emory University in 1986 and is currently a Director for Morgan Stanley Smith Barney. It is his heartfelt goal that the CMTA achieve its stated mission: "to find a cure, to create awareness, and to improve the quality of life for those affected by CMT."



**David A. Chad, MD**  
**Associate Professor of Neurology**  
**Harvard Medical School**

David A. Chad graduated from McGill University medical school in 1974. He completed an internal medicine residency at the Montreal General Hospital (1976), a neurology residency at the University of California at San Francisco (1979), and a fellowship in neuromuscular diseases at Tufts University Medical Center (1982). From 1982 to 2008, Dr. Chad was a member of the Neurology Faculty at the University of Massachusetts Medical School (Worcester) and in August 2008, he joined the neuromuscular unit at Massachusetts General Hospital.

Dr. Chad is a neuromuscular clinician, electromyographer, and educator whose work is focused on the evaluation, diagnosis, care and treatment of patients with neuromuscular disorders, including ALS, peripheral neuropathy, myasthenia gravis, myopathies and muscular dystrophies.



**Ken Cornell, CO, FAAOP**  
**Vice-President**  
**Cornell Orthotics & Prosthetics**

Ken Cornell earned a BA in biology at St. Anselm's College in Manchester, New Hampshire and completed his postgraduate work in orthotics and prosthetics at UCLA in 1980. He has long been active on the board of the New England Chapter of the American Academy of Orthotists and Prosthetists and is a past president. Ken has a particular interest in the orthotic management of CMT and says each patient challenges him to apply his experience, knowledge and skill to help that patient achieve the highest level of activity possible—whether it's playing sports, shopping with family and friends or being able to walk independently around the house without pain.



**William S. David, MD, PhD**  
**Associate Professor of Neurology**  
**Harvard Medical School**

William S. (Bill) David is the Director of the EMG Laboratory and Neuromuscular Diagnostic Center at Massachusetts General Hospital and the Associate Program Director for the Harvard Partners Neuromuscular Medicine Fellowship Program. He is an Associate Professor of Neurology at Harvard Medical School. He received his MD and PhD degrees from the Albert Einstein College of Medicine in New York.

Dr. David's research, clinical activities and educational/administrative

pursuits center on his subspecialty interests of neuromuscular medicine and clinical neurophysiology (EMG), with a particular focus on Amyotrophic Lateral Sclerosis (ALS).



**Florian S. Eichler, MD**  
Associate Professor of Neurology  
Massachusetts General Hospital

Florian S. Eichler recently became director of the Center for Rare Neurological Diseases at MGH, where he is conducting a FDA-funded clinical trial of L-serine in HSN1. He is also the director of the Leukodystrophy

Service at MGH, where he sees patients with a variety of neurogenetic disorders.

Dr. Eichler received his MD from the University of Vienna Medical School in 1997. He then entered a residency in pediatrics and pursued studies on cerebral blood flow and metabolism at the University of Vienna. In 1999, he won a scholarship to study in vivo MR spectroscopy in pediatric patients with metabolic and neurometabolic conditions at Johns Hopkins. He moved to MGH for residency training in Child Neurology and joined the staff there in 2005.



**Vera Fridman, MD**  
Co-Director, CMT Center of Excellence  
Massachusetts General Hospital

Vera Fridman completed her neurology training at the University of Pennsylvania and her neuromuscular fellowship at Massachusetts General and Brigham and Women's hospitals. She subspecializes in both

neuromuscular medicine and electrophysiology, and sees patients with a variety of neuromuscular conditions. Dr. Fridman has a strong clinical and research interest in inherited neuropathies, including CMT.

Following her neuromuscular training, Dr. Fridman pursued a fellowship through the Inherited Neuropathies Consortium, which allowed her to gain experience in the multi-disciplinary care of patients with hereditary nerve diseases. Since 2013, she has been working to build the CMT Center of Excellence at MGH. Her research focus has been on identifying better ways to measure clinical progression in inherited neuropathies.



**Patrick A. Livney**  
Chief Executive Officer  
Charcot-Marie-Tooth Association

Pat assumed the role of Chief Executive Officer (CEO) of the Charcot-Marie-Tooth Association after serving on the Board of Directors for six years. He took office

in December 2011, saying "The CMTA is growing and changing and will continue to aggressively pursue the first treatment for CMT under the STAR initiative."

Pat works diligently to fund the CMTA's groundbreaking research with the singular goal of finding a cure for CMT. To this end, he hosts an annual golf tournament in Chicago, an event that raised more than \$500,000 for CMT research this year.



**Bethany Meloche**  
Director of Social Media  
Charcot-Marie-Tooth Association

Bethany Noelle Meloche, Director of Social Media, began volunteering for the CMTA in 2010. Bethany, who has CMT1A, is a powerful advocate for patients and families affected by CMT. A favorite speaker at

conferences and CMTA branch meetings, Bethany relates honestly and openly the day-to-day struggles of living with CMT and how she tries to rise above them. There are amazing lessons to be learned from buying a slice of pizza for a homeless woman, going on a first date, or—even more frightening for those with CMT—crossing a busy street or getting up off the floor. Bethany encourages everyone to keep tumbling—forward!



**David B. Misener, BSc (HK), CPO, MBA**  
CMTA Advisory Board Member

David B. Misener is an American Board Certified Prosthetist and Orthotist who has been practicing in Albany, NY, since 1998. Originally from Guelph, Ontario, he received his Bachelor of Science in Human Kinetics from the University of Guelph. He then went onto

post-graduate work in Orthotics and Prosthetics at George Brown College, Toronto, Ontario.

Not only does David provide services to people with CMT, he has CMT. He was born with it, as was his son Ethan. David's maternal grandfather was the first known family member with CMT, and David assisted his family in determining the genetic source by working with the Lupski Laboratory at Baylor College of Medicine.



**Elizabeth K. Misener, PhD, LMSW**  
CMTA Advisory Board Member

Elizabeth K. Misener is a licensed master social worker with New York State. She provides coaching on a wide range of issues such as depression, stress management, anxiety, life-work balance and relationships. As a trained social worker, she supports



individuals on their journey of self-discovery through self-reflection and goal setting.

Among Elizabeth's areas of specialization are helping people to improve relationships, reduce stress, encourage personal growth, enhance parenting skills, facilitate decision making, manage time, bolster motivation, clarify objectives, and develop concrete, attainable goals. She has been trained in the technique of Problem Solving Therapy, which is an evidence based short-term intervention for people struggling with depression.

Elizabeth and her husband David are the proud parents of two young boys aged 8 and 6. Her husband David was born with CMT1B, and many of his immediate family members also have CMT1B. Ethan, 8, was also born with CMT1B. She is excited to be part of the CMTA community.



**Christopher Ouellette**  
CMTA Board of Directors

Chris is the Executive Vice President of Operations and Service at Alpha Analytical, an environmental analytical testing laboratory in Massachusetts that employs more than 300 people. He has extensive experience in business operations, strategic planning, production and

quality management and executive leadership, and he plans to put it all to work for the CMTA.

His engagement with CMT began when his nephew was diagnosed with CMT 1A in 2001. As a result, his focus is to proactively develop and support initiatives that will increase CMT awareness, enhance the everyday lives of those with CMT and to someday find a cure for CMT. In 2014, he initiated the first annual Cycle 4 CMT event in Vermont, which raised more than \$70,000 in support of the CMTA's STAR program. His goal is to conduct this event annually with a vision to promote the event on a national level. He looks forward to actively supporting the ongoing fundraising efforts of the CMTA and all initiatives that complement the mission and vision of the association.



**Elizabeth Ouellette, Vice-Chair**  
CMTA Board of Directors

When her son was diagnosed with CMT1A 13 years ago, Elizabeth made a conscious decision to become an active participant in the CMTA, with the goal of making a significant difference in the lives of her son and others affected by this disorder.

Upon moving to California, she obtained an MA in Counseling Psychology, a course of study and training proving invaluable in the understanding

of herself and others, especially in the realm of pain disorders and disabilities. Although she does not have CMT, she has dealt with chronic neuropathic and disabling foot pain for the past 20 years, a condition that has not only helped her to relate to, and empathize with, her son and his CMT, but also to all those who have to manage chronic pain.

As a CMTA Board Member, Elizabeth is making a difference not only in her son's life, but in the lives of all those with CMT. Through her writing, school-based presentations, work with the CMTA's branches and various fundraising events, she continues to build CMT awareness and raise funds needed for research via the STAR initiative.



**Sabrina Paganoni, MD, PhD**  
Associate Professor  
Harvard Medical School

Sabrina Paganoni obtained her MD degree at the University of Milan in Italy and a PhD in neuroscience at Northwestern University in Chicago. She completed her medical training at Harvard Medical School with a residency at Spaulding Rehabilitation Hospital and fellowship at Brigham and Women's Hospital/Massachusetts General Hospital.

Dr. Paganoni's clinical and research activities relate to her subspecialty interests of neuromuscular medicine and clinical neurophysiology (EMG), with a particular focus on Amyotrophic Lateral Sclerosis (ALS) and the rehabilitation of neuromuscular diseases. By combining her training in neuromuscular and rehabilitation medicine, she assists people with neuromuscular diseases in maximizing function and quality of life.



**Jill and Vittorio Ricci**  
Boston, MA CMTA Branch

Jill Ricci joined the CMTA in 2012 by helping to create Vittorio's Circle of Friends. She later joined the CMTA's Boston Branch and became co-leader with Mimi Corrigan. Jill lives in Boxford with her husband Costy, Vittorio and daughter Emma. Her whole family is involved with fundraising and supporting the CMTA.



Vittorio Ricci, a senior in high school, was diagnosed with CMT in 2012. He joined the CMTA to raise awareness and has shared his story in many newspaper articles, *Neurology Now* and *The CMTA Report*. Through fundraising, Vittorio has been invited to the MA State House, and he shared a segment on Boston's Fox 25 News and WHDH TV 7. Vittorio's fundraising page has brought in more than \$110,000 for CMT research.



**Susan Ruediger**  
Director of Development  
Charcot-Marie-Tooth Association

Susan was diagnosed with CMT when she was 18 months old and can trace CMT back six generations in her mother's family. She started the local CMTA Branch in Atlanta, Georgia, after the birth of her first child so she could get more information about her genetic disease.

Susan joined the CMTA staff in January 2010, and, in addition to fundraising activities, she has developed strategic partnerships with companies focused on products and services that increase the quality of life for those with disabilities.

Susan holds a BA from Clemson University and is actively involved in the Atlanta Chapter of AFP (Association for Fundraising Professionals), the Georgia Center for Non-Profits, and the Foundation Center. She is a member of the Junior League of Atlanta where she has served on the Board of Directors.



**Marcia Semmes**  
Director of Print Communications  
Charcot-Marie-Tooth Association

Marcia Semmes brings her experience as attorney, editor, fundraiser and executive director of a nonprofit to the newly created position of director of print communications at the CMTA. Marcia's husband, Clark, is the leader of the Baltimore, MD CMTA Branch, and, like everyone whose life has been touched by CMT, she works with the singular goal of seeing it eradicated.



**Jeana Sweeney**  
Director of Community Services  
Charcot-Marie-Tooth Association

Jeana is the co-leader of the Johnstown, PA, CMTA branch, and she is very enthusiastic when it comes to raising awareness and funds for the CMTA. She has done many things, including softball tournaments, the Archy cookbook, a fishing derby and her all-time favorite, "Stepping it up for CMT" in schools. Jeana is the main point of contact for everything related to being a CMTA branch leader. She helps with any questions related to the website, materials and marketing for branches, and any plans or ideas people may have concerning fundraising. Jeana also visits CMTA branches and speaks about the STAR Program, getting members excited about fundraising and interested in what the CMTA is doing for them!



**Brittany Wright, PhD**  
Postdoctoral Fellow  
National Center for Advancing Translational Sciences

In 2009, Brittany Wright graduated with a Bachelor of Science degree in chemistry from Eckerd College in St. Petersburg, Florida, and started the PhD program in Pharmaceutical Sciences at the Eshelman School of Pharmacy at the University of North Carolina at Chapel Hill. Her graduate research was focused on neuroscience drug discovery where she identified a new molecular therapeutic target for chronic pain. She graduated from UNC with a PhD in Pharmaceutical Sciences with a concentration in chemical biology and medicinal chemistry in December 2013.

In April 2014, Brittany joined the laboratory of Dr. James Inglese at the National Center for Advancing Translational Sciences (NCATS), the newest institute at the National Institutes of Health, where she is focused on assay design and development for CMT1B. Brittany is excited to join the CMTA STAR program and enjoys being part of an organization committed to research for CMT.

In addition to being available for "Lunch with the Experts," Brittany will have information on display about the work being done at NCATS and the NIH.



## Vendor and Sponsor Information

We invite you to visit Brittany and our featured vendors and sponsors to learn more about the products they offer and the contributions they are making to research and to the treatment and management of CMT. (Please note, however, that the CMTA does not endorse the products or services offered by these vendors.)

