



SEPTEMBER 6-8, 2024

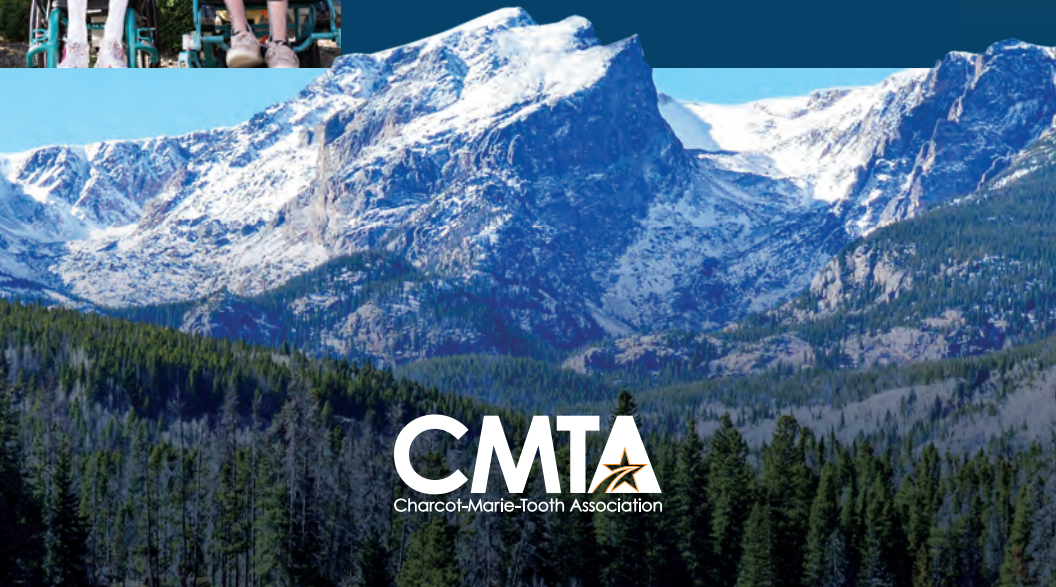
2024 CMTA PATIENT & RESEARCH SUMMIT

summit.cmtausa.org

PRESENTED BY  APPLIED THERAPEUTICS



Virtual Attendees Agenda & Speakers



CMTA
Charcot-Marie-Tooth Association



2024 CMTA PATIENT & RESEARCH SUMMIT

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ABOUT CMTA

The **Charcot-Marie-Tooth Association (CMTA)** is a community-led, community-driven 501(c)(3) nonprofit organization with a mission to support the development of new treatments for CMT, to improve the quality of life for people with CMT, and, ultimately, to find a cure.

As the leading global philanthropic funder of CMT research, CMTA unites the community with clinicians and industry experts to accelerate the advancement of treatments, with investments of nearly \$30 million since 2008.



At the heart of our funding initiatives lies **CMTA's Strategy to Accelerate Research (STAR)**, bringing the best CMT researchers, clinicians, and experts in therapy development together with pharmaceutical and biotechnology companies and patients to accelerate the development of treatments and ultimately a cure for CMT.

CMTA actively works to improve the quality of life for all CMT-affected families. Our comprehensive community-first approach includes educational programs and materials, community and professional conferences, a community-led support network through our extensive **CMTA Centers of Excellence network and Branch system** across North America and worldwide.

To learn more about CMTA, visit cmtausa.org.



SATURDAY, SEPTEMBER 7 AGENDA

- **CLICK ORANGE BUTTON TO JOIN MEETING**



- **ALL SATURDAY ZOOM MEETINGS' ID:**

870 0984 3377

- **PASSCODE:**

345948

- **ALL TIMES ARE MOUNTAIN DAYLIGHT TIME (MDT)**

- **FOR TECHNICAL SUPPORT, EMAIL: info@cmtausa.org**



8:30 a.m. - 9:00 a.m. MDT

CMTA WELCOME ADDRESS

Speaker: Sue Bruhn, PhD

Chief Executive Officer, Sue Bruhn, PhD, will give the opening address which will include CMTA highlights, Summit flow of the day, and much more.



9:05 a.m. - 10:00 a.m. MDT

CMTA CENTER OF EXCELLENCE CLINIC TEAMS (ADULT & PEDI)

Speakers: Vera Fridman, MD & Michele Yang, MD

During this session you will have the opportunity to meet the adult and pediatric CMTA Center of Excellence teams from the University of Colorado and Children's Hospital Colorado. This panel discussion will be led by CMT Clinic directors, Vera Fridman, MD, and Michele Yang, MD. This will be an interactive, open Q and A panel discussion.



10:05 a.m. - 10:45 a.m. MDT

PHYSICAL / OCCUPATIONAL THERAPY: HELP FOR TODAY

Speakers: Clinic Teams from CMTA Centers of Excellence

This session will focus on the most important activities people living with CMT can do daily to help manage CMT. You will hear best practices from physical therapists and occupational therapists who work in the CMT Clinics at the CMTA Centers of Excellence at UC and Children's Hospital Colorado.

ALL SATURDAY, SEPTEMBER 7 SESSIONS



● ZOOM MEETINGS' ID: 870 0984 3377

● PASSCODE: 345948



11:15 a.m. - 11:50 a.m. MDT

SURGERY: CMT SURGICAL OUTCOMES

Speaker: Glenn Pfeffer, MD (via Zoom)

In this session, hear from world renowned foot and ankle orthopedic surgeon, Glenn Pfeffer, MD, on surgical options and outcomes for CMT foot reconstruction surgery. Dr. Pfeffer will discuss who “should” and who “should not” seek CMT surgical interventions.



11:55 a.m. - 12:25 p.m. MDT

PULMONARY: CMT-RELATED BREATHING ISSUES

Speakers: Ashraf Elsayegh, MD & Kenny Raymond

Neuromuscular pulmonologist Ashraf Elsayegh, MD, will share information on CMT-induced neuromuscular respiratory muscle weakness. He will discuss the causes CMT-related respiratory impairment and will offer helpful solutions for navigating care and improving symptoms.



1:25 p.m. - 2:10 p.m. MDT

BRACING/AFO PANEL DISCUSSION

Panel Chair: Ken Cornell, CO

Ken leads a panel discussion on bracing and AFO's for CMT patients. Joining Ken are certified orthotists who serve the CMT community at the University of Colorado and Children's Hospital Colorado. They will share information on cutting-edge bracing options for meeting CMT's unique needs.



2:15 p.m. - 2:40 p.m. MDT

CULTIVATING EMOTIONAL WELLBEING WITH CMT: THE POWER OF PURPOSE

Speaker: Payton Rule

CMT community member and graduate student, Payton Rule, shares findings from a study on CMT Emotional Wellness.



2:45 p.m. - 3:00 p.m. MDT

SMALL STEPS, BIG IMPACT — HOW EACH INDIVIDUAL CAN CONTRIBUTE TO CHANGE

Speaker: Jeana Sweeney

Join us for an inspiring presentation by Jeana as she explores the power of individual actions in supporting CMTA's mission. Together, even the smallest efforts can create lasting change for those living with CMT.

SUNDAY, SEPTEMBER 8 AGENDA

- **CLICK ORANGE BUTTON TO JOIN MEETING**
- **ALL TIMES ARE MOUNTAIN DAYLIGHT TIME (MDT)**
- **FOR TECHNICAL SUPPORT, EMAIL: info@cmtausa.org**



KEYNOTE ADDRESS & STAR WELCOME



- **ZOOM MEETING ID: 853 4061 3682**
- **PASSCODE: 646293**

8:30 a.m. - 9:15 a.m. MDT

KEYNOTE ADDRESS

Speaker: Vittorio Ricci

Vittorio Ricci, CMTA's Boston Branch co-leader, will give the keynote address during this time.

STAR WELCOME: INVESTING IN ALL TYPES OF CMT

Speaker: Sue Bruhn, PhD

CMTA CEO Sue Bruhn, PhD, provides a high-level overview of CMTA's extensive research portfolio and its impact for all of CMT.

SUNDAY BREAKOUT SESSIONS



BREAKOUT SESSION: DEMYELINATING CMT (TYPE 1 AND 4)

- ZOOM MEETING ID: 810 2386 4257
- PASSCODE: 681337

9:30 a.m. – 10:30 a.m. MDT

BREAKOUT SESSION: DEMYELINATING CMT (TYPE 1 AND 4)

Panel Chair: John Svaren, PhD

This panel discussion summarizes current research in demyelinating types of CMT. There will be an opportunity for questions.



BREAKOUT SESSION: AXONAL CMT (TYPE 2)

- ZOOM MEETING ID: 873 1676 3077
- PASSCODE: 784355

9:30 a.m. – 10:30 a.m. MDT

BREAKOUT SESSION: AXONAL CMT (TYPE 2)

Panel Chair: Mario Saporta, MD, PhD

This panel discussion summarizes current research in axonal types of CMT. There will be an opportunity for questions.



BREAKOUT SESSION: UNDIAGNOSED/VUS CMT

- ZOOM MEETING ID: 845 0550 4010
- PASSCODE: 061085

9:30 a.m. – 10:30 a.m. MDT

BREAKOUT SESSION: UNDIAGNOSED/VUS CMT

Panel Chair: Stephan Züchner, MD, PhD

Panelists discuss what a VUS genetic test result means and current research into gene discovery. There will be an opportunity for questions.

SUNDAY, SEPTEMBER 8 AGENDA

- ALL REMAINING SUNDAY ZOOM MEETINGS' ID:

881 6891 2672

- PASSCODE:

597677



11:00 a.m. – 11:20 a.m. MDT

CMT-SORD STORY: GENE DISCOVERY TO CLINICAL TRIAL

Speakers: Sue Bruhn, PhD & Stephan Züchner, MD, PhD

Drs. Bruhn and Züchner tell the story of CMT-SORD from discovery to Phase III clinical trial, and the critical role played by CMTA and our partners GENESIS, the Inherited Neuropathies Consortium (INC), and Applied Therapeutics to accelerate the development of the new potential CMT-SORD treatment. Drs. Bruhn and Züchner will also highlight how this impacts all CMT.



11:25 a.m. – 12:00 p.m. MDT

GENE THERAPY 101

Speakers: Zach Nevin, PhD & John Svaren, PhD

Drs. Nevin and Svaren review some of CMTA's current genetic therapy initiatives and discuss how different genetic therapy approaches can be used to treat CMT.



12:00 p.m. – 12:30 p.m. MDT

PATIENTS AS PARTNERS IN RESEARCH, CLINICAL STUDIES & TRIALS

Panel Chair: Katherine Forsey, PhD

The panel discusses how you can be a patient partner in research and participate in the critical work of accelerating the development of treatments.



12:30 p.m. – 12:35 p.m. MDT

CLOSING REMARKS

Speaker: Gilles Bouchard, CMTA Chairman of the Board

Gilles Bouchard, CMTA's Chairman of the Board, will close out the 2024 CMTA Patient & Research Summit.



SPEAKERS

Gilles Bouchard

Chairman of the Board | CMTA



Gilles Bouchard's son Yohan was diagnosed with CMT1A in 2001. He has been a volunteer advisor to the board since 2007 when he organized the strategic retreat in Palo Alto where the STAR (Strategy to Accelerate Research) program was initially defined and launched. Gilles has been delighted to see how fast the STAR program has progressed since then. Gilles joined the CMTA Board of Directors and assumed the position of chairman in March 2016. Gilles was most recently chair and CEO of Livescribe, a venture-backed company that is the leader in the smartpen market. Previously, he was the CEO of Opnext, a maker of fiber optic components. The majority of his career (17 years) was at Hewlett-Packard, working his way up to executive vice president of Global Operations and CIO, reporting directly to CEOs Carly Fiorina and Mark Hurd. He holds an MS in engineering from UC Berkeley and an executive education degree from Harvard Business School. He was born in France, grew up in the Alps, and loves cycling, mountaineering, and skiing.

Kelli Browning, MSN, APRN, CPNP-PC



Kelli Browning is a dedicated Nurse Practitioner specializing in neuromuscular diseases within the Rehabilitation Department at Children’s Hospital Colorado (CHCO). With a robust nursing background in pediatric intensive care (PICU), post-anesthesia care (PACU), and home health care, Kelli brings over seven years of experience to her role, offering a blend of deep clinical knowledge and compassionate patient care. Kelli completed her Bachelor of Science in Nursing at Creighton University. She then went on to progress her career at CHCO where she built a solid foundation in pediatric nursing. Her extensive bedside experience in the PICU and PACU at CHCO further honed her clinical skills and her ability to handle complex cases with a high level of expertise. She pursued her Master of Science in Nursing at the University of Colorado, focusing further on primary care of the pediatric patient, before joining the Neuromuscular team at CHCO and specializing further in rehabilitative medicine.

In her current role, Kelli employs a multidisciplinary approach to patient care, working collaboratively with Neurologists, Physical Therapists, Occupational Therapists, Orthopedic specialists, Pulmonologists, and Rehabilitation specialists. This collaborative framework ensures comprehensive care tailored to each patient’s unique needs, optimizing treatment outcomes and enhancing overall quality of life.

Kelli is committed to staying at the forefront of research and advancements in neuromuscular diseases, integrating the latest evidence-based practices into her care strategies. Her primary focus is on improving functional abilities and aiding in adaptation to help her patients achieve their highest potential in daily life.

Suzanne (Sue) Bruhn, PhD **Chief Executive Officer | CMTA**



Sue has dedicated her career to patient-focused drug development for the treatment of rare diseases. She’s had a long career in biotech, and seen multiple products advance from research through clinical development and launch onto the commercial market. She believes deeply in the power that patient communities bring to the development process

and is excited to join the largest philanthropic funder of CMT research, and the provider of the most CMT patient services in the world. Sue is a scientist by training, with a degree in Chemistry from Iowa State University, and a PhD from MIT. Prior to joining the CMTA, she had numerous executive leadership roles in pharma and biotech companies, including being a CEO multiple times.

Ken Cornell, CO

Orthotist | Cornell Orthotics and Prosthetics



Ken Cornell is co-founder of Cornell Orthotics and Prosthetics in Peabody, Massachusetts, where managing patients with CMT has been his focus for 25 years. He is the Orthotist at the CMTA Center of Excellence at the MGH Boston location. He is past president of the New England Chapter of the American Academy of Orthotists and Prosthetists. He has

been an educator for dynamic carbon ground reaction AFOs and their functional benefits for patients diagnosed with CMT. With his experience in gait training with AFOs, he manages patients orthopedically to minimize deformity and restore a normal gait pattern with improved balance and propulsion.

Ashraf Elsayegh, MD

Pulmonary Medicine | Cedars-Sinai Medical Center



Dr. Elsayegh is a distinguished physician and researcher based in Los Angeles, California. With over sixteen years of experience, Dr. Elsayegh is a foremost expert in the field of pulmonary medicine as it relates to neuromuscular disease. He currently practices at Cedars-Sinai Medical Center and is an associate clinical professor at UCLA School of Medicine.

His clinical and research interests revolve around respiratory function in the neuromuscular patient with special interest in diaphragm dysfunction. Dr. Elsayegh has authored and published numerous articles and textbooks in the field of pulmonary medicine and pulmonary complications in neuromuscular patients. In addition, he has lectured worldwide on these topics. Dr. Elsayegh has been treating neuromuscular patients, including those with Amyotrophic Lateral Sclerosis (ALS) and Charcot-Marie-Tooth (CMT), for over 15 years. He is an adviser on numerous boards in the fields of pulmonary medicine, critical care medicine, sleep medicine, and neuromuscular disease.

Katherine Forsey, PhD

Chief Research Officer | CMTA



Katherine oversees the CMTA's Strategy to Accelerate Research (STAR) and STAR Advisory Board, a group of 30 of the world's leading CMT clinicians and scientists charged with shaping and delivering the CMTA's research strategies. The CMTA currently has more than 50 active research projects, including sponsored research grants with academic

labs and pre-clinical testing studies with biotech/pharma Alliance Partners. Katherine also curates the CMTA's Patients as Partners in Research initia-

tive, which connects the CMT community with opportunities to get involved in CMT research and increase the body of CMT knowledge. Katherine is a research biologist by training. She completed her PhD and BSc in the UK at the University of York. She also spent time at the University of Cambridge, at the Veterinary Research Institute in the Czech Republic, and at Syngenta working on genetic toxicology.

Hattie Foster RN, BSN, AMB-BC

Clinical Specialty Coordinator Neuromuscular Clinic Department of Rehabilitation Department of Rehabilitation at Children's Hospital Colorado



Hattie is the Nurse Clinical Specialty Care Coordinator in the Department of Rehabilitation at Children's Hospital Colorado. She supports all the neuromuscular clinics, including the Charcot Marie Tooth Clinic. Hattie has been the Neuromuscular Clinical Specialty Care Coordinator for over three years and has previous experience in pediatric home care and the NICU. Hattie is committed to enhancing patient outcomes and experience through expert coordination and compassionate care. Hattie is the main point of contact for the CMT Clinic at Children's Hospital Colorado.

Vera Fridman, MD

Associate Professor, Neurology, CMTA COE Center of Excellence Director University of Colorado's (CU) Anschutz Medical Campus



Vera Fridman is the Associate Professor in the Neuromuscular Section at the University of Colorado's (CU) Anschutz Medical Campus, clinical director of the Charcot-Marie-Tooth Association Center of Excellence at the University of Colorado Hospital.

Melissa Gibbons, MS, CGC

Assistant Professor, Pediatric Genetics and Metabolism University of Colorado School of Medicine and Children's Hospital Colorado



Melissa Gibbons is an Associate Professor at the University of Colorado School of Medicine and a Certified Genetic Counselor at Children's Hospital Colorado. She has been a genetic counselor in the Children's Hospital Colorado Multidisciplinary Neuromuscular Clinics since 2007. Her role in clinic is to provide education about genetic testing to patients and their families. She especially enjoys working with the teenagers in the CMT clinic to help them understand the genetics behind their diagnosis.

Thomas Groennebaek, PhD

Clinical Scientist and a Drug Developer | NMD Pharma



Thomas Groennebaek is a clinical scientist and a drug developer at NMD Pharma, a Danish biotech company dedicated to developing novel treatments for neuromuscular and motor neuron diseases or disorders. Thomas holds a PhD in clinical exercise physiology from Aarhus University. During his PhD, Thomas' research focused on the effect

of ischemic conditioning and exercise on muscle function in patients with cardiovascular disease. After finalizing his PhD studies, Thomas joined NMD Pharma in 2020. At NMD Pharma, Thomas works on the clinical development of NMD670, a novel first in class CIC-1 inhibitor that is being developed for the treatment of Spinal Muscular Atrophy, Myasthenia Gravis and Charcot-Marie-Tooth disease type 1 and type 2. Thomas was part of the team that progressed NMD670 through the pre-clinical and phase 1 safety studies and he continues to work on the clinical development of NMD670 across all indications.

Casey Gruber, CPO

Clinic Manager and Certified Prosthetist Orthotist Evergreen Prosthetics and Orthotics



Casey holds a Bachelor of Science degree in Kinesiology from the University of Maryland and a Master's degree in Prosthetics and Orthotics from Northwestern University in Chicago. He completed his prosthetics and orthotics residencies at various Evergreen clinics around Portland, Oregon. Casey focuses on delivering well-rounded, high-

quality patient care. His approach involves tailoring treatment plans to meet the specific goals and unique needs of each individual. He is particularly interested in exploring how Prosthetics and Orthotics can restore mobility and functional independence, thereby enhancing patients' overall quality of life. Casey and his wife enjoy venturing into Colorado's Rocky Mountains and exploring the state's rich bluegrass music scene. When not working, Casey also enjoys spending time with his three cats (Joni, Leonard, and Pepper) and his dog (Juniper).



Karen Hookstadt

***Occupational Therapist II (Senior) Outpatient Occupational Therapy
Children's Hospital Colorado, University of Colorado Hospital***



Karen Hookstadt has been an occupational therapist for over 30 years. She started her career at Shirley Ryan Ability Lab (formerly Rehabilitation Institute of Chicago) focusing in neurology and spinal cord injury. She then moved to Colorado and accepted a position in Management at Spalding Rehabilitation Hospital with a balance of patient care and management duties for 20 years. Karen returned to patient care at University Hospital and joined the Neuromuscular team 7 years ago.

Carolyn Kelley, DPT, PCS

Children's Hospital Colorado



Carolyn Kelley, DPT, PCS is a physical therapist at Children's Hospital Colorado. Her specialty is pediatric neuromuscular disorders. She works in the neuromuscular clinics at Children's Hospital Colorado, including the CMT clinic, and as a clinical evaluator for neuromuscular trials. She is part of the Children's Hospital SMA newborn screening program and foot management teams. Her professional interest is early identification of children with neuromuscular conditions, early mobility and getting children and their families participating in community programs and adaptive recreation. She is a volunteer with Adaptive Recreation for Childhood Health (ARCH) which is a program that helps kids with physical disabilities participate in sports and adaptive recreation.

Marrisa Lafreniere, MS, CGC

Adult Neurology Genetic Counselor

University of Colorado Anschutz Medical Campus



Marrisa received her Bachelor of Science in Psychology with an emphasis in Biology from the University of California Davis. She then worked as a lab manager in a neuroscience laboratory researching the neurobiology of learning and memory for two years. In 2019, Marrisa moved to Colorado to receive her Masters degree in Genetic Counseling from the University of Colorado Anschutz. After graduation in 2021, she was accepted into a Neurology/Rare disease focused Genetic Counseling Fellowship. After completion of her Fellowship in 2022, Marrisa joined the department as a genetic counselor in the Neurology department at CU Anschutz.

Now MARRISA works closely with patients with various genetic neuromuscular and movement disorders to help them understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease. In her spare time, she enjoys spending time with her dog and her partner, crocheting, baking and snowboarding.

Meghan Moore Burk PhD, PT, DP

Physical Therapist III | Children's Hospital Colorado (CHC) Aurora



Dr. Meghan Moore Burk, PT, DPT, NCS, is a Physical Therapist III at Children's Hospital Colorado (CHC) in Aurora. She earned her PhD in Health Science with a focus on Neurologic Rehabilitation in 2024 from Rocky Mountain University of Health Professions. Dr. Moore completed her Doctorate in Physical Therapy at the University of California, San Francisco, and San Francisco State University. She further honed her expertise through a residency in Neurological Physical Therapy at The Johns Hopkins Hospital (JHH) and served as a teaching assistant and clinical instructor in the University of Delaware program. She is a board-certified Neurological Clinical Specialist.

Dr. Moore's clinical and research interests center on neuromuscular and other pediatric neurological conditions. At CHC, she is part of the Neuromuscular Program for Muscle Disorders and works as a Research Therapist on clinical trials investigating antisense oligonucleotides, gene therapy, and myostatin inhibitors. Her research focuses on the impact of these therapies on patients with neuromuscular diseases, using functional outcomes measures. Dr. Moore is also an accomplished author, contributing to research articles and presenting at national and international conferences. She is actively involved in industry-sponsored advisory boards that shape physical therapy standards of care.

Bipasha Mukherjee-Clavin, MD, PhD

Assistant Professor of Neurology | Johns Hopkins University



Dr. Bipasha Mukherjee-Clavin is an Assistant Professor of Neurology at the Johns Hopkins University School of Medicine. She cares for patients with neuromuscular disorders, with a special focus on patients with peripheral neuropathy, immune-mediated neuropathy, and inherited neuropathy (also known as Charcot-Marie-Tooth disease).

She sees patients in the Johns Hopkins Peripheral Nerve and multidisciplinary Charcot-Marie-Tooth clinics and performs electromyography in the Johns Hopkins EMG lab.

Margaret Murphy-Zane, MD

Pediatric Orthopedics

Children's Hospital Colorado, University of Colorado Hospital



Dr. Margaret Murphy-Zane is an orthopedic surgeon in Aurora, CO and is affiliated with Children's Hospital Colorado. She received her medical degree from Columbia University College of Physicians & Surgeons and has been in practice for over twenty years. She specializes in pediatric orthopedics. She is passionate about helping kids get back to doing all the fun things they want to do. She enjoys getting to know the kids and their families and helping them understand and get through their injury or problem.

Zachary Nevin, PhD

Scientist | Conklin Lab



Zach is a Scientist in the Conklin Lab at Gladstone Institutes where he is combining the versatility of stem cells with the power of new CRISPR technologies to develop gene therapies for demyelinating and axonal forms of CMT. Zach completed his PhD in Genetics and Genome Sciences at Case Western Reserve University.

Riccardo Perfetti, MD, PhD

Chief Medical Officer | Applied Therapeutics



Dr. Perfetti has served as Chief Medical Officer of Applied Therapeutics since August 2018. Before joining the team at Applied Therapeutics, Dr. Perfetti served as a Senior Medical Officer, Vice President and Head of Global Medical Affairs, Diabetes and Cardiovascular Business Unit at Sanofi S.A., a publicly traded pharmaceutical company, from October 2007 to August 2018. Prior to joining Sanofi, Dr. Perfetti served in various roles at Amgen Inc., a publicly traded biopharmaceutical company, including as a Director and Global Development Leader in diabetes, obesity, metabolism and endocrinology from December 2004 to August 2007. He was previously an associate professor of medicine at University of California in Los Angeles and a professor of medicine at the National Institutes of Health (NIH). Dr. Perfetti practiced as an endocrinologist at Cedars-Sinai Medical Center and also served as Director of the Diabetes Research Laboratory and Director of the Outpatient Diabetes Program. Dr. Perfetti received his M.D. and Ph.D. in Endocrinology from University La Sapienza in Rome, Italy and received post-graduate training in endocrinology and molecular biology at NIH.

Glenn B. Pfeffer, MD

Director, Foot and Ankle Surgery Program | Cedars-Sinai Medical Center



Glenn Pfeffer, MD, is director of the Foot and Ankle Center at Cedars-Sinai Medical Center. He is also a co-director of the Hereditary Neuropathy Program and Co-Director of the Cedars-Sinai/USC Gloria Kaufman Dance Medicine Center. Dr. Pfeffer has written numerous scientific articles on orthopedics and has edited seven academic textbooks on the foot and ankle. He has been treating foot and ankle problems in patients with Charcot-Marie-Tooth disease for 25 years. He is a past president of the American Orthopedic Foot and Ankle Society and recently served as president of the California Orthopedic Association. Dr. Pfeffer is frequently interviewed on foot and ankle topics and has been featured on CNN, *Dancing with the Stars*, *Dateline NBC*, *Good Morning America*, and in *The New York Times*.

Caitlin Prechel

Certified Prosthetist Orthotist | RISE Prosthetics + Orthotics



Caitlin practices at Rise Prosthetics and Orthotics in Denver. The use of orthotic bracing, mainly ankle-foot orthoses, can greatly improve mobility. Caitlin specializes in orthotic options that can be pursued based on each individual's muscle strength, range of motion, ankle position, and personal goals.

Kenny Raymond

Head of Communications | CMTA



Kenny was diagnosed with CMT1A in 2002 at 29 years old, after many years of trying to figure out what was going on. He has since dedicated his life to studying CMT and developing a unique way of communicating all things CMT to the patient, healthcare, research, and biopharma/biotech communities.

Kenny is an author, respected advocate, and trusted member of the patient community who continually learns and grows his expertise. He now brings to the CMTA his extensive CMT knowledge and his ability to connect with the community we so proudly serve. Kenny's passion for understanding this disease and his dedication to improving the lives of those who are living with CMT remains as strong as ever. His passion for CMT genetics has led him to pursue an MS degree in genetics from Arizona State University.

Vittorio Ricci

Boston CMTA Branch Co-Leader



Vittorio is 26 years old and was first diagnosed with an unknown type of CMT when he was 14 years old. He later was officially diagnosed with SORD-CMT in 2022, after participating in a research study to identify new types of CMT. His family came to know the CMTA as he went through my diagnosis, and have worked with them for the past 12 years to raise awareness and funds for CMT research. His mother, Jill Ricci, has been the Facilitator for the Boston Branch for several years and recently recruited me as a co-facilitator. Vittorio has been involved in phase II & III clinical trials for Applied Therapeutics' CMT-SORD treatment AT-007, which have been on-going the past three years. Currently, he lives in the Boston area & ironically, works as a lab scientist in the pharmaceutical industry.

Payton Rule

Clinical Psychology PhD Student



Payton Rule is a Clinical Psychology PhD student at Washington University in St. Louis and a National Science Foundation Graduate Research Fellow. Diagnosed with CMT at age five, she is also a proud member of the CMT community. Through her involvement as a member and volunteer in the CMT and broader disability communities, Payton recognized the mental health challenges associated with living with rare diseases. These experiences inspired her research, which aims to identify ways to promote mental health and wellbeing among individuals with CMT and other rare diseases and/or disabilities. In her free time, Payton enjoys playing adaptive sports, spending time with friends and family, and exploring local parks with her dog, Whitt.

Rachel Salzman, DVM

Chief Executive Officer | Armatus Bio



Dr. Rachel Salzman, CEO of Armatus Bio, is an expert in drug development in rare diseases where complex biological and business issues intersect with serious unmet medical need. Prior to Armatus, she was an Executive Vice President at Alcyone Therapeutics, a precision medicines company advancing therapies in serious neurological disorders. Before that she co-founded SwanBio Therapeutics in 2017 and served as Chief Executive Officer and Director through 2019. She was the

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company's President and Chief Portfolio & Development Officer until January 2021. She then founded UltraSquared Bio, a not-for-profit organization dedicated to bringing gene therapies to ultra-rare populations where traditional business cases are not tractable. Prior to her time at Swan, Dr. Salzman was the Chief Science Officer of The Stop ALD Foundation a non-profit employing entrepreneurial approaches and innovative methodology towards effective therapies, cures, and prevention of X-linked adrenoleukodystrophy (ALD), an often-fatal neurodegenerative disease. She holds a B.S. in Animal Science from Rutgers University and a DVM in Veterinary Medicine Oklahoma State University.

Mario Saporta, MD, PHD, MBA, FAAN

Neurologist | University of Miami



Dr. Saporta is a clinician-scientist and associate professor of neurology and human genetics at the University of Miami Miller School of Medicine. He is the medical director of the University of Miami CMTA Center of Excellence and MDA Care Center. Dr.Saporta conducts both clinical and translational research in genetic peripheral neuropathies.

He is the North American principal Investigator for the thePharnext Premier trial for CMT1A and leads a research lab focused on drug discovery and genetic therapy development for peripheral neuropathies using human induced pluripotent stem cell models. He serves on the CMTA Scientific Advisory Board. Dr. Saporta received his MBA in health care management from Fundação Getúlio Vargas, his PhD in neuroscience, his MS in neurology, and his MD from the Universidade Federal do Rio de Janeiro.

Caitlyn Silver, MS, OTR/L

Occupational Therapist | Children's Hospital Colorado



Caitlyn Silver, MS, OTR/L is an Occupational Therapist and Associate Clinical Manager at Children's Hospital Colorado (CHCO) in Highlands Ranch, Colorado.

Mrs. Silver received her Masters in Occupational Therapy from the Medical University of South Carolina, in Charleston, South Carolina. She has been an occupational therapist

at Children's Hospital Colorado for 12 years.

Caitlyn's clinical and research interests include children with neuro-muscular conditions (specifically Charcot-Marie-Tooth), neurodevelopmental conditions, and children with sensory integration dysfunction. Clinically, she has been involved in the development of the Constraint Induced Movement

Continued 

Therapy (CIMT) camp program and the Program for Adolescent Living Skills (PALS) group at CHCO. She has contributed to poster presentations at the Muscle Dystrophy Association. Previously, Caitlyn has worked as an occupational therapist in preschool and adult hospital settings.

John Svaren, PhD

Director, IDD Models Core | University of Wisconsin Waisman Center



John Svaren is a professor in the Department of Comparative Biosciences at the University of Wisconsin and director of the cellular and molecular neuroscience core at the UW Waisman Center, where his laboratory is located. Since 2000, he has focused his research program on the genetic basis of peripheral myelination and the disruptions found in peripheral neuropathies such as CMT. He began working with the CMTA to lead one arm of the collaborative STAR project, which investigated PMP22 regulation to develop assays for drug screening for CMT1A. As the scope of CMTA's mission expanded, he became a member of the CMTA Board of Directors and chair of its Scientific Advisory Board, working with scientists and neurologists in the development and implementation of a multi-front strategy to develop new therapies for common forms of CMT.

Jeana Sweeney

Chief Engagement and Gift Officer | CMTA



Jeana was diagnosed with CMT at the age of 14 and has dedicated her entire professional life to the CMTA community and finding treatments for her daughter and all those who live with CMT. She started out as a volunteer in 2001 and in 2008 she became the director of community services, a position she held for nine years. During that time, she expanded the national branch system, developed Patient/Family conferences, created the successful Walk 4 CMT fundraising program, helped start Camp Footprint, led Awareness Month initiatives and built out the grassroots fundraising campaigns for community/branch leaders.

Jeana was named development director in June 2018. In this role, she spearheads the numerous opportunities for donors, board members, corporate sponsors and pharmaceutical companies to support the mission of the CMTA. She also created the Legacy Society and expanded the Innervators Monthly Giving program. In her spare time, Jeana loves to put on her waders, grab her fishing rod and hit the river to fish for smallmouth bass. The Zara Puppy is her favorite lure.

Robert Will, PT, DPT

***Research Physical Therapist Department Of Neurology
University of Colorado Anschutz Medical Campus***



Robert Will is a Research Physical Therapist at the University of Colorado Anschutz Medical Campus. He focuses on integrating evidence-based practices into patient care. He sees patients in the Multidisciplinary Clinic at UCHealth University Hospital and performs outcome measures for multiple clinical research trials. He collaborates with a diverse team of healthcare professionals to provide comprehensive care and is interested in exploring innovative therapeutic interventions to enhance the quality of life for patients through physical therapy.

Michele Yang, MD

***Associate Professor, Pediatrics and Neurology
Children's Hospital Colorado, Aurora, CO***



Dr. Michele Yang is an associate professor in the section of child neurology at Children's Hospital Colorado. She completed her medical training at the University of Pittsburgh School of Medicine in 1999, followed by a general pediatric residency in 2002 and a child neurology residency in 2005, both at the Children's Hospital of Pittsburgh. She completed an electromyography fellowship at Beth Israel Deaconess Medical Center in Boston and Children's Hospital Boston in 2007. She also completed a pediatric neuromuscular fellowship at the Children's Hospital of Philadelphia and the Hospital of the University of Pennsylvania in 2008. She is board certified by the American Board of Psychiatry and Neurology (ABPN) in neurology with a special qualification in child neurology, by the American Board of Electrodiagnostic Medicine in electromyography, and by the ABPN in neuromuscular medicine. She has been at the Children's Hospital Colorado in the section of child neurology since 2009. Recognizing a need for dedicated care specifically focused on the needs of pediatric CMT patients, she started the multidisciplinary CMT clinic in 2020 with her colleagues in rehabilitation medicine, orthopedics, occupational therapy, physical therapy, and genetic counseling. She and her team are dedicated to understanding the needs of pediatric patients with CMT and the best outcome measures to assess progression.

Stephan Züchner, MD, PhD
Professor and Chair, Department for Human Genetics
University of Miami School of Medicine



Dr. Züchner is a professor of human genetics and neurology at the University of Miami Miller School of Medicine. He serves as the chairman of the Dr. John T. Macdonald Foundation Department of Human Genetics and the co-director of the Hussman Institute for Human Genomics. Dr. Züchner's research interests are focused on identifying genetic variants associated with disease. His lab is internationally renowned for its success in identifying several dozen genes for Mendelian disorders, especially axonal neuropathies, ataxia and spastic paraplegia and for evaluating risk factors for complex genetic conditions, including Alzheimer's disease, Parkinson's disease and obsessive-compulsive disorder. Every day, hundreds of patients around the world get tested and diagnosed for the genes identified in his lab. His pioneering group has promoted genome sequencing methods for disease gene identification in humans, mice, worms and drosophila. Utilizing in-house software solutions, such as the widely used GENESIS platform, he is currently pursuing large-scale exome and genome analysis in multiple Mendelian neurodegenerative disorders to map their complex genetic architecture, including modifier genes.



As the leading global philanthropic funder of CMT research, CMTA unites the community with clinicians and industry experts to accelerate the advancement of treatments, with investments of nearly \$30 million since 2008.



September is CMT Awareness Month!

#LETSBEATCMT

It's time the world hears our stories, and together we will turn up the global volume on CMT awareness!



STEP #1

Use your phone to take photos & videos

STEP #2

Discuss your experience with CMT & share advice for others

STEP #3

Share your content on social media using #LetsBeatCMT

STEP #4

Like & share other posts using #LetsBeatCMT

LEARN MORE & JOIN THE MOVEMENT

CMTAUSA.ORG/LETSBEATCMT





SEPTEMBER 6-8, 2024

2024 CMTA PATIENT & RESEARCH SUMMIT

summit.cmtausa.org

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