SUMMER 2024

THE CMTA REPORT

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CMTA
Charcot-Marie-Tooth Association

CAN AI CURE CMT?

HOW ARTIFICIAL INTELLIGENCE IS REVOLUTIONIZING THE DIAGNOSIS, TREATMENT, AND MANAGEMENT OF CHARCOT-MARIE-TOOTH DISEASE

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Dear CMT Community,

Summer is upon us, and I have been thinking about this change in season and how vital renewal can be as we evolve and move forward. Summer can be a time for change—children move on to a new year and a new grade in school, and many of us take a break to recharge on a summer vacation. Here at CMTA, we continue to evolve as an organization while maintaining our commitment to the community and our passion for research. In that vein, I am thrilled to share some exciting news about the evolution of our Board of Directors, and welcome Wendy Arnone, Patricia "Pat" Verduin, PhD, Bernard Coulie, MD, PhD, and Kevin Marks. These remarkable individuals bring a wealth of experience, a deep personal connection to CMT, and a shared commitment to accelerating our mission.

Wendy Arnone's journey with CMT began with her CMT diagnosis over 30 years ago at a time when research on CMT was nearly non-existent. Her goal to find a cure in her lifetime, combined with her extensive experience as a highly accomplished executive in the insurance industry, will be invaluable to our community. Wendy's leadership roles within UnitedHealthcare, Excellus BlueCross BlueShield, and HealthNow New York Inc., culminating in her role as West Region CEO for UnitedHealthcare's Individual and Employer business, exemplify her ability to drive meaningful change. Her background and dedication to sharing her experiences with CMT to support others make her an inspiring addition to our Board.

Pat Verduin's CMT connection is deeply personal, beginning over 30 years ago with her daughter's CMT1A diagnosis. Recognized for her technical expertise and business acumen, Pat's career is marked by pioneering product innovations that enhance global health and environmental well-being. Her roles in leading Fortune 500 companies and tenure as Chief Science Officer at Colgate Palmolive underscore her ability to lead impactful initiatives. Pat's deep commitment to improving the lives of those with CMT aligns perfectly with CMTA's mission.

Bernard Coulie's journey with CMT started in 1997 when doctors diagnosed his son with CMT1A. His experience as a father and his professional expertise as CEO and President of Pliant Therapeutics brings a unique perspective to our Board. Bernard's dedication to advancing treatments for rare diseases, backed by over 20 years in the biopharmaceutical industry, will accelerate our research initiatives forward. His dual role as a physician and biopharma executive offers invaluable insights into patient care and drug development.

Kevin Marks, personally impacted by an unknown type of CMT, brings his legal expertise and dedication to accelerating CMTA's mission. As Chief Legal Officer at Parker Institute for Cancer Immunotherapy, and with previous leadership roles at the California Institute for Regenerative Medicine and Roche Diagnostics, Kevin's extensive experience in the therapeutics and diagnostics sectors will be instrumental in navigating the legal and regulatory compliance landscapes of our research efforts.

We also express our deepest gratitude to Phyllis Sanders, Alan Korowitz, Elizabeth Ouellette, and Chris Ouellette for their dedicated service on the Board of Directors. Their significant contributions have been pivotal in establishing CMTA as the recognized leader it is today. We are pleased to continue working with them in their new roles as special advisors.

As we welcome these new Board members, we remain steadfast in our commitment to our mission: to support the development of new treatments for CMT, improve the quality of life for those with CMT, and ultimately find a cure. Our new Board members' diverse expertise and personal CMT connections will undoubtedly accelerate our efforts and bring us closer to achieving these goals.

Thank you for your continued support and dedication. Together, we are stronger and more united than ever in our fight against CMT. Together, we will continue accelerating progress and transforming the landscape of CMT research and advocacy. Together, we will achieve a world without CMT. Enjoy your summer!

With gratitude and excitement,

Sue Bruhn, PhD
CMTA CEO

INTRODUCING CHRIS COSENTINO: CMTA'S NEW DIRECTOR OF MARKETING

The Charcot-Marie-Tooth Association (CMTA) is thrilled to welcome Chris Cosentino as our new Director of Marketing, a dynamic leader poised to amplify our mission and impact. Chris joined CMTA in April, bringing with him an impressive track record of success in the nonprofit sector, particularly within the rare disease community.

Chris is no stranger to this field. He previously spent nine years at the Huntington's Disease Society of America (HDSA), where he served as the Director of Marketing and Communications. His extensive experience includes leadership roles at some of the nation's most respected nonprofit organizations, equipping him with the strategic insight and dedication necessary to elevate CMTA's initiatives.

"I am honored to serve the CMT community in this role," said Chris Cosentino. "Working alongside my colleagues, advocates, and partners, we will create opportunities to increase fundraising, education, and engagement. We aim to recruit more individuals to use their platforms to support CMTA's mission."

At CMTA, Chris leads the Marketing and Communications team, which includes Kenny Raymond (Head of Communications) and Sarah Kaider (Digital Marketing Manager). Together, they are committed to producing engaging educational content, enhancing fundraising efforts, and propelling the CMTA brand to new heights.

"Engaging with the CMT community is crucial to our success," Chris emphasized. "I want to leverage our powerful stories to reach beyond the CMT community, raising awareness and fostering new partnership opportunities."

When asked why he joined CMTA, Chris said, "The CMT community immediately inspired me. By building the right strategies, we will reach more people and share incredible stories that garner support in the fight against CMT. The staff here is amazing and motivated, and our goal is to support everyone with CMT while accelerating towards bringing treatments and a cure. It's truly an honor to be part of this team, and I look forward to helping CMTA achieve new heights."

Originally from New York City, Chris and his family reside in North Carolina. He looks forward to connecting with the community at events like Camp Footprint, CMTA Summits, and many other opportunities.

Please join us in welcoming Chris Cosentino to the CMTA family. His leadership and vision will contribute significantly to our vision of a world without CMT.

CMTA ANNOUNCES NEW CENTER OF EXCELLENCE IN THE UNITED KINGDOM

CMTA is excited to announce it has awarded the CMTA Center of Excellence designation to Emma Matthews, PhD, and Niranjanan Nirmalananthan, PhD, at St George's University Hospitals CMT clinic in London.

Staffed by world-renowned expert CMT physicians, clinicians, and researchers, each CMTA Center of Excellence is a patient-focused, multidisciplinary CMT clinic where children, adults, and families affected by CMT receive only the best comprehensive care for their CMT. Through this new center at St George's University Hospitals, Drs. Matthews and Nirmalananthan will support CMTA's focus on improving the lives of those with CMT by providing advanced diagnostic services, personalized treatment plans, educational resources, and support networks that empower patients and their families, fostering hope and community.

"The Charcot-Marie-Tooth Association is delighted to welcome Drs. Emma Matthews and Niranjanan Nirmalananthan as codirectors of the new CMTA Center of Excellence at St George's University Hospitals," said CMTA Director of Community Outreach, Laurel Richardson. "This designation highlights their exceptional expertise and ensures our UK CMT community will have greater access to comprehensive, high-quality care. The addition of this center strengthens CMTA's commitment to improve the lives of those living with CMT."

With the addition of St George's University Hospitals CMT clinic as a CMTA Center of Excellence, CMTA now has 59 centers spanning the globe. We are continuing to add centers all the time. If you are a healthcare provider

specializing in CMT care, we invite you to apply to become a CMTA Center of Excellence at this link: cmtausa.org/cmta-center-of-excellence-application/.

Or, if you would like your provider to become a Center of Excellence, we invite you to share the above application link with them.

Visit the Centers of Excellence directory found on page 26 to find the Center closest to you, or visit our website today: cmtausa.org/coe

THE PEN IS MIGHTIER THAN THE SORD

How a CMT Community Member Rallied Support for FDA Approval

CMT-SORD is a unique type of CMT discovered in 2019 with CMTA funding support. Caused by autosomal recessive mutations of the SORD gene, this axonal type of CMT results from the body's inability to properly metabolize sorbitol—a sugar produced by the body when it metabolizes glucose (blood sugar). Sorbitol then climbs to extremely high levels in peripheral nerve cells, leading to CMT-SORD symptom onset and disease progression.

When the team of researchers led by CMTA's Strategy To Accelerate Research (STAR) Advisory Board Member Stephan Züchner, MD, PhD, discovered CMT-SORD, unbeknownst to them, CMTA Alliance Partner Applied Therapeutics already had a drug that targets how the body produces sorbitol.

Applied Therapeutics has an experimental drug they call "AT-007." They had an active clinical trial with the drug for an unrelated disease when Dr. Züchner's team discovered CMT-SORD. The body normally converts (metabolizes) glucose to sorbitol, then sorbitol to fructose. In CMT-SORD, the body cannot convert sorbitol to fructose. Hence, sorbitol continues to climb as the body converts glucose normally. AT-007, now called "govorestat" (go-vor-eh-stat), targets the body's mechanism for converting glucose to sorbitol.

Leveraging CMTA's research ecosystem of preclinical testing, CMT model development, clinical and therapeutic expertise, and an ability to quickly identify candidates for a clinical trial, Applied Therapeutics was able to progress their drug into a Phase III clinical trial for CMT-SORD in less than two years. Dubbed the INSPIRE trial, the company released positive 12-month interim data in February 2024, then announced its plans to prepare for seeking FDA accelerated approval based on available data. With this news, a CMT-SORD community member sprang into action.

Taking Action

Supported by CMTA, Applied Therapeutics is requesting accelerated approval of its CMT-SORD drug. As part of this process, the company is preparing a pre-NDA (pre-New Drug Application) meeting request with the U.S. Food and Drug Administration (FDA). This is the step in the FDA process where a pharmaceutical company presents data and makes its case for why the FDA should grant a new drug application (NDA). CMT-SORD community member Vittorio Ricci wanted to help.

Vittorio started a change.org petition to ask the FDA to consider the patient and caregiver perspective regarding the benefits of govorestat as they review Applied Therapeutics' request for a pre-NDA meeting. The petition also urged the FDA to accelerate approval of the drug for CMT-SORD. The FDA requires at least 1,000 signatures on a petition of this kind before considering what the petition is asking, and Vittorio had just six days to hit this target.

CMTA proudly shared Vittorio's petition in email and across our social media channels. In less than 4 hours, the petition had more than 1,000 signatures. With six days to go, Vittorio had a new plan: 3,300 signatures—one signature for each person in the US who has CMT-SORD. And the community didn't just respond.

Once word got out that Vittorio had upped the goal, you didn't just respond, you didn't just make your voice heard, you took control! With two days to go, the petition reached 3,300 signatures. By the time Vittorio filed the petition on May 1, more than 3,800 people signed on to urge the FDA to integrate the perspectives of individuals living with CMT-SORD, along with their caregivers, in the evaluation of govorestat for accelerated approval as a first-ever treatment for CMT-SORD.

Accomplishing so much in such a brief period would not have been possible without you. CMTA thanks you for supporting this cause, signing the petition, sharing the petition, and making your voice heard. Although this drug and trial are for CMT-SORD, there is potential real impact for all of CMT.

Should the FDA move this drug into accelerated approval, it will help thousands of CMT-SORD community members. It will also set a precedent by which other potential CMT drugs could benefit. Granting Applied Therapeutics' pre-NDA meeting for CMT-SORD has the real possibility of helping all CMT community members regardless of type. CMTA supports our Alliance Partner as they seek accelerated approval for govorestat to treat CMT-SORD.

HEALTH, FITNESS, AND CMT

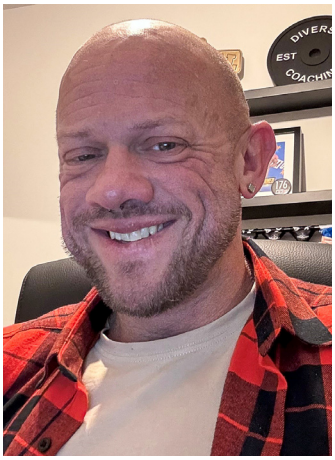
It's well documented that remaining as active as can be while exercising regularly within one's own safety limits is perhaps the best medicine for CMT. Living a lifestyle that is as healthy as possible isn't just a good idea; it's one of the best things those of us with CMT can do to help ourselves manage our CMT. Each of us has our own limitations regarding movement, physical activity, and exercise. Remaining active can look very different for each of us.

It's easy to say, "remain active," or "exercise regularly." Some who have CMT enjoy cycling, swimming, and evening running. Remaining active, however, doesn't necessarily mean daily training for a marathon or competitive bike race. No matter the physical limitations that CMT often deals us, there are ways to enjoy routine exercise while remaining active safely.

From professional bodybuilder to competitive rowing to adaptive exercises to mental resolve, the following four stories are from community members who have turned their CMT into inspiration for overcoming whatever their disease has thrown at them. Their CMT is their strength for managing their disease through physical activity at every level.

John Nixon, CMT1A, UK

Hello, I'm John, aged 44, and I am giving CMT a run for its money, but this wasn't always the case! After suffering from deformed feet, balance issues, struggles with walking, and pain, I was diagnosed with CMT in 1992 at 12 years old. Back then, there was no internet, so I couldn't 'Google' the "How to" of CMT.



John Nixon

I struggled in my late teens and early twenties. I was in a dark place of self-loathing and basking in a pity party, asking myself the question, "why me?" I would fall over, I was very weak, my confidence was shot, and I self-medicated with alcohol, smoking, and junk food. What a catch-22 this was—it was, without a doubt, making my symptoms worse. But, alas, at 28, I decided to be pragmatic.

I joined a gym to strength train and do cardio. This was to increase strength, build muscle, build stamina, improve energy, and hopefully lessen my frequent falls. Although the health workers had encouraged me to stretch daily, they had discouraged me from resistance training claiming that muscle cannot be built when suffering with CMT. As a professional bodybuilder and fitness coach, I can say this is inaccurate.

Sixteen years after starting this journey to take control of my CMT, and now at 44 years of age, although CMT does progress, as it will, I feel the healthiest that I have felt in my adult life. Not just physically, but the bonus is feeling mentally better.

I credit this quality-of-life improvement to living a structured routine of a highly nutritious diet, regular planned exercise of weights and cardio, and keeping away from poor habits such as smoking and alcohol. I cannot do functional training such as CrossFit or triathlons, etc., but I do what is within my capabilities. This involves using weights and cardio safely.

Since starting, I've never looked back.

John Nixon is a CMTA Advisory Board member, fitness coach, bodybuilder, and CMT influencer. Follow John on Instagram: @johnlinecoach.

Julie Stone, CMT2A, USA

My passion for finding CMT-friendly ways to exercise started with my own story. I have CMT2A, and I've always loved fitness. I've been active throughout my life, although, in the past, I've struggled to find exercises that were right for my body. Because of this, I decided to learn more about fitness and CMT.

I turned what was a fitness hobby outward to help others with CMT find safe and effective exercise methods. I've made it my mission to adapt classic exercises to those with CMT at every stage of progression. I strive to build workout plans that match their energy levels and specific needs. Each individual has a unique fitness journey that is ever-evolving. I aim

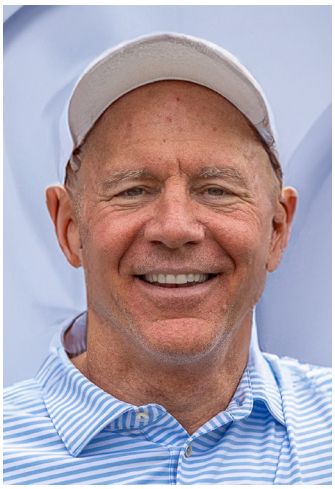

Julie Stone

to make people with CMT feel more stable moving through their everyday lives and feel stronger throughout their whole body. I believe goals are essential to success, but fulfillment comes from learning to love your body during every step of the process.

Julie Stone, NASM CPT, specializes in adaptive exercise, is a CMT influencer, and owner of All Bodies Community. Visit AllBodiesCommunity.com. Follow Julie on Instagram: @cmtdefy.

Steve O'Donnell, CMTX1 (aka CMT1X, CMTX)

No matter where you are in life, it's never too late to start something new. Whether that's a new exercise routine, or healthier diet, it's not really about what you want; it's what you're willing to do to get it.



The secret of your future is hidden in your daily routine. You need to be consistent, dedicated, and put in the work. Do as much as you can with what you have; everybody is different. Everyone can achieve feats they once thought impossible. We must change our mindset and make the extra effort to do so.

It has been proven that your mind triggers you to quit when you have only given 40%. That being said, you can generally do more than you think. To do so, we must remove our governor, stay ahead of the quitting mind, and push past our normal stopping point. Keep moving, go towards your goals no matter what, and do as much as you can with what you have. It is, however, important to listen to your body.

When you're tired, rest, but don't quit. Slow and steady wins the race. To achieve this, you have to have passion; with passion comes commitment; with commitment comes success; and with success, you will get to celebrate. Just do your best, KEEP MOVING, and watch the improvement.

Steve O'Donnell joined CMTA's Board of Directors in 1998 and is the founder and CEO of Therapies for Inherited Neuropathies.

Rebekah Knight, CMT1A, UK

I spent 10 years as a physiotherapist learning how to manage CMT1A after I was diagnosed with it in my first year of university. I started with strength training mainly because I hated all sports and PE growing up and didn't want to be in a team situation in any sport where I might be letting people down or getting in the way. I learned a lot as I started to build my strength and went from struggling to even leg press 40kg to deadlifting over 100kg. During this time, I made videos for CMTA about physical therapy for CMT. I started putting on lottery-funded events with CMTUK to provide education on self-management of CMT and as an opportunity for people with CMT to meet each other and try out some exercises in a safe environment.

In 2021, I started having more problems with my left ankle, and many things

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Steve O'Donnell is renowned for his legendary "Swim for the Cure" across the Chesapeake Bay, an annual event he started in 2002 to raise funds and awareness for CMTA. This 4-mile open-water swim not only highlighted his dedication and endurance but also became a significant fundraising effort, inspiring others and contributing greatly to CMT research. Despite changes in venue over the years, the impact of his swims across the Chesapeake Bay remains a powerful symbol of Steve's resolve never to quit.

FITNESS & CMT

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rowed for Wales at the Home International Regatta last summer, and this year, I was supported by the Para Rowing Foundation to attend a training camp in Portugal, where I trained with coaches and para rowers from around the world.

I post bits of my training online because I know exactly what it's like to get diagnosed with a progressive neurological disease and to be in a state of panic those first few months poking around the whole internet

looking for answers in terms of how it will progress and how it affects people. I regularly get messages from people who've been newly diagnosed and are so pleased to see that although I have limitations, there is still a lot I can do. I am very capable of living a very active lifestyle.

Although not a physiotherapist anymore, I firmly believe in the numerous benefits of exercise. Nowadays, it's much less about "fighting" CMT than being as adaptable as possible and working alongside it.

Rebekah Knight is an adaptive rower, CMT influencer, and former physiotherapy specialist. Follow Rebekah on Instagram: @rebekahknight10.

Rebekah Knight

TO LEARN HOW YOU CAN BECOME AND REMAIN PHYSICALLY ACTIVE while safely enjoying routine exercise, visit: cmtausa.org/exercise

EXPLORE EXERCISE AND PHYSICAL THERAPY FOR INHERITED NEUROPATHIES, produced by Steve O'Donnell: cmtausa.org/tin

EXPLORE THE CMT EXERCISE VIDEO SERIES, produced by Rebekah Knight: cmtausa.org/exercise-series

DOING THE TWO-STEP WITH CMT

BY JEFF SEITZER

Wrapping athletic tape around a callous pad on my bad right foot was the last step in my hour-long preparations. I still had time to do one final check of my gear. Wrong! My ride arrived five minutes early. I grabbed my clubs and the bag with my ultra-groovy supportive devices and rushed out the door. When I arrived at the golf course, I was in momentary shock when I realized I had grabbed my gym bag, not the one with my AFOs. I had no choice but to walk the entire course in what I call my "flats," tennis shoes with only orthopedic insoles. Ugh!

Ironically, I had considered giving up golf altogether not long before. I still played well. All the time and effort planning, organizing, and managing footwear, however, had taken a lot of the fun out of it. And the sore spot on the edge of my bad right foot was only getting worse, despite my efforts to take the pressure off it. Maybe I should take up Scrabble instead?

Blog posts from my fellow CMT community members inspired me not to give up so easily. Many of them had more severe symptoms, and yet they were hiking, running, climbing, you name it, often with much less support than I used to play golf. This got me thinking that maybe I should also be willing to take more chances.

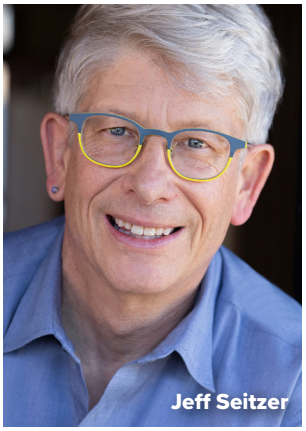
The thinking about it phase was as far as I had gotten. It was life in all its randomness, not conscious choice, which forced me to experiment with how I cope with my condition. This is what I call doing the two-step with CMT. The first time was in graduate school forty years ago.

Late for class one day, running full tilt across campus, my plastic AFO literally shattered. The neurologist I consulted about a replacement suggested that, given my mild symptoms, I should try going without braces. I was over the moon. Why didn't I think of that? Probably because I had been told when diagnosed fourteen years before that without braces, I would end up being severely hobbled.

After fifteen years of trying every conceivable form of footwear, I stumbled (no pun intended) on the CMT Clinic at Wayne State University in Detroit, then in its infancy, where I received much-needed information from Dr. Shy about my condition, CMTX1 (aka CMT1X/CMTX), and received my first SMOs. Two reconstructive foot surgeries a decade later, Dr. Kelekian in Chicago put me back in AFOs. These far more advanced carbonite AFOs helped me avoid injury while regaining my strength.

Fully recovered, I suspected that I relied too much on the AFOs. But I didn't want to take any chances. As an active person, or more accurately, a hyperactive person, avoiding injury was an existential imperative. Losing my mind after only one month of being non-weight-bearing after my reconstructive surgeries convinced me of that.

Walking down the first fairway without the CMT equivalent of Batman's utility belt, I feared the worst: wobbliness, extreme fatigue, AND injury. To my surprise, not only did I leave the course injury-free, but I also played better. I knew that AFOs held my feet too firmly in place to shift my weight effectively as I swung. Without them, however, I assumed that I could not control my swing and would end up spraining an ankle, straining a tendon, or perhaps even breaking a bone. Nothing of the sort occurred. I hit the ball further and straighter than before. And my feet felt like they had the day off without their high-intensity orthopedic companions forcing them into place. The song, "I Feel Like Dancing," silently played through my head as I walked energetically off to the car afterward.


Jeff Seitzer

The CMT two-step sounds like something from The Wizard of Oz. Just tap your heels together, Jeffrey, and you will run barefoot across grassy fields. As if! Step 1, unexpected change, is a quick pivot from panic to momentary joy. Step 2, painful experimentation, is a rocky road, long and drawn out, which, fortunately for me, has led to greater well-being.

The golf course that day had few hills, even around the greens, and did not have yawning sand traps. Wading into the sand at a different course the following week, my bad right foot buckling with each step, and hitting off a steep incline, my ankle making a disturbing cracking sound, gave me pause to think. I had to make some concessions.

At first, I brought along the AFO for my right foot and strapped it on when necessary. This proved unworkable, first because the AFO did not fit into my golf bag, but also because putting it on only to take it off again held everyone up, which is particularly big golf no-no. So, I started wearing an AFO on my right foot, along with a so-called flat on the left foot. The perfect combination.

Besides helping me hit out of the sand and on steep hills, the AFO on my right foot (back foot when swinging) enabled me to pivot even better. I could turn my bad foot from a sideways position to one facing forward, even allowing me to raise my back heel during my follow-through. This enabled me to hit the ball straighter, but it also gave me a few style points, which is always welcome at my age.

Living well with CMT requires a high pain threshold and a willingness to adapt to conditions. This gets old. At the same time, though, it can be an advantage. Studies show that lifelong learning helps keep the aging brain nimble. If so, I am in luck. During a recent visit to the CMT Clinic, now in Iowa City, I learned that weak thumbs are a big problem for those who have CMTX1. If I want to continue playing golf, I will have to completely change how I grip the club. That requires reversing almost sixty years of muscle memory. Bring it, I say!

Jeff Seitzer is the author of a memoir, The Fun Master, *which was a finalist for the Best Book and International Book Awards. It also received Honorable Mention for Book of the Year from the Chicago Writer's Association. He lives in Chicago with his family, where he teaches at Roosevelt and Loyola Universities.*

TIPS AND TRICKS FOR DAILY LIVING

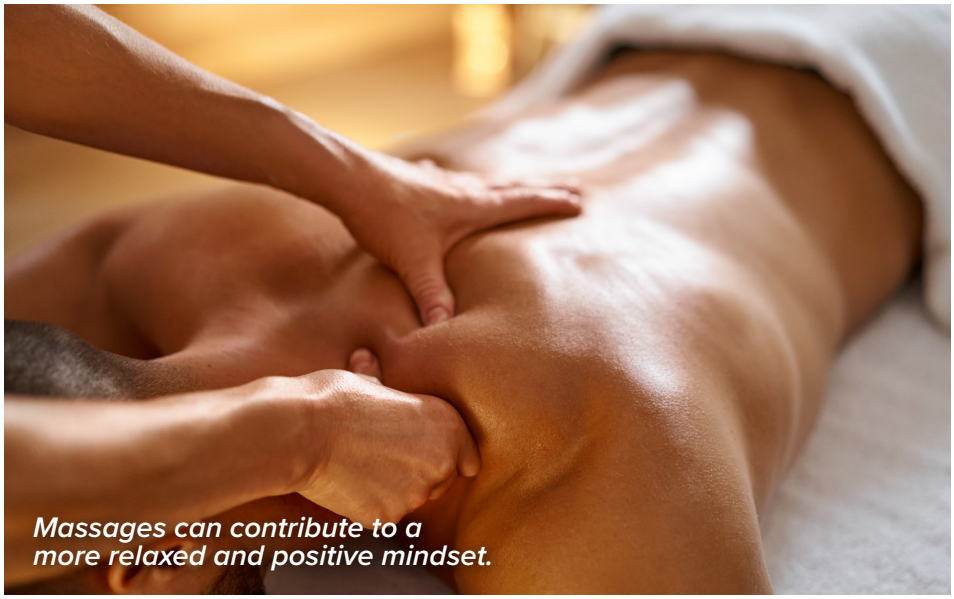
If CMT teaches us nothing else, it teaches us the never-ending need to adapt. From adapting exercise to help maintain our health to adapting the most basic essential tasks, our CMT requires us to adjust. Some adaptations, such as hand controls for driving a car, are complex and expensive. Others are simple, everyday, and readily available tools. Sometimes, an adaptation is just a change in routine.

Living with CMT can come with various challenges, but through shared experiences and practical tips, community members have found ways to enhance their quality of life. Here are some suggestions that have proven helpful for many community members.

One practical idea is to use an office or desk chair to scoot around the house. These chairs are designed to fit through most door openings, making it easier to navigate from room to room without needing constant standing or walking. This can help reduce fatigue and provide a smoother, more efficient way to move about the home. Be mindful, however, of obstacles on the floor, including flooring transitions that could cause the chair to catch and tip over.

Incorporating regular stretching into your daily routine is also highly beneficial. Even when relaxing on the couch, taking a few moments to stretch can help maintain flexibility and reduce muscle stiffness. This simple habit can make a significant difference in overall comfort and mobility.

Use a wheeled office chair to scoot from room to room in your house.



Massages can contribute to a more relaxed and positive mindset.

Massages can be an excellent way to relax tight muscles. Regular sessions, whether self-administered or with a professional, can alleviate tension and promote well-being. These massages soothe the body and contribute to a more relaxed and positive mindset.

To make getting in and out of bed easier, one community member recommends installing a rail on the side of the bed and using a rope ladder down the center. This setup can provide balance and support when pulling yourself up, sitting up from lying down, and getting out of bed.

A powered lift chair stands out as a valuable investment among the various adaptations and tools available. One community member shared that their Stander recliner has been one of the best purchases they've made. These chairs provide crucial support when transitioning from sitting to standing, reducing the strain on muscles and joints. They also offer adjustable reclining positions, enhancing comfort and making finding a suitable posture for relaxation or sleep easier. Incorporating a powered lift chair can significantly improve your mobility, independence, and overall quality of life.

Do you have a tip or trick to share for the next issue of *The CMTA Report*? Please send it to us at info@cmtausa.org

Another approach emphasizes the "3 R's" of caregiving: respect the condition and its stages by learning as much as possible, respect the person by supporting their interests and happiness, and respect yourself as a caregiver by taking time for self-care and recharging.

Incorporating a little sunshine every day can lift your spirits. Singing, laughing, sharing, and turning questions into teachable moments can create a positive atmosphere and improve emotional well-being.

Keep a couple of thick elastic bands handy to maintain muscle tone and strength. Use them for mini resistance exercises with your fingers and hands—at your desk, in your bag, or on the bus.

With gardening or tending to houseplants, sensory loss in the fingertips can make it challenging to feel soil wetness. A soil moisture probe is a practical solution for determining when plants need watering.

Foot care is essential, too. Rolling your feet on a spiky massager roller or using an air compressor massager can alleviate discomfort. Compression socks also help with circulation and reduce swelling.

Experimenting with tools can be beneficial, especially if you have mild weakness. Tools can help form muscle memory with long-term use and remain more manageable even as your strength changes. For example, a favorite jar opener purchased decades ago can still be effective.

Finding a physical activity you love can motivate you to stay active. One community member fell in love with line dancing, supporting their weakened ankles with elastic ankle supports and sturdy shoes, which kept their legs strong for many years.

Lastly, a community member suggests that shoes with motion control technology can provide significant support for those with high arches and weak ankles. Xelero shoes, with their control plate, rollbar, and propulsion elements, have been particularly helpful in reducing strain and enabling longer walks.

Living with CMT is a journey of constant adaptation, but through shared wisdom and practical strategies, we can improve our quality of life and find ways to thrive. By integrating these tips and tricks into our daily routines, we can easily navigate the challenges and maintain our independence. Whether it's a simple change in routine or incorporating specialized tools, each adaptation brings us one step closer to a more comfortable and fulfilling life. Remember, you are not alone—our community is a source of support, strength, and inspiration. Together, we continue to learn, grow, and adapt.

For more about gadgets for everyday living with CMT, please see CMT Gadgets and Assistive Technology with Ashley McElroy, MSM: youtube.com/watch?v=tO73vvYXv0w

With contributions from Angela Graham, San Diego, CA; Ms. Cybell, AL, CMTA Chief Research Officer and community member Katherine Forsey, PhD, York, UK; SunshineSue, Fort Mill, SC; CMTA Miami Branch co-leader Anne Kat, Miami, FL; and Ayda Sanver, North Bethesda, MD.

INNERVATORS HONOR ROLL

ACCORDING TO THE DICTIONARY, an innervator is a nerve stimulator. CMTA INNERVATORS, however, are action-oriented game-changers that sustain CMTA with monthly gifts throughout the year. Life-changing initiatives like Camp Footprint, treatment-focused research, and invaluable education initiatives depend on steady, reliable support from committed and reliable donors. Add your name to the INNERVATORS Honor Roll by visiting cmtausa.org/cmta-innervators and joining today!

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For information on how to make a multi-year pledge or donating securities, email Jeana Sweeney at jeana@cmtausa.org.
CMTA is registered as a 501(c)(3) non-profit organization. **Mail this form to CMTA, PO Box 105, Glenolden, PA 19036**

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THE COMPASS IMPACT: IN THEIR WORDS

I have been an active member of COMPASS for over a year and a half. Finding this community has drastically changed my life for the better. Before getting involved with COMPASS, I felt alone and isolated, and I wanted to push my feelings and experiences with CMT under the rug. Being a part of COMPASS has created a safe space where I feel my unique experiences matter.

I love COMPASS because they understand your deepest personal struggles without you having to explain anything. I will always treasure the connections I have made with those I know already are my lifelong best friends. Because of my involvement with COMPASS, I joined many of my peers and became a camp counselor for Camp Footprint, the only camp in the US for youth who have CMT. Additionally, the in-person connections I've made with everyone are unreal.


Kathy Chau

The COMPASS community fills my cup in so many ways and I am deeply grateful to everyone who is a part of our Funky Feet Tribe.

KATHY CHAU, 27
Salt Lake City, UT

I'm 28 and have lived with the effects of CMT Type 2A since I was six. Until recently, I felt alone in my CMT journey: less able-bodied than those without neuromuscular conditions and more able-bodied than those with more aggressive ones. This no man's land of physical ability—punctuated by unpredictable ebbs and flows, good days and bad—has been difficult and lonely. Following a frustrating flare-up about a month ago, I decided I'd had enough and that I needed more support in my CMT journey. After exchanging emails with CMTA's rockstar director of community outreach, Laurel Richardson, I followed her suggestion.

I connected with COMPASS, a group of other young adults in shoes [and perhaps braces] similar to mine. Days later, I found myself on my first COMPASS call, where I listened to other twenty-somethings exchange stories and perspectives, many of which felt that they had been gleaned by looking through my bedroom window all these years. Our scheduled one-hour call lasted for two. I felt seen on the Zoom call. I was no longer alone.

COMPASS members share similar physical features and the challenges and idiosyncrasies that come with them, like navigating CMT and dating, working when you're chronically exhausted, and feeling self-conscious about wearing our braces without long pants.

Since that first Zoom call, I've deepened connections with my COMPASS peers over the phone and in person. We've shared our dreams, joys, obstacles, music preferences, hobbies, and more. Chiefly, though, we've listened and held space


Ben Bruster

"This no man's land of physical ability—punctuated by unpredictable good days and bad, has been difficult and lonely."

open. Although I'm a new member, COMPASS has already provided me with friends, peer support, plenty of tips and tricks for managing my CMT, and, of course, plenty of banter and dog pics to keep me satisfied.

COMPASS' empathy is palpable, and I firmly believe there's a place for everyone here. I'm excited to see what the future holds, and hopefully, I will meet more COMPASS members at CMTA's Patient and Research Summit this September in Denver, CO.

BEN BRUSTER, 28
Raleigh, NC


Josh Fletcher

"This community has been a pillar of support, providing me with knowledge and experiences highlighting the members' mutual understanding and resiliency."

My life has completely changed since joining COMPASS. COMPASS has helped me navigate the challenges of having CMT, and COMPASS has considerably improved my everyday coping techniques by offering me connections, tools, and, more specifically, customized to CMT's unique problems.

This community has been a pillar of support, providing me with knowledge and experiences highlighting the members' mutual understanding and resiliency. Before joining COMPASS, I frequently had difficulty finding people who could relate to me because of CMT's unique demands and effects. This made me feel alone. Joining COMPASS gave me an instant and deep sense of belonging. Through COMPASS, I've developed close bonds with others who are like me; some are closer than my usual circle of friends.

JOSH FLETCHER,
Salt Lake City, UT

Finding the COMPASS community has truly been so amazing and has had such a positive impact on my mental health. I didn't know any disabled people growing up. Even now, as an adult, I don't have any immediate family members or friends who are disabled. As supportive and understanding as they can be, it's so different connecting with people who have the same or similar lived experiences as me. Finding people who get it makes me feel so seen, heard, and less alone in my struggles and triumphs (because those deserve to be recognized too, no matter how small they may seem to others). I only wish I had found and connected with the COMPASS community sooner!


Sara Jeong

SARA JEONG, 28
Montreal, Canada

WHAT IS COMPASS?

COMPASS, CMTA's young adult group, is not just a community—it's a movement. Over the past year, our members have come together in monthly meetings, delving into discussions, sharing stories, and forging bonds beyond diagnosis. These meetings aren't just about finding support, they're about finding strength in solidarity.

But COMPASS isn't just about meetings—it's about creating unforgettable experiences. Picture this: a happy hour evening at CMTA's Patient and Research Summit in Denver, CO, this September; laughter fills the air as you connect with peers who understand your journey. Moments like these remind us of the power of community and the power to uplift, connect, and empower.

So what are you waiting for? Connect with COMPASS today and be part of something extraordinary.

Follow us on Instagram at @cmtacompass.

To join COMPASS, scan this QR code or visit our landing page at cmtausa.org/compass Together, let's empower each other and build a brighter future for young adults with CMT. Your journey starts here!

COMPASS
NAVIGATING CMT AS A YOUNG ADULT

ACCELERATING RESEARCH AIMED AT BRINGING TREATMENTS AND A CURE

Anonymous Donor Gives $100K to Fund Research by Young Scientists

A very generous $100K given by an anonymous donor will fund new projects by young researchers. Strengthening this capability, CMTA has pledged to match this donation, bringing the total available funding to $200K for projects by young scientists.

"CMTA's Strategy To Accelerate Research (STAR) is powered solely by the generous donations from our community," said CMTA Chief Research Officer Katherine Forsey, PhD. "This anonymous gift is particularly special because it recognizes the potential of young investigators within the CMT research space to contribute to our growing knowledge through new, innovative projects and promotes the application of cutting-edge technologies to CMT. CMTA will use this funding to catalyze new projects and to attract and retain young investigators into the CMT research space."

CMTA is immensely grateful to this anonymous donor for their vision and trust in CMTA and our ability through CMTA-STAR to deliver the greatest possible impact for the CMT community with strategic projects that accelerate the research to bring treatments and, ultimately, a cure.

If you're a young researcher with an innovative, cutting-edge idea that could lead to treatments or a cure for CMT, we invite you to send an introductory email to info@cmtausa.org

Repurposing Drugs for CMT1A

Having an extra copy of the PMP22 gene (three copies instead of two—PMP22 overexpression) causes CMT1A, the most common type of CMT. In previous CMTA-supported research, a team led by CMTA-STAR Advisory Board Chairperson Professor John Svaren, PhD, at the University of Wisconsin Madison found that lowering the overexpression of the PMP22 protein in Schwann cells led to symptom improvement in a model of CMT1A.

Too many copies of the PMP22 gene cause too much of the PMP22 protein in the cells that make peripheral nerve myelin (Schwann cells). This leads to peripheral nerve myelin—the protective sheath of peripheral nerves—functioning poorly, resulting in CMT symptoms and disease progression. Dr. Svaren and colleagues found that targeting the Sterol regulatory element-binding protein (SREBP) can lower PMP22 levels in peripheral nerves. The team is expanding on this research.

With CMTA funding support of $74K, Dr. Svaren and colleagues are administering doses of an FDA-approved drug targeting SREBP in a CMTA-developed model of CMT1A. This new research aims to "turn down" PMP22 expression by affecting SREBP. The team believes turning down PMP22 expression (lowering PMP22 levels in the nerve cells) should improve CMT symptoms. This project is scheduled to conclude by the end of the year.

Nanoparticles for CMT1A, CMT1B, and CMTX1

In a three-year joint project between CMTA and the MDA, researchers led by Alexia Kagiava, PhD, at the Cyprus Institute of Neurology and Genetics are developing nanoparticles to deliver gene therapy approaches for three of the most common types of CMT: CMT1A, CMT1B, and CMTX1 (aka CMT1X, CMTX). The gene mutations causing these three—mutations of the PMP22 gene, the MPZ gene, and the GJB1 gene, respectively, affect the specialized cells that produce and regulate peripheral nerve myelin: Schwann cells.

Delivering gene therapies to Schwann cells is challenging due to the distance these therapies have to travel once delivered and due to the protective blood-nerve barrier. Nanoparticles offer a potential solution because they can cross this barrier, delivering gene therapies to the cells where these genes work.

Since this project began last fall, Dr. Kagiava and colleagues have been working on narrowing down nanoparticle sizes to obtain a size small enough to enter Schwann cells while maintaining the capability to deliver the gene therapy where it's needed. This project illustrates CMTA's commitment to pushing the boundaries of scientific exploration that accelerates the groundwork for many types of CMT.

CMT2A and CMT2F Natural History

With CMTA support of $302,071, an international group of researchers from the Inherited Neuropathies Consortium, led by CMTA-STAR Advisory Board Member Michael Shy, MD, at the University of Iowa, are studying the natural history of CMT2A and CMT2F to learn how these axonal forms of CMT progress over time by measuring changes in various biomarkers in blood, skin biopsies, and "fat-fraction" in calf muscles.

Developed with CMTA support in London by CMTA-STAR Advisory Board Member Mary Reilly, MD, and colleagues, fat-fraction MRI is a new technique that measures fat-to-muscle ratio. As muscles atrophy in CMT, fat replaces the muscle tissue. The fat-fraction MRI technique developed by Dr. Reilly and her colleagues can detect changes in this fat-fraction in calf muscle that correlate with disease progression in as little as twelve months, making this an important biomarker in CMT research. To learn more about the evolution of calf muscle fat-fraction MRI in CMT, see Under the Microscope by CMTA Chief Research Officer Katherine Forsey, PhD, in the Spring 2024 CMTA Report.

This project is taking place over the course of four years, with study

participants undergoing evaluation for two years each. Researchers are actively recruiting patients who have CMT2A or CMT2F. If you have either and would like to participate in this critical research, visit **cmtausa.org/CMT2-natural-history** today to learn how you can join.

Gene Replacement Therapy in CMT4 Subtypes

CMTA announces an investment of $240K into a new gene replacement therapy for CMT4A, with the potential to translate to other types of CMT through a templated approach. Xin Chen, MD, PhD, and CMTA Scientific Advisory Board member Steven Gray, PhD, will lead this important work at the University of Texas Southwestern Medical Center in Dallas, Texas. This investment is part of a key strategic imperative at CMTA to support research with broad impacts for patients.

Recessive GDAP1 gene variants cause CMT4A, a severe demyelinating form of CMT. This project will involve designing and testing an adeno-associated virus 9 (AAV9) vector. Drs. Chen and Gray aim to replace the faulty GDAP1 gene with a fully functional version. This approach will be tested in a model replicating the conditions of CMT4A, providing a crucial step towards potential treatment for this type of CMT. Should this early work prove successful, the researchers believe this approach could be used as a template for other types of CMT, such as CMT4B1 and CMT4D.

"Recent advances in developing desirable AAV capsids, optimizing genome designs, and harnessing modern biotechnologies have dramatically contributed to the growth of the AAV gene therapy field," said Dr. Chen. "With these constantly evolving and improving AAV vector technologies, we believe that studies with relatively rare forms of CMT, such as CMT4A, can provide proof-of-concept for rapidly developing CMT gene therapies. As better AAV vector technology becomes available, we can expand the reach of this 'templated' approach to more forms of CMT."

"Through the Strategy To Accelerate Research (STAR) initiative, CMTA has been at the forefront of accelerating gene replacement therapies for CMT, underscoring our leadership in this critical area of research," said Katherine Forsey, PhD, CMTA Chief Research Officer. "This investment in gene replacement therapy represents a significant step towards developing a roadmap for rapidly translating treatments for CMT into clinical trials. By leveraging advanced AAV vector technologies, we are creating a blueprint for future therapies, bringing hope to the broader CMT community."

DRIVEN BY YOU

These initiatives underscore CMTA's community-led, community-driven role as a leading global philanthropic funder of CMT research aimed at bringing treatments and a cure to our beloved patient community. By uniting the community with clinicians and industry experts, CMTA accelerates the development of new treatments, improves the quality of life for people with CMT, and works tirelessly towards finding a cure for this rare and debilitating disease.

FROM GENE DISCOVERY TO GENE EDITING:
CLOSING THE DIAGNOSTIC GAP AND DEVELOPING CRISPR/CAS9 CAPABILITIES IN CMT

Finding Your Gene

Researchers have discovered over 120 genes linked to CMT; dozens were found with CMTA funding initiatives. Despite this, referred to as the "diagnostic gap," more than 50% of all who have CMT Type 2 still aren't able to obtain genetic confirmation of their disease. Many who fall into this gap have variants of unknown or uncertain significance (VUS).

A VUS is a gene mutation, and scientists haven't yet figured out if it causes something, like CMT, or if it's benign and harmless. With CMTA support, in conjunction with the Inherited Neuropathies Consortium (INC), researchers at the University of Miami, led by CMTA Strategy To Accelerate Research (STAR) Board member Stephan Züchner, MD, PhD, are working to solve these CMT VUS puzzles.

Through expanded data collection and analysis of DNA sequences from almost 2,700 CMT patients in 30 countries and growing, Dr. Züchner and colleagues are using the GENESIS database and analysis platform to understand what a VUS might mean for the CMT patient, and this sometimes leads to the discovery of a new CMT gene. This critical work has led to the discovery of more than 25 genes in recent years, including the CADM3 (CMT2GG), COQ7 (dHMN-COQ7), ITPR3 (CMT1J), and SORD (CMT-SORD) genes. This program has a goal of collecting DNA sequences from 10,000 CMT patients.

Is your type of CMT unknown despite all attempts at genetic testing? If so, CMTA has a research opportunity for you that might lead to finding your gene/type of CMT.

Visit **cmtausa.org/unknown** or scan this QR code with your smartphone:

Fixing Your Gene

Everybody usually has two copies of every gene, with very few exceptions. In many types of CMT, such as CMT2A and CMT2E, the gene with the responsible mutation, the MFN2, and the NEFL genes, respectively, each have one copy with a CMT-causing mutation while the second copy is unaffected. Scientists are developing techniques to "edit out" the mutated copy while preserving the normally functioning copy.

With CMTA support, researchers at the Gladstone Institute, led by CMTA-STAR Board member Bruce Conklin, MD, have demonstrated with CRISPR/Cas9 that it's possible to remove (edit out) the copy of the gene with the responsible mutation while leaving the unaffected copy unharmed and functioning normally. This approach resulted in "curing" affected CMT2E neuron cells in the laboratory.

The Gladstone Institute researchers believe this technology can apply to many other types of CMT. They are currently testing this approach and developing ways to deliver the gene editing capability directly to the nerves in a model of CMT2E while refining these techniques to make them as safe and effective as possible.

Stay current on the latest CMT research by signing up today for our eNewsletter:
cmtausa.org/cmta-enews/

YOU'RE INVITED: PARTICIPATE IN THE LARGEST AND OLDEST CMT BIOBANK

CMTA is investing in creating new stem cell lines to cover more types of CMT and ensure we have representation from both sexes. **We need adults from the CMT community who have specific types of CMT to volunteer to donate a small blood sample and a small skin sample.** The table shows the subtype-related specific mutations we're looking for.

Stem cells can be used to advance the understanding and treatment of CMT in three ways:

- **Disease research:** Study how cells affected by CMT develop and become dysfunctional, and how that might vary.
- **'Clinical trials in a dish':** Test drugs to identify the safest and most promising options for future clinical trials.
- **Cell replacement therapy:** Generate cell types that are affected by CMT, correct their dysfunction, and use these new cells to replace the damaged or dead ones in the patient's body.

Our Partner: NYSCF

CMTA is working on this project with the world-renowned New York Stem Cell Foundation (NYSCF). Our joint mission is to accelerate cures for CMT. NYSCF's independent laboratory is a non-profit accelerator that reduces the cost, time, and risk of developing new treatments and cures. NYSCF uses samples from CMT patients to create stem cells and stores and shares samples and cells with the CMTA's researchers worldwide to help us accelerate the development of treatments.

What Is Involved?

Participation involves providing samples (a small amount of blood, a very small skin sample, and a saliva sample) and health information. Sample collection will be at an NYSCF site in Manhattan, New York, USA. Alternatively, at-home or local collections can also be arranged (within the USA) if travel is a barrier to participation. You will need to be able to provide your genetic test report showing one of the specific mutations listed below. Due to the requirements of the collection process, we can only accept applications from persons living within the USA.

Which CMT Types Are Needed?

We need contributions from each of the subtypes' specific mutations given in this table:

How Do I Take Part?

Please follow the link below to the expression of interest form. The expression of interest should take no more than 5 minutes to complete. Completing the expression of interest form does not commit you to participating in the study or guarantee that you will be eligible to participate.

If selected, you will be contacted by NYSCF, which will confirm your information, determine eligibility, and provide full details about the study procedures. If you decide to participate, NYSCF will ask you to sign a consent form and arrange your sample collection.

Details provided on this form will be accessed only by the CMTA staff and NYSCF staff involved in this study. By completing this form, you are allowing CMTA and NYSCF staff to securely view and store your information and contact you for the purposes of this study.

We may not be able to contact non-selected applicants due to the volume of applications, but we are hugely grateful for your interest in this study.

Secure expression of interest form:
forms.office.com/r/m0LuE1TxJV

To view this opportunity on our website, visit:
cmtausa.org/biobank

or scan this QR code with your smartphone:

SUBTYPE	SPECIFIC MUTATION	BIOLOGICAL SEX
CMT1A	PMP22 gene duplication	Female
CMT1A	PMP22 gene duplication	Male
CMT1B	MPZ (p.Thr124Met) aka T124M	Female
CMT1B	MPZ (p.Arg98Cys) aka R98C	Female
CMTX1 (aka CMT1X/CMTX)	GJB1 del mutation	Male
CMTX1 (aka CMT1X/CMTX)	GJB1 (p.Ser26Leu) aka S26L	Male
CMT2A	MFN2 (p.His361Tyr) aka H361Y	Male
CMT2A	MFN2 (p.Arg364Trp) aka R364W	Male
CMT2A	MFN2 (p.Thr105Met) aka T105M	Male
CMT2A	MFN2 (p.His361Tyr) aka H361Y	Female
CMT2A	MFN2 (p.Thr105Met) aka T105M	Female
CMT2E	NEFL (p.Asn98Ser) aka N98S	Male

UNDER THE MICROSOPE

BY KATHERINE FORSEY, PhD
CMTA Chief Research Officer

In "Under the Microscope," CMTA Chief Research Officer, Katherine Forsey, PhD, takes a closer look at topics related to CMTA's Strategy To Accelerate Research (STAR). Dr. Forsey oversees STAR and CMTA's STAR Advisory Board, comprising over 30 world-leading experts in CMT who provide scientific input, evaluate ongoing or proposed CMTA-funded research projects, and guide CMTA's research strategy. Through STAR, CMTA currently has more than 50 active research projects, including sponsored research grants with academic labs and preclinical testing studies with biotech/pharma Alliance Partners.

CAN AI CURE CMT?

In this issue, I have placed Artificial Intelligence (AI) under the microscope to share several ways this rapidly advancing digital tool is used to support patients with Charcot-Marie-Tooth disease (CMT) and to accelerate the development of treatments.

The advances in AI over the last decade have been astonishing. They have significantly accelerated the discovery of new treatments, improved diagnostic accuracy, increased our ability to discover biomarkers, facilitated personalized medicine approaches by analyzing large-scale biological data, and uncovered intricate disease mechanisms.

Every three months or so, there seems to be a new announcement in biology driven by an AI tool, and most recently, the launch of AlphaFold 3, developed by DeepMind. AlphaFold 3 represents a significant leap forward in predicting the 3D structures of biomolecules (proteins, DNA, and RNA). This is crucial because the 3D structure of biomolecules largely determines their function and interactions within living organisms. What used to take somebody years to figure out and explain can now be done in less than one day. But why is this important for CMT?

Understanding Disease Mechanisms

CMT is a group of inherited neurological disorders characterized by peripheral nerve damage, leading to muscle weakness and sensory loss. By accurately predicting the 3D structures of biomolecules relevant to CMT, such as those involved in peripheral nerve function and maintenance, AI tools like AlphaFold 3 can provide insights into the underlying molecular mechanisms of the disease. This deeper understanding is essential for developing targeted therapies that address the root causes of CMT. In CMT, gene mutations lead to disruptions in the normal function of peripheral nerves. If we can visualize these disruptions via AI tools, we can reveal potential treatment targets.

How Else Could AI Impact CMT Patients and The Development of New Treatments?

Early Diagnosis: AI algorithms can analyze medical records, genetic data, and patient symptoms to aid in the early diagnosis of CMT. By diagnosing the disease earlier, patients can receive support with disease management sooner, potentially slowing disease progression.

Personalized Treatment Plans: AI can analyze large datasets of patient information to develop personalized treatment plans based on individual characteristics and disease progression. This can lead to more effective and tailored interventions for CMT patients. For example, if AI picked up an increased frequency of falls being added to a patient's medical records, it could flag for an assessment by an orthotist to discuss options to improve stability.

Treatment Discovery and Development: AI can streamline the drug discovery process by analyzing vast amounts of biological and chemical data to identify potential drug

AI AND CRISPR IN CMT

The CMTA-funded Conklin Lab at the Gladstone Institutes, UCSF, are world-leading CRISPR specialists and use AI to develop new approaches for therapeutic genetic editing of CMT diseases. Bruce Conklin, MD, said, "We use AI to analyze genomes of patients with CMT to identify the types of edits that will eliminate disease genes. Since there are thousands of potential edits for each, we need AI to analyze the experimental results that will lead us to the edits predicted to give the best outcome for the most patients. We also use AI to analyze microscopic images of human neurons from CMT patients to determine the effectiveness of therapeutic edits."

MACHINE LEARNING IN CMT

Impactful medicines are difficult to discover, especially in the case of progressive neuropathies like CMT. CMTA Alliance Partner Avicenna Biosciences, a Durham, NC-based biotech company, created a machine learning platform capable of identifying novel and optimized clinical candidates related to known drugs, albeit drugs with limitations holding back their impactfulness. A core area of focus is rare neurodegenerative syndromes, and Avicenna is excited about using these new techniques to impact CMT patients.

candidates for CMT treatment. AI-driven virtual screening of compounds can accelerate the identification of promising candidates for further testing.

Clinical Trial Optimization: AI algorithms can optimize clinical trial design by identifying suitable patient populations, predicting treatment responses, and optimizing trial protocols. This can reduce the time and costs associated with clinical trials for CMT treatments. Within clinical trials, scientists use AI to analyze data from clinical assessments such as calf fat-fraction measured by MRI, which I wrote about in the Spring 2024 issue of The CMTA Report. These assessments measure how CMT affects the body over time, and monitoring them during a clinical trial can tell researchers if the treatment is working to slow disease progression.

Remote Monitoring and Management: AI-powered wearable devices and remote monitoring systems can track CMT patients' symptoms and disease progression in real time. This continuous data collection provides valuable insights into the effectiveness of treatments and enables early intervention when necessary.

Genomic Analysis and Detection of New Types of CMT: Researchers use AI to analyze genomic data to identify specific genetic mutations associated with CMT and predict how disease-causing (pathogenic) they are. This information aids in understanding the underlying mechanisms of the disease and in developing targeted therapies. A notable example is the CMTA-supported GENESIS program and how their MAVERICK platform has helped scientists discover over 100 rare disease genes, resulting in over 200 journal paper publications, including many new CMT genes.

Building Better AFOs: Prof Joshua Burns, PhD, Tegan Cheng, PhD, Elizabeth Wojciechowski, PhD, and Joyce Zhanzi Wang, PhD, at the University of Sydney School of Health Sciences & Children's Hospital at Westmead, Sydney, NSW, Australia, shared how they use AI to design better ankle-foot orthoses (AFOs). "For several years, our team of engineers and clinician-scientists have been working on a revolution in the design and production of AFOs using AI and 3D printing. We've shown our 3D-printed AFOs are lighter and perform just as well as traditional AFOs for children with CMT. Now, we are using AI to understand better how AFOs are manually made and digitizing this process to improve functional design. We are also integrating wearable sensors to create smart AFOs and using AI to monitor patients with CMT in their daily lives. This research aims to improve the AFO experience of children and adults with CMT and related neuropathies."

Patient Support and Education: AI-driven virtual assistants and chatbots can provide support and education to CMT patients, answering questions, providing information about the disease, and even offering emotional support. The trick with any such platform is the questions you ask. My personal favorite is ChatGPT by OpenAI. Try asking ChatGPT, "What can I do to slow disease progression in CMT?," or throw in a list of your symptoms and ask it for a diagnosis (always be sure to contact your own doctor if you have any medical concerns). The ChatGPT data source currently includes information only up to May 2023 when using the free version. Regularly check cmtausa.org for the latest CMT news and sign up for CMTA's free research eNewsletter to receive the latest research news straight to your inbox.

AI AND WEARABLES

CMTA has funded several global studies led by the Inherited Neuropathy Consortium (INC) at CMTA Centers of Excellence to study various digital measures of gait, balance, and physical activity in CMT patients and track how they change over time. These tools are now available in clinical trials to help monitor any treatment or intervention's effect.

AI AND FINDING GENES

Stephan Züchner, MD, PhD, co-founder and CEO of The Genesis Project Foundation, whose GENESIS program CMTA funds, shares his story... "A large amount of genetic information from CMT and related rare disease patients is being analyzed in our research projects. AI software helps us tremendously to interpret this data. Examples include AI-supported quality assessment, identification of DNA variants that alter RNA transcripts (RNA splicing), and especially AI that classifies changes into either benign (not disease-causing) or pathogenic (CMT-causing). The latter was developed in-house and is called MAVERICK. It has been a huge help to identify new disease genes as well. MAVERICK is like a trusted assistant to our scientists, who make the ultimate decisions. We expect these novel tools to improve our analysis and even detect patterns that we humans have not—this can greatly accelerate scientific progress in CMT."

Can AI cure CMT?

By leveraging AI technologies in all these ways, healthcare providers, researchers, and patients can work together to improve the diagnosis, treatment, and management of Charcot-Marie-Tooth disease, ultimately leading to the development of treatments and better patient outcomes.

AI, no doubt, will play a starring role at almost every stage along the journey to cure CMT, and in many ways, it already is!

EXPLORING THE MANY NAMES OF CMT2A

BY KENNY RAYMOND

When it comes to CMT, and especially CMT genetics, science is approaching warp speed. In December 1990, there wasn't a single known CMT gene. By Christmas Eve 1993, there were three. By May 1, 1998, with CMTA funding support, researchers had found six CMT genes. By the end of 2011, marking the end of the first 20 years of CMT gene discovery, researchers had discovered 55 CMT genes. Today, 13 years later, scientists have discovered another 79 genes, and dozens with CMTA funding support, bringing the total to 134 known CMT genes CMTA has cataloged. Sometimes, a discovery is a correction to a previous discovery.

Scientists publish their findings in a research paper when they discover a new CMT gene. The research paper is referred to as "published literature." The phrase "in the literature" or "in the published literature" refers to published research papers. The researcher or group of researchers who discover a new CMT gene pick the name for their discovery. There's a CMT naming nomenclature scientists and researchers use as a guideline for naming their discovery—the CMT gene and its mutation, and this name becomes known as the subtype name, such as CMT2A, for example. This is where our story begins.

Discovering the Elusive CMT2A Cause

Before scientists found the first CMT gene, three basic types of CMT existed: CMT1, CMT2, and CMT3. Whether somebody had CMT1 or CMT2 depended on their nerve conduction study (NCS) results. If nerve conduction speeds were slower than 38 meters/sec, amplitudes were somewhat reduced, and each nerve was the relative same, it was CMT1. If nerve conduction speeds were faster than 38 meters/second, amplitudes were significantly reduced, and there was variability between various nerves, it was CMT2. CMT3 was diagnosed when CMT started in infancy. CMT3, today, is no longer used. CMT1 and CMT2, however, are, and the nerve conduction criteria that separate the two from one another are the same today as they were when first published in 1963.

The gene for CMT1A was the first that scientists discovered, and it was found in 1991. Scientists had figured out by this time that there would likely be more than one CMT gene. This first discovery, a duplication of a tiny segment of chromosome 17, 17p11.2-p12, to be exact, predated the exact gene, the CMT1A-causing PMP22 gene duplication, by a year.

Discovering the exact gene involved in CMT1A might seem like a correction to the chromosome segment duplication discovered a year prior. However, the chromosome segment duplication is the reason for the PMP22 gene duplication. The 1st discovered the duplication, and the 2nd clarified the gene.

With the CMT1A discovery coming in 1991, the cause for what CMT researchers and scientists had already dubbed CMT2A remained elusive for 10 years. Then, in 2001, the breakthrough everybody struggled to make was published. Scientists working in Japan at the University of Tokyo discovered a mutation in the KIF1B gene and linked their discovery to CMT2A. Finally, we had the cause for CMT2A. Or did we?

Genetic Roommates

The CMT2A-causing KIF1B gene mutation was discovered in just one family. While this was truly a breakthrough discovery, scientists had difficulty finding this KIF1B mutation in others diagnosed with CMT2A. Try as they may, it just was not happening. CMTA Strategy To Accelerate Research (STAR) Board Member Stephan Züchner, MD, PhD, tells the story in a CMT4Me podcast of how, despite all of his efforts, he could not find this KIF1B mutation in anybody who was thought to have CMT2A. Instead, he found a mutation in a different gene at the same genetic address: the Mitofusin2 gene (MFN2).

In 2004, after identifying his MFN2 discovery as the actual culprit for CMT2A, Dr. Züchner published his findings. Although published literature does not explain this discrepancy between KIF1B and MFN2 in the linkage to CMT2A, the confusion likely stems from the two gene's cytogenic address.

A gene's cytogenic location (its address) is the distinct location within DNA where a gene lives. Both the KIF1B and the MFN2 genes live on chromosome 1. Specifically, both genes live at 1p36.22. Chromosomes are divided into two basic parts (segments): a short arm (p) and a long arm (q). The "p" and "q" are always lowercase. Each arm is subdivided into bands (a number) and sub-band

(another number), with the band and sub-band separated by a decimal point.

In cytogenic location expression, 1p36.22, for example, the first number (1 – 22, or an X or Y) is the chromosome, and everything starting with either a "p" or a "q" and after is the specific location within the chromosome. Putting it all together, 1p36.22 is the house number, street, city, and postal code for both the KIF1B and the MFN2 genes. They are literal genetic roommates.

The Confusion Sets In

After discovering the KIF1B association with CMT, we had a genetic confirmation for CMT2A. Things were great for a few years. Then came the MFN2 discovery, which was proven to be the actual CMT2A gene. One would think that this was it, that KIF1B would see itself out, that MFN2 was the way. This would be the easiest outcome, but CMT is never easy.

At some point after Dr. Züchner's MFN2 discovery, CMT2A, as a subtype name, split into two. The published literature does not give a date, but the KIF1B-associated CMT2A discovery became known as CMT2A1, and the MFN2-associated CMT2A discovery became known as CMT2A2.

CMT2A1 was the name used for KIF1B because this discovery came 1st. CMT2A2 was used for MFN2 because this discovery came 2nd. Remember, though, that both actually referred to the same thing, to the same type of CMT. Everybody became used to this bit of confusion, and all was well for a few years until a new MFN2 discovery in 2011 changed it all up.

CMT2A, regardless of which gene we're talking about (KIF1B or MFN2), is caused by an autosomal dominant mutation in its associated gene. To add to the CMT2A confusion, scientists discovered an autosomal recessive CMT-causing mutation in the MFN2 gene. The scientists who made this discovery named it CMT2A2B. If the autosomal dominant MFN2 discovery is called CMT2A2, it makes sense that the autosomal recessive discovery should be called CMT2A2B, right? The "B" indicates that the discovery follows Dr. Züchner's original MFN2 discovery.

With the help of CMTA funding, 134 known CMT genes have been cataloged since 1991.

Let's review. 2A1 equals autosomal dominant KIF1B. 2A2 equals autosomal dominant MFN2. 2A2B equals autosomal recessive MFN2. Now that we have a handle on the origins of the many names, there's another monkey wrench to throw into the mix.

Clarifying the Confusion

Everybody was used to the confusion of CMT2A1, CMT2A2, and CMT2A2B. The CMT community wasn't a huge fan, but they had a handle on it. The published literature doesn't give a date, but by c.2013, CMT2A2 morphed into CMT2A2A. CMT2A1 remained in association with the KIF1B gene. With the autosomal recessive MFN2 discovery known as CMT2A2B, in what was likely an effort to clear up all the confusion, the autosomal dominant MFN2 discovery morphed into CMT2A2A.

For a period of time, we had CMT2A1, CMT2A2, and CMT2A2A, all referring to the same type of CMT. CMT2A2 and CMT2A2A are the same things linked to the same mutations in the same gene. Another way to look at CMT2A2 and CMT2A2A, hopefully not confusingly, is autosomal dominant MFN2-associated CMT, which we can shorten to AD-MFN2-CMT. Who am I kidding? That's even more confusing! Am I right?

By the late 20-teens, scientists, researchers, CMT experts, and practicing clinicians alike were trying to come up with a solution to this CMT2A multiple-name conundrum. One paper suggested scrapping the current naming conventions favoring an inheritance pattern-gene format, as in AD-MFN2-CMT. While this works great for scientists, researchers, doctors, etc., asking somebody who has CMT to explain what AD-MFN2-CMT is to somebody who's never heard of CMT is a huge ask, especially with complex gene names such as the SH3TC2 gene, which would translate to AR-SH3TC2-CMT (AR = autosomal recessive). It's so much easier to say and explain CMT4C. Have no fear. A solution is on its way.

At some point, and again, the published literature doesn't provide a date; CMT2A1, CMT2A2, and CMT2A2A were merged into simply CMT2A, which is where we are today. ClinGen officially retracted the KIF1B association with CMT in 2020. Today, any internet search returns for CMT2A, CMT2A1, CMT2A2, and CMT2A2A all refer to the same thing—they are synonymous with one another. Any internet search return or mention of CMT2A caused by the KIF1B gene is synonymous with CMT2A caused by the MFN2 gene, but the KIF1B association should be disregarded. CMT2A2B is still CMT2A2B—no changes for this one.

Although the KIF1B association with CMT has been retracted, the KIF1B gene remains on some CMT genetic test panels. CMT genetics experts advise that should a CMT genetic test result include a KIF1B mutation, the MFN2 gene needs to be re-examined and in a much closer analysis, especially if the MFN2 gene was not included in the test. While there are KIF1B gene mutations known to cause other diseases, scientists no longer consider the KIF1B gene to have any connection to any neuromuscular disease, including CMT.

Beyond the CMT2A confusion, two additional subtypes are caused by mutations of the MFN2 gene. In total, various mutations of this gene are associated with four types of CMT: CMT2A, CMT2A2B, CMT2B4, and HMSN-6A. HMSN is the acronym for Hereditary Motor and Sensory Neuropathy. Despite its name, this is a type of CMT. Although four different types are associated with the MFN2 gene, each is caused by different mutations within the gene.

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BRANCH NOTES


Seattle Branch

DENVER, CO
The Denver, CO branch gathered in person on Saturday, February 10th, with 14 members in attendance. During the meeting, they welcomed Occupational Therapist Karen Hookstadt from CMTA's Center of Excellence at the University of Colorado. Karen shared resources, techniques, and exercises for thriving with hand and movement issues associated with CMT. Members shared their stories and approaches to personal health care, life improvement, and enhancement. Karen helped show how living and thriving, despite our challenges, is possible. The Denver, CO branch enjoyed meeting in person.

JACKSONVILLE, FL
The Jacksonville, FL branch welcomed guest speaker Mackenzie Meyer, CPO, LPO, to their in-person meeting on March 2nd. She shared information about lower limb prosthetics and custom ankle-foot orthotics with the branch members. The group closed out their meeting by discussing future fundraising plans. The Jacksonville branch looks forward to their next in-person meeting on June 15th.


Jacksonville Branch

HAWAII
The Hawaii branch met virtually on April 17th to welcome guest speaker Victoria Berezovich. Victoria is the Edmonton, Alberta branch leader, and she is a passionate advocate and community member dedicated to raising awareness, providing support, and fostering understanding of CMT. Victoria shared about CMT positivity and the power of the community. The Hawaii branch is excited about the newfound CMT community.

BOSTON, MA
The Boston branch held its first meeting of 2024, with 11 people joining virtually on March 8th. The members caught up as a group and offered insights into orthotics, exercise, and staying active. Boston branch members look forward to meeting in person this summer for a branch social.

NEW MEXICO
The New Mexico branch met virtually on February 10 with 8 members in attendance. They enjoyed an outstanding presentation by special speaker Ashley McLeroy, MSME, who spoke about assistive technology. She covered numerous assistive technology areas and shared helpful gadgets. The New Mexico branch was thrilled to learn about the resources available to them in their local area. Members are looking forward to their next meeting on May 18th.

WILMINGTON, NC
The Wilmington, NC branch met on May 4th with 16 people in attendance to hear an informative presentation by Kelsey Komyathy. Kelsey is a pharmacist at the Duke University Hospital in Durham, and she is a CMT community member. Kelsey's presentation included valuable information on the recently revised CMT neurotoxic drug list. Many members of the branch are interested in meeting up regularly for coffee. The branch welcomed new members and enjoyed catching up as a group.

WESTCHESTER, NY
The Westchester, NY branch met on March 16th and was delighted to welcome Cheyenne Alfino. Cheyenne specializes in Parenting and Relationship Counseling, and she is a CMT community member. She led the members through a discussion on CMT and emotional wellness. Members said the meeting was "encouraging, emotional at times, and uplifting." The Westchester group enjoyed learning from Cheyenne.

SEATTLE, WA
The Seattle branch met on April 13th and welcomed members in person and virtually. Each member brought gadgets and accessibility aids they found helpful for a CMT show and tell. The members concluded the meeting by creating emergency preparedness mini-kits for people with disabilities. The Seattle branch is looking forward to their next meeting.

MANITOWOC, WI
The Manitowoc, WI branch met on February 27th with 20 members in attendance for an informative and productive meeting with Tiffany Grider, MS, LGC, as a guest speaker. Tiffany is a licensed genetic counselor at the University of Iowa Health Care and sees CMT patients there with Dr. Shy at CMTA's Center of Excellence. She shared about the importance of genetic counseling and allowed time for members to ask questions. The Manitowoc branch was thrilled that Tiffany was able to join them.


Wilmington NC Branch

CMT2A
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Conclusion

If CMT is nothing else, it is confusing, and the CMT2A naming saga lives up to CMT's confusing ways. Out of the many CMT2A names, CMT2A, CMT2A1, CMT2A2, and CMT2A2A, one unifying name has emerged: CMT2A. Today, each of the formers is known simply as CMT2A. When researching symptoms, any finding of CMT2A1, CMT2A2, or CMT2A2A refers to and is synonymous with CMT2A. If you've been diagnosed with CMT2A1, CMT2A2, or CM2A2A, the diagnosis is the same as CMT2A, and each is interchangeable with one another. Today, we have only two simple CMT2A subtypes: CMT2A, of course, and CMT2A2B. What's the difference?

CMT2A is caused by autosomal dominant mutations of the MFN2 gene, and autosomal recessive mutations cause CMT2A2B. Beyond these two, different autosomal dominant mutations in this gene cause the type of CMT called HMSN-6A, and different autosomal recessive mutations in this gene cause the early onset and often severe CMT2B4.

CMT isn't about genes, per se. Rather, CMT is about certain mutations in certain genes. Having a mutation in a gene that has CMT-causing mutations doesn't necessarily mean the mutation is causing CMT. It takes the right mutation, and when multiple subtypes are caused by mutations in a single gene, such as with the MFN2 gene, the mutation itself drives the subtype diagnosis. It's important to note that not every mutation in a CMT gene causes CMT. Some mutations are benign and harmless.

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ALL CMTA BRANCHES CAN BE ACCESSED ONLINE AT www.cmtausa.org/branches

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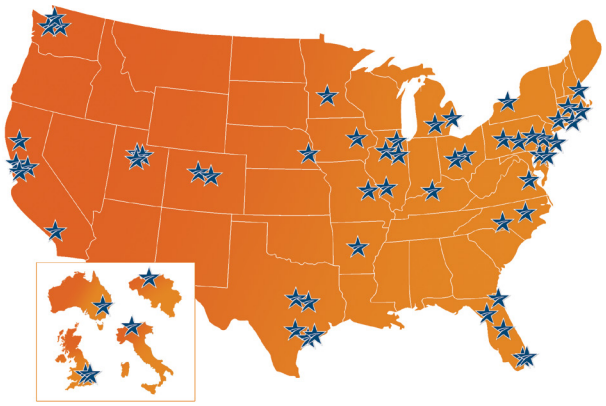
INTERESTED IN STARTING A BRANCH IN YOUR AREA?
Contact CMTA Director of Community Outreach Laurel Richardson at **laurel@cmtausa.org**

CMTA CENTERS OF EXCELLENCE

CMTA's mission is 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. One of the many ways we implement this mission is by sponsoring patient-focused, multi-disciplinary Centers of Excellence CMT clinics. World-renowned CMT care specialists and researchers staff each CMTA Center of Excellence, ensuring those living with CMT receive only the best comprehensive care for themselves and their loved ones.

CMTA Centers of Excellence affiliated with the Inherited Neuropathy Consortium (INC), marked below with an asterisk, goes further by collecting and recording genetic, biological, and other data from individuals with CMT as part of CMTA-funded research. For more information, visit cmtausa.org/coe.



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SALT LAKE CITY (PEDIATRIC)
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Children's National Hospital
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CMTA's Strategy to Advance Research (STAR) Advisory Board is overseen by Katherine Forsey, PhD, CMTA's chief research officer, and comprises a Scientific Advisory Board (SAB), a Therapy Expert Board (TEB) and a Clinical Expert Board (CEB). Each plays a critical role in furthering CMTA's 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.

The SAB provides scientific input for ongoing and proposed projects, the TEB evaluates the translational quality of ongoing and proposed projects, and the CEB provides expert guidance and support to CMTA's Alliance Partners regarding clinical trial planning and delivery. CMTA expanded its Advisory Board by two members in 2023-24 (denoted by an asterisk on the list below) to reflect the evolving needs of STAR.

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University of Miami

Stephan Züchner, MD, PhD, SAB
University of Miami

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Charcot-Marie-Tooth Association

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WHAT IS **CMT?**

Named after the three doctors who first described it in 1886: Charcot (shar-coh), Marie, and Tooth, Charcot-Marie-Tooth disease (CMT) is an inheritable peripheral neuropathy that includes many motor and/or sensory neuropathies, axonopathies, myelinopathies, and neuronopathies.

Due to the effects on the nerves, people with CMT suffer lifelong progressive muscle weakness and atrophy of the arms and legs, and/or progressive sensory loss; and CMT can affect other parts of the body. There is no treatment or cure for this debilitating and often overlooked disease.

CMT leads to problems with balance, walking, and hand use. CMT can cause foot drop, chronic nerve pain, chronic muscle and joint pain, abnormal reflexes, fatigue, tremors, sleep apnea, hearing loss, breathing difficulties, and much more.

Early signs of CMT can be toe-walking, especially in children; frequent trips and falls, frequent ankle sprains, and difficulty with handwriting, tying shoes, or buttoning a shirt.

Visit CMTA's What is CMT webpage today: cmtausa.org/cmt

The Charcot-Marie-Tooth Association 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 more than $24 million since 2008.

EMPLOYEE MATCHING GIFTS are a type of corporate giving program that essentially double an employee's initial donation to an eligible nonprofit organization.

CMTA is a registered partner with **DOUBLE THE DONATION,** a company that makes it easy to find out if your employer will match your donation to CMTA.

Visit **doublethedonation.com/cmta** or scan the code to enter your company name to get started. If your employer has a matching gift program, Double the Donation will provide information about eligibility, match limits, and the procedure for submitting a matching gift application!