WINTER 2024

THE CMTA REPORT

Charcot-Marie-Tooth Association

cmtausa.org

From Research to Reality:
A New Era in CMT

Strategy To Accelerate Research
SPECIAL EDITION

Chief Research Officer, Katherine Forsey, PhD at CMTA's 2024 Patient and Research Summit in Denver, CO

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WINTER 2024
CMTA-STAR SPECIAL EDITION

THE CMTA REPORT | WINTER 2024

Kenny Raymond, *Editor*
Karlyn Rosen Aires, *Designer*

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Email CMTA at info@cmtausa.org

Charcot-Marie-Tooth Association

P.O. Box 105
Glenolden, PA 19036
(800) 606-CMTA (2682)
FAX (610) 499-9267

cmtausa.org

Charcot-Marie-Tooth Association

Dear CMT Community,

A LETTER FROM SUE, OUR CEO

As we reflect on 2024, we're thrilled to share this Winter CMTA-STAR Special Edition, showcasing a year that highlights the essential synergy that research and community can achieve in accelerating progress toward our vision of a future without CMT. This edition is a testament to the impact that we can achieve together, supporting CMTA's Strategy To Accelerate Research (STAR) program and making bold choices on both strategic and scientific fronts.

One transformational change this year is the landmark CMTA-INC Strategic Alliance, with an unprecedented $1.2 million to keep the Inherited Neuropathies Consortium (INC) strong and accessible as its NIH funding winds down. This partnership builds a bridge between laboratory discoveries, drug companies, and community members, positioning CMTA at the forefront of accelerating treatments across all of CMT. At the same time, our ongoing investment in preclinical models and tools, along with clinical trial readiness, moves us closer to delivering transformative options worldwide.

Across these pages, you'll also read about innovative steps forward in genetic therapies, groundbreaking partnerships, and the expansion of opportunities for patients to participate in clinical research. Each article tells a story of commitment—from the scientists in our global network to the thousands of supporters who make this work possible. This year, we continued to break new ground, investing in projects that not only target specific types of CMT but also have the potential to advance treatment options for all affected by CMT. We are not waiting for "someday"; we are working on real solutions today.

Every gain and each success are a direct result of our community's unwavering and generous support. United, we're setting new standards for what's possible in CMT. Please consider a donation to power our mission and bring genuine hope to every person impacted by CMT.

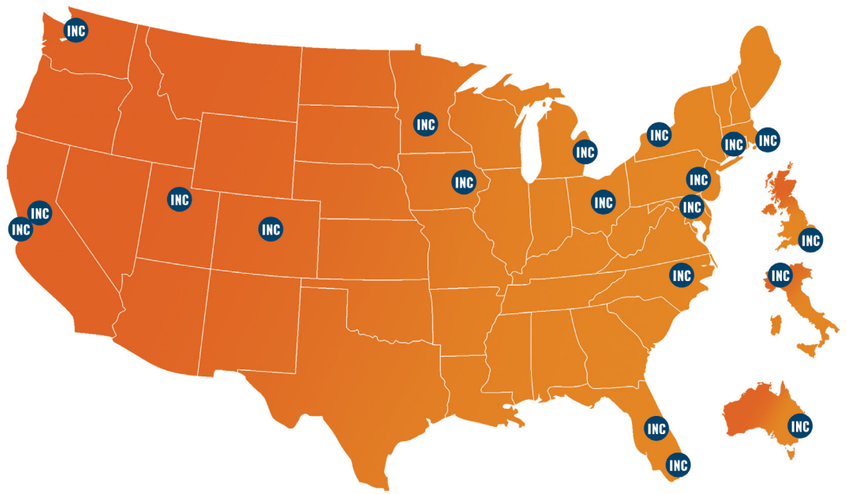
Thank you for being part of this journey. Together, we're accelerating forward, one breakthrough at a time.

With gratitude,

Sue Bruhn, PhD
Chief Executive Officer

A BOLD NEW STRATEGIC ALLIANCE BETWEEN CMTA AND INC:
CREATING A WORLD WITHOUT CMT

CMTA has formed a landmark alliance with the Inherited Neuropathy Consortium (INC), positioning CMTA at the forefront of CMT research and patient care worldwide. Through CMTA's $1.2 million annual funding commitment, this partnership strengthens a global network dedicated to CMT and empowers CMTA as the partner of choice for pharmaceutical companies engaged in clinical trial design. The CMTA-INC strategic alliance will drive research progress from the lab to the clinic, accelerating trials and treatments that offer real hope to the entire CMT community.



The CMTA-INC strategic alliance tackles essential research areas—from preclinical studies and biomarker development to gene discovery, natural history, and clinical trial readiness—ensuring robust support across every stage of drug development. As NIH funding for INC winds down, CMTA's commitment keeps the entire INC network solid and accessible, maximizing impact for researchers, patients, and drug developers. This collaborative model also fosters a new generation of researchers and clinicians, supporting the future of CMT-focused research and care.

INC'S IMPACT

- More than **8,300 patients followed** for up to 15 years
- **30 new CMT genes** identified by INC, including SORD and RFC1
- **300 publications** that advance the understanding of CMT
- **Developed Natural History** on 1A, 1B, 1J, X1, 2A, 2E, 4C, 4B, CMT-SORD
- Developed several **validated outcome measures and biomarkers.**
- Trained more than **20 Principal Investigators**

DRIVING PROGRESS AT EVERY LEVEL OF RESEARCH AND PATIENT CARE

This alliance deepens CMTA's long-standing support for INC, a partnership rooted in a shared mission to advance research and improve care. INC's comprehensive approach—spanning natural history studies, outcome measures, and drug development support—provides crucial tools that accelerate therapies from trials to treatment, tools pharmaceutical partners depend on to bring innovative medicines to market.

"Through the CMTA-INC strategic alliance, we address the needs of every individual affected by CMT,

Inherited Neuropathy Consortium

regardless of type," says CMTA CEO Sue Bruhn, PhD. "CMTA's dedication to comprehensive research, care, and support for all CMT types drives us forward. This partnership strengthens that commitment. By sustaining INC's critical work, we are taking powerful steps towards delivering the research our community deserves. This alliance represents a new chapter for the CMT community, offering real help today and genuine hope for tomorrow. We are proud to be at the forefront of shaping a new era of CMT drug development."

A NEW ERA IN CMT RESEARCH

With this collaboration, the CMTA-INC strategic alliance charts the course for sustainable, impactful progress in CMT research and care, changing the trajectory for individuals affected by CMT everywhere.

"INC's journey always centers around empowering patients through research and clinical innovation," says INC Principal Investigator Michael Shy, MD. "Our alliance provides a sustainable path forward, ensuring each of our over 20 international sites continue the vital work that brings us closer to transformative treatments and a world without CMT. Together, we bridge an essential funding gap and open new doors for collaboration and scientific discovery."

INVESTING IN CMT'S FUTURE

The CMTA-INC strategic alliance represents a new era for CMT research, setting a global standard for collaborative, patient-focused research that uniquely attracts biopharma partners. By sustaining critical funding across INC's network, CMTA is at the forefront of clinical trial readiness, from diverse patient populations and extensive natural history data to validated outcome measures and established trial protocols drug companies need. This alliance offers an ideal pathway for biopharma to accelerate therapies from the lab to patients.

Gilles Bouchard, CMTA Chairman of the Board of Directors, emphasized, "Thanks to CMTA's Strategy To Accelerate Research (STAR) investments over the past decade, 100% funded by the CMT community, we now have a rich ecosystem and pipeline in CMT research, and we are starting to see promising clinical trials emerging. The CMTA-INC alliance is setting the stage for the next phase of STAR, where we engage with pharmaceutical partners in clinical trial design to support testing potential treatments with patients. This is truly an exciting time."

With CMTA and INC working hand-in-hand, the future of CMT research enters a new era—one where collaboration and scientific innovation promise better outcomes for all with CMT. This alliance solidifies CMTA's leadership role in bridging clinical research and patient care, expanding access to clinical trials, and ultimately shaping a future where effective treatments are within reach. CMTA strengthens its mission through this partnership and delivers a powerful message to the global CMT community: together, we're creating a world without CMT.

FROM PATIENTS TO RESEARCH:
CMTA'S iPSC BIOBANK

One key area of research focus for CMTA is to support the creation of tools that can help scientists screen therapies quickly to determine the best molecules to move forward with in drug development. As part of this important objective, CMTA has created a collection of stem cell lines for "disease in a dish" studies of seven different types of CMT. The cell lines were made directly from small skin biopsies or blood samples donated by CMT patients at CMTA Centers of Excellence. The donor samples were converted to stem cells at the New York Stem Cell Foundation (NYSCF) using the highly innovative induced pluripotent stem cell (iPSC) technology, whose significance has been recognized with a Nobel Prize.

iPSCs are powerful tools for studying diseases because 1) they have the genetic profile of a patient with CMT, 2) they can be maintained indefinitely and used for many studies, and 3) they can be converted to cell types in the body that are relevant to the disease process. For example, CMT1A iPSCs can be converted to Schwann cells that make the myelin sheath of peripheral nerves. CMT2A iPSCs can be converted to motor neurons for study, as CMT2A impacts the peripheral nerve axon, which begins with the neuron.

The iPSC collection is part of CMTA's Preclinical Toolbox of models to study CMT and test potential treatments. NYSCF maintains CMTA's iPSC toolbox, currently consisting of 21 cell lines representing CMT1A, 1B, X1 (aka CMT1X, CMTX), 2A, 2E, 2Y, and 4J. Twenty seven drug developers and research groups around the world have thus far used these cell lines.

More information about CMTA's iPSC collection can be found at https://nyscf.org/research-institute/repository-stem-cell-search/.

DRIVING GLOBAL PROGRESS:

ACCELERATING CMT RESEARCH FOR ALL

IN 2024, CMTA'S STAR PROGRAM continued to push the boundaries of science, bringing together a global network of researchers, clinicians, and the global CMT community to accelerate treatment development. From genetic therapy to drug discovery, our therapeutic approaches are designed to offer hope to everyone affected by CMT. With every CMTA-funded innovative project, discovery, and clinical trial, we are one step closer to transforming lives.

None of this would be possible without the generous support of our community, and together, we are driving the future of global CMT research.

GENETIC DISCOVERY: UNLOCKING NEW PATHWAYS FOR CMT TREATMENT

Over 40% of people with CMT do not know the genetic cause of their disease. CMTA's commitment to genetic discovery is unparalleled, and we're changing the landscape of CMT research by funding the GENESIS platform. Over the last decade, scientists collaborating via the GENESIS genomic research platform and database have identified critical genes responsible for many undiscovered types of CMT, providing the roadmap for future therapies. This groundbreaking work is vital not only for those with CMT today but also for future generations.

- **CMT Gene Discovery:** Since its inception, the GENESIS platform has driven new gene discoveries across CMT. For families with no clear genetic diagnosis, these breakthroughs mean more precise testing, diagnosis, and the potential for targeted therapies. Key to this progress is our collaboration with Stephan Züchner, MD, PhD, the GENESIS founder and CMTA-STAR Advisory Board member. His pioneering work, including advancements leading cutting-edge sequencing technology like Long Read Sequencing, has uncovered genes such as SORD and RFC1, linking these discoveries to new paths for treatments.
- **CMT-SORD:** A significant milestone in our genetic discovery efforts was identifying mutations in the SORD gene responsible for CMT, using cutting-edge sequencing technology that unveiled the mutation that had previously been hiding in the genome. Dubbed "CMT-SORD," this discovery almost immediately unlocked the potential for a targeted treatment pathway for this unique type of CMT.
- **Accelerated Path to Treatment:** After identifying the SORD gene, CMTA's STAR program sprang into action, leveraging our established global clinical trial readiness ecosystem. This collaboration enabled a rapid progression from discovery to clinical trials. Govorestat, an experimental drug developed by CMTA-STAR Alliance Partner Applied Therapeutics, emerged quickly as an encouraging potential treatment for CMT-SORD. Applied Therapeutics' ongoing Phase III INSPIRE trial for CMT-SORD has shown that reducing sorbitol levels can improve symptoms, illustrating the real impact of this research.

From the first gene to the last, CMTA is dedicated to advancing genetic discovery so that everyone with CMT can know their type, unlocking pathways for new treatments and, ultimately, a cure. The success of the CMT-SORD discovery highlights the power of collaboration and CMTA's strategic investments. Alongside our genetic discovery work, CMTA's focus on clinical trial readiness ensures that breakthroughs like CMT-SORD can move swiftly from the lab to patients. By uncovering the genetics of CMT, we open doors to treatments that have the potential to change lives. None of this would be possible without your support. Join us on this important journey. Together, we can make a difference.

NOT SOMEDAY. TODAY: CMTA | STAR DRIVES TRANSFORMATIVE CMT RESEARCH

GENETIC THERAPY: CORRECTING THE ROOT CAUSE OF CMT

Genetic therapy holds the remarkable potential to treat CMT at its source, aiming to correct the underlying genetic mutations that cause the disease. Through global collaborations with leading scientists, CMTA-STAR is advancing multiple genetic therapy projects:

- **CMT1A:** Through a multi-site collaboration with scientists at the University of Wisconsin-Madison, Gladstone Institutes at UCSF, and Ospedale San Raffaele in Italy, CMTA are supporting the development of genetic therapy aimed at silencing the overproduction of the PMP22 protein, which is the hallmark cause of CMT1A. This therapy has the potential to stop disease progression and change the lives of thousands of people living with CMT1A.
- **CMTX1 (aka CMT1X, CMTX):** Progress is being made at the Cyprus Institute of Neurology and Genetics, where researchers use cutting-edge nanoparticle technology to deliver genetic therapies for CMTX1 to the specific cells in the body that are affected. This novel approach could be adapted for many other forms of CMT that affect the same cell type, opening doors for new treatments across the board.
- **CMT2A:** At UCSF's Gladstone Institutes, scientists led by CMTA-STAR Advisory Board Member Bruce Conklin, MD, use CRISPR technology to target the MFN2 gene mutations that cause CMT2A. By "editing out" harmful mutations with CRISPR, this approach has the potential to "fix" the MFN2 gene, offering new treatment possibilities.
- **CMT4A, 4C, 4B, and 4J Genetic Therapies:** Ongoing research is focused on developing genetic therapies for these rare CMT subtypes. These innovative projects aim to address the specific genetic

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ACCELERATING CMT RESEARCH FOR ALL

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mutations associated with each, and their successes can potentially create a template for future genetic therapies across many types of CMT that could ultimately benefit the entire CMT community.

Genetic therapy offers more than hope. It offers the possibility of correcting the genetic causes of CMT, bringing us closer to long-term, transformative solutions for many in the CMT community. CMTA supports projects across type 1, 2, and 4 CMTs because each type needs a different approach and, if successful, could be replicated for many subtypes of CMT. By supporting these efforts, you're helping to turn groundbreaking research into real-world treatments that can change lives.

SMALL MOLECULE THERAPIES: EXPANDING TREATMENT POSSIBILITIES

Small molecule therapies offer a promising approach for treating CMT by targeting the proteins and cellular processes involved in the disease. These tiny compounds can easily enter cells to address issues caused by CMT, making them crucial for improving cell function. This strategy allows for potential treatments that can be fast-tracked to patients, benefiting many types of CMT.

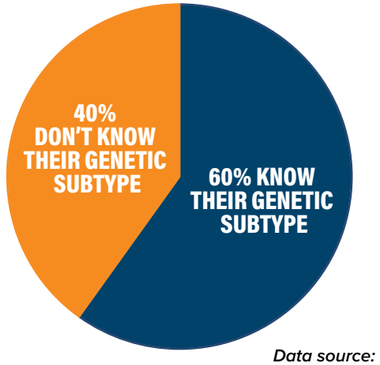
- **CMT1A & CMT1B:** Researchers are repurposing existing FDA-approved drugs to reduce PMP22 protein levels and improve proteasome function (the system that recycles proteins in our cells), offering hope to individuals with both CMT1A and CMT1B. This research, led by Jordan VerPlank, PhD, at the Uniformed Services University, could rapidly bring effective treatments to the community.
- **Activating the Unfolded Protein Response for CMT1B:** Researchers at the Ospedale San Raffaele in Milan, Italy, led by CMTA-STAR Advisory Board Member Maurizio D'Antonio, PhD, are exploring a promising approach to treat CMT1B by activating the body's natural mechanisms to address misfolded proteins. This research focuses on identifying small molecules that enhance the unfolded protein response (UPR), which helps clear harmful proteins caused by mutations in the MPZ gene. By restoring healthy nerve function through this innovative pathway, the team aims to improve symptoms and quality of life for individuals with CMT1B.
- **CMT2E ASO:** Researchers at the University of Miami are developing antisense oligonucleotide (ASO) therapies to target mutations in the NEFL gene responsible for CMT2E. An ASO is like a piece of genetic tape that sticks to a faulty gene to help fix its problems. This approach aims to correct the underlying genetic defect, offering an encouraging avenue for CMT2E community members.

These small molecule therapies, although diverse, share a common goal: to improve the lives of those living with CMT. By targeting the underlying biological mechanisms of the disease, we're laying the groundwork for treatments that could bring real, meaningful change. Your support allows us to explore these innovative treatments and bring hope to many with CMT.

CLINICAL TRIAL READINESS: PAVING THE WAY FOR FUTURE THERAPIES

As we move closer to developing viable treatments, we must be ready to test these therapies in clinical trials. CMTA-STAR invests heavily in preparing for these trials, ensuring the infrastructure is in place when a breakthrough is ready to deliver to the community.

- **Clinical Trial Outcome and Biomarker Development:** Collaborating with the Inherited Neuropathy Consortium (INC), we are supporting the development of critical outcome measures and biomarkers to measure disease progression, especially for types like CMT1A, CMT1B, CMT2A, CMT2F, and CMTX1. These could include things measured in blood samples, or imaging technology like MRI, or functional measures like how far you can walk in a certain number of minutes. These efforts will allow us to more quickly and accurately assess if a potential treatment is working, streamlining the clinical trial process.



Data source: CMTA Patients as Partners in Research Platform

- **Natural History Studies:** CMTA has invested substantially in extending the largest-ever CMT1A Natural History study. The Accelerate Clinical Trials in CMT (ACT-CMT) is an international CMT1A natural history study conducted at five INC clinical sites in the US, UK, and Italy. By gathering long-term data on the progression of CMT1A, ACT-CMT aims to identify key biomarkers and clinical outcomes that will accelerate future clinical trials.
- **Global Clinical Network:** With 65 CMTA Centers of Excellence spanning 6 countries—more than 20 are INC sites, we are creating a global network of clinical trial hubs, ensuring that every person with CMT, no matter where they live or what type of CMT they have, has access to cutting-edge therapies and patient care services. These networks are crucial in accelerating new treatments and moving us closer to clinical trials that benefit the entire CMT community.

Our clinical trial readiness efforts ensure that when the time comes, we are prepared to bring new therapies to those who need them most—efficiently, effectively, and globally. By contributing to CMTA's mission, you are vital in expanding access to the latest innovations for all.

PRECLINICAL MODELS AND TOOLS: LAYING THE FOUNDATION FOR FUTURE SUCCESS

To fast-track the discovery of new therapies, CMTA funds the development of preclinical models and tools that allow scientists to test treatments before moving to human trials. These models help ensure that only the most encouraging therapies reach the next stage.

- **CMT1A:** Researchers at the University of Antwerp are pioneering a human-derived myelin organoid model for CMT1A. This model replicates human peripheral nerve myelin, allowing scientists to test potential therapies in a more accurate biological setting. Such advances bring us closer to finding a therapy to repair the myelin sheath of people with CMT1A.
- **Dental Stem Cell Model for CMT:** Led by Esther Wolfs, PhD, at FIERCE Lab in Belgium, this innovative project utilized human dental pulp stem cells (hDPSC) to create a new model for CMT1A. The research gathered stem cells through community support, allowing for the creation of patient-derived Schwann cells which are implicated in many types of CMT. While focused on CMT1A, the team received tooth donations from donors with other types of CMT, including CMT1E, HNPP, CMTX1, CMT2A, CMT2C, CMT2T, and CMT4A, allowing the development of models for these types of CMT. This highlights how a project for one type of CMT can drive advancements for many others, reinforcing our commitment to all individuals affected by CMT.
- **CMT2A:** Developing stem cell lines and in-the-dish models for CMT2A is another critical focus of CMTA's efforts.

These models enable researchers to better understand the disease and test therapies in realistic environments, ensuring that treatments for CMT2A and other forms of CMT are well-founded and effective.

These preclinical tools provide the foundation for future breakthroughs, allowing researchers to explore new ideas and test potential treatments that could ultimately change the lives of everyone affected by CMT. Your contributions are essential for this vital research, helping to build a brighter future for all with CMT.

CLINICAL TRIALS: ADVANCING TREATMENT FOR ALL CMT SUBTYPES

Clinical trials are essential for translating research discoveries into effective treatments. CMTA-STAR is proud to support several ongoing and upcoming clinical trials aimed at addressing various CMT types. These trials represent a critical step forward in developing therapies that can improve the lives of individuals living with CMT.

- **CMT-SORD: INSPIRE Trial:** CMTA-STAR Alliance Partner Applied Therapeutics is currently conducting the Phase III INSPIRE trial for CMT-SORD. This study investigates the efficacy of govorestat, an experimental treatment that aims to reduce sorbitol levels in patients, potentially alleviating symptoms associated with this unique type of CMT. Early results have shown promise, reinforcing the importance of this trial for the CMT community.
- **HSN: SENSE Trial:** Led by CMTA-STAR Advisory Board Member Mary Reilly, MD, the ongoing Phase II SENSE trial at University College London, in London, UK, is investigating a new treatment for a type of CMT called Hereditary Sensory Neuropathy 1 (HSN1). Recruitment began in August 2023, with plans to enroll 50 participants who will receive either the trial drug or a placebo in a double-blind study design. Currently, 29 participants have been enrolled. Dr. Reilly and colleagues report no serious side effects from the placebo or drug groups.
- **Multiple Types of CMT:** SYNAPSE-CMT Trial: CMTA-STAR Alliance Partner NMD Pharma is conducting a Phase II trial, known as SYNAPSE-CMT, which aims to evaluate a small molecule inhibitor targeting the neuromuscular junction in patients with various types of CMT. This trial signifies a commitment to exploring treatments that can simultaneously benefit many different types of CMT.

These trials are critical in advancing our understanding of CMT and developing effective treatments. They illustrate CMTA's dedication to fostering collaboration among researchers, clinicians, industry partners, and CMT community members, ensuring that the voices of those affected by CMT are heard in the research process. By supporting these initiatives, you're helping ensure new therapies reach the community, providing hope for all living with CMT.

JOIN US:

DRIVING GLOBAL IMPACT FOR THE CMT COMMUNITY

Every advancement CMTA makes in research is a step forward for the entire CMT community. Our CMTA-STAR program, powered by a global network of collaborators and community donors, is leading the way in finding solutions for CMT that will impact people worldwide. From genetic therapy to drug discovery, from biomarkers to preclinical models, we are building the future of CMT treatment—together.

None of this would have been possible without your support, as well as your participation. As we approach the end of 2024, we ask you to join us. Your contributions directly fuel these advancements, bringing hope to everyone with CMT. Now is the time to join us and drive progress forward—together, we can make a lasting difference in the lives of everyone affected by CMT.

CMTA'S GLOBAL COLLABORATION

HUBS OF CMTA PARTNERSHIP:

The Boston Area *includes 11 Alliance Partners and 5 iPSC licensees as well as a CMTA Center for Excellence*

The San Francisco Bay Area *includes 4 Principal Investigators, 5 CMTA-STAR Advisory Board Members, 4 Alliance Partners, 2 iPSC Licensees. and 3 CMTA Centers of Excellence.*

APPLIED THERAPEUTICS
New York City, NY
Drug Developer, CMTA-STAR Alliance Partner, CMT-SORD INSPIRE Trial

Charlotte Sumner, MD
Baltimore, MD
CMTA-STAR Advisor, CMT2C Expert

Michael Shy, MD
Iowa City, IA
CMTA Board of Directors, CMTA-STAR Advisor, INC Principal Invesitgator

Steven Gray, PhD
Dallas, TX
CMTA-STAR Advisor, CMT Genetic Therapies Research Scientist

Bruce Conklin, MD
San Francisco, CA
CMTA-STAR Advisor, CRISPR Genetic Therapies Scientist

Stephan Züchner, MD, PhD
Miami, FL
GENESIS Founder, CMTA-STAR Advisor, CMT Genetics Scientist

- **CMTA-STAR Alliance Partner**
- **CMTA-STAR Toolbox iPSC Licensee**
- **CMTA-STAR Advisory Board Member**
- **CMTA-STAR Funded Researcher**
- **CMTA Center of Excellence**
- **INC-Affiliated CMTA Center of Excellence**

CMTA-STAR ALLIANCE PARTNERS: DRIVING GLOBAL COLLABORATION TO ACCELERATE TREATMENTS FOR CMT

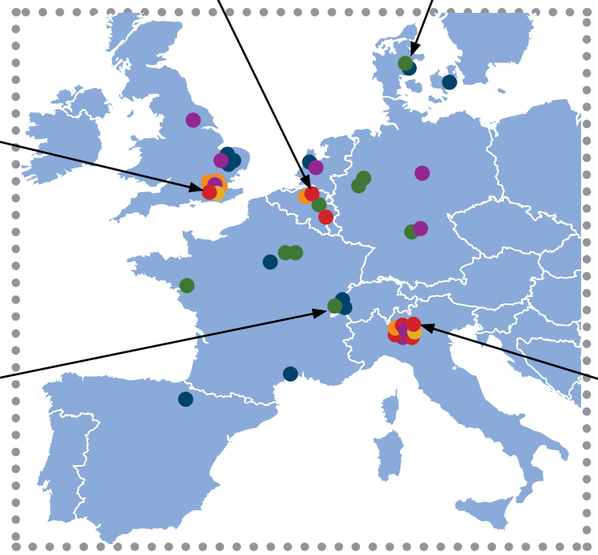
The CMTA-STAR Alliance Network unites 53 partners across ten countries, creating a thriving ecosystem of biotech, pharmaceutical, and research organizations dedicated to advancing patient-focused drug development for CMT. Through this global alliance, partners collaborate with CMTA experts to access cutting-edge CMT models, receive strategic support, key opinion leader and patient input, and accelerate progress toward clinical trials. This collective momentum, driven by the efforts, expertise, and commitment of all involved, accelerates the pathway to a cure and reflects our mission to bring therapies closer to reality for everyone impacted by CMT.

Kleopas Kleopa, MD
Cyprus
Developing Genetic Therapies with CMTA Funding

Vincent Timmerman, PhD
Antwerp, Belgium
Discovered the Genetic Cause of CMT1A. Current CMTA-Funded Organoid Project

NMD PHARMA®
Aarhus, Denmark
Drug Developer, CMTA-STAR Alliance Partner, Conducting CMT-SYNAPSE Clinical Trial

Mary Reilly, MD
London, UK
CMTA Center of Excellence Clinic Director, CMTA-STAR Advisor, HSN SENSE Clinical Trial



NOVARTIS
Basel, Switzerland
Drug Developer, CMTA-STAR Alliance Partner, CMT1A Genetic Therapy

Alessandra Bolino, PhD
Milan, Italy
CMT4B Genetic Therapy, Peripheral Nerve Society President-Elect

A JOURNEY FROM DISCOVERY TO IMPACT:
THE CMT-SORD STORY

In May 2020, a groundbreaking discovery emerged in CMT research. With CMTA funding, a team of scientists at the University of Miami led by Stephan Züchner, MD, PhD, working with the innovative GENESIS genomic research platform, identified mutations in a gene called SORD as the cause of a potentially treatable type of CMT. Now known as "CMT-SORD," which is short for Sorbitol Dehydrogenase Deficiency, the significance of this discovery cannot be overstated.

Unbeknownst to the GENESIS researchers, by uncovering the genetic cause of CMT-SORD, they unlocked the potential for a targeted treatment, transforming a previously unidentified type of CMT into one with a clear therapeutic pathway. The SORD gene is part of a well-known metabolic pathway that breaks down a specific sugar to create energy in the body. Leveraging this knowledge and the SORD gene discovery, CMTA's research network quickly transitioned from identification to action, catalyzing the development of a potential treatment.

CMTA-STAR Mobilizes

Upon identifying the gene mutation causing CMT-SORD, CMTA's Strategy To Accelerate Research (STAR) partners quickly mobilized. Through STAR, CMTA's broad clinical trial readiness ecosystem, which includes the Inherited Neuropathy Consortium (INC) and its sensitive outcome measures developed previously with earlier CMTA-STAR funding, enabled CMT-SORD to accelerate quickly into clinical trials with reliable, functional measures. These trials were conducted by Applied Therapeutics, who had an experimental drug, govorestat, that offered an encouraging solution.

Applied Therapeutics began developing govorestat in 2017 for galactosemia, a disease completely different from CMT-SORD—but in a twist of fate, the two share some key biology: a pathway that metabolizes glucose to another sugar, sorbitol, and then to fructose. But in CMT-SORD, the body can't metabolize sorbitol through this pathway, so it accumulates in the body, eventually damaging the peripheral nerves and leading to CMT symptoms.

When news of the CMT-SORD discovery broke in the scientific journal Nature Genetics, it quickly grabbed the attention of Applied Therapeutics, which had specialized expertise in the SORD gene and its role because of their ongoing clinical studies in galactosemia with govorestat. Recognizing the opportunity, CMTA partners quickly mobilized to support Applied Therapeutics with clinical trial readiness tools for their drug in CMT-SORD.

The Impact of Community and Patients as Partners

In record time, leveraging the power of CMTA's clinical trial readiness ecosystem, scientists collaborated to ensure a smooth transition from discovery to clinical testing. Because some information about govorestat was already known, given its trial in galactosemia, the company could rapidly move into a trial with people based on these preparatory efforts.

CMTA's Patients as Partners in Research platform rallied, taking quick action to share details of the opportunity to participate in this impactful research with the CMT community. CMTA hosted two patient focus groups with CMT community members and the Applied Therapeutics team to ensure patients' views on the proposed treatment, features of the trial design, and the burden of disease were considered. CMTA also helped accelerate the recruitment of patients for the clinical trial. As a result, the CMT-SORD clinical Pilot Study launched in June 2021 with 100% recruitment, just 13 months after the CMT-SORD discovery was announced in May 2020.

Many important things were learned from the CMT-SORD Pilot Study. The study showed that govorestat could lower sorbitol levels and that sorbitol levels drive disease severity. The higher the sorbitol levels, the more severe and further progressed the CMT-SORD symptoms. Lowering sorbitol levels improved CMT symptoms. With these results, a Phase III trial was announced in December 2021, and CMTA's Patients as Partners in Research rallied again to support recruitment.

From these combined efforts, Applied Therapeutics achieved full enrollment of 56 study participants by October 2022, going from discovery to Phase III clinical trial with full enrollment in just over two years—a previously unimagined achievement.

The Impact of CMTA-STAR's Investments in Clinical Trial Readiness

The functional tests measuring govorestat's impact on CMT-SORD patients resulted from CMTA's investments in and collaboration with INC. These tests, including the CMT Functional Outcomes Measure (CMT-FOM), which assesses how treatments impact patients' functionality, and the CMT Health Index (CMT-HI), which evaluates the impact on patients' well-being, have been instrumental in measuring the effects of govorestat on CMT-SORD. CMTA-STAR's forward-thinking investments ensured these outcome measures were in place, allowing the CMT-SORD clinical trials to advance from gene discovery to assessing clinical trial effectiveness and safety at an unprecedented speed.

The acceleration of CMT-SORD into clinical trials exemplifies CMTA's collaborative approach and strategic research investments. The impact of CMTA-STAR's clinical trial readiness ecosystem is evident in govorestat's entry into the Phase III INSPIRE trial in less than two years. Utilizing outcome measures developed through

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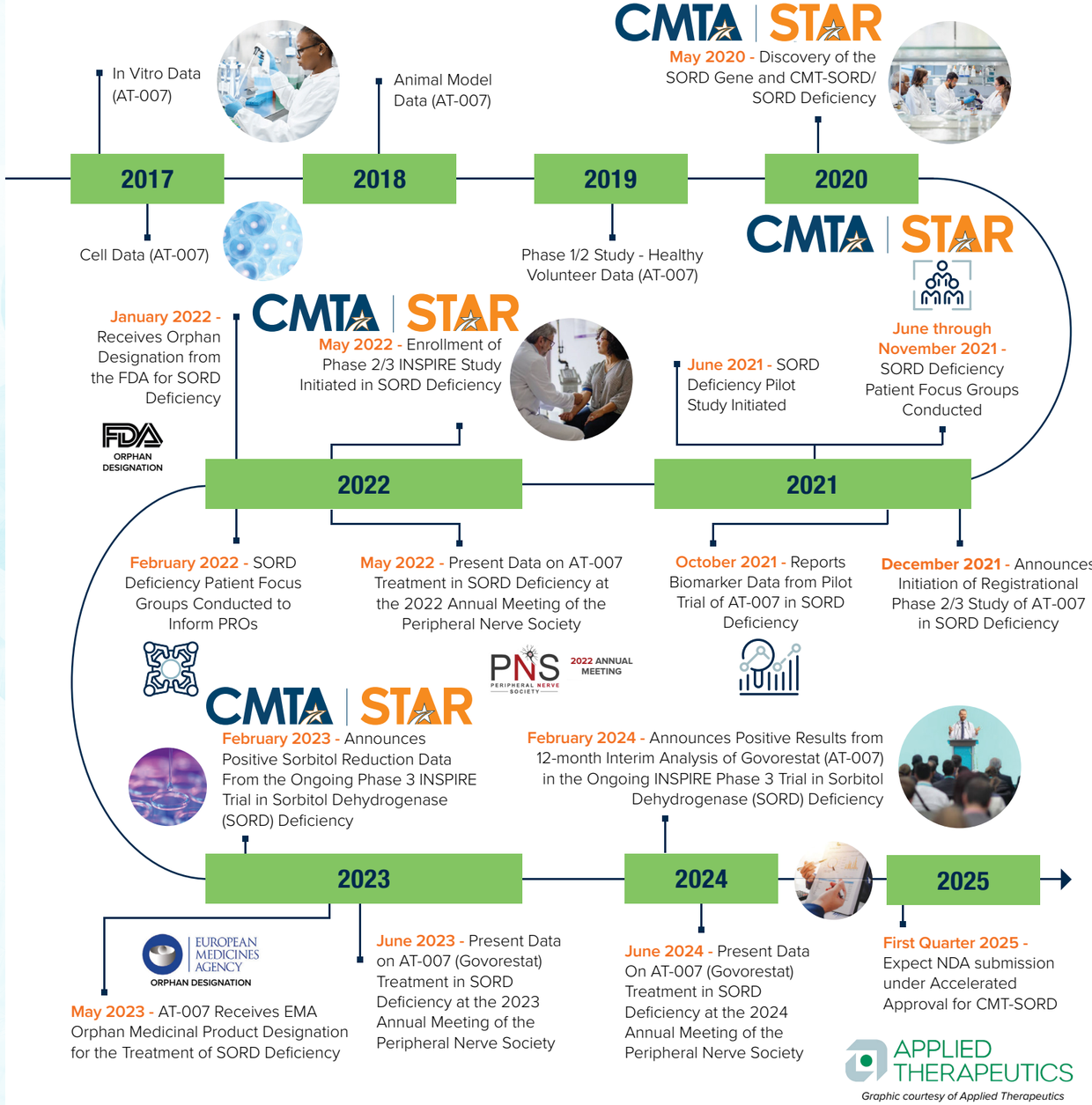
CMT-SORD/SORD DEFICIENCY DEVELOPMENT
TIMELINE SHOWS CMTA-STAR'S IMPACT



Graphic courtesy of Applied Therapeutics

TESTING FOR CMT-SORD

If your healthcare provider has diagnosed you with CMT2, axonal CMT, or distal hereditary motor neuropathy (dHMN, aka HMN): subtype unknown, you might have CMT-SORD. Genetic testing can be performed. However, there is also a specialized urine test your doctor can order.

THE CMT-SORD STORY

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CMTA-STAR initiatives, the ongoing trial released encouraging interim results in February 2023, marking a significant milestone in the quest for treatment.

From Hope to Reality

For the CMT community, every breakthrough offers a ripple of hope. CMT-SORD's record-setting speed of advancement matters not just for those with this type of CMT but for all affected by CMT. Should govorestat receive approval to treat CMT-SORD, it would set a precedent as the first approved treatment for any CMT and pave the way for future treatments for all patients with CMT. Our investments in clinical trial readiness have paid off, and the pathway has been proven and can be used by other treatments in the pipeline.

The CMT-SORD story is a testament to the impact and transformative power of research, advocacy, and community. It shows that with concerted effort and unwavering community support, we can move from discovery to treatment, from hope to reality. The impact of these efforts is an example of how targeted research and strategic partnerships can lead to life-changing outcomes. CMTA's commitment to accelerating research and empowering patients continues to drive us forward, ensuring that each step brings us closer to a future without CMT.

Your Support Makes Progress Possible

None of this would be possible without the support and participation of our dedicated community. The generosity of individuals like you power every discovery, every clinical trial, and every step toward treatment. Please consider donating to CMTA to sustain and accelerate this vital work as we close the year. Together, we can bring hope and real change to every person impacted by CMT.

NEW FACES STRENGTHEN CMTA'S COMMITMENT TO ACCELERATING CMT RESEARCH

We're thrilled to welcome two exceptional individuals to the CMTA research team: Stephen Lin, PhD, our new Director of R&D Alliances, and Cassidy Miller, MHSA, who joins us as Research Project Manager. Their talents and dedication strengthen our mission to accelerate treatments for CMT.

As Director of R&D Alliances, Dr. Lin (Steve) brings a wealth of expertise to CMTA-STAR's Preclinical Toolbox. With his background in managing extensive stem cell collections and preclinical programs, Steve will oversee our invaluable array of animal models and patient-derived stem cell lines, which are critical in advancing potential CMT therapies.

Steve's prior experience spans prominent institutions, including StemCells, Inc., Thermo Fisher Scientific, and the California Institute for Regenerative Medicine (CIRM), where he managed preclinical development programs that drove scientific breakthroughs. Holding a PhD in Biochemistry from Washington University in St. Louis, Steve now calls the San Francisco Bay Area home, where he lives with his wife, twin sons, and daughter.

Cassidy joins us as Research Project Manager, providing crucial support across CMTA's research pillar, including our Strategy To Accelerate Research (STAR) program, STAR Advisory Board, and our Patients as Partners in Research platform. Cassidy, who started her CMTA journey as a volunteer counselor at Camp Footprint, has a special connection to the CMT community. Her dedication and compassion led her to become a long-standing Cabin Captain, where she continues to inspire young campers.

Cassidy holds an MBA in Health Services Administration, with a minor in Nutrition and Dietetics, from Marywood University, and her previous experience includes healthcare coordination and lab administration. Originally from Cleveland, OH, Cassidy now resides in Scranton, PA.

"The evolution of our research program has opened the door to bringing Steve and Cassidy on board," said Katherine Forsey, PhD, CMTA Chief Research Officer. "Steve's expertise in toolbox development and preclinical testing will enhance the accessibility of our partners to explore CMT therapies using these models. Cassidy's organizational skills and passion will be instrumental in managing our growing research portfolio and streamlining day-to-day operations. Their combined expertise and enthusiasm will greatly benefit the CMTA-STAR ecosystem."

FUEL BREAKTHROUGHS IN CMT RESEARCH—

SUPPORT THE STAR FUND

Join us in making a transformative impact for the CMT community!
If you would like to directly support a research initiative with a multi-year donation pledge or appreciated annuities, directing your support to the STAR Fund powers groundbreaking studies, accelerates new treatments, and brings real hope to families affected by CMT.

By supporting CMTA's Strategy To Accelerate Research (STAR), you become part of the driving force behind global collaborations, life-changing therapies, and real progress towards a cure. To explore these impactful giving opportunities, please contact Jeana Sweeney at jeana@cmtausa.org.

Together, let's bring breakthroughs within reach and create a brighter future for all those affected by CMT.

THE POWER OF ONE:
HOW ONE FAMILY'S ACTION IS ACCELERATING CMT4C RESEARCH

Sometimes, all it takes is one person to make a significant difference. Bob Paulsen, a seasoned tech entrepreneur and father, embodies this truth. His journey from a concerned parent to a key supporter of cutting-edge genetic research for CMT4C is a testament to the power of determination and action, in conjunction with the broad network and reach of CMTA. Thanks to his efforts, we are moving closer to finding treatments and cures for CMT, beginning with a newly approved genetic therapy project—"Project Foresee."

THE TURNING POINT: From Scoliosis to a CMT4C Diagnosis

Bob's journey began, like many parents whose child has CMT, with his son Quinn's early health struggles. From the age of three, Quinn's scoliosis and muscle weakness were apparent, but for years, the Paulsens were met with confusion and misdiagnoses. It wasn't until late summer 2023, after comprehensive testing, that they finally received a definitive diagnosis: Charcot-Marie-Tooth disease type 4C.

Though devastating, the diagnosis also gave Bob a sense of direction. With his company recently sold and more time to focus on Quinn's future, Bob turned his attention to finding answers and solutions. This led him to CMTA in the fall of 2023, where he learned about groundbreaking research and opportunities to make a difference for Quinn and others facing CMT.

FUNDING HOPE: Bob Paulsen's Commitment to CMT4C Research

Bob's commitment paved the way for Project Foresee, a project plan developed in collaboration with CMTA-STAR Advisory Board members and CMTA leadership, to fund IND-enabling studies for a potential genetic therapy treatment for CMT4C. This new project, coordinated by CMTA and led by CMTA-STAR Advisory Board Member Steven Gray, PhD, at the University of Texas Southwestern, marks an essential step toward clinical trials, bringing hope to those with CMT4C. These studies were made possible by previous CMTA investments to develop genetic therapy through funded programs in the Cyprus-based laboratory of Kleopas Kleopa, MD, who is also working on this project.

The project team has already completed the first steps, including getting positive feedback from the FDA that the project plan appears sound. The next step for the project will be IND-enabling studies required before testing in humans to evaluate potential risks and help to determine the right dose for human trials. Led by Dr. Steven Gray, these tests mark a significant milestone toward an IND (Investigational New Drug) application, which is a request to the FDA that asks for approval to test a new treatment in people. Ultimately, the project's outcome will be to test the CMT4C genetic therapy in CMT4C patients in a clinical trial under an IND.

The Paulsen Family

Based on this detailed project plan from CMTA, Bob moved quickly as he realized the potential impact he could make. Inspired by the organization's work and determined to accelerate progress, he spearheaded efforts to raise substantial funds for Project

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The Paulsen's "Foresee Gala" raised $250,000 for CMTA-STAR's Project Foresee.

THE POWER OF ONE

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Foresee. In June 2024, the Paulsen family hosted a “Foresee Gala” in his hometown of Denver, Colorado, raising $250,000 to support this CMTA-STAR project directly. More importantly, this event increased awareness of CMT4C through various channels to reach well over 1,000 new supporters.

A Community Effort

Bob’s action demonstrates that one person’s dedication can create a ripple effect. His efforts have rallied a network of donors and the broader CMTA community. By connecting with scientists like CMTA-STAR Advisory Board Chairperson John Svaren, PhD at the University of Wisconsin-Madison, who introduced him to CMTA, and Dr. Gray, Bob is helping to accelerate research and bring much-needed treatments closer to reality.

CMTA CEO Sue Bruhn, PhD, remarked: “I am inspired by Bob’s commitment to advancing genetic therapy for CMT4C. His dedication is helping us take crucial steps towards a treatment that could change the lives of those with CMT4C. This project reflects the power of one person to make a real difference in the fight against CMT.”

Looking Ahead: The Road to a Cure

Quinn’s journey, like that of so many others affected by CMT, is ongoing. But with champions like Bob Paulsen driving progress, hope is on the horizon. His story is a powerful reminder that it only takes one person to create change and bring hope to many others. The money raised at the Foresee Gala is already making an impact, but the work is far from over. Seeing this project through will require a significant fundraising effort and take several years.

As we close out the year, we invite our entire CMTA community to join Bob in supporting Project Foresee, which could transform lives. With your help, we can continue to fund critical projects like the CMT4C genetic therapy study and push forward the treatments that will one day cure CMT. Together, we can make a difference—one person, one project, one donation at a time.

CELEBRATING THE LEGACY OF STEVEN S. SCHERER, MD, PhD:

SCIENTIST, CARE PROVIDER, ADVOCATE, FRIEND

CMTA wishes to honor Steven S. Scherer, MD, PhD (or just “Steve” to all who know him), a long-time member of the CMTA Board of Directors and the Advisory Board for Strategy To Accelerate Research (STAR) who retired from Penn on July 1st. While he may be stepping away from patient care, Dr. Scherer will remain deeply involved in CMTA, and will continue the clinical research to realize the goal of finding treatments for CMT. His contributions have had a lasting impact on CMT.

To date, Dr. Scherer has authored more than 190 original scientific papers and 73 reviews, many of which have been instrumental in advancing our understanding of CMT. His groundbreaking discovery of the first X-linked gene in CMT—the CONNEXIN 32 gene (aka GJB1)—paved the way for subsequent studies in CMT genetics on how gene mutations lead to various forms of CMT. In addition to leading the work from his own laboratory, Dr. Scherer has worked collaboratively with many colleagues, including many of whom are well-known to the CMTA community (Drs. Charles Abrams, David Herrmann, Kleopas Kleopa, Davide Pareyson, Mary Reilly, Michael Shy, John Svaren, and Stephan Züchner) to advance the diagnostics and potential therapeutic avenues of CMT, thereby influencing the direction of research for decades. Dr. Scherer wishes to thank CMTA, which has supported many of these efforts.

In addition to his research, Dr. Scherer has also performed service to a variety of organizations. In particular, he has held numerous positions in the Peripheral Nerve Society, including serving as president from 2017 to 2019,

Dr. Steven S. Scherer

and was the first-ever recipient of the Alan J. Gebhart Prize for Excellence in Peripheral Nerve Research.

Roots in Michigan and Beyond

Dr. Scherer’s passion for science and medicine has deep roots. His father was a science teacher, and his mother was a nurse. He grew up in Milford, Michigan, and attended the University of Michigan, where his journey into the world of neurology began. He was strongly influenced by Dr. Stephen Easter, who later became his PhD thesis advisor and lifelong friend. As anyone who knows him will confirm, Dr. Scherer and his wife, DeAnn, remain dedicated to the Wolverines. He proudly wears maize and blue, and seldom misses a chance to cheer on his team.

As he transitions from his clinical duties, Dr. Scherer is looking forward to spending more time with his family and having adventures with DeAnn. They recently spent a month in the Rocky mountains, a well-deserved opportunity to recharge.

A Lifelong Commitment to CMT

Reflecting on his impact, Jeana Sweeney, CMTA’s Chief Engagement and Gift Officer, notes, “Dr. Steve Scherer transformed patient care into something extraordinary. Having known Steve for over twenty years, I have had the privilege of seeing firsthand—and hearing directly from patients—their immense respect and admiration for him.”

For Jeana, Dr. Scherer’s influence reaches beyond his clinical skills. “I deeply respect him for the exceptional care he provides to those living with CMT and his unwavering dedication to CMTA. His heartfelt commitment has left an enduring impact on our entire community, and his legacy will continue to inspire and strengthen us for years to come,” she shared.

A close colleague and friend, Michael Shy, MD, reflects on their early years together. “I first met Steve in

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Progress through Unity: One Community, One Vision

TURN HOPE INTO REALITY – DOUBLE YOUR IMPACT!

ALL GIFTS BEFORE 12/31/24 WILL BE MATCHED, UP TO $500,000. GIVE TODAY!

Imagine a future where CMT is not just treatable but curable. What would that mean for you and your loved ones? For millions living with this disease, it would be life-changing.

Your generosity has already made a significant impact, and thanks to supporters like you, CMTA is making strides towards this future every day. Your contributions help fund groundbreaking research, bringing us closer to a world free from CMT.

As a valued community member, we want to remind you that there's still time to **double your impact!** Your donation today will be matched, amplifying your support for families like Molly's and Zolana's.

Molly's daughter Quinn was diagnosed with CMT1B before her 2nd birthday, and while the diagnosis was scary, CMTA has been a lifeline for their family. Quinn now has access to a supportive community that provides her with encouragement and vital resources. At age 7, she's made incredible progress, parking her walker and running around with her friends for the first time. With your help, we can continue to fight for a future where Quinn and others like her no longer need walkers or AFOs.

Zolana, diagnosed at age 7, dreams of becoming an artist. Although she worries that CMT may cause setbacks, she finds hope in CMTA's Strategy To Accelerate Research (STAR) and is determined to live her life without limitations. Together with her sister and cousin, she even published a cookbook to raise funds for CMT patients, inspiring others to think creatively about how they can make a more significant impact.

Thanks to the generosity of people like you, Quinn, Zolana, and countless others find hope and are empowered by CMTA's fight for a brighter future. **Together, we can turn hope into reality.**

Make your year-end gift today, and remember, your donation will be **matched** to make an even greater difference for those living with CMT.

YES—I WANT HELP CURE CMT! HERE IS MY DONATION OF:

◯ $50 ***becomes $100*** ◯ $100 → ***becomes $200*** ◯ $250 → ***becomes $500*** ◯ $500 → ***becomes $1000*** ◯ $1000 → ***becomes $2000*** ◯ Other: $_____

◯ Please charge my credit card. ◯ Visa ◯ MasterCard ◯ American Express ◯ Make my gift a recurring Monthly Donation.

◯ Check enclosed, payable to the Charcot-Marie-Tooth Association.

◯ I am interested in learning more on how to make a major gift or a multi-year pledge. ◯ I would like information on donating securities.

Name_____ Email _____

Card #_____ Exp. Date_____ CVV#_____

Signature_____ Billing Address_____

City_____ State_____ Zip_____ Phone_____

Dedicate this gift to: _____ *Honoree's Email:*_____

The Charcot-Marie-Tooth Association (CMTA) is a registered 501(c)(3) nonprofit organization.
Your donation is tax-deductible to the fullest extent allowed by law. Tax ID: 22-2480896.

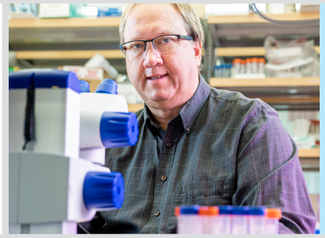
Use the enclosed envelope to mail this form to: CMTA, PO Box 105, Glenolden, PA 19036

DONATE ONLINE AT: www.cmtausa.org/curecmt

EMPOWERING PROGRESS: HOW YOUR DONATION DRIVES CMTA'S MISSION

$10
Delivers a "Box of Sunshine" to newly diagnosed youth.

$30
Delivers new patient kits to a CMTA Center of Excellence.



$50
Funds foundational research needed to tell if a new treatment is working in a clinical trial.

$100
Supports the next generation of cutting-edge researchers.

$200
Adds new models to the CMTA's Preclinical Toolbox needed for CMT drug development.

$500
Funds AAV genetic therapy research.

$750
Funds one camper to have a life-changing week at Camp Footprint.

CMTA
Charcot-Marie-Tooth Association

DONATE ONLINE AT: www.cmtausa.org/curecmt

$1,000
Funds CRISPR genetic therapy research.

$2,500
Supports nanoparticle genetic therapy research.

$5,000
Supports research into new outcome measures that accelerate clinical trials.

$10,000
Supports a foundational Natural History study that paves the way for a clinical trial.

$20,000
Funds new iPSC stem cells that biotech companies can use to test new therapies/treatments.

$50,000
Drives a clinical trial to become a treatment that's available to patients.

Donations are tax-deductible to the fullest extent allowable by law.
Figures are estimates only. This list is not intended to be comprehensive.

STEVEN SCHERER

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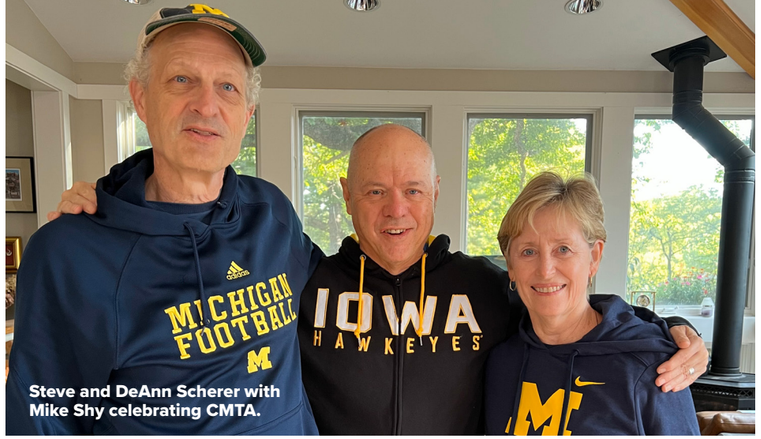
the late 1980s when we were both studying the molecular biology of PNS myelination in the lab of John Kamholz. In retrospect, this lab was one of the great incubators for CMT research over the next 30-40 years, as fellow young investigators also included Larry Wrabetz and Laura Feltri. There is really no one like Steve. His love and knowledge of peripheral nerve, its morphology, and function are unsurpassed."

He continues, "Steve would let you know if he didn't believe your science or if your data was unconvincing. However, he would do anything for you as a friend, and you knew he was always there for you in your corner. This even included trying to help me improve my writing style over the years. There are only a few dear friends one gets to have during life. I am proud to say that Steve was and is one of mine."

As Jeana and Dr. Shy highlight, Dr. Scherer's impact extends far beyond his research papers. To the CMT community, he is a compassionate advocate and an irreplaceable presence whose expertise and dedication have touched countless lives. Whether through his clinical work, his leadership within CMTA, or his ongoing research, Dr. Scherer's contributions will continue to impact his beloved CMT community.



Steve and DeAnn Scherer with Mike Shy celebrating CMTA.

A Legacy of Dedication and Impact

As Dr. Scherer steps back from his clinical role, his presence will be missed by countless patients, colleagues, and friends touched by his work, compassion, and dedication. Though retiring from patient care, he will remain a guiding force as a CMTA-STAR Advisory Board Member and as a member of CMTA's Board of Directors. His contributions to CMT research and patient care have set a standard that will guide the field for years. We are grateful for all he has given to the CMT community, and his unwavering commitment inspires us. Thank you, Dr. Scherer, for a lifetime of dedication—we look forward to seeing your legacy continue to shape the future of CMT.

CMTA BRANCHES

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

ALABAMA
Northern Alabama
Kimberly Parry
757-235-6260

ARIZONA
Phoenix Area
Christina Fisher
623-742-8921

ARKANSAS
Central Arkansas Area
Becky Bandy
832-689-1089

CALIFORNIA
Antelope Valley Area
Donna Murphy
661-317-6332
Danielle Metzger
661-317-6533
Los Angeles Area
Alani Price
310-710-2376
John Ramos
951-318-5669
Cheyenne Alfino
747-232-4604
Orange County Area
Beth Dorin
949-929-2908
San Diego Area
Kendall Trout
760-632-5654
San Fran/Bay Area
Lisa Weiner
415-994-3744

COLORADO
Denver Area
Ron Plageman
303-929-9647

CONNECTICUT
Hartford
Roy Behlke
239-682-6785
North Haven
Lynne Krupa
203-288-6673

DISTRICT OF COLUMBIA
Washington, DC
Steven Weiss
Kimberly Hughes
301-962-8885

FLORIDA
Destin Area
Ted Spring
850-368-1097
Jacksonville Area
Tim Nightingale
904-504-1953
Stephanie Burkhalter
904-710-3771
Miami Area
Norma Levy
Anne Katz
Jessica Villalon
miamifloridacmtabranch@cmtausa.org
Naples
Roy Behlke
239-682-6785
Palm Beach County
Ilene Meckley
palmbeachcmtabranch@cmtausa.org
Sarasota Area
Rachel Rivlin
941-284-0766
Tampa Bay Area
Edward Linde
813-712-4101

GEORGIA
Atlanta Area
Jeannie Zibrida
404-307-6519

HAWAII
Honolulu Area
Monica Rocabado
831-207-5050

IOWA
Iowa City Area
Jeffrey Megown
319-981-0171

ILLINOIS
Chicago Area
Doreen Pomykala
815-351-1328

INDIANA
Fort Wayne Area
Aimee Trammell
574-304-0968
Northwestern Indiana
Ashley Pomykala
815-341-6931

KANSAS
Kansas City Area
Tammy Adkins
314-608-6889
Aron Taylor
913-744-5674

KENTUCKY
Southeastern Kentucky
Chloe Shaffer
southeasternkentucky-cmtabranch@cmtausa.org

MAINE
Portland Area
Branch Leader Needed
Call 800.606.2682

MARYLAND
Baltimore
Sarah Wilson
301-615-9589

MASSACHUSETTS
Boston
Jill Ricci
978-887-1014
Vittorio Ricci
978-476-5369

MICHIGAN
Central Michigan Area
Megan Berger
517-256-5854
Jonah Berger
303-827-4218

MINNESOTA
Minneapolis Branch
Lynn Anne Groebner
952-393-3188
Angela Christensen
612-695-3864

MISSOURI
Kansas City Area
Tammy Adkins
314-608-6889
Aron Taylor
913-744-5674
St. Louis Area
Payton Rule
618-401-4822
Amanda Rule
618-698-3039

NORTH CAROLINA
Charlotte Area
Mona Doshi
980-339-8560
Scott Roehrig
980-297-1449
Durham Area
Jeanne Boehlecke
919-942-7909
Wilmington Area
Laurel Richardson
910-515-8488

NEBRASKA
Lincoln Area
Brandon Lederer
402-680-0502

NEW JERSEY
Central New Jersey
Mark Willis
732-252-8299

NEW MEXICO
Albuquerque Area
Gary Shepherd
505-296-1238

NEW YORK
Buffalo Area
Maryann Ciskal
716-435-3899
Long Island
Alessandro Cacciani
772-882-8319
Syracuse Area
Michael Casey
315-898-6819
Westchester Area
Beverly Wurzel
Frank Wurzel
201-224-5795

OHIO
Cincinnati Area
Josh Marsh
330-268-6436
Cleveland Area
Jessica Diamond
216-570-6432
Columbus Area
Angela McCabe
937-831-5968

OREGON
Portland
Rachel Tessier
portlandocmtabranch@cmtausa.org

PENNSYLVANIA
Bucks County Area
Julie FitzGerald Schell
315-573-3919
Chester County
Carol Aruffo
610-405-9291
Harrisburg
Erin Weierbach
717-379-7504
Northwestern Area
Joyce Steinkamp
814-833-8495
Pittsburgh
Erin Wood
pittsburghcmtabranch@cmtausa.org

SOUTH CAROLINA
Columbia Area
Zack Boyd
803-622-6565
Kyle Bryant
803-378-6202
Greenville Area
Rebecca Lauriault
864-918-2437

TENNESSEE
Central Tennessee Area
Brittney Grabiel
423-213-2336

TEXAS
Austin Area
Nate Halk
512-415-6097
Dallas/Fort Worth
Stephanie Jackson
dallascmtabranch@cmtausa.org
Houston Area
Tami Delmark
houstoncmtabranch@cmtausa.org

UTAH
Orem Area
Melissa Arakaki
801-494-3658

VIRGINIA
Southwestern Virginia
Karen Brown
540-558-5043
Central Virgina
Karen Dyer-Smith
434-882-7030

WASHINGTON
Seattle Area
Denise Snow
206-321-1261
Emily Osborne
425-220-4225

WISCONSIN
Madison Area
Debi Weber
608-712-8709
Manitowoc Area
Barry Hett
920-388-9992

CANADA
Western Canada
Victoria Berezovich
westerncanadacmta-branch@cmtausa.org
Toronto Area
Branch Leader Needed
Call 800.606.2682

MEXICO
Guadalajara, Jalisco Area
Tomas Luis Lopez Valenzuela
+52 1 33-18-28-17-07
Guadalupe Valenzuela Cazares
+52 1 33-17-94-53-21

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.

ARKANSAS

LITTLE ROCK (PEDIATRIC)
Arkansas Children's Hospital
Clinical Director: Aravindhan Veerapandiyan, MD
Appts: 501-364-1850

CALIFORNIA

LOS ANGELES (ADULT & PEDIATRIC)
Cedars-Sinai Medical Center
Clinical Directors: Richard A. Lewis, MD, and Melody Badii, MD
Appts: 310-423-4268

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Stanford Children's Health*
Clinical Directors: John Day, MD, PhD, and Ana Tesi Rocha, MD
Appts: 650-723-0993

PALO ALTO (ADULT)
Stanford Neuromuscular Program*
Clinical Director: John Day, MD, PhD
Appts: 650-723-6469

SAN FRANCISCO (ADULT)
University of California, San Francisco
Clinical Director: Mark Terrelonge, MD
Appts: 415-353-2273

SAN FRANCISCO (PEDIATRIC)
University of California, San Francisco
Clinical Director: Alex Fay, MD, PhD
Appts: 415-353-7596

COLORADO

AURORA (ADULT)
University of Colorado*
Clinical Director: Vera Fridman, MD
Appts: 720-848-2080

AURORA (PEDIATRIC)
Children's Hospital Colorado
Clinical Director: Michele Yang, MD
Appts: Alison Ballard, 720-777-3907

CONNECTICUT

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Connecticut Children's Medical Center, Farmington*
Clinical Director: Gyula Acsadi, MD, PhD
Appts: Nanci Stolgitis, RN, 860-837-7500

NEW BRITAIN (ADULT & PEDIATRIC)
Hospital for Special Care
Clinical Director: Kevin J. Felice, DO
Appts: Sharon McDermott, 860-612-6305

NEW HAVEN (ADULT)
Yale School of Medicine
Clinical Director: Sasha Zivkovic, MD
Appts: 203-785-4085

FLORIDA

ORLANDO (PEDIATRIC)
Nemours Children's Hospital*
Clinical Director: Omer Abdul Hamid, MD
Appts: 407-650-7715

GAINESVILLE (ADULT & PEDIATRIC)
University of Florida
Clinical Director: James Wymer, MD, PhD
Appts: 352-294-5400

JACKSONVILLE (ADULT)
Mayo Clinic Florida
Clinical Director: Christopher Lamb, MD
Appts: 904-953-0853

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MIAMI (ADULT)
University of Miami*
Clinical Director: Mario Saporta, MD
Appts: 305-243-9173

MIAMI (PEDIATRIC)
Nicklaus Children's Hospital
Clinical Director: Migvis Monduy, MD
Appts: 786-624-2154

ILLINOIS

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Northwestern Memorial Hospital
Clinical Director: Daniela Maria Menichella, MD, PhD
Appts: 312-695-7950

University of Illinois at Chicago
Clinical Director: Charles K. Abrams, MD, PhD
Appts: 312-996-4780

Rush University
Clinical Director: Ryan D. Jacobson, MD
Appts: Sherri Taylor-Kennedy, 312-942-4500

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HSHS St. Elizabeth's Hospital
Clinical Director: Raghav Govindarajan, MD
Appts: 618-641-5803

IOWA

IOWA CITY (ADULT & PEDIATRIC)
University of Iowa Hospitals & Clinics*
Clinical Director: Michael E. Shy, MD
Appts: 319-384-6362

KENTUCKY

LOUISVILLE (ADULT)
University of Louisville
Clinical Director: Zeng Y. Wang, MD, PhD
Appts: Anson Ashburn, 502-588-4800

MARYLAND

BALTIMORE (ADULT & PEDIATRIC)
Johns Hopkins University*
Clinical Directors: Charlotte Sumner, MD, and Bipasha Mukherjee-Clavin, MD, PhD
Appts: (Adult) 410-614-1196, (Pediatric) 410-955-4259

MASSACHUSETTS

BOSTON (ADULT & PEDIATRIC)
Massachusetts General Hospital*
Clinical Director: Reza Seyedsadjadi, MD
Appts: Tamika Scott, 617-726-3642

MICHIGAN

ANN ARBOR (ADULT & PEDIATRIC)
University of Michigan
Clinical Co-Directors: Gary Gallagher, MD, and Brett McRay, MD
Appts: 734-936-9020

MINNESOTA

MAPLE GROVE (ADULT)
University of Minnesota*
Clinical Director: David Walk, MD
Appts: 763-898-1000

MISSOURI

ST. LOUIS (ADULT & PEDIATRIC)
Washington University School of Medicine
Clinical Director: Stefanie Geisler, MD
Appts: 314-362-6981

NEBRASKA

OMAHA (ADULT)
Chi Health Clinic Neurology Institute
Clinical Director: Yu-Ting Chen, MD
Appts: 402-717-0070

NEW JERSEY

HACKENSACK (ADULT & PEDIATRIC)
Hackensack University Medical Center
Clinical Director: Florian Thomas, MD, PhD
Appts: Annerys Santos, 551-996-1324

NEW YORK

ROCHESTER (ADULT & PEDIATRIC)
University of Rochester*
Clinical Director: David Herrmann, MD
Appts: 585-275-2559

NORTH CAROLINA

CHAPEL HILL (ADULT & PEDIATRIC)
University of North Carolina*
Clinical Director: Rebecca Traub, MD
Appts: 984-974-4401

CHARLOTTE (ADULT & PEDIATRIC)
Atrium Health Neurosciences Institute
Clinical Director: Urvi Desai, MD
Appts: 704-446-1900

OHIO

COLUMBUS (ADULT & PEDIATRIC)
Nationwide Children's Hospital
Clinical Director: Zarife Sahenk, MD, PhD
Appts: 614-722-2203

COLUMBUS (ADULT & PEDIATRIC)
Ohio State University, Wexner Medical Center*
Clinical Director: Michael Isfort, MD
Appts: 614-293-4969

PENNSYLVANIA

HERSHEY (PEDIATRIC)
Penn State Health Hershey Medical Center
Clinical Director: Ashutosh Kumar, MD
Appts: 717-531-2908

PHILADELPHIA (ADULT)
Hospital of the University of Pennsylvania*
Clinical Director: Steven Scherer, MD, PhD and Chafic Karam, MD
Scheduling for Clinic Visits: Kari Roberts, Kari.Roberts@pennmedicine.upenn.edu
Scheduling for Research Visits: Pooja Patel, 215-898-0180

PHILADELPHIA (PEDIATRIC)
Hospital of the University of Pennsylvania*
Clinical Directors: Sabrina Yum, MD
Appts: Hannah Borger, 215-590-1719

PITTSBURGH (PEDIATRIC)
Children's Hospital of Pittsburgh
Clinical Director: Hodas Abdel-Hamid, MD
Appts: 412-692-6106

TEXAS

AUSTIN (ADULT)
Austin Neuromuscular Center
Clinical Director: Yessar Hussain, MD
Appts: 512-920-0140

DALLAS (PEDIATRIC)
University of Texas Southwestern
Clinical Director: Kaitlin Batley, MD
Appts: 214-456-2768

BEDFORD (ADULT 16+)
Kane Hall Barry Neurology
Clinical Director: Sharique Ansari, MD, MPH
Appts: 817-267-6290, option 4

HOUSTON (ADULT)
Baylor College of Medicine
Clinical Director: Thomas Lloyd, MD
Appts: 713-798-2273

HOUSTON (ADULT)
Houston Methodist Hospital
Clinical Director: Jun Li, MD
Appts: 713-441-3763

UTAH

SALT LAKE CITY (ADULT & PEDIATRIC)
University of Utah*
Clinical Director: Russell Butterfield, MD, PhD
Appts: 801-585-7575

SALT LAKE CITY (PEDIATRIC)
Primary Children's Hospital
Clinical Director: Russell Butterfield, MD, PhD
Appts: 801-213-7756

SALT LAKE CITY (PEDIATRIC)
Shriner's Hospital
Clinical Director: Russell Butterfield, MD, PhD
Appts: 801-536-3564

WASHINGTON

SEATTLE (ADULT)
University of Washington Medical Center*
Clinical Director: Michael Weiss, MD
Appts: Gail Schessler, 206-598-7688

SEATTLE (ADULT)
Swedish Neuroscience Institute
Clinical Director: Christyn Edmundson, MD
Appts: 206-320-3494

SEATTLE (PEDIATRIC)
Seattle Children's Hospital
Clinical Director: Seth Perlman, MD
Appts: Kara Smith, BSN, 206-987-6678

WASHINGTON, DC

(ADULT)
Medstar Georgetown University
Clinical Director: Nicholas Streicher, MD
Appts: 202-444-1774

(PEDIATRIC TO AGE 21)
Children's National Hospital
Clinical Director: Diana Bharucha-Goebel, MD
Appts: Kathleen Smart, 202-476-6193

INTERNATIONAL LOCATIONS:

AUSTRALIA

WESTMEAD (PEDIATRIC)
The Children's Hospital at Westmead*
Clinical Director: Manoj Menezes, MD
Appts: (02) 98451325
daralyn.hodgson@health.nsw.gov.au

BELGIUM

B-2650 EDEGEM (ADULT & PEDIATRIC)
Antwerp University Hospital
Clinical Director: Prof. Dr. Peter De Jonghe
Appts: +32 3 821 34 23
Neuromusculaire@uza.be

UNITED KINGDOM

LONDON (ADULT)
University College London Hospitals*
Clinical Director: Mary M. Reilly, MD
Appts: Mariola Skorupinska, (0044)2034488019
mariola.skorupinska@uclh.nhs.uk
St. George's University Hospital
Clinical Directors: Niranjanan Nirmalananthan, PhD, and Emma Matthews, PhD
Email: nervemuscle@stgeorges.nhs.uk

ITALY

MILAN (ADULT & PEDIATRIC)
C. Besta Neurological Institute*
Clinical Director: Davide Pareyson, MD
Appts: +39-02-70631911
sara.nuzzo@istituto-besta.it

THAILAND

BANGKOK
Siriraj Hospital
Clinical Director: Oranee Sanmaneechai, MD
Appts: +66-24192660

Email CMTA at info@cmtausa.org

Charcot-Marie-Tooth Association

P.O. Box 105
Glenolden, PA 19036
1-800-606-CMTA (2682)
FAX (610) 499-9267

cmtausa.org

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 nearly $30 million since 2008.