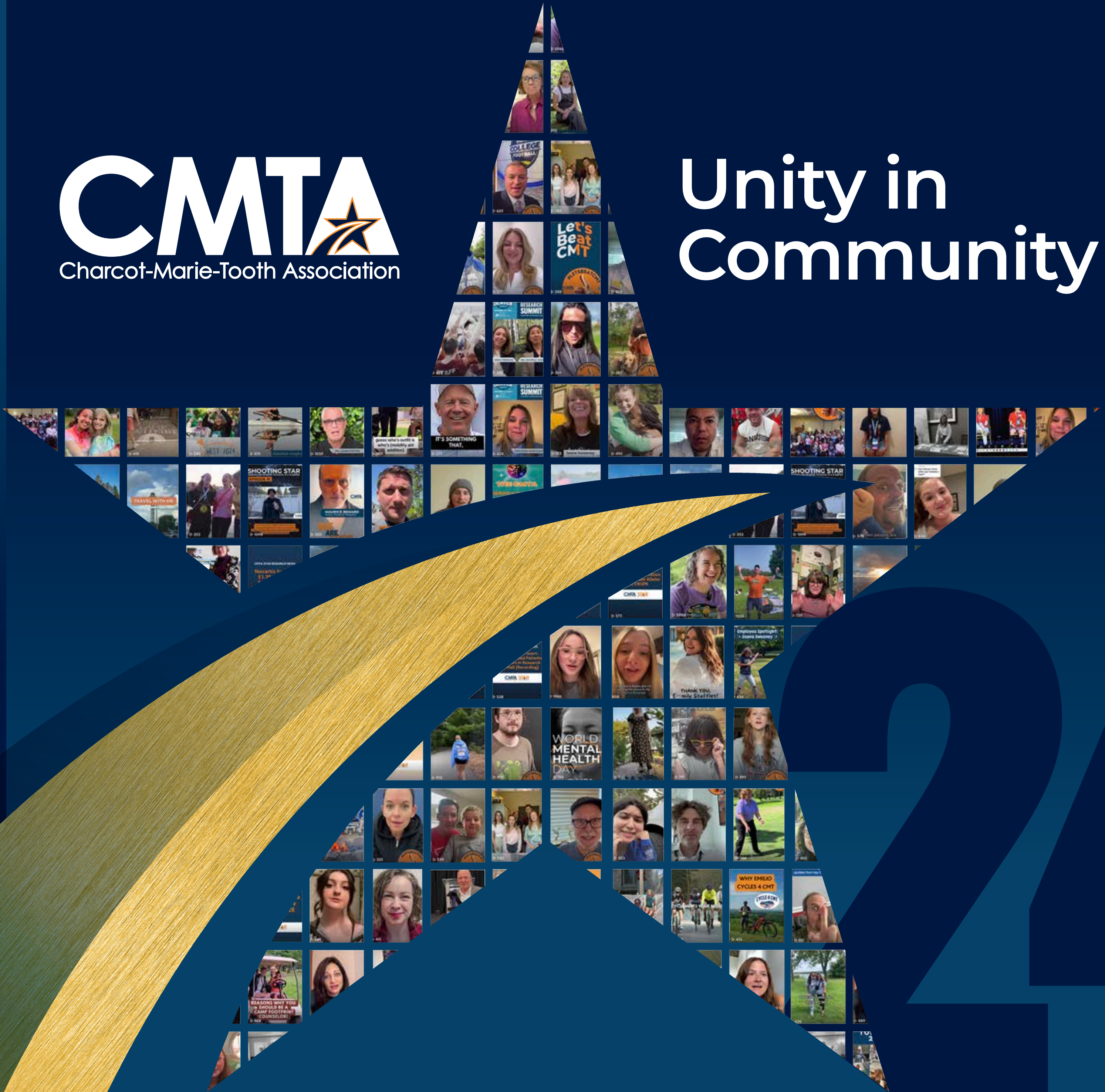
2024 YEAR IN REVIEW

CMTA
Charcot-Marie-Tooth Association

Unity in Community



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CMTA Chief Executive Officer's Message

Big Changes, Bigger Impact

Dear CMTA Family,

Thank you for all your incredible support throughout 2024 where we saw big changes, and we made an even bigger impact.

One transformational change this past year was 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.

Since 2008, CMTA has invested nearly $30 million towards accelerating CMT research. 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.

The power of connection and community was on full display at the **2024 CMTA Patient & Research Summit** in Denver, Colorado. With more than 450 attendees, both in-person and virtual, the CMTA Summit provided updates on cutting-edge research that benefits everybody living with CMT and offered a space for members of our community to come together and build lasting relationships.

Camp Footprint continues to be a transformative experience for young people with CMT. This summer, campers embraced leadership roles, formed deep friendships, and found strength in being their true selves, surrounded by others who understand their journey.

At CMTA, we remain focused on accelerating research, empowering patients, and fostering the next generation — whether young campers making lifelong connections, young adults stepping into community leadership through our **COMPASS Young Adult Program,** or pioneering scientists driving treatments for CMT.

In 2024, we welcomed five new CMTA Centers of Excellence to our global network of patient-focused, multi-disciplinary CMT clinics staffed by world-renowned expert CMT physicians, clinicians, and researchers.

We also continue to evolve as an organization where this year we welcomed new professionals who bring a wealth of experience, and a shared commitment to accelerating our mission.

Looking ahead to 2025, there is a wealth of exciting opportunities for our community to come together, to learn, and to connect with one another. Our calendar is brimming with engaging community events, from quarterly informative ***Lunch and Learn*** webinars, to local Branch meetings, to the largest global gathering of CMT community members: our flagship **Patient and Research Summit in Indianapolis this September.** These community events serve as vital forums for education, advocacy, and support; and I invite each and every one of you to participate and make your voices heard, whether in person or virtually.

Thank you for your continued support!

Sue Bruhn, PhD
Chief Executive Officer
Charcot-Marie-Tooth Association

CMTA and INC Forge Groundbreaking Alliance to Accelerate Comprehensive Research Across All CMT Types

The **Charcot-Marie-Tooth Association (CMTA)** announced a landmark alliance with the **Inherited Neuropathy Consortium (INC). With a $1.2 million annual funding commitment, this partnership positions CMTA as the partner of choice for CMT research and for pharmaceutical companies involved in drug development and clinical trials.**

CMTA and INC are accelerating progress from the lab to the clinic, advancing trials and treatments that offer hope to the CMT community.

This strategic alliance strengthens CMTA's longstanding support for INC, rooted in a shared dedication to advancing CMT research on the path to a cure. With NIH funding for INC concluding in 2024 after 15 years due to an NIH rule, CMTA's investment will support biomarkers, clinical outcomes, natural history studies, and other critical tools for clinical trial readiness. This funding also enables genetic testing, new gene discovery, and the training of future CMT researchers, creating tools that pharmaceutical partners rely on to bring innovative medicines to market.

A Commitment to Every Person with CMT

CMTA's mission is to improve the lives of all those affected by CMT. By uniting the patient community, researchers, clinicians, and drug developers, the CMTA-INC alliance accelerates progress toward effective treatments. **CMTA CEO Sue Bruhn, PhD,** noted: "Sustaining INC's work through the new CMTA-INC strategic alliance means we can address the needs of every individual affected by CMT, regardless of type. This transformational partnership represents a new chapter for the CMT community, offering real help today and genuine hope for tomorrow. We are proud to play this role in shaping a new era of CMT research."

CMTA CEO, Sue Bruhn, PhD

Michael Shy, MD, Principal Investigator of INC and a pre-eminent leader in the CMT clinical research space, shared his vision: "INC's journey has always been about empowering patients through research and clinical innovation. This new alliance provides our work with a sustainable path forward, ensuring that each of our over 20 international centers can continue the vital research that brings us closer to transformative treatments and a world without CMT. This partnership bridges an essential funding gap and opens new collaboration and scientific discovery opportunities. Together, we're building a future that changes the trajectory of CMT research and patient care."

Michael Shy, MD, Principal Investigator of INC

A New Era for CMT Research

The CMTA-INC strategic alliance sets a bold precedent in CMT research, drug development, and patient care. **Gilles Bouchard, Chairman of the Board of Directors for CMTA,** 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."

CMTA Chairman of the Board, Gilles Bouchard

About the Inherited Neuropathy Consortium (INC)

INC is a collaborative network of CMT investigators from multiple academic medical centers who currently follow over 8,300 individuals with CMT. Since its inception in 2009, INC has published over 300 manuscripts defining the natural history of multiple CMT subtypes and developing the clinical outcome measures and biomarkers that allow investigators to perform these natural history studies and make the subtypes **"Clinical Trial Ready."** INC has identified thirty new genes that cause CMT to date and trained more than twenty outstanding young investigators devoting their careers to CMT research.

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**

CMT-SORD

Sorbitol Dehydrogenase Deficiency

Supported by CMTA, **Applied Therapeutics** requested accelerated approval of its CMT-SORD drug, govorestat. 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).

Vittorio Ricci

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 the meeting. The petition also urged the FDA to accelerate approval of govorestat 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 via email and across social media channels. Actors **Maurice Benard** and his son **Joshua** of the daytime drama *General Hospital* also lent support on social media.

In less than 4 hours, the petition had more than 1,000 signatures. With six days to go, Vittorio had a revised goal: 3,300 signatures — one signature for each person in the U.S. who has CMT-SORD. The CMT community didn't just respond — it 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.

FDA, Prioritize Patient Input in Review of Potential First Treatment for CMT-SORD

4,823

verified signatures

Sign this petition

The Issue

We, the undersigned, appeal to the U.S. Food and Drug Administration (FDA) to integrate the perspectives of individuals living with CMT-SORD, along with their caregivers, in the evaluation of govorestat (AT-007), a promising candidate for the first-ever treatment of this rare genetic condition. CMT-SORD, also known as SORD Neuropathy or SORD Deficiency, affects 3,300 people in the US and there are currently no treatments.

Our message to the FDA is clear:

CMT-SORD is a debilitating axonal neuropathy, and the diagnosis is crushing. CMT-SORD demands effective treatment. CMT-SORD represents a significant unmet medical need, causing profound suffering and disability for those afflicted and their families. The lack of approved treatments exacerbates the challenges faced by patients living with this rare neuromuscular disease, leaving them with limited options for managing symptoms and no options for slowing disease progression. Patients with CMT-SORD begin to show symptoms of difficulty walking in the teenage years and continue to lose their lower limb function over the next two decades of life

Maurice Benard who portrays Sonny Corinthos on ABC's General Hospital — along with his son, actor-musician, Joshua — supported CMTA's initiatives through social media.

Structure of the SORD protein.

What Is CMT-SORD?

CMT-SORD, also known as **Sorbitol Dehydrogenase Deficiency,** is a unique type of CMT. Identified in May 2020 with funding from CMTA, this type of CMT is caused by mutations in the SORD gene. These mutations result in the build-up of sorbitol, a sugar that becomes toxic to nerves when it accumulates in the body. This buildup leads to symptoms commonly associated with CMT, such as muscle weakness, sensory loss, and challenges with mobility, balance, hand use, and more.

CMT-SORD is the most commonly occurring autosomal recessive axonal type of CMT and affects approximately 3,300 people in the US and 60,000 worldwide.

Camp Footprint

Celebrating its ninth year as the only sleep-away camp in the U.S. exclusively for kids with CMT, **Camp Footprint** offers more than just a week of fun— it's an inspirational experience where campers can be their authentic selves, often for the first time. CMTA National Youth Programs Manager and Camp Footprint Director **Jonah Berger** captured the impact perfectly: "I watch firsthand every year how being around those who understand CMT has an unbelievable impact on the campers and staff that can only be defined as life-changing. The inclusive environment allows campers to shed the weight of feeling different, replacing it with a deep sense of community."

This year's Olympic-themed events fueled friendly competition and teamwork while fostering unity. The camp's lasting impact extends far beyond the week itself, with many campers continuing the friendships and connections they forged long after the campfires have faded.

Thanks to a grant from **Bass Pro Shops**, Camp Footprint received Wicked Ridge M-370 crossbows with AccuDraw, BOG tripods, and other essential gear, allowing even campers with severe CMT to participate.

The community's dedication, from the donations of Big Shot Targets to the efforts of camp parents who helped deliver the equipment, reflects the spirit of Camp Footprint: ensuring every camper gets to do every activity and that no camper is left out.

Camp Footprint reached new heights in 2024, offering a transformative experience for 152 campers across two coasts — seventy-one campers in the West (Oregon) and eighty-one campers in the East (Pennsylvania). Supported by 135 dedicated counselors and program staff across both Camps, Camp Footprint continued to serve as a life-changing experience for campers and camp staff alike. For many, it was their first time meeting others with CMT, as 30 new campers joined the Camp Footprint family this summer. **Learn more at www.cmtausa.org/campfootprint**

2024 CMTA Patient & Research Summit

Denver, Colorado

The 2024 CMTA Patient and Research Summit in Colorado was a remarkable gathering of more than 450 attendees, both in person and virtually, making it one of the largest CMTA Summits to date. Held from September 6-8 in Denver, the event welcomed participants from across the U.S., six other countries, and virtual attendees from ten countries, with over 60% attending for the first time.

CMTA CEO **Sue Bruhn, PhD,** captured the essence of the weekend: “The CMTA Patient and Research Summit embodied our core focus to provide Help for Today and Hope for Tomorrow. Beyond the practical solutions for managing everyday life with CMT, it created a unique space for attendees to connect, share, and build lasting relationships with fellow community members, exhibitors, and speakers.”

From the moment attendees arrived on Friday and participated in a “dine-around” across Denver’s vibrant restaurant scene, a sense of community was undeniable. For many, this was their first experience meeting others with CMT face-to-face, a powerful reminder that they are not alone in their journey.

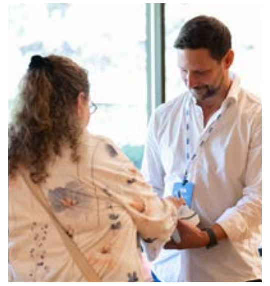
One attendee captured this sentiment perfectly: “I came away with an improved understanding of and hope for current research initiatives, ways to live with CMT, and a profoundly increased feeling of connection to the CMTA Community.”

Saturday’s sessions were packed with educational presentations from CMT specialists, clinicians, and fellow community members. Topics ranged from the latest in symptom management to community perspectives on navigating life with CMT. Sunday’s focus shifted to research, with world-renowned CMT scientists sharing groundbreaking advancements in potential CMT treatments and ongoing projects aimed at finding a cure.

On Sunday, attendees learned about the significant achievements in CMT research through the **CMTA’s Strategy To Accelerate Research (STAR)** program. With more than 50 active research projects, including 35 sponsored initiatives, STAR has invested $30 million to accelerate research in treatments for all forms of CMT. This year alone, nine new projects were approved, targeting both demyelinating and axonal forms of CMT. The collective impact of this research is moving us closer to treatments that will benefit the entire CMT community. One of the standout achievements shared was the rapid progress of CMT-SORD research.

Discovered through CMTA’s ongoing genetic discovery initiative, CMT-SORD advanced from gene discovery to a Phase III clinical trial in just two years, thanks to CMTA’s commitment to ensure the infrastructure was in place to start trials as soon as the science allowed. CMTA-STAR Alliance Partner **Applied Therapeutics,** conducting the trial, reported encouraging 12-month data earlier this year, fostering hope for the entire CMT community.

Breakout sessions provided updates on research for demyelinating CMT, axonal CMT, and undiagnosed forms of CMT (unknown genetic cause), highlighting the expansive reach of CMTA-STAR’s funded research. These sessions reinforced CMTA’s commitment to accelerating treatments that will bring hope to the entire CMT community. Attendees left confident, knowing that every project supported by the CMTA brings us closer to effective treatments for all CMT, not just for the few.



Sunday’s keynote speaker, 26-year-old **Vittorio Ricci,** provided an inspiring example of the power of community and advocacy. Diagnosed with the CMT-SORD in 2021 after years of uncertainty, Vittorio shared his journey from his initial diagnosis at age 14 to receiving genetic confirmation to his involvement in a Phase III clinical trial and advocacy for govorestat, a potential first-ever treatment for CMT. *(See page 4)*

As we look ahead to next year’s Summit, scheduled for September 5-7, 2025, in Indianapolis, Indiana, we remain committed to providing **Help for Today and Hope for Tomorrow.**

A Special Thank You to Our Sponsors and Exhibitors

CMTA would like to extend our heartfelt gratitude to the sponsors and exhibitors of the 2024 Patient & Research Summit.

ACCELERATION SPONSOR
Applied Therapeutics

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ActioBiosciences
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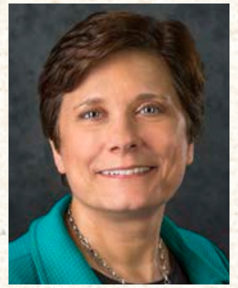
New CMTA Appointments

CMTA Board of Directors

WENDY ARNONE



Wendy is a highly experienced executive with a clinical background, and has held leadership roles within insurance providers including UnitedHealthcare, Excellus BlueCross BlueShield, and HealthNow New York Inc. Wendy most recently served as West Region CEO for UnitedHealthcare's Individual and Employer business, a $10 billion region encompassing thirteen states with over three million members. She brings an incredible wealth of healthcare industry expertise to the CMTA Board of Directors, along with significant leadership experience and financial acumen.

BERNARD COULIE, MD, PhD

Bernard Coulie brings a wealth of experience to the CMTA Board. As the CEO and President of Pliant Therapeutics, a NASDAQ-listed biopharmaceutical company focusing on rare and life-threatening fibrotic diseases, Bernard has dedicated over 20 years to the biopharmaceutical industry in both executive leadership as well as board of director roles. His personal connection to CMT, dating back to 1997 when his son was diagnosed with CMT1A, fuels his commitment to advancing treatments for the disease. Bernard holds an MD and PhD from the University of Leuven, Belgium, and an MBA from the Vlerick Management School, in Leuven, Belgium. He brings invaluable insights as a physician, an executive, and an experienced drug developer.

KEVIN MARKS

Kevin Marks joins CMTA's Board with a distinguished legal background in the drug development space. Currently serving as Chief Legal Officer at Parker Institute for Cancer Immunotherapy (PICI), Kevin oversees legal, intellectual property, and human resources. Prior to PICI, he held leadership positions at the California Institute for Regenerative Medicine (CIRM) and Roche Diagnostics where he served as Vice President and Head of Legal and Compliance. Kevin brings extensive experience in the therapeutics and diagnostics sectors, legal and compliance issues, and sponsored research. He earned his law degree from Villanova University after completing his undergraduate studies at Lafayette College.

PATRICIA VERDUIN, PhD

Patricia Verduin, PhD, is renowned for her combination of technical depth and business acumen. Dr. Verduin boasts a distinguished career dedicated to pioneering product innovations that enhance global health and environmental well-being. With a PhD in Food Chemistry from Rutgers University and an MBA from Fairleigh Dickinson University, her leadership spans prestigious board of director roles in Fortune 500 companies, including FMC Corporation, Ingredion Corporation, and Monsanto, among others. Her tenure as Chief Science Officer at Colgate Palmolive, where she led a global team of scientists, engineers, designers, and regulatory and compliance specialists to develop innovative product solutions, underscores her commitment to driving impactful change, aligning seamlessly with CMTA's mission of improving the lives of all who have CMT.

CMTA-STAR Scientific Advisory Board

CHARLOTTE J. SUMNER, MD

Charlotte J. Sumner, MD, is a Professor of Neurology, Neuroscience and Genetic Medicine at Johns Hopkins University School of Medicine in Baltimore, MD. She is a Daniel Nathans Scientific Innovator and the Vice Chair for Clinical Research in the Department of Neurology. Dr. Sumner is also President of the Peripheral Nerve Society.

New CMTA Staff Appointments

Chris Cosentino
Director of Marketing

Kenny Raymond
Head of Communications

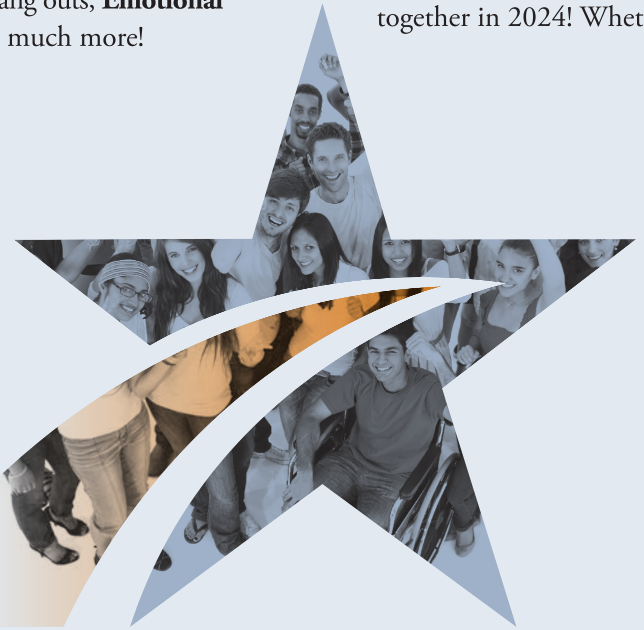
Cassidy Miller, MHSA
Research Project Manager

CMTA COMMUNITY OUTREACH

Building Community — Highlights from 2024

by Laurel Richardson
CMTA Director of Community Outreach

Community is and always has been the heart of CMTA, and 2024 was no exception. We embraced a ***"unity in community"*** theme early in the year and that feeling carried us through twelve months of wonderful connection via **education meetings, support group (branch) gatherings,** two different weeks of **Camp Footprint,** a **Summit** weekend, **COMPASS Young Adults** hang outs, **Emotional Support Group meetings, youth outings,** and much more!



Each year CMTA offers many opportunities to get involved with and get connected to the organization. If learning more about CMT is your thing, one of the ways you can expand your knowledge is by attending an education meeting like the quarterly ***Lunch & Learn* webinars** and by attending the annual **Patient & Research Summit.** This year over 1,000 community members and their families joined us for education meetings virtually and in-person.

New to our programming line-up this year and with the aim of supporting the mental health wellness side of CMT, we hosted monthly (virtual) **Emotional Support Group (ESG) meetings.** These meetings were facilitated by mental health professionals who are part of the CMT community and understand the challenges of living with a progressive condition.

CMTA Branches embodied the 2024 theme of *"unity in community."* With 91 branch leaders (14 new leaders), 65 branches (6 new), and over 105 branch meetings, the branch footprint reaches far and wide. Every branch meeting offers members a unique opportunity to learn about local resources, share struggles and triumphs, hear what is happening at CMTA and most importantly, meet local CMT community members.

Not to be outdone by the adults, the kids had an amazing time getting together in 2024! Whether at **Camp Footprint** or on a **Youth Outing** (held in Dallas, Baltimore and Denver), the kiddos know they never have to feel alone in their CMT journey. When they weren't getting together in person, they were able connect and engage with the youth community via Instagram and TikTok.

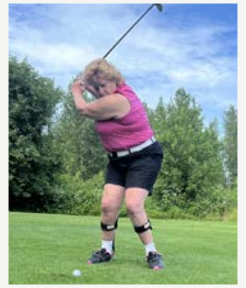
The **COMPASS young adult program** grew continuously throughout the year with opportunities to connect virtually each month on Zoom. There were also a few in-person meet ups with COMPASS members gathering in Los Angeles, Salt Lake City and Denver. The enthusiasm and energy from this group is contagious and we can't wait to watch them grow.

As we reflect on a year full of unity, connection, and education, one of the things we are most proud of is that we awarded the **CMTA Center of Excellence** distinction to five neuro-muscular neurologists. Finding CMT specialists who can provide comprehensive care to the CMT community is one of the most important parts of our mission as an organization. We are proud to add these new specialists to this growing network of clinicians.

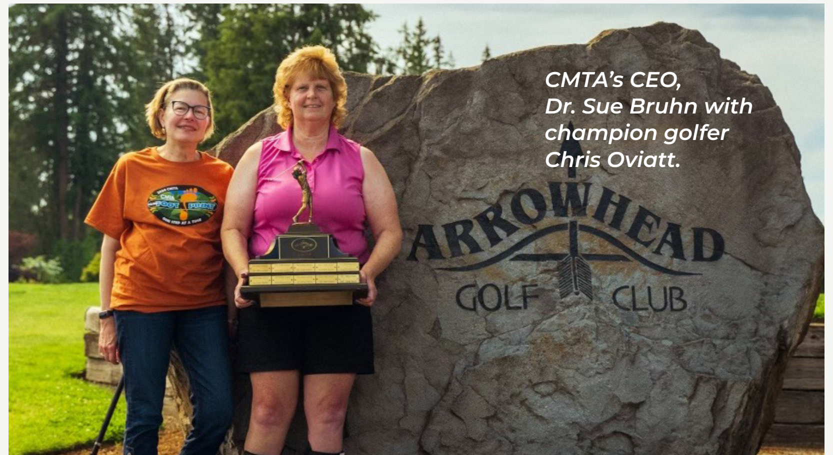
Emotional Support Groups

CMTA hosts monthly **Emotional Support Groups** on Zoom. Each meeting provides support and wellness for the CMT Community to discuss mental health topics related to their experiences with CMT. Register for an upcoming Emotional Support Group by visiting **cmtausa.org/events.**

Golfer Chris Oviatt Shares Her CMT Journey



Chris Oviatt is a champion golfer from Oregon who was diagnosed with Charcot-Marie-Tooth disease (CMT) in 1987. Chris talked with CMTA's CEO, **Dr. Sue Bruhn,** about her journey with CMT and how she never let it stop her from playing the game she loves. Watch her story on CMTA's YouTube Channel: ***Golfer Chris Oviatt's Journey with #CMT***



CMTA's CEO, Dr. Sue Bruhn with champion golfer Chris Oviatt.

Dr. Scherer Retires

CMTA honors **Steven S. Scherer, MD, PhD,** a long-time member of CMTA's Board of Directors and Advisory Board for Strategy To Accelerate Research (STAR), who retired from Penn Medicine in July. 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, where 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.

Dr. Steven Scherer

COMPASS Young Adult Program

COMPASS, CMTA's young adult program, 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 that go beyond a diagnosis. These meetings aren't just about finding support — they're about finding strength in solidarity and shared experience.

COMPASS isn't just about meetings — it's about creating unforgettable experiences!

Learn more at cmtausa.org/compass

CMTA's Gadget Store on Amazon

In 2024, the Charcot-Marie-Tooth Association (CMTA) launched a comprehensive **Amazon storefront** featuring assistive gadgets and products for the CMT community. CMTA earns a commission on most of the items on the storefront but is not responsible for the fulfillment, and/or quality of any of the unique products listed. To learn more about the store, **visit cmtausa.org/gadgets**

New CMTA Branches

CMTA Branches truly embodied the spirit of unity in 2024. With 91 branch leaders (14 new), 65 branches (8 new), and over 105 meetings, these groups offered members the chance to share resources, celebrate triumphs, and connect with others in their area. Whether learning about local services or hearing the latest CMTA updates, Branch members forged meaningful relationships that strengthened their connection to the CMT community.

Learn more at cmtausa.org/branches

USA

Indianapolis, IN
Springfield, MO
South Louisiana
Salt Lake City, UT
Northwestern Indiana
Palm Beach County, FL

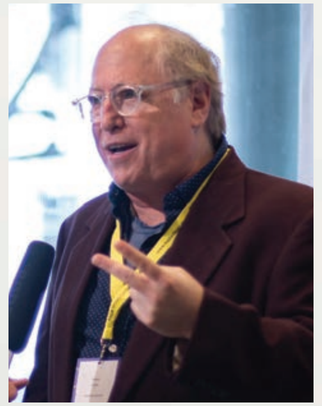
INTERNATIONAL

Eastern Canada
(Toronto, Canada)

#LetsBeatCMT: A Global Awareness Campaign

CMTA launched the **#LetsBeatCMT** campaign for **CMT Awareness Month** in September. This initiative featured daily videos, personal stories, photos, and infographics, bringing unprecedented engagement from the global CMT community. Well-known personalities such as **Adam Zucker** (CBS Sports) and Emmy Award-winning writer/producer **Danny Zuker** *(Modern Family)* lent their support, amplifying the campaign's reach and impact.

Adam Zucker



Danny Zuker

CMTA COMMUNITY OUTREACH

Expanding Expert Care:

CMTA
Charcot-Marie-Tooth Association
Centers of Excellence

CMTA added five new neuromuscular neurologists to its global network of **Centers of Excellence** in 2024. These specialists provide comprehensive, patient-focused care and are a vital part of CMTA's mission to improve the lives of individuals with CMT.

To find a CMTA Center of Excellence near you, visit cmtausa.org/coe

Duke University
Durham, North Carolina

Natalie Katz, MD

Virginia Commonwealth University
Henrico, Virginia

Nicholas Johnson, MD

University of Kentucky
Lexington, Kentucky

Nakul Katyal, MD

Mahidol University Siriraj Hospital
Bangkok, Thailand

Oranee Sanmaneechai, MD

St. George's University Hospital, NHS Foundation Trust
London, England

Niranjanan Nirmalananthan, PhD and Emma Matthews, PhD

CMTA RESEARCH

CMTA-STAR: Accelerating Research, Empowering Patients

by Katherine Forsey, PhD
CMTA Chief Research Officer

Since 2008, CMTA has invested nearly **$30 million** to accelerate CMT research. In 2024, **CMTA's Strategy To Accelerate Research (CMTA-STAR)** program continued to unite researchers, clinicians, and the global CMT community, accelerating treatment development through multiple approaches, including gene therapy and drug discovery. Each CMTA-funded project, discovery, and clinical trial brings us closer to transforming lives and offering hope to everyone affected by CMT.

Driving Research Forward

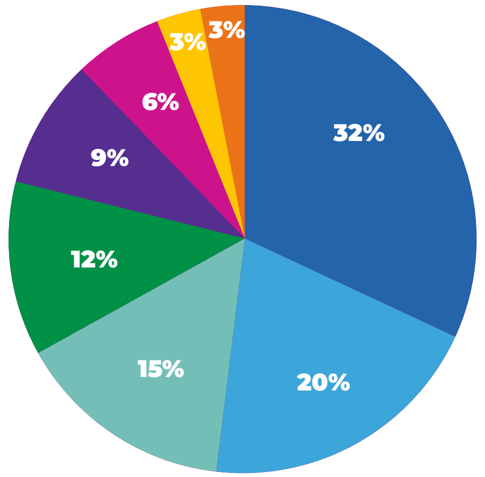
In 2024, CMTA funded 34 projects across 14 CMT subtypes and supported genetic discovery initiatives for the 40% of the community who do not yet know the genetic cause of their CMT. With the launch of the **CMTA-INC** strategic partnership, our efforts now encompass all known and currently unidentified CMT subtypes.

The total value of these projects reached **$9,701,422,** with eight new projects initiated and eight completed this year. Summaries of all projects are available in the ***CMTA Winter Report 2024*** and on CMTA's website.

Our leadership in research and clinical trial readiness is evident in the increasing number of trials underway or on the horizon *(see page 20)*. CMTA has created an ecosystem connecting the CMT community, researchers, and clinical trials, attracting new CMTA-STAR Alliance Partners and accelerating progress toward the first approved treatments for CMT.

CMTA-Funded Research Projects

2024 active sponsored research projects by type of approach



	PROJECT BY TYPE OF APPROACH	NUMBER OF PROJECTS
🟦	Genetic therapies incl. delivery	11
🟦	Natural history and biomarkers	7
🟩	In vitro model	5
🟩	Small molecule	4
🟪	Gene discovery	3
🟪	INC partnership	2
🟨	Animal model	1
🟧	Clinical trial	1

TOTAL VALUE OF PROJECTS
$9,701,422

NUMBER OF PROJECTS FUNDED BY CMTA
34

CMT SUBTYPES COVERED BY A CMTA-FUNDED PROJECT
100%

New Gene Discoveries

In 2024, CMTA added **seven new CMT-causative genes** to its growing genetics catalog: *DHX9, INSC, NARS1, NDUFS6, RFC1, SACS,* and *TECPR2.* **This milestone expanded the catalog to 136 genes and 170 subtypes.** A discovery on chromosome 13 (SACS gene – CMT-SACS) filled a gap in the catalog, as CMT disease genes are now found on all chromosomes except the Y chromosome.

CMTA'S GENETICS CATALOG:

136 CMT Genes and 170 Subtypes

The discovery of CMT-SORD highlights the impact of genetic discovery. Funded by CMTA and published in ***Nature Genetics*** in 2020, this breakthrough has advanced to a Phase III clinical trial led by **Applied Therapeutics,** a CMTA-STAR Alliance Partner. This trial could deliver the first FDA-approved treatment for CMT, paving the way for future advancements.

Preclinical Toolbox: Laying the Foundation for Therapies

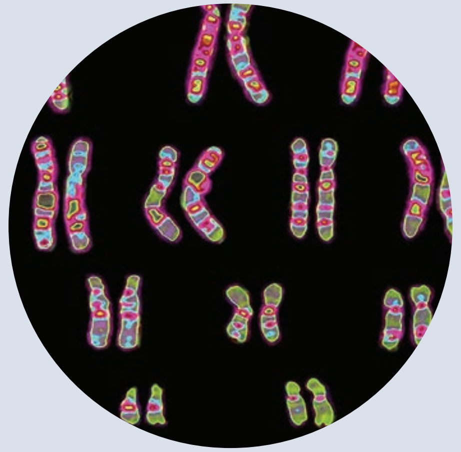
CMTA's Preclinical Toolbox accelerates therapy development by providing researchers with models and tools to test treatments before human trials. These include genetically engineered models for subtypes like **CMT1A, CMTX1, CMT2A,** and **CMT2E,** as well as patient-derived cell lines.

In 2024, six new stem cell lines were added to the toolbox, bringing the collection to 17 lines across seven subtypes and two isogenic controls. Since its launch, **34 organizations have utilized CMTA's Biobank,** driving research and development forward.

CMTA also collaborated with eight companies on preclinical testing in CMT1A, 2A, and 2E, conducting ten studies. These studies ensure only the most safe and effective therapies proceed to human trials.

2024 Pre-Clinical Toolbox At-A-Glance

- **10 studies** conducted in 2024
- **8 Alliance Partners** tested with CMTA
- **6 new stem cells lines** added to CMTA's Biobank
- **3 CMT Subtypes covered** CMT 1A, 2A and 2E



Since the inception of CMTA's Pre-Clinical Toolbox, ***34 organizations*** *have used CMTA's stem cell lines for Research and Development.*

CMTA-STAR Alliance Partners: Leading Clinical Trials

CMTA-STAR Alliance Partners made significant strides in the 2024 Clinical Trials Pipeline:

Applied Therapeutics | APPLIED THERAPEUTICS
CMT-SORD
A Phase III trial that could set a regulatory precedent as the first FDA-approved treatment for CMT.

NMD Pharma | NMD PHARMA NEUROMUSCULAR DISORDERS
CMT Types 1 and 2
A Phase II trial launched at the CMTA Center of Excellence in Austin, Texas, with sites in the USA and Europe. Focused on improving neuromuscular junction function.

Elpida Therapeutics | ELPIDA THERAPEUTICS
CMT4J
Preparing for an interventional gene replacement therapy trial.

UCL Queen Square Institute of Neurology | UCL Queen Square Institute of Neurology UCL
SENSE Trial
A Phase II trial testing Serine for HSN-1, with methods that may inform outcome measures for other CMT trials.

Actio Biosciences | actio BIOSCIENCES
CMT2C
Phase I recruitment set to begin in early 2025.

Novartis | NOVARTIS
CMT1A
Investigating siRNA-based approaches to reduce PMP22 expression.

Armatus Bio, Inc. | ARMATUSBIO
CMT1A
Exploring AAV-based methods to lower PMP22 levels.

Sarepta Therapeutics | SAREPTA THERAPEUTICS
CMT1A and CMTX1
Exploring early-stage programs focused on genetic therapies.

Shaping the Future of CMT Research

In 2024, CMTA hosted two in-person **CMTA-STAR Advisory Board meetings:** one in Dallas,. TX and one in Montreal, QC, Canada. The Montreal meeting was alongside the **Peripheral Nerve Society** annual meeting.

CMTA | STAR
Charcot-Marie-Tooth Association | Strategy To Accelerate Research

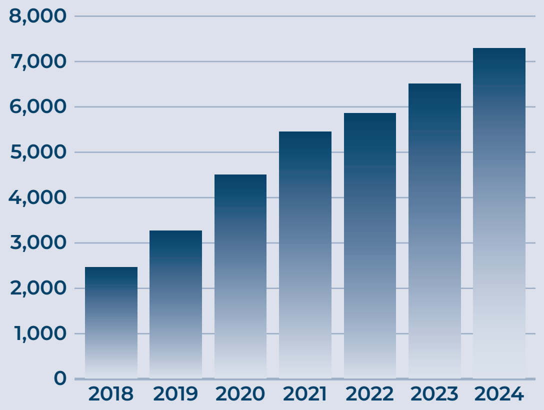
These gatherings assessed new projects and prioritized those with the greatest impact. Monthly virtual meetings ensured continuous progress and provided vital feedback to researchers and companies. The CMTA-STAR Advisory Board consists of more than 30 world-leading CMT research scientists and clinicians.

PATIENTS as PARTNERS in RESEARCH

Charcot-Marie-Tooth Association

CMTA's Patients as Partners in Research program continues to empower the community to drive progress. With **over 7,000 profiles created — 932 added in 2024 alone** — the platform represents the largest global patient-reported CMT data set. To date, it has supported over 31 studies, focus groups, and clinical trials across 49 subtypes.

Total Number of Patient Partner Profiles



Key Metrics At-A-Glance

- **Over 7,000 patient profiles** created
- **49 CMT Subtypes** represented
- **More than 31 studies** supported

Join Us!

Your participation fuels progress. Register, build your profile, and explore studies in your area at **cmtausa.org/pap**, or scan this code.

A Salute to Our Incredible Community of Donors!

by Jeana Sweeney
CMTA Chief Engagement and Gift Officer

Reflecting on 2024, I am overwhelmed with gratitude for your incredible generosity and dedication, which have powered our mission in extraordinary ways. This past year wasn't just about raising funds — it was about coming together as a community to make a meaningful difference and bring hope to everyone living with CMT. Because of your support, we've advanced research, funded critical programs, and expanded resources for our growing community. Together, here's what we accomplished:

2024 Match Challenges: A Community United for Change

Our community rallied like never before, transforming **$755,000** in matching opportunities from anonymous donors into **$1.51 million raised** for CMT research, programs, and support. These dollars represent action, impact, and real hope. Might you be our next matching donor?

Sustained Support Through Monthly Giving: The Power of Our 'INNERVATORS'

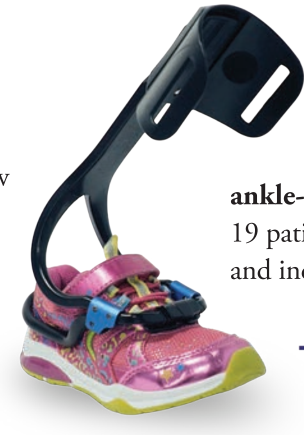
Our **IN*NERV*ATORS** — monthly donors who stand at the heart of our mission, helped us achieve incredible milestones. With 80 new donors joining this inspiring group in 2024, the impact of consistent, ongoing support has never been clearer.

See our call to action on page 28.
Learn more at cmtausa.org/innervators

Foundations, Sponsorships, and Corporate Partnerships

We were honored to receive **six grants totaling $182,150,** and **partner with ten corporate and biopharma sponsors** who supported us in driving transformative change:

- **Camp Footprint:** Their contributions helped provide free attendance for 151 youth at Camp Footprint, including covering travel expenses for 37 campers, ensuring no child was left behind.



- **Innovative Partnerships:** Collaborations, like those with **TurboMed Orthotics,** provided *free* **ankle-foot orthoses (AFOs)** to 19 patients, empowering mobility and independence.

TurboMed ORTHOTICS USA

- **Patient & Research Summit:** Their generosity brought the 2024 Summit to life, providing education, connection, and empowerment to hundreds of attendees. Nine companies showcased cutting-edge products and resources for the CMT community.

Pictured above: Kenny Raymond, CMTA Head of Communications; Daniel Brennan, NMD Pharma; Sue Bruhn, CMTA Chief Executive Officer; and Dottie Caplan, Applied Therapeutics at the 2024 CMTA Patient & Research Summit in Denver, CO.

2024 Donor Impact on Research

Your generosity fueled remarkable progress in **CMTA's Strategy To Accelerate Research (STAR)** program. With **$4.2 million** donated in 2024, you've supported advancements in gene therapy and small molecule therapies, offering new hope for effective treatments targeting the root cause of CMT.

Bequests: A Legacy of Generosity

We were deeply honored to receive **$333,192** through five generous bequests. These gifts reflect the lasting commitment of individuals ensuring a brighter future for the CMT community.

Supporting Life-Changing Programs

- **Patient Support Fund:** Provided travel assistance for six patients to undergo expert surgery in Los Angeles, transforming their quality of life.
- **Bracing Fund:** Established in late December, this fund will provide essential AFOs to individuals in need, fostering independence and mobility.

This year demonstrated the incredible power of what we can achieve together. As your Chief Engagement and Gift Officer, and on behalf of the entire development team — **Karen, Mary,** and **Nicole** — we are immensely proud of all we've accomplished as a community. For us, these roles are more than jobs — they are callings. We are honored to serve as the voices and advocates for this extraordinary community, and with every gift we receive, we are deeply humbled by your generosity and unwavering commitment.

Let's carry this momentum into 2025 and make it a year of even greater impact. Together, we can continue to rally around CMTA's mission and achieve incredible things for everyone affected by CMT.

The Foresee Gala: Unmasking the Cure for CMT4C

In June 2024, the **Paulsen family** *(pictured above)* hosted the **Foresee Gala** in Denver, Colorado, **raising $250,000 to support Project Foresee.** This event not only raised vital funds but also significantly increased awareness of CMT4C, reaching over 1,000 new supporters through various channels.

Led by **Bob Paulsen,** Project Foresee is a CMTA-sponsored initiative advancing the most promising gene therapy for CMT4C. The goal is clear: to follow established steps toward curing the first handful of patients within the next three years.

Impact Through Peer-to-Peer and In-Person Events

From galas to golf tournaments, heartfelt personal asks, and even the sweetest lemonade stands, your creativity and passion have raised essential funds to support the CMTA. Each effort, no matter the size, makes a meaningful difference. They prove that even the smallest acts of kindness can have a big impact.

Your dedication shines a spotlight on CMT, reminding us of the incredible power of community. Thanks to fundraisers like you, we're stepping into 2025 with renewed hope, unstoppable momentum, and a shared commitment to make an even greater impact.

To learn more visit cmtausa.org/waystogive

CMTA Visits Broadway: *Hell's Kitchen*

CMTA supporters attended a Broadway performance of ***Hell's Kitchen*** on September 28. Special thanks to **Gary** and **Helene Lenet** for their leadership in this initiative, bringing together over 50 guests and raising an incredible **$45,000** for CMTA's mission.

As Gary shared during the evening, "We draw inspiration from the Broadway show *Hell's Kitchen,* which tells of **Alicia Keys'** remarkable journey, rising from her roots with the support of her friends, community, and a devoted music teacher. Just like this, with our friends and community, we remain committed to standing with you as we fight for a future where CMTA is a key part of the cure."

Thank you to everyone who attended and donated. Together, we are shaping a future for you and for generations we may never meet.

Editors Note: *Mrs. Lenet passed away in October. Our hearts are with Gary and the Lenet family.*

Tuesday, December 3, 2024: #GivingTOESday

Each year, CMTA puts its spin on the popular *#GivingTuesday* initiative with **#GivingTOESday.** More than **$80,000** was raised for CMTA on the 2024 global day of giving, but any day can be a *'TOESday'!*

To learn more, visit cmtausa.org/givingtoesday

CMTA Fundraiser: The Coldiron Derby Party

The **Coldiron Derby Party** is an annual fundraising event hosted by CMTA Board Member **David Coldiron** to support CMTA. The event celebrates the Kentucky Derby while raising much-needed funds and awareness for CMTA. This year's event raised **$ 65,000.**

The Coldiron family

TOP 5 WALK & CYCLE 4 CMT EVENTS

Walk & Cycle 4 CMT: A Year of Unstoppable Momentum

This year, **CMTA's Walk & Cycle 4 CMT** community **raised $417,386,** with **1,559 participants** nationwide. The power of connection and shared purpose was on full display at each of these local events.

PRESENTED BY APPLIED THERAPEUTICS | SPONSORED BY NOVARTIS

BOSTON **Walk & Cycle 4 CMT**
74 Participants / $53,098 Raised

PHILADELPHIA **Walk 4 CMT**
133 Participants / $51,224 Raised

CENTRAL NEW JERSEY **Walk & 4 CMT**
208 Participants / $49,618 Raised

WASHINGTON DC **Walk 4 CMT**
56 Participants / $30,517 Raised

SAN DIEGO **Walk 4 CMT**
80 Participants / $17,254 Raised

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BECOME AN INNERVATOR

JOIN CMTA'S MONTHLY GIVING PROGRAM

CMTA INNERVATORS are action-oriented game-changers. They sustain CMTA with monthly gifts throughout the year. Life-changing initiatives like Camp Footprint treatment-focused research, and invaluable educational initiatives depend on steady, support from committed and reliable donors.

If you want to make a lasting difference in the CMTA community, please sign up to be part of the new ***INNERVATORS*** program and leave an indelible footprint on the lives of present and future generations.
Join now. It's easy, effortless, and everlasting!

To learn more, visit
cmtausa.org/innervators

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