SUMMER 2023

THE CMTA REPORT

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Charcot-Marie-Tooth Association

Charcot-Marie-Tooth Association

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Unexpected Consequences

SPERM DONOR WITH CMT GENE PASSES IT ON TO MULTIPLE FAMILIES

BY MARCIA SEMMES

The letter from the sperm bank landed like a bomb in the midst of the families who had conceived children with Donor X: He had tested positive for one of the genes that cause Charcot-Marie-Tooth disease (CMT), a rare neuromuscular disease that affects one in 2,500 people.

The February 28 letter from the sperm bank asked Donor X to submit to genetic testing after one of his offspring tested positive for a likely pathogenic variant in the DMN2 gene. While he had no CMT symptoms—and no family history of the disease—he agreed. The sperm bank also recommended that the biological mother be tested. Only the donor's results were positive.

Based on this result, the letter continued, all offspring of Donor X have a 50 percent chance of inheriting his gene variant and are at risk for symptoms if the variant is determined to cause disease.

Single mother Erica B., 47, got one of those letters. Two of her four children—Frankie, 11 and George, 7—were conceived using Donor X's sperm. Both have a rare form of CMT—Dominant Intermediate B, a group of CMT subtypes in which nerve conduction does not indicate either axonal CMT or demyelinating CMT. Instead, nerve conduction study results are somewhere in between, or intermediate, a term that refers only to nerve conduction and not to disease severity or progression.

Donor X was an open donor, meaning that his biological children are allowed to contact him when they turn 18. This also meant that there was a portal for parents who had children with the same open donor. After Frankie was born, a family contacted Erica through the portal to offer her a vial of Donor X's sperm they no longer needed. She didn't hesitate: She wanted full siblings, and at the time there was no reason to say no.

Erica knows of 10 kids from seven families who share Donor X's DNA. Many of them have met at Erica's house, spending weeks together and visiting Disney World daily. All of the kids are very much alike, Erica says, sharing personalities, looks and mannerisms. They call each other "diblings," a mashup of donor siblings.

> "Diblings" George and Frankie share a rare form of CMT.

Frankie first exhibited CMT symptoms at the age of 2. Over the years, his symptoms grew worse, as they do with a progressive disease. Frankie was formally diagnosed with CMT in December 2022, George in February, less than two weeks before the sperm bank letter arrived. Erica tested negative.

Today, both brothers are legally deaf and live with apraxia, severe nerve pain and sensory issues. Frankie also has severe breathing issues. While Frankie wears a hearing aid, George communicates solely by signing and the whole family knows American Sign Language. Finding the silver lining in this particular cloud, Erica says that "Frankie teaches us what is coming for George." In the last 18 months, Frankie has lost almost all muscle tone and started "toe-walking," a typical CMT symptom.

What's extraordinary about the boys is their ability to participate in everyday life. As Erica put it, "Their lives are greatly impacted but not destroyed." They are in a special Little League, swim like fishes, golf and play bocce ball. George happily spends hours at a time in the sandbox. But any of those activities can set off paroxysms of pain so bad that Frankie is afraid he's dying. Both take gabapentin with mixed results, and Erica is concerned that Frankie's dose keeps getting upped. "You know where that leads," she said.

Erica has two other sons as well: William, 27, an automotive journalist, lives at home with the family, providing crucial support and care. She recently adopted baby Joey, now 8 months, who was born with fetal alcohol syndrome. With the love and care this remarkable family provides, Joey is improving—and he keeps Frankie happy and distracted. "I am forever grateful for Joey allowing us to show him what love and family truly mean," she said.

While Erica clearly adores all her children, she said she wouldn't have used Donor X's sperm for George if she had been informed about the CMT. "I wouldn't have purposefully had a child who had to go through so much pain."

Erica is a fighter—and a fierce advocate for her kids. Her advocacy extends to everyone with CMT, and her hope is that by being open about her family's story, "The specialists and the donors feel compelled to help find treatment for the painful and debilitating symptoms that CMT causes."

In a happy footnote to the family's story, Frankie was recently accepted to Camp Footprint, the country's only sleepaway camp for kids who have CMT. Fully funded by the CMTA, Camp Footprint (motto "One Step at a Time") gives kids with CMT a chance to participate in all the regular camp activities like swimming, horseback riding, sports, campfires and music. But they'll also get the experience of connecting to strangers who quickly become friends because they share the same physical and emotional challenges. Campers get the chance to feel understood, to be normal and to blend in instead of standing out.

TECH ENTREPRENEUR BRINGS ENTHUSIASM, BUSINESS TRACK RECORD TO CMTA BOARD

Tech entrepreneur David Apple joined the CMTA Board of Directors on April 25, saying he is thrilled to become part of the generation that will find treatments and cures for CMT.

Apple's CMT journey began in January when his 2-and-a-half-year-old son Ari was diagnosed with CMT1A, the result of a de novo mutation. As with many members of the CMT community, getting the diagnosis took more than a year of unwavering determination and persistence, scheduling initial appointments and second opinions from over a dozen specialists. None of them suspected that CMT was the cause of Ari's symptoms.

Following Ari's diagnosis, David turned his anguish into action with the dual goal of setting Ari up for success in the short term and working toward a cure for CMT in the mid-term.

CMTA Board President Gilles Bouchard said, "The Board is excited to welcome David Apple. His enthusiasm and relentless energy, combined with a remarkable business track record, will be a very valuable asset in our fight against CMT."

David, who is French American, lives in San Francisco with his wife and two kids. He spent the last decade of his career in tech, notably as an early employee of two startups—Notion and Typeform—that grew into "unicorns," companies that reach a valuation of $1 billion. He is currently the chief business officer of ScorePlay, and an active angel investor and start-up advisor.

David has a BS in mechanical engineering from The Johns Hopkins University and an MBA from Imperial College London. He is currently completing an online biotech course to better understand potential treatments and cures for CMT.

CMTA ADDS DRUG DEVELOPMENT EXPERT MARTINE GARNIER TO STAR ADVISORY BOARD

The Charcot-Marie-Tooth Association added to its already deep bench of CMT experts March 13, appointing Martine Garnier, PhD, EMBA, to its Therapy Advisory Board (TEB). Garnier, the director of drug discovery and development, alliance project manager and founder of MG Pharma Consulting, said she is "proud to be joining the STAR Advisory Board to contribute to the CMTA's patient-focused mission and vision by helping push the boundaries of science to deliver the next-generation life-changing medicines to CMT patients."

The TEB is one of the three legs of CMTA's STAR Advisory Board, along with the Scientific Expert Board and the Clinical Expert Board. The TEB evaluates the translational quality of ongoing and proposed projects, meaning how quickly scientific discoveries can be "translated" to treatments. It focuses on the pharmacological approach of disease-area project team efforts, the potential to form strategic alliances with pharmaceutical partners and preclinical strategy. The TEB may also take an active role in managing and monitoring strategic alliances.

Garnier brings to the TEB 20 years of experience leading drug discovery and clinical development projects within pharma, biotech and contract research organizations, with expertise in small molecule therapeutics and neuroscience. She spent 10 years doing international academic research in France, the United States and Italy.

Throughout her career, Garnier has worked on a wide range of target classes and disease areas, including oncology, antibiotics and rare diseases. She joined GSK Neuroscience R&D in 1995, contributing to establishing GSK pain research and psychiatry R&D expertise in Milan and Verona Centers of Excellence for Drug Discovery. She also supported several biotech projects, advancing more than 20 candidates and preclinical assets from lead optimization to clinical stages.

Garnier received her PhD in molecular pharmacology and toxicology from Paul Sabatier University in Toulouse, France, and her Laureate Executive MBA from HEC Paris. She has received research grants and awards from Georgetown University Medical Center, Telethon Italia and Fondation HEC and has written more than 33 peer-reviewed publications.

SUMMER WITH CMT

SUMMER IS A SEASON FOR MOVEMENT

whether it's driving cross-country, traveling by tram, swimming, golfing or any one of a myriad of outdoor activities. Members of the CMT community can do all of those—but some may need certain accommodations before breaking out their swimsuits, dusting off their bicycles, or taking that cross-country road trip. In this issue, we bring you tips for making the most of summer and all its glories.

SWIMMING WITH CMT

BY JONAH BERGER

Before I became a father with CMT, I worried about certain traditional "Daddy" moments. One of the biggest was walking my child into the ocean for the first time. It's hard for me to walk on my own in moving water and uneven sandy ground, much less while carrying a child.

My family vacationed at Lake Michigan last year. I call it the ocean because, with the waves and tides, it basically is. My wife Megan was feeding my infant son Artie, and my daughter Amelia (Millie Rose) wanted to go into the water. The time had come to face the fear. I took off my braces and held her hand and we walked—funky style—down to the water.

When we got to the water's edge, I instinctively sat down. And without missing a beat, Amelia sat down in front of me between my legs. We sat there for a while and the water rose up over our feet. She loved it. Every few minutes, she would say, "Let's go farther Dada." And I would take a butt scoot or two forward. And the waves would come up and get more of us wet.

I had my arms around her and anytime I let go for any reason, she would grab my arms and yell "HOLD ME DADA!" In that moment, I didn't feel disabled. I wasn't lacking for anything. I was her dad. Her dad who has to sit instead of stand in shallow water. Her dad who had his arms around her so no harm would come to her. She wasn't accommodating me. She didn't know any different. To her, this is life. This is fatherhood, and the look in her eyes told me that it was fine. All she wanted or needed.

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Jonah and his daughter Millie Rose

SWIMMING WITH CMT

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And when the time came for her to go all the way in, I gladly handed her over to Grandma and Uncle Mickey. No sadness that I couldn't be the one to take her, but joy at the needed reminder that we are all so unique. And when we receive acceptance and pure love, our differences aren't burdens we carry around. They can just be who we are. Pretty cool to see how some of the parts of fatherhood that I was most dreading have turned out to be the most precious. Thanks, Millie Rose.

Jonah is the CMTA's national youth programs manager and the director of Camp Footprint.

TAKE PRECAUTIONS FOR FUN IN THE SUN

BY BRITTANY BAGGETT, RN, BSN

While summer is great for outdoor activity and fun, sunny weather increases the risk of dehydration, heat illness and injuries. People with CMT may also be at particular risk for foot injuries.

Staying hydrated is vital to keeping the heart pumping regularly and ensuring muscles work correctly. Dehydration puts added stress on muscles and organs. The best way to prevent dehydration is to keep a water bottle close and take frequent sips. Eating fruits and vegetables is another great way to hydrate.

Failure to hydrate can lead to heat exhaustion or heat stroke. The former can typically be treated quickly by getting out of the heat as soon as symptoms—profuse sweating, nausea or vomiting and headache—occur. If symptoms last longer than an hour, worsen, or vomiting occurs, seek medical attention immediately.

Heat stroke occurs when the body can no longer regulate its temperature due to prolonged exposure to the heat or strenuous exercise in the heat. During heat stroke, the body can reach temperatures of 103 degrees Fahrenheit or higher. Signs and symptoms of heat stroke include dry, hot skin, dizziness and confusion. Heat stroke is a medical emergency that should be treated immediately.

Heat illnesses can be prevented by taking a few simple steps: Wear light-colored, breathable clothing and take frequent breaks in the shade. Check the local weather forecast and plan your day accordingly.

Foot care is also important for summer exercise. People who suffer from CMT are more prone to foot injuries and an untreated minor sore can quickly become a big problem, especially if it becomes infected. Foot injuries can be prevented by following a few simple rules. Check feet frequently to monitor old sores and watch for new ones. Scope out the trail BEFORE hiking to determine difficulty and maintenance. Map information and trail maintenance updates are usually accessible online. Keep your feet dry, and wear supportive footwear.

Finally, caregivers play a crucial role in the safety of their loved ones and are their greatest advocates. During the summer, caregivers help prevent heat illnesses, dehydration and foot injuries by frequently offering water and monitoring their loved ones for any changes in behavior.

Brittany is a freelance writer with seven years of nursing experience, a mother, wife and outdoor enthusiast. Her father was diagnosed with CMT 10 years ago, and she has symptoms but has not been formally tested.

NAVIGATING PRAGUE WITH CMT: STEPS, TRAMS AND HILLS, OH MY!

BY RACHAEL SOUZA

I left my home state of Rhode Island to move to the Czech Republic last year. I made the decision when the pandemic finally felt like it was ending, and I realized that I was about to turn 30 working a job I hated and living in the same state I grew up in. I wanted, no, I needed a change, and despite the challenges posed by living in a foreign country with CMT, I'm glad I made it.

In Prague, I signed up at The Language House (TLH) for a four-week TEFL (Teaching English as a Foreign Language) accreditation course promising hands-on learning, job assistance and visa assistance. I learned a lot and gained practical teaching experience that made the transition to paid teaching much easier.

TLH also accommodated my CMT, putting me in more accessible student housing than the typical hostel on a steep hill they normally provide. But I still had to climb three flights of stairs to get to my flat. They also gave me a key to use the elevator at the school and arranged the lessons I needed to teach so I wouldn't have to travel outside the school to complete them. In the past, I wouldn't have disclosed my disability and would have just dealt with whatever was given to me.

Rachael in Prague

A Prague tram

To get around in Prague, I either walk or take the tram to my destination. I strategically rented an apartment across the street from a tram stop. It takes me less than two minutes to walk to it. The tram runs every 10 minutes or so, so even if I miss one, another arrives in no time. There are trams all over the city, making it easy to get around without a car.

There are two common types of trams. The more modern version, which is more frequent, is handicap accessible. At most stops, they pull up close enough to the curb that it is just a small step up to enter the tram.

The older version, to my dismay, has several steps up to enter. Getting on the older tram, I use the railings on the door to pull myself up and the handlebar on the wall to make it to the top.

I can usually get on both types of tram with only minor issues, though I've fallen or struggled more than once, most often when it is raining. My fellow passengers tried to help me, not knowing that they literally needed to pick me up and place me on my feet. Otherwise, I can't get the support I need from their outstretched arms to pull myself up.

Once on the tram, I try to find a seat. The moment I sit down I feel the anxiety growing in my stomach as I fear someone older or more visibly disabled or pregnant will board the tram and make me move. I spend these rides yelling inside my head I am disabled! I am allowed to sit here! Yet, I often get up if asked.

My disability is invisible when I wear pants or tall boots that hide my braces. People think I just walk funny. In Prague, I don't speak the language so a part of me fears that even if I told them I was disabled and could sit there they wouldn't understand me. I could flash my brace but that feels invasive and embarrassing. A friend suggested I write on an index card in Czech that I'm disabled, but I haven't done so.

Often, I feel like the people who ask for my seat need it more than I do. I can stand on a tram if I have to, I have before. I do need to grip the railing tightly, with both hands, and am still more likely to fall than most people, but still. I can make it work, so why bother arguing?

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TRAVELING WITH CMT

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Once, in Dublin, Ireland, a few friends and I boarded a double decker bus. At a stop, several people disembarked, and a handicapped seat opened up. My feet were aching from the walking we did around the city and trying to keep up with my able-bodied companions. It was a relief to sit for a moment and give my feet a break. The relief didn't last long.

A young woman and her mother boarded the bus with a stroller and stood in the wheelchair area across from me. They began whispering about how handicapped seats are reserved for the elderly and for mothers. I often think of that young woman's words while I sit on the trams in Prague. It's why I tell myself over and over again that I am disabled, I am allowed to sit in the handicapped seats.

Even though the trams in Prague have been a challenge for me on occasion, I still enjoy how easy they are to use and how reliable they are. I don't ever want to go back to driving myself everywhere like I did in the United States. The public transportation at home isn't nearly as reliable, clean or accommodating as it is in most of Europe. Just one more point in favor of moving to Europe.

Photos courtesy of Rachael's blog. Read more about her adventures abroad at www.peculiargenespeculiarplaces.com.

Matt takes a swing

GOLFER WITH CMT1X PURSUES TOURNAMENT DREAMS

BY SARAH KAIDER

Matt and his fiancée Mariah Trapp

Des Moine, Iowa, native Matt Meeks, 32, has been playing golf since he was a child. It's not just a sport for him, though; it's a passion that he has pursued despite being diagnosed with CMT1X. His persistence paid off, and today he's waiting to hear if he has been accepted to compete in the U.S. Adaptive Open, which will be held July 10-12.

Matt hopes to inspire others with CMT to take up golf. "There are so many great opportunities out there for people like us," he said. "Things are not always as bad as they seem. Sure, you're not able to do buttons, but that's just how it goes. You have to continue to move forward with a great support system around you and find ways to continue to do the things that you love."

Matt's golf career began when his mother urged him to find a sport he enjoyed. A family friend recommended golf, and Matt was hooked after attending a summer camp.

After winning a local tournament at Woodland Hills in Des Moines, Matt qualified for the Amateur Athletic Union national Championship in 2001 when he was just 10 years old. He took seventh place at the national championship and went on to win the state championship in the 14-and-under age bracket.

In high school, Matt played varsity golf for all four years. He dreamed of joining the Air Force, but his hopes were dashed when he found out that he had CMT, rendering him ineligible for military service.

After this setback, Matt played golf for his local community college while earning an associate degree in information technology and network administration. He also continued to play in local and corporate tournaments. He thought that his competitive golf career would end as his symptoms worsened, but he discovered the U.S. Adaptive Open, a golf tournament for golfers with disabilities.

CMT forced Matt to adapt his game. Since he cannot rely on his ankle strength, he uses his upper body more for his swing. He recommends using a wide shoe and having a proper fitting for a grip. A proper grip helps someone with fine motor and grip difficulties keep control of the club. He uses a multi-compound grip for driver 3, wood, hybrid, or driving iron, while for all his irons, he uses a mid-size grip without taper.

Matt's fiancée, who will become his wife on June 9, has been his constant support through all his struggles. "She has been wildly supportive through absolutely everything," he said. "She allows me to pretty much go golf whenever I want, and I think that's all us guys can really ask for."

Matt's story is one of perseverance and determination. He has not let CMT stop him from doing what he loves. He is excited about the opportunities available for people with disabilities and hopes that his story will encourage others to pursue their passions.

Sarah is the CMTA's digital marketing manager.

HAND CONTROLS MAKE SUMMER DRIVING SAFER

BY CLARK SEMMES

In my defense, it was dark. My wife and I were on vacation, in a rental car. As I pulled out of a parking space in a crowded lot, my foot somehow got stuck under the brake pedal.

My feet and my brain don't communicate well. Sometimes they don't communicate at all. It's because of my CMT.

As I struggled to free my foot and press down on the brake pedal, the rental car slowly rolled backward toward a row of parked cars. I was too slow. Before I knew it, I heard the unmistakable sound of metal hitting metal. I had hit a parked car. I cursed my CMT and got out to survey the damage.

Luckily, the damage to both cars was minimal. The damage to my confidence was not. It was now obvious that my CMT was impairing my driving ability. That time I got lucky, and no one was hurt. The next time I might not. I could forgive myself for injuring myself. Injuring someone else was out of the question. It was time to take action. It was time to get hand controls.

A number of companies manufacture hand controls that can be installed in one's vehicle.

I started with a query on Facebook: What is the process for getting hand controls installed on my car? The responses were immediate, supportive and helpful. God bless the CMT community. The first thing I learned was that the process is different in every state. For me, living in Florida, the process began at a place called Grace Adaptive Driving where I would receive instruction on how to drive with hand controls.

I'll admit, on the day of my instruction, I was very nervous. It had been almost 50 years since I took driver's ed in high school. Was I too old to learn a new way to drive? What if I couldn't get the hang of it? What if I failed the class? Would they take away my driver's license? My CMT causes my hands to shake slightly. When I get nervous, it gets worse. That day I could barely hold a cup of coffee.

My fears began to subside the moment I met my instructor, Carlos. The chill vibes he emitted calmed my nervous energy. I immediately liked him. Carlos began by giving me a battery of cognitive tests. My body may be slightly broken, but my mind is still clear and I whizzed through the tests. (The only one that gave me pause was reciting the months of the year backward. Try it. It's not as easy as it sounds.)

With my cognitive abilities confirmed it was time to hit the road. We climbed into Carlos' light truck. He gave me some quick instructions and then we were off. I will not lie, after 50 years of accelerating and braking with my feet, it felt strange to do both with my hands. I instinctively kept braking with my right foot. But with concentration and Carlos' gentle reassurance, I began to adapt.

Carlos kept a careful eye on my progress and his hand close to the steering wheel. As we circled the parking lot and then pulled out onto a real roadway my nerves returned and my hands shook more. But by intensely focusing on my task and listening carefully to Carlos' instructions, I managed to avoid any accidents or major mishaps. As the day wore on, Carlos' hand moved away from the steering wheel, and I slowly relaxed. In time, I was even able to hold a conversation.

Just as soon as I felt comfortable with the right-handed hand control, Carlos threw me a curve. We changed cars. The new car had hand controls on the left. I was back to square one as I struggled with this new alignment, but soon it felt comfortable too. We tested two other hand-control models that day before I settled on the first one, which was the simplest and least expensive. That's not to say it was cheap—all in, the controls cost around $3,000, offset by a $1,000 rebate from Honda.

At the end of the day, Carlos congratulated me on graduating, and we arranged for the installation of my hand controls at a place called Mobility Works. The final state step in the process is to visit the DMV and take a driving test using my new equipment. I am currently practicing for this final hurdle. I am sure when I visit the DMV, I will be nervous and my hands will be shaking, but I have a new motto I borrowed from the military, "Adapt and overcome."

Clark Semmes is a member of the CMTA Advisory Board.

CMT Patients Urge Faster Clinical Trials During FDA Listening Session

BY SARAH KAIDER

CMT patients and their families urged the Food and Drug Administration to expedite clinical trials for potential treatments at a Zoom Patient Listening Session April 18. They also expressed the need for treatments that will slow the progression of muscle wasting.

FDA Patient Listening Sessions provide a critical platform for patients and their caregivers to share their perspectives on issues that affect their health and well-being. By sharing their experiences with the FDA, patients can help inform the development of medical products that better meet their needs. After one such session on cystic fibrosis, the FDA used the insights gained to approve Kalydeco, which has been shown to improve lung function in patients with cystic fibrosis.

These sessions illustrate FDA's understanding that patients and their caregivers are essential partners in the drug and medical device development process. Engaging with patients early in the development process can lead to better health outcomes and more patient-centered medical products.

Dr. David Hermann, a member of the CMTA's Therapy Expert Board and Clinical Expert Board, summarized and defined CMT and its symptoms for the session. He shared patient-reported quality of life data, economic burden data, what current care looks like for CMT and pre-clinical approaches. He cited an ascorbic acid study, which didn't result in a treatment, but helped inform how to conduct clinical trials and the limitations of current outcome measures. He also explained how the Inherited Neuropathy Consortium was born out of the need for these measures and shared examples of work being done to find CMT biomarkers.

Giving a patient perspective, former CMTA Branch Leader Alanna Huber shared her family's diagnostic odyssey. She lost her job because she had to care for her 15-year-old son Carter, who has a severe case of CMT1A. Carter's unique disease progression resulted in scoliosis and chest cavity surgery, with over $157,000 in medical costs, excluding surgery and genetic testing. Huber's testimony highlighted both the need for effective treatments and the financial burden on families living with CMT.

Joe Hogan, 62, also shared his CMT journey, which includes hand and foot deformities and foot amputation. He spoke about being bullied due to walking, running and balance problems and choosing not to have children to avoid passing the disease along. He emphasized the need for the FDA to expedite treatments for CMT, which would improve quality of life for him and other CMT patients.

By sharing their experiences and perspectives with the FDA, CMT patients and their families helped raise awareness of the disease and the urgent need for effective treatments. The insights gained from this session will inform the development of biomarkers, outcome measures, and new targets for drug development.

The CMTA and other patient advocacy organizations actively ensure that the patient's perspective is at the center of research and development efforts for CMT. As part of that effort, the CMTA's Patients as Partners in Research program allows patients with CMT to contribute feedback directly to researchers through online surveys, patient focus groups and research studies.

Join the CMTA'S Patients as Partners in Research program at bit.ly/3x3euW1.

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APPLIED THERAPEUTICS ANNOUNCES POSITIVE SORBITOL REDUCTION DATA FROM INSPIRE TRIAL

Applied Therapeutics, Inc. recently announced positive sorbitol reduction data from its ongoing global Phase 3 INSPIRE trial. The INSPIRE trial is a double-blind, placebo-controlled registrational study evaluating the effect of once-daily oral AT-007 in approximately 50 patients aged 16-55 with SORD Deficiency in the United States and Europe.

SORD Deficiency (also called SORD Neuropathy or CMT-SORD) is a debilitating hereditary axonal neuropathy caused by mutations in the sorbitol dehydrogenase gene, leading to an inability to metabolize the sugar sorbitol. This inability results in the accumulation of high levels of toxic sorbitol, which causes motor neuron degeneration and loss of mobility and motility.

AT-007 (govorestat) is a central nervous system penetrant aldose reductase inhibitor, which blocks conversion of glucose to sorbitol. It has previously been shown to reduce sorbitol levels in an open-label pilot study in patients with SORD Deficiency.

In a pre-specified interim analysis of the ongoing Phase 3 INSPIRE trial, AT-007 reduced sorbitol levels by a mean of approximately 52 percent (or approximately 16,000ng/ml) over 90 days of treatment (p<0.001 vs. placebo) in patients with SORD Deficiency.

At baseline, the mean blood sorbitol level of SORD patients was approximately 29,000ng/ml, with a range of approximately 22,000ng/ml-38,000ng/ml.

The INSPIRE study will continue in blinded format to the 12-month interim clinical outcomes assessment. If the primary clinical outcome measure (10-meter-walk/run) reaches statistical significance at 12 months, the study will be completed and unblinded. If not, the study will continue in blinded format to 24 months, and clinical outcomes will be assessed again in a final statistical analysis. AT-007 continues to be safe and well tolerated to date.

"The results of the interim sorbitol analysis are quite compelling, and we believe the reduction in sorbitol level with AT-007 is clinically meaningful," said Riccardo Perfetti, MD, PhD, chief medical officer at Applied Therapeutics.

"The role of sorbitol in disease pathogenesis in SORD Deficiency is clear," said Michael Shy, MD, Director of the Division of Neuromuscular Medicine at Carver College of Medicine, University of Iowa Medical Center, and Principal Investigator on the INSPIRE Phase 3 trial. "The reduction in sorbitol levels seen thus far with AT-007 is impressive and is predicted to translate into clinical benefit."

NEW DTX TREATMENT FOR CMT1A DIRECTLY TARGETS EXCESS PMP22

DTx Pharma is developing a treatment for CMT1A that directly targets the cause of the disease—excess peripheral myelin protein 22 (PMP22). The treatment, DTx-1252, reduces the amount of PMP22 in the myelin-producing cells that are affected by CMT1A.

PMP22 is a protein needed to make the protective covering around nerves—or myelin sheath—which helps transmit signals to the muscles. A large body of data from animal models of CMT1A suggests that reducing PMP22 levels allows the myelin sheath to form properly. This leads to improved transmission of nerve signals and restoration of muscle function.

DTx Pharma is conducting the required safety studies in animals to support an Investigational New Drug (IND) application to the Food and Drug Administration. This is needed to allow the testing of DTx-1252 in humans prior to the first human clinical trial, which will test both the safety of DTx-1252 and how well it reduces the amount of PMP22 protein. The resulting data will support longer-term studies to evaluate improvement in sensory and motor function in CMT1A patients.

To support development of DTx-1252, the company is working closely with members of the Inherited Neuropathy Consortium (INC) and patient associations, including the CMTA. Patient participation in natural history studies such as the ongoing observational study ACT-CMT (Accelerate Clinical Trials in Charcot-Marie-Tooth Disease) helps support the design of clinical trials evaluating DTx-1252.

Katherine Forsey, PhD, the CMTA's chief research officer, said the CMTA looks forward to supporting the development of DTx-1252 for the treatment of CMT1. That is the most common subtype of CMT, affecting approximately 1 in every 5,000 people. "The exciting progress in the development of DTx-1252 aligns with the CMTA's mission to accelerate the development of new treatments for CMT. We look forward to sharing further updates from DTx with the CMT community," Forsey said.

UNDER THE MICROSOPE

*In “Under the Microscope,” **CMTA Chief Research Officer Katherine Forsey, PhD,** takes a closer look at issues related to the CMTA’s Strategy to Accelerate Research. Katherine is a research biologist by training and holds a doctorate in biology from the University of York in the area of expression, activity and localization of metabolic enzymes during preimplantation embryo development. She and members of her family have CMT1A. She lives in Yorkshire, England with her husband and two children.*

Antisense Oligonucleotides

ASOs (antisense oligonucleotides) are a type of medication that works by altering how specific genes in the body are expressed, or turned from DNA into a functional protein. They are short man-made pieces of genetic material designed to bind to a specific target messenger, RNA (mRNA).

ASOs can be delivered through intravenous injection, subcutaneous (just beneath the skin) injection or orally. Importantly, ASOs can cross the blood-nerve barrier following subcutaneous injection, a difficult challenge for other potential CMT treatments. ASOs are still a relatively new class of drug, and more research is needed to fully understand their safety and efficacy in different patient populations. Through our Strategy to Accelerate Research (STAR), the CMTA is investing in and supporting the development and testing of ASO-based treatments.

ASOs work by targeting the genetic mutations that cause the disease. They can be designed to either enhance the production of functional proteins that are lacking in CMT, or to block the production of abnormal or over-abundant proteins that are harmful to the nerves.

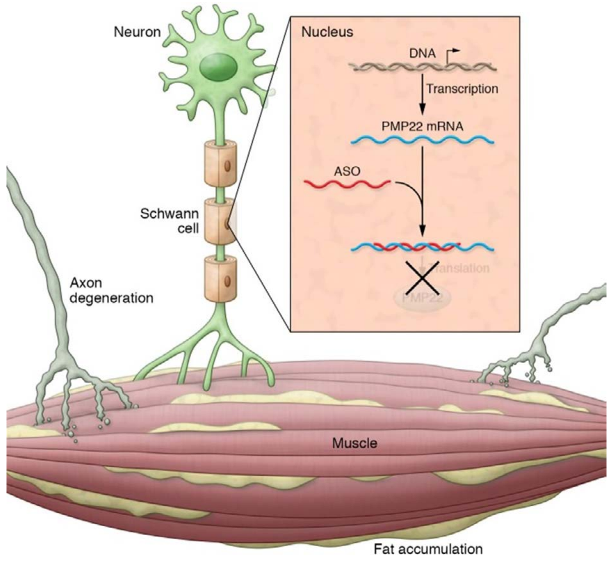
EXAMPLES OF ASOs USED TO TREAT CMT

- **In 2018, Ionis Pharmaceuticals, a CMTA Alliance Partner, showed that PMP22 ASOs reversed CMT1A features in animal models. Their results support the use of ASOs as a potential treatment for CMT1A and elucidated potential disease and target engagement biomarkers for use in future clinical trials.**
- **In collaboration with the CMTA, Shift Pharmaceuticals is currently carrying out preclinical testing of ASOs to treat CMT1A. The findings from the project will be critical in advancing research that will lead to clinical trials for CMT1A patients. See here for more on these tests.**

For example, ASOs can be designed to treat CMT1A, the most common form of the disease, which is caused by a duplication of the PMP22 gene. ASO treatment can reduce the levels of PMP22 mRNA produced by gene duplication. This can lead to increased myelination (the process by which nerve fibers are insulated), improved nerve conduction speeds and a reduction in CMT symptoms. ASOs could also potentially be used to treat several other types of CMT.

ASO therapies have been approved to treat other human diseases since 1998. They have demonstrated positive results in humans with debilitating neurodegenerative diseases such as spinal muscular atrophy (SMA) and familial amyloid polyneuropathy (FAP). Nusinersen (Spinraza), an ASO-based treatment for SMA, was approved in 2016 and 2017 by multiple regulatory authorities worldwide.

ASOs can be designed to selectively target any RNA sequence, meaning it is possible to target patient-specific mutations that are causing disease. The first custom-made ASO therapy, Milasen, was developed and used to treat a patient with an ultra-rare form of Batten disease in 2018, providing proof of concept that mutation-specific therapy was feasible, safe and effective.



ASO BINDING TO PMP22 mRNA

CMT1A damages the myelin sheath (demyelination) that over time leads to degeneration of axons and fat accumulation in muscle. ASO binding to PMP22 mRNA may prevent demyelination but may not reduce existing axonal degeneration and fat accumulation in muscle, supporting the use of ASOs as early as feasible.

CMTA FUNDS PRECLINICAL WORK ON MODULATING UNFOLDED PROTEIN RESPONSE IN CMT1B MICE

The CMTA announced a $33,000 grant on April 25 that will enable Maurizio D’Antonio, PhD, to establish if molecules that selectively activate the ATF6 pathway of the unfolded protein response (UPR) can engage targets in the peripheral nerves of CMT1B mouse models.

If so, D’Antonio and his team at San Raffaele Scientific Institute in Milan, Italy, will perform a time-course study to identify the best dosing strategy.

CMT1B neuropathy is caused by a mutation in the myelin protein zero (MPZ/P0) gene. It is one of the four most prevalent types of CMT, causing around 5 percent of cases. The mutated protein causes deficits within the myelin sheath, which insulates the nerve and is needed for the efficient transmission of nerve impulses. Retention of the mutant protein within cells and activation of cellular stress pathways such as the UPR are common disease-causing mechanisms among MPZ mutations.

D’Antonio and his team have been studying the role of the UPR in demyelinating neuropathies for many years, and the CMTA has funded previous research in this area. This has enabled the team to collect strong evidence that the UPR pathways are a good target for therapies to treat CMT1B and CMT1A.

Maurizio D’Antonio, PhD

“Modulation of the UPR is emerging as an exciting therapeutic option for many conditions, including demyelinating types of CMT,” D’Antonio said. He added, “Thanks to the CMTA funding we will establish if targeting the ATF6 pathway is a viable option for CMT1B. Positive results will allow us to quickly move to preclinical trials in different CMT1B models with selective activators of ATF6.”

D’Antonio will also explore the possibility of combinatorial therapies targeting multiple UPR pathways simultaneously, which may result in a synergistic beneficial effect. Positive results would stimulate the development of these compounds for clinical trials.

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ACCORDING TO THE DICTIONARY, an innervator is a nerve stimulator. According to the CMTA, an Innervator is an action-oriented donor who gives monthly to the organization, sustaining STAR (Strategy to Accelerate Research), our premier research initiative, as well as important community initiatives like Camp Footprint, Patient/Family Conferences and educational programs. All of these programs require steady, reliable support from committed donors. Please join the Innervators today!

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Enzo Perla
Matthew Kopecky

Wayne and Cathy Rogers—"Happy 50th Wedding Anniversary!"
Bob and Kathy Krafty
Tom and Terry Manning

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Brent Trout

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Peggy Twohig and Harry Wise—"Happy Wedding Day!"
Karen Orehowsky

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EVAN'S FAMILY ARE PROUD INNERVATORS because it helps them remain positive when it comes to Evan's future with CMT. That positivity is contagious, as seen in this picture of Evan with an award from his YMCA swim team for his dedication, grit and season-long positivity! We believe that "Hope lies in dreams, in positivity, and in the courage of those who would like to make dreams a reality!" —JIE GONG

CAMP COUNSELORS VOLUNTEER FROM SEA TO SHINING SEA

A group of veteran camp counselors known as Top Gun came together in March for a weekend of planning for the first-ever Camp Footprint West June 12 to 17.

We call them Top Gun because they are the best of the best. They've volunteered their time and energy for the youth of the CMTA for years now, never asking for money or credit. They simply show up and devote themselves to the mission. And when asked to come together for a weekend to help plan Camp Footprint 2023, they all arrived with smiles on their faces and ideas in their heads.

We spent the weekend exploring the new camp location—B'nai B'rith Camp in Otis, Oregon. Located on the banks of a gorgeous lake five miles from the Pacific Ocean, the scenery is unreal. We talked about how to run camp at the highest level of efficiency, fun and community. We worked on the camp schedule, the cabin dynamics, the camp theme and the components of family connectivity that Camp Footprint has always worked toward.

Top Gun at Camp Footprint West

The level of creativity was inspiring. These amazing volunteers are so completely tied to the success of our campers, staff and our program. Never satisfied with the status quo, this group continued to discuss and debate the past and future of Camp Footprint.

As the director, I left the weekend with two distinct feelings. First, with an optimism for this coming summer that is simply unmatched. Our program and campers are in the best of hands. And second, with an appreciation yet again for how much of this amazing organization is run on volunteer power. We wouldn't be where we are without these devoted souls. So thanks to Top Gun, the Camp Footprint staff, our Youth Council, our Advisory Board, our Board of Directors, our Branch Leaders, and the many other wonderful volunteers of the CMTA. With all of our hearts, we thank you for all that you do.

—Jonah Berger, CMTA National Youth Programs Manager & Camp Footprint Director

INTRODUCING OUR NEW GIFT-PLANNING WEBSITE

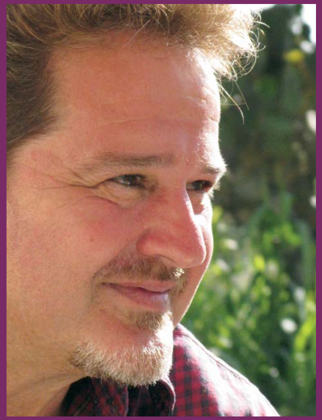
CMTA LEGACY SOCIETY

Visit our new and easy-to-use planned giving website at www.cmtausa.org/legacy to learn how you can make an extraordinary commitment to help further our mission.

To learn more or to have a confidential conversation, please call Jeana Sweeney, Chief Engagement and Gift Officer at 800.606.2682 or Email jeana@cmtausa.org

For over a decade, David Tannenbaum has answered readers' questions in *The CMTA Report*, helping them work through issues relating to living with CMT. We will miss David's gentle wisdom and wish him well in his future endeavors.

WHAT'S ON YOUR MIND? ASK DAVID.



David Tannenbaum has an LCSW degree and has been a psychotherapist in New York City for more than 30 years, specializing in helping others with the task of growing emotionally and spiritually through physical challenges. "My CMT has been my greatest challenge and my best teacher in life," says David.

As this is my last "Ask David" column, I wanted to share with readers what I have learned over the years.

Those of us who experience life with CMT have much in common. We have a shared vulnerability, never quite feeling "balanced on the ground." This lack of balance creates a feeling of insecurity simply being in the world. Even though bracing and other devices can help us feel steadier, our real work is finding balance and strength within our souls. As we reach deeper levels of acceptance throughout our lifetimes, we see how so much of our suffering comes from feeling that things should be different from the way they are. Comparing ourselves to others is a futile habit that can keep us perpetually upset.

Focusing on what we can do as opposed to our limitations can lift us out of depression. Feeling gratitude for small everyday blessings is a practice that is worth its weight in gold.

We are all one-of-a-kind souls with much to give the world. Every day is an opportunity to rise above our limitations and show others that our real strength comes from who we are—our willingness to share our story and the courage to keep going—and not in how we walk.

Maybe our suffering has deepened our compassion for others, something the world needs now more than ever. I truly believe that our CMT journey gives us an opportunity to choose how we feel about our challenges at every given moment. This doesn't mean that we have to be "positive" all the time. It simply means that after we allow ourselves to feel whatever we need to feel, we pick ourselves up and go forward. After we experience grief or loss there is often an opening of the heart to experience something new.

Connection strengthens the life in us, sometimes by discovering that others need us, other times by discovering that our love matters to someone more than we realize or that someone loves us just the way we are.

The biggest lesson CMT taught me is that my fearful thoughts are projections of a future that I don't control and that they are usually wrong. Living without fear is the secret of life. Catch those fear thoughts as soon as they arise. Just say "hello" to them and dispassionately let them leave. Come back to the present, and live your life in the best way you know!

In closing, I wanted to take this opportunity to thank the CMT community for allowing me the privilege of sharing what I have learned over the years in the hopes of helping others navigate a stony path filled with ups and downs to live as fully as possible!

Warmly,

David Tannenbaum

In Third Year, Dance 4 CMT Raises More than $30,000

HI! I AM AVA LITTLE FROM DEARBORN, MICHIGAN.
I was diagnosed with CMT1A when I was 3 years old (I'm 11 now). I also have many family members of all ages who have CMT, so it is a big part of our life.

My family and I chose to host a dance this year because I wanted to educate others about CMT, fundraise for the CMTA and have lots of fun while doing it. Since this was our first year hosting a dance, my first step was to invite my family, friends and classmates.

Next year, my family and I hope to open up the dance to other youth with CMT and their families in Michigan. Organizing the dance took some planning with my parents but it was really fun. Of course, I chose blue and orange as our dance colors and was in charge of creating an amazing playlist. Our local Jet's Pizza even provided pizza for all of our guests. Everyone was so generous in helping us raise money toward a cure for CMT. My favorite part of the night was being able to join in the Zoom and see all of the other dances that were being hosted across the nation.

Ava with her parents, Lisa and Matt, and brother Ethan

We are so proud of Ava and all of the CMT youth. It has been so inspiring to see Ava connect with other youth as they empower each other. The youth organization of CMTA was wonderful in helping us every step of the way and was always just an email away to answer any of our questions. Overall, hosting the dance was a very special and memorable experience for our family, and we look forward to Dance 4 CMT 2024 and continuing the journey toward a cure. —***AVA'S PARENTS, LISA AND MATT LITTLE***

AIR FORCE VET WITH CMT DEFIES THE ODDS THROUGH EXERCISE

Peggy Leger's trainer, Jerrica McKinley, says that when Peggy first came to her in August 2021, she was nervous about taking her on as a client. Peggy has CMT, and both of them knew there would be challenges, particularly with balance. Today, though, Jerrica says there isn't much Peggy can't do during her sessions. Her balance is much better, and she has even gained muscle mass, which is rare in people with CMT.

Peggy's CMT journey began after the birth of the first of her three daughters in 1981. She started going to the gym to lose the baby weight and noticed that she couldn't do several exercises that others could do easily. Her primary care physician ran tests but found nothing. She was in the Air Force, which sent her to a neurologist, who also ran countless tests. This time they yielded an answer—she had CMT. Peggy worked for the Air Force as an AC/refrigeration tech, and doctors feared that her low nerve conduction meant that the equipment she worked on might shock her and she wouldn't feel it. She was retrained for a new job working on computers.

During Desert Shield/Desert Storm, Peggy's commanding officer sent her to the hospital to see if she could go into combat. The C.O. kept sending her back to try to requalify. After Peggy's third trip to the hospital, she left the military.

Peggy went to the Disabled American Veterans (DAV), a nonprofit, and filed for disability. The first determination only qualified her for a small percentage of her salary, and her DAV representative encouraged her to decline the offer. She did and he refiled the paperwork. The second offer was 90 percent. She was approved for permanent and total disability in 1993 and for Social Security less than a year later.

As Peggy got older, she could see her muscles wasting away and feel her strength decreasing. She decided to start walking to try to build up her legs. It worked, and in 2015 she walked a half marathon. At her next doctor's visit, she excitedly told the doctor about her victory. The doctor threw cold water on it, telling Peggy that walking long distances could cause more harm than good and recommending that she only walk up to three miles at a time. This became Peggy's daily routine, though it felt like a defeat.

Even the three-mile walk became more difficult over time, and Peggy reached a point where she was tripping more and more and suffering more injuries. While walking one morning, she crossed paths with a friend, who recommended Jerrica as a personal trainer.

Peggy and Carrie Leger

Since starting to train with Jerrica, Peggy has increased her quality of life in ways she never thought possible. Her strength, flexibility, grip, and balance all improved, thanks to working out with weights and other adapted exercises. Even her mental health improved, raising her confidence and ability to take care of everyday activities.

In early 2023, Jerrica was looking for ways to promote her business and highlighted Peggy as one of her success stories. "I cannot accurately describe the happiness it brought me when she told me she wanted to use my story," Peggy said, adding, "I'm proud of the things I've accomplished and cannot wait to see where it goes from here."

Bringing things full circle, Peggy's middle daughter Carrie, who also has CMT, now joins her in the gym. They have a friendly competition to see who can do each exercise better.

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BRANCH NOTES


Jacksonville Branch

SAN FRANCISCO, CA

The San Francisco/Bay Area branch held a hybrid meeting on April 22 with 11 people attending in person and three people joining via Zoom. Members connected over shared experiences in their CMT journeys. They also talked about local resources and shared CMTA updates. They plan to meet regularly throughout the year.

NEWBURY, CT

The Newbury branch met on March 25 to catch up with each other and plan for future meetings and in-person gatherings. They talked about attending Boston branch meetings and the CMTA Patient & Research Summit in Boston in November.

JACKSONVILLE, FL

Fifteen members of the Jacksonville branch welcomed guest speaker Dr. Christopher Lamb to their meeting March 4. Dr. Lamb is the director of the CMTA Center of Excellence at Mayo Hospital in Jacksonville. He talked about CMT and shared information about being seen at Mayo. Attendees also shared their CMT journeys, and everyone agreed it was wonderful being back in person. The branch will meet quarterly this year.

ATLANTA, GA

More than 30 members of the Atlanta branch welcomed neuromuscular specialist Dr. Garcia Santibanez to their meeting March 25. Dr. Santibanez sees CMT patients at Emory University School of Medicine. She talked about the care her clinic provides and answered questions about CMT. Members also compared notes on their favorite gadgets to help with activities of daily living. The branch will hold another WALK 4 CMT in Atlanta this year.

CHICAGO, IL

The branch met in person on April 29 with seven people in attendance. They shared their CMT journeys, talked about local resources, planned meetings for the year, and heard CMTA updates. Branch leader Doreen Pomykala will begin offering monthly virtual visits for those interested in connecting regularly. There will also be occasional in-person meetings. The Chicago branch also met virtually on March 7 with 14 people in attendance. They welcomed guest speaker Elizabeth Meisner, PhD, LMSW, who gave a wonderful presentation titled, "Tools for Mental Health." She shared easy everyday strategies to improve mental health. The branch also did some meeting planning for the rest of the year.

BOSTON, MA

Nine members of the Boston branch met virtually March 8 to hear guest speaker Craig Zeltsar, a long-standing member of the Boston branch and recent member of the CMTA Board of Directors. He spoke about his involvement with the CMTA, as well as that of his son Evan, who is a member of the Youth Council. Vittorio Ricci was introduced as new co-leader of the Boston branch, and members talked about the upcoming Walk 4 CMT and the CMTA Summit in Boston in November.

WESTCHESTER, NY


Ashley McLeroy

The Westchester, NY Branch welcomed guest speaker Ashley McLeroy to a virtual meeting on March 4. Ashley has extensive expertise in the field of adaptive equipment and shared an informative presentation titled "Tech for Daily Living." She spoke about adaptive devices for the kitchen, bathroom, garden and laundry to help make chores easier for those with CMT. The branch will hold additional meetings throughout the year.

BUCKS COUNTY, PA

Eight members of the Bucks County branch met virtually on March 25. They spent time catching up and making meeting and fundraising event plans for the year. They shared updates and resources available in the area. They started planning for the Valley Forge, PA Walk 4 CMT to be held in September or October. They also plan to hold an in-person gathering in August.

STATE COLLEGE, PA

The State College, PA branch met virtually on March 19 with six people in attendance. They shared their CMT journeys and talked about CMT resources. They also talked about genetic testing options and challenges. Members plan to meet regularly on Zoom and may hold an in-person meeting in late summer.

Recruitment Still Open for CMT&Me Study

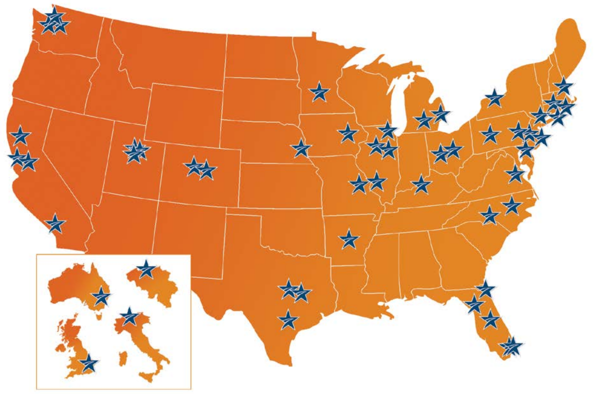
Recruitment for the CMT&Me study sponsored by Pharnext and Vitaccess—with the support of the CMTA—is ongoing. The aim of the study is to collect real-time data directly from patients, who provide researchers information on what it's like to live with CMT to help them develop better treatments.

The study also aims to find out how treatment can improve patient quality of life and slow CMT progression. Data is collected directly from patients using the app, CMT&Me, which was developed in a collaboration among scientific experts, patient advocacy groups and people living with CMT. The app is available in English, German, French, Italian and in culturally adapted Spanish language versions for both Spain and the United States.

The study currently has over 3,100 participants, with more than a third of those based in the United States. The research and data collected from the 4-year-old study has informed multiple publications and manuscripts on topics including depression, work impact and symptom burden in patients with CMT.

More information for study participants can be found at https://vitaccess.com/cmt-and-me/ and by following the CMT&Me Facebook page where data nuggets and webinars are also shared.

CMTA CENTERS OF EXCELLENCE



One of the CMTA's primary missions is improving the quality of life for those with CMT and one way we accomplish this mission is by sponsoring patient-centric, multi-disciplinary CMT clinics, staffed by some of the highest quality CMT clinicians and researchers in the world. Through these CMTA Centers of Excellence, children, adults and families affected by CMT can be assured of receiving comprehensive care by a team of CMT experts.

Although all the Centers of Excellence listed below are all comprised of multi-disciplinary teams of CMT specialists, the CMTA's Centers of Excellence affiliated with the Inherited Neuropathy Consortium or INC (marked with an asterisk), go a step further by collecting and recording genetic, biologic and other data from people with the disease.

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Clinical Director: Richard A. Lewis, MD
Appts: 310-423-4268

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Clinical Directors: John Day, MD, PhD, and Ana Tesi Rocha, MD
Appts: 650-723-0993

PALO ALTO (ADULT)
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Appts: 650-723-6469

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Clinical Director: Alex Fay, MD, PhD
Appts: 415-353-7596

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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

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Appts: Sharon McDermott, 860-612-6305

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*** Denotes the CMTA Centers of Excellence that are included in the National Institutes of Health Inherited Neuropathy Consortium (INC). The INC is co-sponsored by the CMTA and the Muscular Dystrophy Association. Worldwide more than 10,000 patients with CMT have been enrolled in protocols, and their data, de-identified to protect patient privacy, is housed in a common repository. As a result of this collaboration, a new CMT evaluation scale for children has been established, along with a new evaluation system for adults, and an infrastructure has been developed to perform natural history studies and clinical trials for CMT.**

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

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FAX (610) 499-9267

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CMT PATIENT MEDICATION ALERT:

Definite high risk (including asymptomatic CMT):

- Taxols (paclitaxel, docetaxel, cabazitaxel)
- Vinca alkaloids (Vincristine)

Moderate to significant risk:

- Amiodarone (Cordarone)
- Arsenic Trioxide (Trisenox)
- Bortezomib (Velcade)
- Brentuximab Vedotin (Adcetris)
- Cetuximab (Erbitux)
- Cisplatin and Oxaliplatin
- Colchicine (extended use)
- Dapsone
- Didanosine (ddI, Videx)
- Dichloroacetate
- Disulfiram (Antabuse)
- Eribulin (Halaven)
- Fluoroquinolones
- Gold salts
- Ipilimumab (Yervoy)
- Ixabepilone (Ixempra)
- Lefluonamide (Arava)
- Lenalidomide (Revlimid)
- Metronidazole/Misonidazole (extended use)
- Nitrofurantoin (Macrodantin, Furadantin, Macrobid)
- Nitrous oxide (inhalation abuse)
- Nivolumab (Opdivo)
- Pembrolizumab (Keytruda)
- Perhexiline (not used in US)
- Pomalidomide (Pomalyst)
- Pyridoxine (mega dose of Vitamin B6)
- Stavudine (d4T, Zerit)
- Suramin
- Thalidomide
- Zalcitabine (ddC, Hivid)

Uncertain or minor risk:

- 5-Fluouracil
- Adriamycin
- Almitrine (not in US)
- Chloroquine
- Cytarabine (high dose)
- Ethambutol
- Etoposide (VP-16)
- Gemcitabine
- Griseofulvin
- Hexamethylmelamine
- Hydralazine
- Ifosfamide
- Infliximab
- Isoniazid (INH)
- Lansoprazole (Prevacid)
- Mefloquine
- Omeprazole (Prilosec)
- Penicillamine
- Phenytoin (Dilantin)
- Podophyllin resin
- Sertraline (Zoloft)
- Statins
- Tacrolimus (FK506, Prograf)
- Zimeldine (not in US)
- a-Interferon

Negligible or doubtful risk:

- Allopurinol
- Amitriptyline
- Chloramphenicol
- Chlorprothixene
- Cimetidine
- Clioquinol
- Clofibrate
- Cyclosporin A
- Enalapril
- Glutethimide
- Lithium
- Phenelzine
- Propafenone
- Sulfonamides
- Sulfasalazine

The medications listed above are potentially toxic to CMT patients. Vincristine has been proven hazardous and should be avoided by all CMT patients, including those with no symptoms. Taxols also pose a high risk to people with CMT. The remainder of the medications listed above present varying degrees of potential risk for worsening CMT neuropathy. Before taking any medication or changing medication, all CMT patients should make sure the treating physician is fully aware of their medical condition.

WHAT IS **CMT?**

More than 3 million people worldwide have CMT. It is one of the most commonly inherited nerve disorders and affects the motor and sensory nerves.

CMT is slowly progressive, causing the loss of muscle function and/or sensation in the lower legs and feet, as well as hands and arms.

People in **all ethnic groups may be affected by CMT.**

CMT is genetic, but it can also develop as a new, spontaneous mutation.

CMT can vary greatly in severity, even within the same family.

CMT causes structural deformities such as high-arched or very flat feet, hammertoes, hand contractures, scoliosis (spinal curvature) and kyphosis (rounded back).

CMT can also cause foot drop, poor balance, cold extremities, cramps, nerve, muscle and joint pain, altered reflexes, fatigue, tremor, sleep apnea, hearing loss and breathing difficulties.

CMT rarely affects life expectancy.

Some medications are neurotoxic and pose a high risk to people with CMT, notably Vincristine and Taxols. See full list (at left) of medications that may pose a risk.

More than 100 different genetic causes of CMT have been identified.

Many types of CMT can be determined by genetic testing. Please consult with a genetic counselor (www.nsgc.org) or your physician for more information.

Although there are no drug treatments for CMT, a healthy diet, moderate exercise, physical and/or occupational therapy, leg braces or orthopedic surgery may help maintain mobility and function.

The CMTA-STAR research program and extensive partnerships with pharmaceutical companies are driving remarkable progress toward delivering treatments for CMT, bringing us closer to a world without CMT.