

THE

CMTA Report

SPRING 2019

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A Message from CEO Amy Gray



DEAR FRIENDS,

In the last issue of *The CMTA Report*, I told you about the CMTA's efforts on the gene therapy front and expressed my hope that gene therapy for CMT will become a reality in the near future. That future is now: In this issue, you'll read the exciting story of how members of the CMTA's STAR network of researchers came together to set up the first gene therapy trial for CMT2D. While the trial involves just one person—6-year-old Caroline Fletcher—the lessons learned from it could ultimately benefit other 2D patients (page 4).

We are also thrilled to announce we are co-funding a CMT Type 1 Gene Therapy Project with the Muscular Dystrophy Association. The project is geared at developing a treatment for CMT1X, and, if successful, could have lessons that apply to all demyelinating types—all Type 1s and most Type 4s (page 11).

In this issue of *The CMTA Report*, you'll also hear from two of our younger community members, Vittorio and Olivia, about their perspectives as patients living with CMT. There is valuable information about pulmonary rehab in our Ask the Expert section, and so much more, including the list of local CMTA Branches and Centers of Excellence where you can receive expert care from a CMT-trained neurologist in a multi-disciplinary setting.

Finally, we are proud to unveil in this issue our refreshed logo. As you will read on page 16, we have refreshed our visual identity as an organization to demonstrate our renewed commitment to the community. We will be unveiling other transformations later this year, including our new website. Our aim is to provide the CMT community with the most comprehensive resources possible.

Your contributions make our progress possible. Thank you for all of the ways you support the CMTA.

A handwritten signature in black ink that reads "Amy Gray". The signature is fluid and cursive.

AMY GRAY, Chief Executive Officer

WELCOME TO THE CMTA ADVISORY BOARD, KEN CORNELL

Ken Cornell, CO, is co-founder of Cornell Orthotics and Prosthetics in Peabody, Massachusetts, and the orthotist at the CMTA Center of Excellence at Massachusetts General Hospital in Boston. He has focused on managing patients with CMT for 25 years. Ken is past president of the New England Chapter of the American Academy of Orthotists and Prosthetists. He has also been an educator for dynamic carbon ground reaction AFOs and their functional benefits for patients diagnosed with CMT. With his combined experience in gait training with AFOs, he manages patients orthotically to minimize deformity and restore a normal gait pattern with improved balance and propulsion.



A CLINICAL TRIAL FOR CAROLINE



BY MARCIA SEMMES

At 6 years of age, Caroline Fletcher weighs just 20 pounds, most of it seemingly in her huge brown eyes and masses of curly brown hair. Her limbs are slimmed by the effects of Charcot-Marie-Tooth disease, which keeps her confined to a wheelchair. Like all forms of CMT, Caroline's disease is progressive. At least, it always has been. But the Fletcher family has new hope for Caroline's future. If all goes well in the next 12 months, she'll be taking part in a one-person clinical trial that could slow or stop progression of her disease, buying time for additional therapies to be explored.

Caroline has CMT2D, one of the rarest and most devastating forms, occurring in only a few thousand people in the United States. It affects her arms and legs. Unlike most other types, it also affects her breathing. That's why her grandfather, Dr. Stephen Fletcher, a pediatric neurosurgeon in Houston, began exploring treatment options five years ago. After some preliminary research on the field, Fletcher emailed Rob Burgess, PhD, at The Jackson Laboratory (JAX) in Bar Harbor,

Maine, asking if he could help Caroline. Burgess told him, "I'm a mouse guy. I'm sorry but I just do disease in mice. I don't have any clinical capacity." Burgess put Fletcher in touch with the CMTA, which led to the collaboration among the Fletcher family, the CMTA, JAX and gene therapy expert Scott Harper, PhD, a principal investigator in the Center for Gene Therapy at Nationwide Children's Hospital in Columbus, Ohio.

Harper was working on something called "knockdown" therapy. Typically, gene therapy employs a single injection of an engineered virus that contains the gene needed to correct a deficiency. Knockdown therapy, on the other hand, gets rid of the expression of the bad gene. Caroline has a de novo mutation in her GARS gene and the protein it produces is toxic. Harper told Fletcher he would look into it and in the course of his research he came across Dr. Burgess' work on mice. Some six months after Burgess first got the call from Caroline's grandfather, he got a call from Harper, who told him: "I know how to fix this and we should try it in your mice." Burgess agreed and the road to Caroline's clinical trial began.

Burgess had already developed a mouse with a mutation in the GARS gene, albeit a slightly different one than Caroline's. He set to work developing a mouse with her exact mutation. While Burgess was doing that, Dr.

Harper began designing and optimizing RNAi sequences. RNAi, which stands for RNA interference (RNAi) is a mechanism for sequence-specific gene silencing that recognizes mutant genes and targets them to be degraded. The Food and Drug Administration approved the first-ever RNAi sequence in August 2018.

The knockdown therapy—precisely an allele-specific-knockdown strategy using RNAi, processed through a microRNA shuttle vector and delivered by AAV9—worked on both mutations of the gene in mice. According to Dr. Burgess, when the mice received the therapy the day they were born, neuropathy was almost completely prevented. The benefits declined with time, he explained: The longer researchers waited to give the therapy, the less benefit was realized. But even when delivered after the onset of neuropathy, mice still showed signs of benefit. The mice did not show significant regeneration and it is unclear whether human subjects will regain function. Burgess noted, though, that unlike mice who are simply put back in their cages after receiving the gene therapy, Caroline will have additional treatments available to her, like occupational and physical therapies.

Burgess cited a number of compelling reasons for choosing Caroline for the clinical trial. One was the fact that her grandfather, who does *in utero* surgeries on fetuses with spina bifida and also does research, was a “catalyst” for the clinical trial. Another is that Caroline has a “technically amenable mutation that is easier to target,” Burgess said. A third is the severity of her case: Dr. Jerry Mendell, who assessed Caroline in his clinic at Nationwide Children’s Hospital in January 2018, described her case as being more like spinal muscular atrophy.

“It will be a race against time to see if we get it through the FDA” in time for Caroline to benefit, Burgess said. Dr. Mendell, who Burgess called “one of the leaders in gene therapy trials for children with neuromuscular diseases,” will be the principal investigator on Caroline’s clinical trial.

To get FDA approval, the researchers will have to file an Investigational New Drug (IND) application, which is a request for FDA authorization to administer an investigational drug to humans. The researchers are currently gathering materials for a pre-IND inquiry. They will have to show that the proposed gene therapy worked in their models, provide

tion. They hope to file the IND this spring.

Caroline’s trial is just the first step, though. “We got this far with a treatment specific to Caroline’s mutation,” Burgess said, noting that the RNAi sequence developed by Dr. Harper only matches Caroline’s mutation. But if this approach works for her, the knockdown approach could apply to other CMT2D cases. Providing further support for the CMTA’s decision to support Caroline’s clinical trial, the Harper and Burgess labs just received notice of an award from the National Institutes of Health for a related approach that would apply to any GARS

At the age of nine months, Caroline stopped crawling and could no longer hold things.



plans for toxicology and manufacturing and produce a clinical plan for treating Caroline.

Approval for the clinical plan in a single-person clinical trial doesn’t take as long as approval for a typical clinical trial, Burgess explained, because there won’t be lots of exclusion criteria with just one subject. In addition, the bar for toxicology is much lower because researchers won’t have to show, for example, that it’s safe for pregnant women. “The big challenge” Burgess said, “is justifying that it’s worth doing.” In general, he said, the more severe the disease is, the easier it is to make that showing. Burgess and his fellow researchers are awaiting their last few mouse model results, which they’ll then submit for publica-

tion, showing that their work has merit and longevity beyond the need to treat Caroline. If those results are positive, researchers could go on to try a “knockdown and replace” approach, which would get rid of good and bad copies of a given gene and then “replace the good stuff.” That general strategy might then apply to other types and other diseases like ALS and Huntington’s. “It’s an incremental process,” Burgess said, “and each one of these is a step forward and lowers the bar for the next step.”

Clinical trials and gene therapy approaches are expensive. If you’d like to contribute to Caroline’s clinical trial, visit www.cmtausa.org/cure-caroline/. ★

To learn more about the CMTA’s gene therapy work, visit www.cmtausa.org/research/star-gene-therapy/.

CLINICAL TRIALS: A PRIMER



THE CMTA'S STRATEGY TO ACCELERATE RESEARCH (STAR) continues to gather momentum, culminating in the clinical trials required before any drug or medical device reaches the market. CMT clinical trials currently include the two mentioned in this issue: Acceleron's phase two trial for ACE-083 and the single-person clinical trial for CMT2D.

More clinical trials are on the way. To help readers understand what's happening, we offer this primer on the Food and Drug Administration's drug approval process. For a more in-depth look, see www.fda.gov/ForPatients/Approvals/default.htm. To see what clinical trials are recruiting patients with CMT, visit www.clinicaltrials.gov.



The FDA's drug approval process involves five basic steps. The discovery/concept phase is the first, as research for a new drug or device begins in the laboratory. In Step 2, the pre-clinical phase, drugs and devices undergo laboratory and animal testing to answer basic questions about safety. Clinical trials follow, then FDA review and finally FDA Post-Market Safety Monitoring.

Clinical trials are simply studies, or trials, in humans. They occur only after researchers (or developers) complete the FDA's

Investigational New Drug (IND) process, which requires them to submit animal study data and toxicity data, manufacturing information, clinical protocols (study plans) for studies to be conducted, data from any prior human research, and information about the investigator.

While preclinical research answers basic questions about a drug's safety, clinical trials study the ways the drug will interact with the human body. They are designed to answer specific research questions related to a medical product and follow a spe-

cific study plan, or protocol, developed by the researcher or manufacturer. Before a clinical trial begins, researchers review existing information about the drug, then decide who qualifies to participate (selection criteria), how many people will participate, how long the study will last, whether there will be a control group, how the drug will be given to patients and at what dosage, how to assess the results and how the data will be reviewed and analyzed.

Clinical trials follow a standard progression, starting with early, small-scale Phase 1 studies

FAST TRACK

In cases involving serious conditions with unmet medical needs, the FDA can "fast track" the drug approval process in order to get important new drugs to patients sooner. Once a drug receives Fast Track designation, early and frequent communication between the FDA and a drug company is encouraged throughout the entire drug development and review process. The frequency of communication assures that questions and issues are resolved quickly, often leading to earlier drug approval and access by patients.

CMTA partner Acceleron Pharmaceuticals announced in November 2018 that the FDA had granted Fast Track status to its ACE-083, an injectable drug intended to increase muscle mass and strength. Commenting on the designation, Robert K. Zeldin, MD, Chief Medical Officer of Acceleron, said: "Results from our Phase 2 trials have shown that patients treated with ACE-083 experience robust increases in muscle volume. If our ongoing clinical studies show that ACE-083 also improves functional outcomes and confirm the favorable safety profile observed thus far, the Fast Track process could help us work with the FDA to deliver it to patients as quickly as possible." Preliminary results from Part 2 of the trials are expected by the end of 2019 for CMT.

lasting several months and involving 20 to 100 volunteers with the disease. Phase 1 studies are designed to assess safety and dosage. Approximately 70 percent of drugs move on to the next phase.

Phase 2 studies have up to several hundred people with the disease/condition and can last from several months to two years. Their purpose is to examine the drug or device's efficacy and side effects. Some 33 percent of drugs move on to the next phase. Phase 3 trials look at the drug's efficacy and monitor subjects for adverse reactions. Some 300 to 3,000 volunteers with the disease participate in Phase 3 studies, which last from one to four years, with 25 to 30 percent of drugs moving on to Phase 4. Several thousand volunteers with the disease take part in the Phase 4 study, which looks at the drug's safety and efficacy.

FDA REVIEW

Once the FDA receives an NDA, the review team decides if it is complete. If not, the review team can refuse to file the NDA. If it is complete, the review team has six to 10 months to make a decision on whether to approve the drug.

If the FDA determines that a drug has proved safe and effective for its intended use, it works with the applicant to develop and refine prescribing information, or "labeling." Labeling accurately and objectively describes the basis for approval and how best to use the drug.

In many cases, issues remain to be resolved before the drug is approved for marketing. Sometimes the FDA requires the developer to address questions based on existing data. In other cases, FDA requires additional

CLINICAL TRIALS FOR RARE DISEASES



Clinical trials for rare diseases, which affect fewer than 200,000 people in the United States, present additional challenges.

Populations are small, limiting opportunity for study and replication. Genetic disorders like CMT are often characterized by wide range of severity, clinical presentation and rate of progression. Rare diseases are often poorly understood and natural histories incompletely described. Diagnosis is often difficult, with years between presentation and diagnosis. Many rare diseases are serious or life-threatening, and many who have them have unmet medical needs. Regulatory and drug development precedent is often lacking, as are outcome assessment tools.

The FDA is currently funding studies on the possibility of using natural history models to augment the need for placebo arms in studies of drugs that target very rare diseases, where trial recruitment can be challenging. "We believe these important studies will provide key information about how these rare diseases develop and progress and can ultimately help in the development of models of disease behavior that can make the development process more efficient," FDA Commissioner Scott Gottlieb, MD, said in announcing the grants. "We've been working overtime to develop models that can simulate the behavior of placebo arms in the setting of very rare diseases, where recruiting for clinical trials can be especially hard."

If a drug developer has evidence from its early tests and pre-clinical and clinical research that a drug is safe and effective for its intended use, the company can file a New Drug Application (NDA) to market the drug. The FDA review team thoroughly examines all submitted data on the drug and makes a decision whether to approve it. The NDA's purpose is to demonstrate that a drug is safe and effective for its intended use in the population studied. Along with clinical results, developers must include proposed labeling, safety updates, drug abuse information, patent information, data from studies conducted outside the United States, institutional Review Board compliance information and directions for use.

studies. If the NDA doesn't contain sufficient data for the FDA to determine the safety and effectiveness of a drug, it may organize a meeting of one of its Advisory Committees to get independent,

expert advice and to permit the public to make comments. These Advisory Committees include a Patient Representative who provides input from the patient perspective. ★

CMTA-Funded Research Database for CMT2 Identifies New Disease-Causing Mutations

BY MARCIA SEMMES

While researchers have identified more than 90 mutated genes that cause CMT, approximately 50 percent of CMT2 patients do not yet have a definitive genetic diagnosis. Dr. Stephan Züchner at the University of Miami is working to change that, spearheading an ambitious project to identify new disease-causing mutations in patients seen in the Inherited Neuropathies Consortium (INC). The INC is an integrated group of academic medical centers, patient support organizations, and clinical research resources dedicated to conducting clinical research in different forms of CMT and improving the care of patients. Funded primarily by the National Institutes of Health (NIH) with supplemental funding from the Charcot-Marie-Tooth Association and the Muscular Dystrophy Association, the INC plays a key role in developing the infrastructure necessary to evaluate CMT therapies.

“The work in Züchner’s lab aligns perfectly with the CMTA’s Strategy to Accelerate Research (STAR),” CMTA CEO Amy Gray said. “Funding this project is another way the CMTA supports the development of therapies for the CMT community,” she added, explaining, “As we discover more of the genetic causes of the unidentified forms of

CMT, we can strategically target them with potential therapies.”

Züchner and the team of University of Miami and INC researchers use purpose-developed software, the GENESIS platform, to perform large-scale exome and genome analysis on the DNA of patients with CMT2 (among others), which primarily affects the axons of motor and sensory neurons. Up to half of all individuals with CMT have CMT2, or approximately 1 in 5,000 people.

For those who haven’t kept up with the rapidly changing field of genomics, determining the order of DNA building blocks (nucleotides) in a person’s genetic code is called DNA sequencing. Two other methods—whole exome sequencing and whole genome sequencing—rely on new technologies that allow for rapid sequencing of large amounts of DNA. These approaches are known as next-generation, or

Next-generation sequencing has transformed the genetics field.

next-gen, sequencing. Next-generation sequencing has sped up the process, taking only days to weeks to sequence a human genome, while reducing the cost.

With next-generation sequencing, it is possible to sequence large amounts of DNA, such as all the pieces of an individual’s DNA that provide instructions for making proteins. These pieces, called exons, are thought to make up some 2 percent of a person’s genome.



Dr. Stephan Züchner

Together, all the exons in a genome are known as the “exome,” and the method of sequencing them is known as whole exome sequencing. Because most known mutations that cause disease occur in exons, whole exome sequencing is thought to be an efficient method to identify possible disease-causing mutations. Ultimately, however, the field is gradually moving to whole genome sequencing, an approach that produces nearly 10 to 20 times more data. This is partially driven by the observation that DNA variations outside the exons can affect gene activity and protein production and lead to genetic disorders—variations that whole exome sequencing would miss. Many more patients will potentially receive a genetic diagnosis when whole genome sequencing analysis is fully established.

As Züchner explains it, “Next-generation sequencing has transformed the genetics field. We use clinical, bioinformatics, and molecular approaches to study the outcome of large scale exome and whole genome sequencing projects in pursuit of identifying and understanding the function of known and novel disease genes for neuromuscular and neurodegenerative disorders.”

The massive amounts of data Züchner and his colleagues collect goes into two user-friendly databases: the GENESIS genomics analysis platform and the Inherited Neuropathy Variant Browser, where CMT neurologists and scientists worldwide can analyze and share their CMT data with ease and add detailed clinical observations. They can also leave suggestions for new CMT genes to be added and rate the pathogenicity of any genetic variants. The project is equally

important for diagnosis recommendations and for continued discovery research. Each new cause of CMT yields important clues and potential drug targets for CMT2. Even though different genes are mutated in different CMT2 forms, there is increasing evidence of underlying linkages between diverse types of CMT2 that will enable therapies that can work with multiple forms of the disease. ★

Stephan Züchner, MD, PhD, MD (hc), FAAN, is a Professor of Human Genetics and Neurology at the University of Miami Miller School of Medicine, Chairman of the Dr. John T. Macdonald Foundation Department of Human Genetics and the Co-Director of the Hussman Institute for Human Genomics. He is also the Co-Chair of the CMT and Related Disorder section of the Peripheral Nerve Society and a member of the CMTA's Scientific Advisory Board. Dr. Züchner has been involved in the discovery of more than 50 disease-causing genes, including the MFN2 gene, which accounts for more than 20 percent of all CMT2.

ACE-083 GETS ORPHAN DRUG DESIGNATION

The FDA granted orphan drug designation to Acceleron Pharmaceuticals' ACE-083 for CMT on March 5. The FDA Office of Orphan Products Development grants this designation to advance the evaluation and development of safe and effective therapies for the treatment of rare diseases or conditions affecting fewer than 200,000 people in the U.S. Under the Orphan Drug Act, the FDA may provide grant funding toward clinical trial costs, tax advantages, FDA user-fee benefits, and seven years of market exclusivity in the United States following marketing approval by the FDA.

The orphan drug designation does not alter the standard regulatory requirements and process for obtaining marketing approval.

CMTA CENTERS OF EXCELLENCE

www.cmtausa.org/coe

CMTA CENTERS OF EXCELLENCE are patient-centric, multidisciplinary CMT clinics where children, adults and families affected by CMT can be assured of receiving comprehensive care by a team of CMT experts. The Centers roughly correspond to the 21 international sites that make up the NIH Inherited Neuropathies Consortium (INC)—a group of academic medical centers, patient support organizations and clinical research resources sponsored in part by the CMTA. The centers will become even more important as the CMTA begins clinical trials, which will depend on how much we know about the “natural history” of CMT—how different types of CMT progress over time and whether novel medications are slowing the course of the disease. Much of that information will be supplied by the Centers of Excellence.



CMTA CENTER OF EXCELLENCE

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*These Centers of Excellence are not part of the INC.

INTERNATIONAL

The Children's Hospital (Westmead, Australia).....	Dr. Manoj Menezes
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C. Besta Neurological Institute (Milan, Italy)	Dr. Davide Parryson
University of Antwerp (Edegem, Belgium)	Dr. Jonathan Baets

Monthly Giving Benefits CMTA and Donors

Two CMTA branch leaders are leading more than just their branches: They're also setting an example for the community on how to help the CMTA through monthly giving.

Scheduled monthly giving helps the CMTA plan ahead and make decisions about which research projects to fund. It also helps the giver budget and plan—and make donations before year's end, when holiday gift-giving considerations can cause a cash crunch.

Central New Jersey Branch Leader Mark Willis describes his monthly giving as “a unique situation.” His company has a huge United Way drive each October, he explains, when donations are matched by 50 percent. Employees can designate a specific charity to receive their donations, “If you choose this route,” he says, “100 percent of your donation plus the company's 50 percent match goes directly to your selected organization and not to the general United Way Community Safety Net. It is even paid in one lump sum while my deductions are spread over the whole year.”

Mark describes his monthly giving as a win/win/win: “It's great for budgeting; it's taken before I see it; and it grows to more than I gave myself! How awesome is that?”

This is not Mark's total giving to the CMTA: He does what he can to support his friends' efforts and the New Jersey Walk 4 CMT. “However,” he says, “this regular donation assures a bonus to the CMTA and makes the bulk of my giving easier and more affordable.”

Erie, PA Branch Leader Joyce Steinkamp agrees: “Monthly donations are my unwavering fight for the CMTA's mission—to support the development of new drugs to treat CMT, to improve the quality of life for people with CMT, and, ultimately, to find a cure.”

Joyce finds that “Monthly giving is the easiest way to stand up and fight, without fail, for STAR and the mission of the CMTA. I dream of a world without CMT, and I believe STAR is our best hope of getting there.”

If you want to make a lasting difference in the CMTA community, please sign up to be part of the new INNERVATOR monthly giving program and leave an indelible footprint on the lives of present and future generations.

Join now at www.cmtausa.org/monthly.

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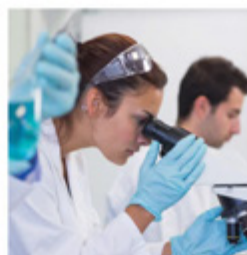
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Please sign up to be part of the new monthly giving INNERVATOR program and leave an indelible footprint on the lives of present and future generations.

When you join as a CMTA INNERVATOR, you will receive:

- A CMTA INNERVATOR welcome packet
- A one-year printed subscription to *The CMTA Report*
- Special mention in *The CMTA Report*
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Sign up at: www.cmtausa.org/monthly

CMTA and MDA Co-Fund Study On Gene Replacement Therapy for CMT1X

The CMTA and the Muscular Dystrophy Association (MDA) announced March 1 that they have jointly awarded a research grant totaling \$276,430 over three years to Kleopas Kleopa, MD, professor and senior consulting neurologist at the Cyprus Institute of Neurology and Genetics, Cyprus School of Molecular Medicine, in Nicosia, Cyprus. Dr. Kleopa is a world-renowned expert on gene replacement therapy for CMT1X, the second-most-common form of Charcot-Marie-Tooth disease.

Mutations in the gene coding for the gap junction beta-1 protein (GJB1), also known as connexin 32 (Cx32), are associated with the X-linked form of CMT (CMT1X), which affects approximately 1 in 25,000 people. Using this grant funding, Dr. Kleopa will perform critical, proof-of-concept studies to test whether delivery of the Cx32 gene using an adeno-associated virus (AAV) vector can improve symptoms in a mouse model of CMT1X as well as determine the optimal route for delivery of the therapy.

AAV vector technology has been shown to be a safe and effective delivery vehicle for “corrective” gene replacement therapy in both preclinical and clinical studies. There have been several successful applications of AAV vector gene replacement therapy for genetic diseases of the nervous system. For example, AveXis, a Novartis company, currently has a Biological License Application (BLA) under review by the Food and Drug Administration (FDA) for their gene replacement therapy for spinal muscular atrophy (SMA), which

has shown encouraging results in a Phase 1 clinical trial.

“The study is yet another step on the road to realizing our hope that in the not-too-distant future, gene therapy for CMT will be a reality beyond the lab,” said CMTA CEO Amy Gray. “This project builds upon our prior joint efforts to take gene therapy for CMT1X to the next phase, but even more importantly, if successful, learnings will be applied to all demyelinating forms of CMT (Type 1 and most Type 4).”

“Gene therapy continues to show promise for the treatment of neuromuscular disease,” said Amanda Haidet-Phillips, PhD, one of the MDA’s scientific portfolio directors. “MDA is hopeful that this new, translational work may pave the way for gene therapy to be developed for CMT and we are grateful for the partnership of the CMTA to help accelerate this important study forward.”

With previous MDA-funded support, Dr. Kleopa pioneered a gene therapy approach to treat the X-linked form of CMT, showing that a single spinal injection of the Cx32 gene was associated with production of normal protein in nerves and improvement of peripheral nerve health and motor performance in a mouse model of CMT. In a follow-up study co-funded by the MDA and the CMTA, he examined whether repeated injections in mice led to increased protein levels and tested whether treatment at later stages of the disease led to improvement like that seen for treatment in the early stages.

The target cell type for this therapy is the Schwann cell, which generates the insulating myelin



Dr. Kleopas Kleopa of the Cyprus Institute of Neurology and Genetics

sheath around peripheral nerves. The challenge for CMT1X and other demyelinating forms of CMT is optimizing delivery of the gene to Schwann cells. In previous studies, Dr. Kleopa employed a different type of viral vector to deliver the Cx32 gene, but for this new study he will adapt this approach to AAV, which has been more widely used in the nervous system and shown promise in clinical studies for other diseases. The project will test several types of AAV and different injection paradigms to determine the best method to restore the function of Cx32 in Schwann cells. Positive results may help advance development of treatments for other types of CMT affecting Schwann cells, as a similar AAV approach can be applied to CMT1A and other subtypes of CMT1.

Dr. Kleopa recently joined the CMTA’s Scientific Advisory Board, along with three other gene therapy experts, reflecting the field’s expanding importance in the search for a cure for CMT.

The MDA and the CMTA have been working together since 2016 to advance CMT research, therapy development and clinical care, and to increase understanding about the disease by improving education for children and adults affected by CMT, medical professionals, and the public. ★

QA ASK AN EXPERT

Does pulmonary rehabilitation work for patients with CMT?

My name is Aimee. I am an occupational therapist in Washington state, working with a patient who has a form of CMT with respiratory complications. My main question is: Do you know of anyone who has done any studies about simple exercises to help encourage better breathing in patients with phrenic nerve involvement? Also, I have heard that scoliosis is often associated with CMT. What does this have to do with only one phrenic nerve being involved, and are there case reports of anyone having bilateral paralysis of the phrenic nerves?

Dr. Ashraf Elsayegh answers:

Pulmonary rehabilitation has proved beneficial in patients with obstructive pulmonary disorders and restrictive pulmonary disorders. While I am aware of no studies on the benefits of exercise on neuromuscular patients, there are suggestions by multiple health-care professionals worldwide that pulmonary rehabilitation should have some benefit for this population. I have started to send some of my neuromuscular patients to our pulmonary rehabilitation center to see if and how they benefit.

One of the goals of a pulmonary rehabilitation program is upper (and lower) extremity strengthening. To exercise the remaining respiratory muscles (intercostal muscles and the diaphragm), there are commercially available devices (expiratory muscle strength trainers like the EMST and The Breather) that may be beneficial. The theory is that using these devices, which require one to breathe deeply against a pressure, will strengthen those specific muscles. However,

there are no studies that I am aware of that look at the CMT population and usage of these devices. There are a few very small studies in the ALS (Amyotrophic Lateral Sclerosis) population from which we can extrapolate some data for the CMT population. The data in ALS patients was obtained mostly from small studies (15–25 patients) in Florida. The results of the studies thus far have revealed a mild improvement in maximal expiratory pressures and a mild improvement in cough ability. The devices were well tolerated by the patients. However, further larger studies are needed before a firm conclusion can be reached. There is a larger study currently ongoing now. It is important to note that neuromuscular patients should only use these devices once a day for a total of five breaths (three to five times per week), with the device set at 50 percent of maximal expiratory pressure.

The benefits of conscious breathing—the technique of taking deep breaths such as in yoga or meditation—have not been studied. However, it falls into the same category as pulmonary rehabilitation programs: There may be some minimal benefit, but it has not been confirmed as of yet. Again from personal experience, I have noticed that it is very beneficial to overcome anxiety-related breathing issues in this patient population.

In neuromuscular disease, phrenic nerve damage is a significant concern as it innervates the diaphragm. As far as phrenic nerve paralysis, in general unilateral



nerve paralysis is secondary to trauma or surgical damage. Bilateral phrenic nerve paralysis is almost always due to neuromuscular disease. Bilateral damage is common in neuromuscular diseases such as ALS, but much less so in CMT.

Scoliosis is much more common in CMT patients. Although scoliosis rarely affects phrenic nerves, it produces its own set of breathing problems. Severe scoliosis is associated with development of restrictive lung pathology, which leads to shortness of breath and respiratory failure. Therefore, patients with significant scoliosis should be monitored carefully with pulmonary function testing for early detection of respiratory issues. ★



Ashraf Elsayegh, MD, FCCP, is an expert in the field of pulmonary medicine as it relates to neuromuscular disease and a member of the CMTA Advisory Board. He currently practices at

Cedars-Sinai Medical Center in Los Angeles and is an associate clinical professor at UCLA School of Medicine. His clinical and research interests revolve around respiratory function in the neuromuscular patient, with special interest in diaphragm dysfunction. Dr. Elsayegh has treated neuromuscular patients, including those with ALS and CMT, over the last 15 years.

From Diagnosis to Marathon to Lab in Five Months: *A Patient's Perspective*

I am getting old. I'm 21, three years into a five-year bioengineering degree at Northeastern University in Boston, and I feel old.

My back hurts, I can't keep up with all the new trends, and I've begun sighing the way my grandfather does when people get a little too chatty. I may not be forward-stooping, "walked-a-mile-uphill" old, but things are changing too fast for me to keep up with. I don't think anyone can really keep up with all of it anymore. But what keeps me filled with youthful glee are the recent events that have occurred with my CMT.

In August 2018, I visited Dr. Michael Shy's CMT clinic at the University of Iowa for the third time. I learned that his team had successfully identified the mutation that caused my formerly undiscovered subtype of CMT Type 4. They explained that a deletion of a single DNA base pair on Chromosome 15 sparked my nerve damage. Since my first visit three years ago, what little muscle that remained on my calves had been gnawed to the bone by nerve damage. Fortunately, there were few signs of the neurological damage progressing any further.

I never really cared about what type I had until I learned what type I had. A certain aura of uncertainty hung around my condition since I was diagnosed seven years ago. Like some weird four-headed thing washed up on a dirty beach, I kept asking myself "What is it?" A lot of my fear about CMT came from what I did not know about it. It was like being trapped in a dark forest: I wasn't scared of the dark, I was scared of what could be lurking in the dark.

But now I can look it in the face and see how fortunate I am. It seems to be a fairly mild type and I met another older gentleman at the clinic with the same subtype who was just as physically active as anyone else.

Less than a week after that visit to Iowa, I ran my first full marathon in Vermont. (I know what you're thinking: "Oh no, is he one of those insufferable people with a 26.2 sticker on his rear windshield now?" The answer is no: I share a car with my younger sister, who has not run a marathon, and I don't want her taking credit for my marathon while she's out and about.) I had trained for the previous five months to run 26.2 miles across Southern Vermont into Western Massachusetts. I ran in a pair of Turbomed braces and the biggest knee brace I could find on my left leg to prevent my chronic runner's knee from flaring up. Luckily, I held my body together with enough Velcro, plastic, compression tape and naiveté to get me across the finish line in six hours.

I've never craved McDonald's chicken nuggets and fries more in my entire life than when I got home after finishing. I also couldn't walk for two days after. I stepped off of a curb too quickly and it felt like every fiber in my quadriceps disintegrated down to the molecular level. I definitely see myself doing this again eventually, but I'm probably going to try to cross something else off my bucket list first.



I started a six-month internship at a biopharmaceutical company called Acceleron Pharmaceuticals in January. Located in Cambridge, Massachusetts, the company is currently in phase two trials of a muscle-growing drug that targets several neuromuscular diseases, including CMT. I'm working in the drug formulations department alongside a team of scientists to develop the proper formula to ensure the drug works as efficiently as possible. I'm just learning all of the lab protocol now, but I hope to take on a small project in the department in the coming months. It's a very odd position to be in. My job is to

I always imagined being diagnosed and hearing the news of a treatment for CMT happening over the course of my lifetime, not over five months.

(continued on page 26)

WALK THIS WAY: Morristown, New Jersey

Hosting a Walk 4 CMT creates a huge impact, generating crucial funds for research and building community. CMTA Board Member Herb Beron, a veteran of numerous CMTA fundraisers, affirms: "Bringing local communities together for a common cause is critically important. You see it with so many charities, starting at the grassroots level and building out awareness. Much of the work that happens in smaller organizations like the CMTA needs to be on a local level. Friends and family rally and turn out for CMTers in their hometown."

That's just what happened last September for the very first time in Morristown, New Jersey. Central New Jersey Branch Co-Leader Mark Willis explained, "Doing a walk was a long-time dream for me and often discussed at branch meetings. Ours is a very transient group and membership is very fluid, so finding someone to lead such an event was always problematic. My life as a person with CMT, working a wonderful but demanding job and having some serious health issues in my immediate family, just meant I could not do it all myself. Then Andi Cosby came into the CMTA and she rolled out a program and found a location. How could you say no to such energy?"

"Walks are about so much more than walking," CMTA National Events Manager Andi Cosby says. "They bring local patients, family and friends together to empower one another. Participants set their own fundraising goal, and there is no registration fee." Andi, who

joined the CMTA in December 2017 with 15 years' experience in walk campaigns, told *The CMTA Report*: "We want everyone to be able to be a part of these energizing events. The CMTA strives to provide turnkey materials for successful fundraising at all comfort levels."

In preparation for the Morristown event, the CMTA sent save-the-date postcards to CMTA members in the area. The national organization also sent emails and posted on social media to promote Morristown's Walk 4 CMT. Mark asked branch members to spread the word as well.

Participants registered at

www.Walk4CMT.org, which

they used to create their own fundraising pages to solicit donations from family, friends and co-workers. Walk leaders Lori Mattheis and Mark secured food and entertainment for all to enjoy.

The result was an afternoon of family fun with more than 100 walkers in attendance, all of them invested in finding a cure for CMT. Donuts, snacks and drinks were donated, as well as entertainment by a DJ, face painters and a balloon artist. Past Camp Footprint attendees encouraged other kids to come to camp in 2019. Newly diagnosed CMT patients were welcomed and shared experiences and information with fellow CMTers. It was a fun, moving event that generated over \$13,000 in revenue for the CMTA.

"Walks and runs can be intimidating for people with CMT, but a Walk 4 CMT is something special for us," Mark said. "The majority of the people walk just like you! Some don't even walk," he added, "They ride their scooters. This was so much more of a community than any other fundraising event I've attended."

Herb Beron, who took part in the Morristown walk, offered valuable advice about the ease of getting started as a fund raiser: "It's all trial and error. Nobody is born with the skill to organize a fundraiser, but we've all been solicited enough to know what works and what does not. Don't be shy—people in general want to give to charity, and they're most likely to give to things that have special meaning to them. Tell as much of your story as possible—again, the more you can make emotional connections with potential donors, the better. Also, always mention corporate matching programs as most big companies will match their employees' charitable contributions. And lastly, often smaller companies will want to get involved, especially if the cause is local. Knock on doors of local businesses and see if they'll sponsor the event. There's usually an opportunity for publicity ... they'll be thrilled to have their names out there."



In addition to the Morristown walk, Walk 4 CMT events were held in 30 different locations nationwide in 2018, bringing together more than 1,500 people who care about finding a cure for CMT.

In 2019, the CMTA wants to help create more opportunities for CMT patients and caregivers to come together to bond, show comradery and fundraise for the critical research that will fund treatments, and ultimately a cure. CMTA staff provide turnkey resources for anyone who wants to host a Walk 4 CMT event. There's even an online tool box with step-by-step instructions for a successful event: www.cmtausa.org/walk4cmt-toolbox. To learn more, email Andi at andi@cmtausa.org. She can help even if you're experienced at holding walks: Baltimore branch leader Clark Semmes reports that for the last Oxford "Funathlon," Andi came up with the brilliant idea of diverting the walk past a local ice cream parlor and negotiated \$2 cones with the owner.

The CMTA provides turnkey resources for anyone who wants to host a Walk 4 CMT event: www.cmtausa.org/walk4cmt-toolbox. To learn more, email Andi Cosby at andi@cmtausa.org.

Special thanks to the 31 amazing Walk Leaders who volunteered their time and efforts to make the Walk 4 CMT events possible!

Extra applause to the walks with over 100 attendees and those that raised over \$10,000!

TOP ATTENDANCE

1. Peaks Island, ME—Marie Louie
2. Chicago, IL*—Jay Pate
3. Morristown, NJ*—Lori Mattheis & Mark Willis
4. Parkland, FL*—Lara Rustici
5. Lakewood, OH—Erin Black
6. Washington, D.C.*—Maureen Dufresne & Steve Weiss
7. Los Angeles, CA*—Alani Price
8. Pittsburgh, PA—Debbie Czarnecki
9. Palo Alto, CA*—Ori Bash & Tau O'Sullivan
10. Dallas/Ft. Worth, TX—Thomas Roderiguz

TOP FUNDRAISERS

1. Palo Alto, CA*—Ori Bash and Tau O'Sullivan
2. Washington, D.C.*—Maureen Dufresne & Steve Weiss
3. Atlanta, GA—Jeannie Zibrida
4. Parkland, FL*—Lara Rustici
5. Milwaukee, WI—Frank Gess
6. Chicago, IL*—Jay Pate
7. Boston, MA—Jill Ricci
8. Morristown, NJ*—Lori Mattheis & Mark Willis
9. Los Angeles, CA*—Alani Price
10. Tampa, FL—Vicki Pollyea

*Indicates Walk Sites that made both Top Attendance and Top Fundraiser lists.





CMTA UNVEILS UPDATED LOGO

For more than 35 years, the CMTA has supported the CMT community and invested in the most promising treatment-oriented research. The CMT community has been with us every step of the way. That is why, when we embarked on refreshing our logo and visual identity, we reached out to members of the CMTA’s community for input and guidance on how best to showcase our mission, goals and the people we serve.

Community members responded loud and clear. Words like “progress” and “momentum” came up time and time again. The word “community” and the sense these members had about being a part of a close-knit family has never been stronger. One called the CMTA “a beacon of hope,” explaining, “Through STAR, we have seen so much

progress in research that the hope for a cure is real!” Another member commented, “We feel a part of a community that is actively working with the CMTA to end this disease.”

The CMTA’s new logo, which we are revealing for the first time in this issue of *The CMTA Report*, is only the third version in our 35-year history. We believe our updated look captures the progress of our Strategy to Accelerate Research (STAR) and the dedication of our unwavering supporters who have backed us every step of the way. The STAR and its arch, which have been a part of the CMTA’s logo since 2008, have been rotated forward to highlight more than a decade of progress and breakthroughs in CMT research. The STAR shines brightly, its vibrant orange representing the energy and exuberance of the CMT community.

“Through STAR and our incredibly generous and engaged members, encouraging advancements have been made in the development of treatments for CMT,” said Amy Gray, CEO of the CMTA. “Our new logo represents our promise of even more breakthroughs at an accelerated pace. We will not rest until we put an end to CMT.”

The launch of the new logo follows yet another landmark year of research progress. With more than 20 pharmaceutical and biotechnology partners, the world’s top CMT scientists collaborating through STAR, and a community powering our efforts, the CMTA is leading the way to a world without CMT.

In addition to a new logo, the CMTA will be launching a new website later this year. Stay tuned for more information in the near future. ★

LOGOS THROUGH THE YEARS

A contest for the first logo was announced in the Fall 1994 *CMTA Report* and the winning design was announced in Spring 1995 *CMTA Report*.



The original logo was replaced with a version that incorporated the star in the 2010 web redesign. The new logo appeared in the January/February *CMTA Report*.



The CMTA’s Strategy to Accelerate Research (STAR) was launched in 2008 and the STAR Logo first appeared in April 2008. The STAR Logo has been updated to a stand-alone star for 2019.



“Through STAR, we have seen so much progress in research that the hope for a cure is real!”

Lace-up High Tops (Self-Prescribed)

BY OLIVIA LONGORIA

“She walks freakishly.”

I had heard whispered variations of this statement my entire life. But hearing it from Dr. Glenn Pfeffer, a world-renowned surgeon I had driven 10 hours to meet, added an extra dose of disappointment.

I was diagnosed with Charcot-Marie-Tooth disease at age 9. It seems like most of my life has been spent traveling across the country, seeing specialists and hoping to find a solution. These trips always ended in a disappointing drive home, empty-handed.

I was always ashamed of my disease. I made sure to be last in a crowd so no one would pay attention to the way I walked. I skipped homecoming because I couldn't wear high heels. After two failed surgeries and seven broken bones, I gave up. I self-prescribed lace-up high tops to help me pick up my feet. Although this worked enough to get me through a school day without falling (most of the time), my mom never gave up the search for a solution.

One night during the spring of my sophomore year of high school, I sat in front of the computer crying tears of happiness. My mom's years of research had finally led to an answer—a foot and ankle specialist who had developed a surgical treatment for CMT. By the end of the month I had scheduled an appointment, driven to Los Angeles, met Dr. Pfeffer, and after his jarring opening statement, learned that he would change my life.

Dr. Pfeffer, a CMTA Advisory Board member, is one of the most interesting, dedicated, and genuine people I have ever met. He spoke in rhetorical questions, often



Olivia, 17, is a high school senior. She lives in El Paso, Texas, where she and her mother, JoAnn are CMTA branch co-leaders. She'll be attending Tulane University in the fall.

answering them himself. Every so often, he looked up from his glasses to ensure I was following along. I walked into his office expecting to be asked to do things I cannot do and then sent home without a treatment. But seeing “Charcot-Marie-Tooth Center” on the wall of his office gave me something I had almost forgotten how to feel: hope. During the concluding moments of my appointment, he offered me something no one else had: improvement. He told me he could get me from a D- in walking to a B+.

The next year of my life was spent in and out of the hospital,

undergoing a tendon transfer surgery in which the tendons from my toes, the only ones that work, were cut, split, and moved to the top of my foot. I also had a calcaneal osteotomy, partial ankle fusion, my Achilles tendon lengthened and my plantar fascia cut—quite the mouthful of medical terminology, I know. At the same time, I was doing my school work from home on a makeshift schedule to allow me to stay in the International Baccalaureate program while spending only 30 minutes a week with my teachers.

During recovery, I had many expectations of what would

(continued on page 27)

JIM LEA'S LEGACY

James McKenzie (Jim) Lea died in December 2016, but at the CMTA his memory will live forever. Jim, the inventor of the Therm-a-Rest self-inflating camping mattress, left a substantial portion of his estate to the CMTA, ensuring that children with CMT will grow up with the hope of a world without the disease. You could almost say that he created a cushion against the hard ground for the CMTers who come after him.

Jim was born in Tacoma, Washington, on October 22, 1920. His family loved the outdoors and regularly went camping and hiking.



Jim first noticed signs of his CMT in his thirties, but he wasn't diagnosed until his late fifties. After that, he managed it by ignoring it, working around his difficulties and getting ahead with his day. It was the same sort of can-do attitude that led him to create the Therm-a-Rest (and found an outdoor equipment company) after his 30-year career at the Boeing Company was terminated in the largest company-wide layoff in history.

The nonagenarian came late to the CMTA, attending his first meeting in Seattle in 2012. But once he was in, he was all in, spreading awareness about CMT

and distributing flyers about the disease at his 90th birthday party. Jim encouraged young people with CMT to “stay strong, accept the condition, and find alternative ways to achieve your goals.”

Jim made sure that his deep connection to the CMTA was honored when he died by including the organization in his will. Gifts like Jim's to the CMTA's Legacy Society help fund research that is already beginning to yield a harvest of promising results, with more on the horizon. ★

To learn more or to have a confidential conversation, please call CMTA Director of Development Jeana Sweeney at 800-606-2682 x106.

Plant the seeds of change and watch the chances of a cure for CMT grow.

By investing in the CMTA's Legacy Society, you can ensure that children with CMT will grow up with the hope of a world without CMT.

A bequest can be made in a number of ways:

1. You can donate a specific dollar amount or asset to the CMTA.
2. You can donate a percentage of your estate to the CMTA.
3. You can donate to the CMTA from the balance—or residue—of your estate.
4. You can designate the CMTA to receive specific assets.

Making a gift to the CMTA's Legacy Society helps fund research that is already beginning to yield a harvest of promising results, with more on the horizon. With your kindness and generosity, the promise of a brighter future is close at hand. What will your legacy be?

*With Your Kindness,
the Promise of a
Brighter Future for so many
is Close at Hand*

cmtausa.org/legacy

CMTA LEGACY SOCIETY
This is Your Legacy

CMTA
Childhood Muscle-Energy Association

To learn more or to have a confidential conversation, please call Director of Development Jeana Sweeney at 800.606.2682

SMILE AND GIVE OTHERS SUPERPOWERS

BY ADAM BELLCORELLI

As children we dream of having superpowers. We want to lift boulders with one hand and move things with our mind. We want to fly around our neighborhood or disappear from sight.

When I was a kid, I spent so much time falling down, my superhero name could have been Captain Gravity. I have CMT, which messes with the signals my brain sends to my muscles. So whether I was standing in the shower or walking down a staircase, I would abruptly experience gravity. This may seem like a terrible superpower. The “super” part was going on inside my mind. Faster than a sack of potatoes, I

This article was excerpted from a TEDxOshkosh Talk that can be viewed at www.youtube.com/watch?v=paemv2oTetY.

would accept that I was falling, then softer than a fluffy pillow, land as gently as possible. Once the falling ended, I would decide how urgent my next move needed to be, based on damage.

These sudden stops, and the increasing weakness throughout my body, led my mom and me to investigate less “leg-dependent” modes of transportation. I began using a wheelchair as a teenager, and my friends quickly saw it as an extension of me, like a new pair of shoes. I began to notice, though, that others often had different reactions, and I thought

maybe I’d found my new superpower: making people stare. My friends and I would walk and roll everywhere in town and they would ask if I noticed everyone staring at us. I’d tell them it’s because we’re awesome.

A few years ago, while being interviewed for Humans of Oshkosh, I was asked, “What’s the first thing people notice about you?” I said, “My wheels.” The interviewer pressed harder, asking, “What is it that people respond to?” “Oh, it’s my smile,” I told her, adding, “I’m addicted to people and just seeing people makes me

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Virginia, CMT Advocate

smile. People respond to that immediately. In spite of the bits of me that don't work right, I know that if I am doing okay in this moment, I can smile about life."

I developed this attitude thanks to two great philosophers in my life, Aristotle and my mom. Aristotle teaches an appreciation for moderation in all things, avoiding extremes, whether positive or negative. My mom would say, "You may have it rough; some have it better/some worse, so enjoy whatcha got." My mom also told me that having a disability doesn't mean doing less, it means working harder and doing more.

Early in my life I learned I had two gifts: a clearly visible disability and an obnoxiously optimistic attitude.

My two gifts, together, have meant that people are not intimidated by me but I have everyone's attention, even if just for a moment. Sometimes people are uncomfortable in that moment, but they are receptive. And in that moment, I am not just smiling because I am happy. I smile at people, or rather, I smile TO people, liberating fears and challenging them to embrace their power.

As a young man, I crashed my car swerving to avoid a possum. In that moment, I learned that crashing is not unlike falling, and I was good at that. I had to accept that the crash was happening, try to land as gently as possible, and make my next move, based on the damage. Once the crashing had stopped, I turned off the radio. I saw that my friend and I were knocked around but okay, and neither of us could get out of the mangled car. I looked down and my dumb right knee had whacked a piece of metal on my steering

wheel. This sliced a clean hole in my leg. I could see tendons, but there wasn't much blood.

It didn't take too long for the police and paramedics to show up, and because I'm from a small town, I knew the officer who walked up to my shattered window. He shined a flashlight in the car then on my knee. I asked, "How's YOUR night going, Pat?" He smiled, looked me in the eye and said, "Better'n yours." Sgt. Yost's smile told me it was going to be okay, so I smiled back.

**WHEN YOU GIVE
A SMILE TO
ANOTHER PERSON,
YOU CREATE
A TINY, SAFE
COMPASSIONATE
COMMUNITY...**

When you give a smile to another person, you create a tiny, safe, compassionate community in that instant. And since everyone's dealing with something, by living intentionally, every moment, by not letting a wheelchair, or

flesh wound, or a weakness of any kind slow us down, we are able to help others believe they too are powerful, in spite of their barriers.

One afternoon in college, having just finished my finals, I was eating lunch and a student stopped to talk to me. He said, "You don't know me, but you are the reason I passed my classes this semester." I must have looked surprised or confused because he went on to explain, "I'm from southern Illinois and once it got cold, I'd make every excuse to stay in my warm bed, then I'd look out the window and see you pushing your chair through the rain or snow and I'd feel silly. I don't know your name but we live in the same building, have classes in the same hall across campus, and you would show up each day with a smile on your face; so what's my deal?" That made me smile.



A smiling Adam Bellcorelli

I have no idea what path that student took after we sat and talked that day but I'm confident he's better off than he would have been if I weren't smiling in the rain. Sometimes your smile is a lifeboat, and sometimes it's a rocket ship and it is always powerful beyond measure.

In a world where you can be anything, be kind, be connected. Smile and give others super-powers. ★

Adam, 40, lives in Oshkosh, Wisconsin, with his wife and two kids, 10 and 6. He is a business consultant with Change Management Communications Center, where he teaches new managers how to better motivate their people and how to develop leadership skills.



WHAT'S ON YOUR MIND? *Ask David.*

Dear David,

I am a 40-year-old mom with CMT. I have a beautiful 15-year-old daughter who is beginning to experience some balance issues, as well as the usual foot drop symptoms. I feel blessed because she seems so well-adjusted, has lots of friends and wears her orthotics proudly. I was a mess at her age. The problem is my husband. I do most of the chores around the house and am often fatigued, but he expects me to push through it. He gets angry when I say that I am exhausted and not up to accompanying him when he feels like going out. Interestingly, my daughter doesn't have a problem standing up and saying no to him. He will say things to me like: "Stop feeling sorry for yourself," or "You're just using your CMT as an excuse to get out of going on family outings." Even friends of mine seem frustrated when I sometimes say no to participating in some social activities. No one seems to understand what it is like for me to live with CMT especially since I look "normal."



Write to David at info@cmtausa.org.

David replies:

First let me congratulate you on raising a great kid who obviously is not allowing her CMT to stop her from feeling good about herself. I understand your frustration, but unless others have walked in our shoes or "orthotics," they have no idea of the kind of fatigue we experience on a daily basis. A truly sensitive or empathetic spouse would at least try to understand and take your word for it. It is unfortunate that your husband is not in this category. I have realized over the years that it is important to let go of any guilt or shame around our own limitations. Accepting your limits helps you go beyond them. Our own self-acceptance helps us realize that we do not have to apologize to anyone for what we can or cannot do on any given day. Freedom is being able to stand up for yourself and express clearly and firmly what is okay and what makes you uncomfortable. Standing up to your husband or "friends" might sound something like: "It's hurtful that you don't

understand when I tell you I am exhausted and I wish you would simply believe me." When I feel pushed to participate or say yes when I really mean no, I am usually irritable, and not good company anyway.

It might also be true that your husband has a difficult time seeing you struggle because it brings up his fears about your CMT. Men tend to dislike and avoid feeling vulnerable, so his fears concerning your condition could manifest as anger, which he is more comfortable expressing than his worry. By the way, sometimes we simply don't feel like pushing ourselves and that's okay too. There is always a balance between participating in activities and saying no to them, but our level of activity is strictly our own decision to make. As I have said in the past, "Stand up for your right to sit down." ★

David Tannenbaum has an LCSW degree and has been a psychotherapist in New York City for the past 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.

Donate and support the CMTA in the fight against the progressive and devastating effects of CMT

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To give a gift of stock or learn about leaving a legacy gift to the CMTA, please call or email Jeana Sweeney, 800-606-2682 x106 / jeana@cmtausa.org.

Have You Seen Jeana Sweeney's New Hat?

Until recently, Jeana Sweeney was the much-beloved director of community services for the CMTA. In that role, she doubled the number of branches, helped start a camp for kids with CMT, spearheaded Patient/Family Conferences and in general made the world a better place for anyone with CMT.

Jeana is still making the world a better place for people with CMT, but she has a new hat: She's now the CMTA's director of development. That means she's responsible for everything related to raising money for the CMTA—including the major gifts program, the annual fund, planned giving, Walk 4 CMT events and national campaigns.

Jeana previously held multiple senior positions at the CMTA and brings to her role more than two decades of experience in fundraising and community service.

If you'd like to make a donation—or just compliment Jeana on her new hat—you can reach her at jeana@cmtausa.org or 800-606-2682 x106.



Don't Fear the Chair!

BY CLARK SEMMES

When my uncle fell and broke his hip at the age of 93, it fell on my aunt and my mother to care for him. While they dreaded the thought of moving him to a nursing home, they knew it was the only option. Their fear was that the loss of independence and the ability to care for himself would leave him depressed. In fact, the exact opposite happened. My uncle loved the nursing home and was only too happy to let others cook his meals and do his laundry. My mother soon added a new selection to her oft-repeated list of life lessons. This one was short and simple and went something like this: “By the time you need to go to a nursing home, you will be happy to be there.”

Recently, I have been traveling from my home in Florida to Philadelphia every three weeks to take place in a CMT clinical drug trial. While I enjoy contributing

to the search for a treatment for CMT, I do not always love schlepping through airports. In fact, I hate it. My two least favorite things in the world are walking long distances and standing and waiting. Airports require both. I hate waiting in line to be screened



by the TSA, and I really hate walking down long terminals to locate my gate. Invariably, my knees ache and my ankles are wobbly by the time I arrive.

When I mentioned this to my wife, she suggested that I ask for a wheelchair. I always knew a wheelchair was available if I wanted it, but for some reason I resisted using it. I guess I was afraid that people would think I was abusing the system. But on my last trip home from Philly, after participating in a long series of strength tests, I spotted a wheelchair stand and made a beeline towards it. It might have been the best decision I ever made.

Within minutes I was whisked through security with only a perfunctory examination of my clothing and luggage and rolled down a long terminal that had functioned as a torture chamber for me in the past. I must admit I felt a little like royalty. While others had to struggle and sweat, I barely had to lift a finger.

So now my whole attitude towards wheelchairs has changed and I have a new life lesson to pass on: Do not fear the wheelchair. By the time you need it, you will be happy it is available. ★

BRANCH LEADER SPOTLIGHT: Corey Dalfrey, Southern Louisiana

While not an overly religious person, Southern Louisiana Branch Leader Corey Dalfrey has been amazed at the many small miracles that have led him and his family to where they are today. Corey, his wife Danielle and their two sons live in Prairieville, Louisiana. The couple, who were both born and raised in the tiny town of Fordoche in South Louisiana, have been married for 19 years. Danielle is a nurse and Corey served in the U.S. Air

Force as a petroleum expert for 20 years. He was deployed all over the world, including Iraq, Kuwait, Saudi Arabia, Qatar and Oman—“everywhere but Afghanistan.” While in the military, Corey noticed that his hands were often numb and tingling, a condition that military doctors diagnosed as carpal tunnel syndrome. He wore combat boots most of the time and thinks now that they may have stabilized his ankles and prevented some of the injuries people with CMT experience.

Corey’s son Jagger, now 10, frequently tripped and fell as he was growing up. Like many people with CMT, he was labeled “clumsy.” In time his feet started to turn in. After a neurologist diagnosed Jagger with CMT, Corey had a “light-bulb moment” that led to his own diagnosis (through genetic testing) with CMT1A after more than a decade of being misdiagnosed.

Corey retired from the military in 2014 and began teleworking for the Department of Defense in Houston. When his employer told him that he had to move from Louisiana to Houston, he and Danielle decided that with Jagger’s CMT they would be better off staying put. They stayed and Corey started a small business called CD and Sons LLC that does petroleum-related surveys.

Because the closest CMTA branch was some 300 miles away, they also started the South Louisiana Branch, covering Baton Rouge, New Orleans, Lafayette, and parts of Mississippi. Jagger

had surgery on his feet in June 2018 and the Dalfreys had their first branch meeting the following month.

More than 40 people attended that first meeting, including two women who drove 2 1/2 hours from Mississippi and a family that Corey’s mother-in-law recognized from her church. In one of those small miracles that many with CMT will recognize, the family’s 16-year-old daughter also had CMT. The family was especially curious about Jagger’s surgery, and just a few months later, Jagger’s doctor performed the exact same surgery on the daughter—a midfoot osteotomy, calcaneal osteotomy, tendon trans-

fer of peroneus longus to the peroneus brevis tendon, and calcaneal osteotomy with plantar fascia release.

“Funny how things work out,” Corey says, adding, “If I hadn’t resigned, we would have moved to Houston and we would have never met

this family and possibly never changed her life either. So, our branch has not even been going for one year yet, and we have changed two young kids’ lives.”

The new branch has also raised \$1,300 selling T-shirts and taking Facebook donations from friends and family and will hold its first walk in September on the campus of Louisiana State University.

And, in another small miracle, Jagger was released from PT following his surgery and physical therapy and no longer needs to wear AFOs. ★

“...our branch has not even been going for one year yet, and we have changed two young kids’ lives...”



Rear, Talon and Corey
Front, Danielle and Jagger



CMTA Branches

Most CMTA Branches can be accessed
online at www.cmtausa.org/branches

ALASKA
Anchorage Area
Megan Rodgers
907-244-2100

ARKANSAS
Little Rock
Candice Cargile
501-516-5588

ARIZONA
Phoenix Area
Christina Fisher
623-742-8921

CALIFORNIA
Antelope Valley Area
Donna Murphy
661-317-6332
Danielle Metzger
661-317-6533
Los Angeles Area

Alani Price
310-710-2376
Sacramento
Holly Stevens
408-203-8804
Rashid Thomas
916-947-5377
Ernie Hinds
916-205-5682
Michael Huff
408-674-1281
San Diego Area
Annette Van Veen
760-473-5014
Kendall Trout
760-632-5654
South Bay Area
Ori Bash
408-829-4562
Tau O'Sullivan
916-806-2173

COLORADO
Denver Area
Ron Plageman
303-929-9647
Dick Kutz
303-988-5581

CONNECTICUT
Hartford
Roy Behlke
239-682-6785
Kait Lair
412-427-7281
North Haven
Lynne Krupa
203-288-6673

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

FLORIDA
Central Florida
Linda Davis
Mitch Davis
863-875-4239
Jacksonville Area
Tim Nightingale
904-504-1953
Stephanie Burkhalter
904-710-3771

Melbourne Area
Clark Semmes
410-350-4812
Naples
Roy Behlke
239-455-5571
Sarasota Area
Rachel Rivlin
Manuel Goldberg
941-870-3326
Tampa Bay Area
Vicki Pollyea
813-251-5512
Edward Linde
813-712-4101
West Palm Beach
Phil Lewis
561-307-0100
Eileen Martinez
561-901-5566

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Atlanta Area
Jeannie Zibrida
404-307-6519

IOWA
Iowa City Area
Jeffrey Megown
319-981-0171

ILLINOIS
Chicago Area
Jay Pate
630-888-4673
Doreen Pomykala
815-351-1328

INDIANA
Fort Wayne Area
Aimee Trammell
574-304-0968

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Kansas City Area
Tammy Adkins
314-608-6889
Aron Taylor
913-744-5674
Springfield Area
Jessica Brantner
417-468-8049
Jessica Hardy
417-434-1656

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704-904-2828
Durham Area
Jeanne Boehlecke
919-942-7909

MASSACHUSETTS
Boston
Mimi Works
617-913-4600
Jill Ricci
978-887-1014

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Baltimore
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Easton
Clark Semmes
410-350-4812

MAINE
Portland Area
Mary Louie
207-450-5679

MICHIGAN
Grand Rapids Area
Cabrielle Rudisill
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Amy Reynolds
616-916-6732

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Central Minnesota
Jo Smith
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Minneapolis Area
Duane Hodges
612-325-5448

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Kansas City Area
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Aron Taylor
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Jo Koenig
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Shelly McMahon
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Jessica Diamond
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Central New Jersey
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732-252-8299
Jacqueline Donahue
732-780-0857

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Gary Shepherd
505-296-1238

NEVADA
Las Vegas Area
Martha Boadt
231-852-4251

NEW YORK
Buffalo Area
Peter Morris
716-866-3519
Maryann Ciskal
716-435-3899
Kristen Braun
716-270-3095
Syracuse Area
Michael Casey
315-439-9325
Upstate New York
Elizabeth Misener
David Misener
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Westchester Area
Beverly Wurzel
Frank Wurzel
201-224-5795

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Jo Koenig
513-607-2822
Cleveland Area
Heather Hawk Frank
440-479-5094
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541-846-8525
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503-310-7229 (M)

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Chester County
Ashley Trout
484-364-9334
Harrisburg
Erin Weierbach
717-379-7504
Johnstown Area
J.D. Griffith
814-539-2341
Jeana Sweeney
814-269-1319
Northwestern Area
Joyce Steinkamp
814-833-8495
Pittsburgh
Debra Czarnecki
412-331-6744

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Kyle Bryant
803-378-6202
Greenville Area
Rebecca Lauriault
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Amanda Jenkins
864-313-2872

TENNESSEE
Nashville Area
Bridget Sarver
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Teresa Shoaf
615-772-8810

TEXAS
Austin Area
Nate Halk
512-415-6097
Dallas/Fort Worth
Amy DeSilva
770-826-6007
Thomas Roderiguz
817-913-8477
El Paso Area
Jo Ann Longoria
915-526-0676
Olivia Longoria
915-491-0786
Houston Area
Kristin Leard
713-516-8630
Benjy Hershorn
832-731-0121
Meredith Wells
832-264-7312

UTAH
Orem Area
Melissa Arakaki
801-494-3658

VIRGINIA
Fredericksburg
Leigh Van Doren
540-370-1968
Suffolk Area
Jordan Harness
843-303-0648

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

WEST VIRGINIA
Charleston Area
Karen McClure
304-548-4413

WISCONSIN
Madison Area
Debi Weber
608-712-8709
Milwaukee Area
Lois Hawkins
414-249-0390

CANADA
Southern Ontario
Kelly Hall
519-843-6119
Toronto Area
Linda Scott Barber
416-997-5084

Interested in starting a branch in your area?

Contact CMTA Director of Community Outreach
Laurel Richardson at laurel@cmtausa.org.



CMTA BRANCH NEWS

MELBOURNE, FL

On November 17, 2018, the Melbourne, FL CMTA Branch had a great meeting featuring guest speaker Bethany Meloche. The meeting was held in the Aquarina Beach Club overlooking the ocean, and Bethany joined in from London via FaceTime. A lot of great CMT tips were shared, including using KT tape for ankle support (available at Marshalls) and the NuStep recumbent cross trainer for exercise.

★

TAMPA BAY AREA, FL

Two dozen people turned out for the Tampa Bay Area's winter 2019 branch meeting, including one new member and two snowbirds who migrate to meetings every winter. Plans for 2019 include the 5th Annual Walk-n-Roll Picnic and Fundraiser 4 CMT. Members also talked about upcoming community events at the CMTA, shared research updates, and shared resources with the group.

★

SPRINGFIELD, MO

Laurel Richardson, CMTA Director of Community Outreach, attended the Springfield, MO CMTA Branch meeting on February 23. She shared STAR research updates and information on upcoming community programs with the 18 members present. She also talked about how to be "in communication" with the CMTA. The meeting was a classic example of CMTers helping fellow CMTers with best practices for navigating life with CMT!



Springfield, MO Branch members

CHARLOTTE, NC

The Charlotte branch invited all North and South Carolina branches to join a conference call with Dr. Rebecca Traub on January 27. Dr. Traub is the clinical director at the new CMTA Center of Excellence at the University of North Carolina in Chapel Hill. She shared a neurological perspective on CMT and provided updates on recent research advances, including current clinical trials. She also talked about the new COE and the services it provides. While a conference call is not a typical branch meeting format, it was very well received.

★

RESEARCH TRIANGLE AREA, NC

Members and spouses of the RTP (Raleigh/Durham) Branch gathered on February 16 to talk about the mobility challenges that CMT poses while traveling. The guest speaker was Steve Tuten, CEO of Southern Leisure Tours, who has years of experience taking seniors on tours, sometimes with canes, walkers or wheelchairs. Two women with walkers went with him to Iceland. Steve suggested a number of workarounds for travel challenges and encouraged branch members to take trips, even if they cannot do everything the group does. Members also shared tips and experiences from travels near and far.

★

CINCINNATI, OH

Twenty people turned out on a cold Saturday morning in January as the Cincinnati branch welcomed guest speaker Jonah Berger. Jonah, a CMTA Advisory Board member and director of Camp Footprint, shared inspirational stories of his life with CMT. His stories resonated with everyone, reminding them that they are not alone on their CMT jour-



Jonah Berger poses with Cincinnati Branch Co-Leaders

neys. Branch members "highly recommend" Jonah's book, "He Walks Like a Cowboy."

★

COLUMBUS, OH MINI-CAMP REUNION

On Saturday, January 26, a group of Camp Footprint campers and counselors led by CMTA Camp Director/Youth Leader Jonah Berger, came together in Ohio for a mini-reunion. Campers and counselors bowled, ate, played games and laughed the afternoon away. Branch leader Jess Diamond planned the event for Camp Footprint family members, who joked that they had never seen each other in heavy coats before. It was a great reminder of how amazing the Camp Footprint family is and how much fun is had any time you get the tribe of the funky feet together!

★

NASHVILLE, TN

The Nashville branch welcomed Dr. Ryan Castoro to its February 23 meeting. He discussed the strength study at Vanderbilt that is currently recruiting CMT1A patients. Penny Powers offered a physical therapy perspective on CMT and branch leaders shared CMTA updates, including news of three Patient/Family Conferences in 2019. The Nashville branch earned a "pizza party" for its amazing fundraising efforts in 2018 and will have that celebration at a park in May. Members also starting talking about their upcoming Walk 4 CMT, which will take place on September 21 at Long Hunter State Park.

VITTORIO

(continued from page 13)

prepare the drug to reach potential patient prospects while simultaneously being part of that pool. It's an amazing opportunity. My original plan was to graduate and work in prosthetic development. However, having some great opportunities to work in cell and protein development, I am prepared to take any challenge that comes my way.

Not long after being diagnosed with CMT and starting

high school, I remember walking through a hallway thinking, "I bet in a few years, I won't even notice the leg braces at all." That wasn't true. I still notice them and other people do too. I'm playing with the same hand I was dealt at the start. But even pocket deuces can turn into a winning hand. I was officially diagnosed, ran a marathon, and then earned an internship developing a potential treatment all within a few months. I could chalk all that up to work ethic

(if I were selfish), an amazing support system (if I were selfless), or luck (if I were honest) but I don't really want to deconstruct it. I want to enjoy it.

Things are moving very quickly. I always imagined being diagnosed and hearing the news of a treatment for CMT happening over the course of my lifetime, not over five months. It can be overwhelming, but I look forward to being overwhelmed if it means this kind of progress for me and everyone with CMT. ★

STUDENTS AT BOSTON COLLEGE HIGH SCHOOL RAISE FUNDS FOR THE CMTA

A mite box is a box used to collect coins for charitable purposes. Students at Boston College High School in Boston, Massachusetts, use a mite box to take weekly collections for local, national, and international nonprofits, a sign of the school's commitment to the world beyond its doors and an effort to be ever mindful of those in need. On January 3, the students of Boston College High School collected \$1,000 for the CMTA.



OLIVIA

(continued from page 17)

change. Would people notice? Would I be more confident? The results that came with the tremendous physical changes didn't have anything to do with people noticing I didn't "walk weird" anymore. They had everything to do with me confronting the internal battle I had kept hidden my whole life. The more physical progress I made, the more I learned about myself. When you're bedridden, on heavy narcotics for pain, and need help putting your clothes on, it's very easy to get trapped in frustration. Being forced to ask for help allowed me to finally gain what I was lacking in understanding. That took more of a toll than any broken bone or operation ever could.

Over the course of a year, I went through a physical transformation of getting new feet and learning to walk again. I also

traded shame and embarrassment for empathy and acceptance. I found purpose in understanding that my condition makes me who I am. I've even become the co-leader of the El Paso CMTA Branch, which did not exist in our city until this year. Most importantly, I've learned that it wasn't the operation that gave me confidence. The surgical gift of new feet was only a vehicle for being able to feel at peace with myself.

While I'm thankful for their support in my first 16 years of life, I no longer wear my lace-up high tops. I retired them

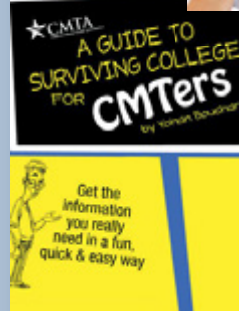
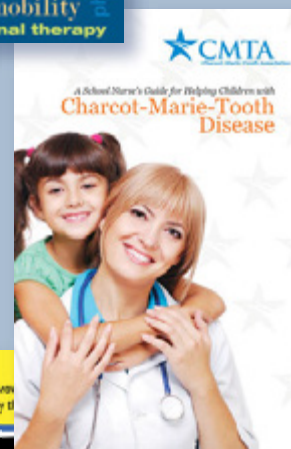
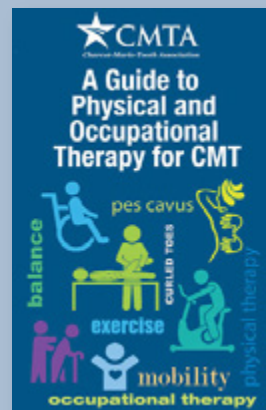
to a shelf at the top of my closet to make room for the new kinds of shoes I am now able to wear. My experience taught me I can overcome any obstacle—and use it as a platform to make a difference. I no longer look down at my Converse All Stars and worry about falling, I look up in front of me, excited for the future. ★



Olivia's former kicks

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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 (Erbix)
Cisplatin and Oxaliplatin
Colchicine (extended use)
Dapsone
Didanosine (ddI, Videx)
Dichloroacetate
Disulfiram (Antabuse)
Eribulin (Halaven)
Fluoroquinolones
Gold salts
Ipilimumab (Yervoy)
Ixabepilone (Ixempra)
Leflunomide (Arava)
Lenalidomide (Revlimid)
Metronidazole/Misonidazole
(extended use)
Nitrofurantoin (Macrochantin,
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-Fluorouracil
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)
α-Interferon

Negligible or doubtful risk:

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



The Charcot-Marie-Tooth Association
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WHAT IS CMT?

- ▶ More than 2.8 million people worldwide have CMT, which 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.
- ▶ Men and women 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's 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.