

THE

CMTA Report

SUMMER 2019

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IN THE
FAMILY

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OUR 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.
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WE ARE FAMILY...



DEAR FRIENDS AND FAMILIES,

In many ways, CMT is a family affair: When one person has it, the whole family is affected. Ideally, the family gets involved, researching doctors and devices, providing caregiving and moral support and raising funds for CMT research. That's why we decided to take a look in this issue at the many different branches of the CMTA family tree—the patients, the families that support them and the doctors who treat them while looking for a cure.

We're thrilled to feature a family that has made the search for a cure for CMT2 its mission and is challenging others to match their commitment. You can read about Bob and Gail Buucks' path to giving and their \$1 million challenge grant in a new feature we're calling "Why We Give" on page 14.

In many families, multiple generations have CMT. Except for cases of spontaneous mutation, CMT is an inherited disease. The CMTA Facebook page recently exploded with comments over whether members of the same family can have different types. Suffice to say: opinions differed. But the question is one of fact, not opinion, so we asked genetic expert (and CMTA Advisory Board member) Carly Siskind for a definitive answer on page 5.

Facebook ignited another controversy among members of the CMTA last spring. Television psychologist Dr. Phil claimed that an able-bodied woman dating a disabled man can be his lover or his caregiver, but not both. That seemed a little oversimplified, so we asked our own expert, psychotherapist David Tannenbaum, CMTA Advisory Board member and author of the "Ask David" column in *The CMTA Report*, what he thinks. You'll find his opinion on page 17.

We're also pleased to spotlight a family with multigenerational CMT in a piece by wise-beyond-her-years Marlyce Hunsaker, 15, who tells us why she believes she has been blessed to have a family that has CMT—and how the friends she made at Camp Footprint last year have become a second family.

Finally, because three of the doctors on the CMTA Board of Directors are such important members of our CMT family, we took a look at the men behind the lab coats. You'll learn who used to drive a taxi in Philly, who is an amateur geologist and who's a Star Trek fan on page 8.

We hope you enjoy reading about members of the CMT family. If you'd like to meet some of them, make plans to attend one of our next "family reunions," aka CMTA Patient/Family Conferences, either in St. Louis on Saturday, September 7 or Atlanta on Saturday, November 2. Hope to see you there!

Sincerely,

A handwritten signature in cursive script that reads "Amy Gray".

AMY GRAY, Chief Executive Officer

ALL IN THE FAMILY

BY MARLYCE LONGHURST

My family has had CMT for generations.

Six people in my family have CMT, starting with my great grandmother Jackie, then my grandfather Mont, my uncle Branch, my uncle Keelen, and my mother. CMT goes back so far that we aren't even sure where it originates. I inherited CMT from my mother, who inherited it from her father, Mont Hunsaker. He inherited it from his mother and

so on. CMT has become a normal thing for my family, so we know how to adapt to it. We all get our orthotics from Hanger Clinic, mostly because my mother Amberly and my uncle Branch both work for them. [Branch Hunsaker is the West zone recruiter for Hanger while Amberly Jensen is an office administrator in Hanger's Murray Utah, clinic. Branch was the CMTA's "I'm a Star" Winner in 2015 and his wife Emily is a former branch leader.]

While we all have CMT 1A, we have different experiences. Over the years, medical advances have led to new and different treatments, so even though my mother and I have the same type of CMT, we have different abilities. When my mother had her surgeries, medical technology was not as advanced, which is why she isn't as functional as my doctors hope I will be after my surgeries.

One of the best benefits that I have gotten from CMT was my experience at Camp Footprint. I met so many wonderful people and learned so much about CMT and how other people live with it. At Camp Footprint, I went rock climbing, ziplining, boating and paddle boarding, and I learned archery. Even though it has almost been a year since I was last at camp, I remain in contact with many of the fabulous people that I

met there. This August, I will be attending camp for the second time. I am extremely excited because being around so many people who understand my situation helps me cope with it. I make so many wonderful friends and memories when I am with them.

Most people I have met who have CMT have different styles of AFOs. Some of us have patterns

on ours, others just have plain white ones or even metal. But we all understand each other's situation and we are always there for each other. I have been blessed with a family who has CMT. They have all been there for me through all the hard times and the times when I had no confidence. When I first

started wearing AFOs, I was ashamed of them. All I wanted was to make it all go away. I hid my condition and my AFOs and tried to act as if they weren't even there. But I know now that CMT is not something to be ashamed of. It is part of who I am. I may have CMT for the rest of my life, and it may be challenging at times, but I know that I am not alone in facing this. I know that if I put my best foot forward, I can lead a happy and successful life, CMT and all. ★

Marlyce, 15, had five surgical procedures on her feet on May 30, including plantar release, tendon transfer and muscle lengthening.

"I have been blessed with a family who has CMT. They have all been there for me through all the hard times..."



Three generations of CMT: (left to right) Branch, Mont, Marlyce and Amberly

WE ARE FAMILY: *BUT DO WE HAVE SAME TYPE OF CMT?*

BY CARLY SISKIND, MS, CGC



Genetic testing has come a long way in the last decade. We went from testing one gene at a time for \$1,000 apiece to being able to test for 40 to 100 genes for ... well, that depends on your insurance, but much less money. Overall, this is a huge advancement in genetics, family planning, community building and finding answers, but there are some issues that arise while looking at all of these genes.

Let me back up and give my basic primer on genes. We have ~20,000 genes in the body, of which we have two copies, one from mom and one from dad. Each gene has a specific function in the body. Genes are made of DNA, which is essentially a code—a series of letters that, when spelled correctly, let the gene do its function. The letters of the code are A, C, G and T. They are arranged in a specific order in the gene. People can have CMT when there is a change in the gene—if a C becomes a G, if a couple of letters are missing or if a whole gene is duplicated or missing.

There are currently about 100 genes that are known to cause CMT when there are one or more changes in the gene. There will probably be 100 more genes found to cause CMT before everyone has a genetic diagnosis. Right now, about 80 percent of the time people with demyelinating forms of CMT will get an answer through genetic testing. That number is only about 40 percent for people with axonal forms of CMT.

When doing genetic testing, there are three possible outcomes:

- **Positive:** there was a change in the gene that is known to cause CMT.

- **Negative:** there were no changes found in any of the genes.
- **Variant of uncertain significance (VUS):** a change was found in the gene, but it is unknown if it is disease-causing or part of normal human variation.

Coming back to my point about there being issues that can come with looking at all of these genes—the more genes that are sequenced, the more likely it is to find one or more variants of uncertain significance. A VUS is not a diagnosis—there is not enough information to say if it is disease-causing or not. If a person has a disease-causing variant in

(continued on page 16)

CMTA Board Funds Gene Therapy Trial for CMT4C, Study on Wearable Sensors for Clinical Trials

The CMTA Board of Directors voted at its March meeting to award \$120,000 to Dr. Kleopas Kleopa to develop a gene therapy for CMT4C, a demyelinating neuropathy caused by autosomal recessive mutations in the *SH3TC2* gene, expressed specifically in myelinating Schwann cells.

Dr. Kleopa recently showed that a lentiviral gene delivery by intrathecal injection can provide a partial therapeutic benefit in a CMT4C model. In this two-year project, Kleopa will explore the possibility of delivering



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

the therapeutic *SH3TC2* gene by a clinically translatable adeno-associated virus (AAV9) vector. The new work will test this approach in the *Sh3tc2*^{-/-} mouse model of CMT4C at early and late stages of neuropathy. The study will

provide the proof of principle required for clinical translation of CMT4C gene therapy.

The CMTA's scientific reviewers said the proposal could provide a potentially translatable therapy for CMT4C and that, if successful, the CMTA would likely grant additional funding to develop the next steps, such as toxicology and biodistribution.

They noted, however, that the study is an early investigation and that a pathway of commercialization (and therapeutic use) was not yet detailed.

Market potential for adoption is low, proposal reviewers said,

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

Cedars-Sinai Medical Center (Los Angeles).....	Drs. Robert Baloh and Richard Lewis
Children's Hospital of Philadelphia (Philadelphia).....	Dr. Sabrina Yum
Children's Hospital of Pittsburgh (Pittsburgh)*.....	Dr. Hodas Abdel-Hamid
Connecticut Children's Medical Center (Farmington).....	Dr. Gyula Acsadi
Hospital for Special Care* (New Britain, CT).....	Dr. Kevin J. Felice
Johns Hopkins University (Baltimore).....	Dr. Thomas Lloyd
Lucile Packard Children's Hospital at Stanford (Palo Alto).....	Drs. John Day and Ana Tesi Rocha
Massachusetts General Hospital (Boston).....	Dr. Reza Seyedsadjadi
Nationwide Children's Hospital (Columbus)*.....	Dr. Zarife Sahenk
Nemours Children's Hospital (Orlando).....	Dr. Richard Finkel
Northwestern Memorial Hospital (Chicago)*.....	Dr. Daniela Maria Menichella
Ohio State University (Columbus).....	Dr. Amro Stino
Oregon Health & Science University (Portland)*.....	Dr. Chafic Karam
Rush University (Chicago)*.....	Dr. Ryan D. Jacobson
Stanford Neuroscience Health Center (Palo Alto).....	Dr. John Day
University of Florida (Gainesville)*.....	Dr. James Wymer
University of Illinois at Chicago (Chicago)*.....	Dr. Charles K. Abrams
University of Iowa (Iowa City).....	Dr. Michael Shy
University of Miami (Miami).....	Dr. Mario Saporta
University of Michigan (Ann Arbor)*.....	Dr. Dustin Nowacek
University of Minnesota (Maple Grove).....	Dr. David Walk
University of Missouri (Columbia).....	Dr. Raghav Govindarajan
University of North Carolina (Chapel Hill)*.....	Dr. Rebecca Traub
University of Pennsylvania (Philadelphia).....	Dr. Steven Scherer
University of Pittsburgh Medical Center (Pittsburgh)*.....	Dr. Sasha Zivkovic
University of Rochester (Rochester, NY).....	Dr. David Herrmann
University of Texas Southwestern (Dallas)*.....	Drs. Susan Iannaccone and Diana Castro
University of Utah (Salt Lake City).....	Dr. Russell Butterfield
University of Washington (Seattle).....	Dr. Michael Weiss
Wayne State University (Detroit).....	Dr. Jun Li

CLINICAL DIRECTOR

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

INTERNATIONAL

The Children's Hospital (Westmead, Australia).....	Dr. Manoj Menezes
The National Hospital for Neurology & Neurosurgery (London, England).....	Dr. Mary Reilly
C. Besta Neurological Institute (Milan, Italy).....	Dr. Davide Pareyson
University of Antwerp (Edegem, Belgium).....	Dr. Jonathan Baets

THE CMTA FUNDING PROCESS

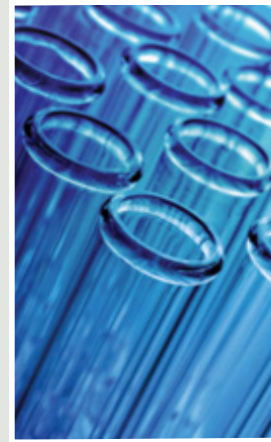
The CMTA Board does not accept unsolicited proposals, but it occasionally seeks out targeted proposals in specific disease areas, then employs a protocol aimed at reaching a funding recommendation within six weeks. Under that protocol, STAR Advisory Board members, including both Scientific Expert Board members and Therapeutic Expert Board members, conduct a first-level scientific review on translational projects brought to, or identified by, the CMTA. The initial pre-proposal is limited to three pages and must include specific aims.

STAR Advisory Board members then either reject the proposal or ask for a full proposal from the investigator, including a detailed budget. Applicants whose projects are green-lighted for a full proposal then have two weeks to submit it. All full proposals must include a research plan with preliminary data, budget, timelines and information about the investigative groups involved.

Once a full proposal is received, the Scientific Expert Board evaluates its scientific merit and the Therapeutic Expert Board confirms that there is a therapeutic opportunity/asset that will benefit the CMTA STAR therapy development mission. Each advisory board review is completed in two weeks or less, then both recommendations go to the CMTA Board of Directors.

Project proposals are scored in two stages, the first emphasizing scientific excellence and the completion of milestones and the second evaluating the translational and commercialization potential of the project. To read more about the scoring criterion, visit www.cmtausa.org/our-research/for-researchers/funding/.

At the end of the six weeks, the CMTA Board of Directors either approves or rejects the project. When a proposal is funded, the CMTA documents, agrees and signs off on the research plan with the research investigator's institution.



given the small patient population and anticipated high cost. This may limit use to academic proof of concept trials unless a common vector with adaptability across Schwann cell disorders can be developed that solves the issues of efficiency in delivery and expression. Studies targeting the cross-CMT adaptability of gene delivery and cell-specific expression are to be encouraged, they noted.

THE WEARABLE SENSOR STUDY

Also at its March meeting, the CMTA Board awarded \$109,799 for a one-year study that will examine the use of wearable sensors to measure outcomes in clinical trials involving individuals with CMT1A. The sensors will be tested for reliability, validity and responsiveness.

The sensors were recently used in a study that successfully

used anti-sense oligonucleotides to knock down peripheral myelin protein 22KDa (*PMP22*), substantially reversing the phenotype of the rat and C22 mouse models of CMT1A, including measures of strength and balance (hind limb grip strength and time on the rotating rod). The ability to measure changes in strength, motor function and balance over short periods of time in early-phase human trials is essential in identifying potential therapeutic agents. This study will provide the initial data regarding sensor-derived parameters that is necessary to plan a larger multisite study to develop this outcome measure for future clinical trials.

The board voted to commit funding to Phase 1 of the proposed study. A second phase to evaluate outcome measures over a one-year period (requiring funding of \$85,946) will be considered

later if the Phase 1 study demonstrates clear and useful results.

Both of the CMTA's scientific reviewers found significant merit in the study's potential to advance the ability to refine functional outcome measures on an individual patient basis. There is significant research potential to begin translating findings from working with CMT animal models at a preclinical level to a comparable level of sophistication in patients. Patient heterogeneity will be a confounding factor, the reviewers said, with a relatively small patient group (appropriate for a first pilot study) selected broadly from a wide age range, but similar level of disease burden. Reviewers expressed "some confidence" that the investigators have established a track record of work and funding in other neurological movement disorders on this approach. ★

THE DOCTORS

IN THE 19TH CENTURY, the three doctors most closely associated with CMT were the ones who gave it their names: Drs. Jean-Martin Charcot, Pierre Marie and Howard Henry Tooth. In the 21st century, three of the doctors most closely associated with CMT are on the CMTA's Board: Drs. Steven Scherer, Michael Shy and John Svaren.

Many know them by name or by reputation; some are fortunate enough to be their patients. While the CMTA has a growing army of researchers trying to find treatments and cures for CMT, the three Dr. S's are the generals. They lead the fight. Along with other board members, they decide which research projects to fund.

Most of those in the CMT community know the three men's professional qualifications. Their CVs each run in the double digits and collectively they have well over half a century of CMT research experience. But who's behind the lab coats? Clark Semmes takes a look.



Dr. Michael Shy

Mike Shy grew up in Bethesda, Maryland, where his father was an academic neurologist at the National Institute of Neurological Disorders and Stroke (NINDS).

The family later moved to Philadelphia, where his dad served as the chair of the Neurology Department at the University of Pennsylvania. Mike's mother was a pianist who left the field to raise her two children.

Mike attended Harrington High School in Philadelphia, where his main hobbies were playing baseball and trying to get a date. He went on to major in history at the University of Pennsylvania. Mike's father died of a heart attack at the age of 47 while Mike was still in college. Post-college, Mike worked as a Volunteer in Service to America (VISTA) in northwest Minnesota, a cab driver in Philadelphia and a preschool teacher before attending medical school, first at the Free University of Brussels and then at the University of Albany.

Dr. Shy first met Steven Scherer when they worked

together at a UPenn lab in the early nineties. In 1996, Mike left Philadelphia for Wayne State University in Detroit, where he opened the first clinic for CMT patients in the country. Today Mike runs the CMT Clinic at the University of Iowa. His wife Rosemary, a pediatrician, also works at the clinic.

For Dr. Shy, one of the most rewarding aspects of his job has been the fulfillment of the hope from the nineties that researchers would find the causes of CMT. Today, more 100 genes causing CMT have been identified and clinical trials are underway that will eventually lead to a treatment.

While researchers can be very competitive, Mike is happy to report that the researchers working on CMT are a unique group of excellent collaborators and close friends.



Dr. Michael Shy

Dr. Steven Scherer

Steve grew up in the small town of Milford, Michigan, where his father was a public school teacher who eventually became superintendent of schools. As a child, he loved watching University of Michigan basketball games on television, and he can still name many of the U of M players from that era. From a tender age, he knew he wanted to attend the University of Michigan.

At the U of M, Steve majored in zoology. He first learned about CMT in grad school when he discovered a textbook called “Peripheral Neuropathy” that captured his imagination. Steve went on to attend the University of Michigan medical school and in 1985 he completed his studies with a combined MD/PhD.

Dr. Scherer was a neurology resident at the University of Pennsylvania when he met Dr. Mike Shy. They shared an interest in CMT and other hereditary neuropathies. Since then they have worked together on numerous projects and papers.

Dr. Scherer first heard about the CMTA in the early 1990s, when it was headquartered close to his home. Back then, he says, the CMTA was small; he got involved with the hope of bringing more rationality and science to the organization. He has since witnessed the CMTA grow exponentially and assume a leadership role in the CMT community. Steve points to the creation of the INC database, which organized the identification of patients and collection of data about them, as a major milestone.

Steve’s hobbies include geology, bicycling and gardening with his wife, DeAnn, with whom he will celebrate 38 years of marriage



Dr. Steven Scherer

this summer. They have four grown children, two girls and two boys. Steve will turn 64 next March, but he has no immediate plans to retire, though he has promised DeAnn he will wind up the bulk of his work by the time he reaches 70.

Dr. John Svaren

John Svaren is from the tiny town of Arlington, South Dakota, population <1,000. He is the youngest of six children born to a

Lutheran pastor father and school teacher mother. In a small high school where talent was not necessarily a prerequisite, John played football and basketball and ran track. Off the field, he played tuba in the school band and sang in his church choir.

John met his wife Laura at summer camp when he was 18 years old. They have been married for 33 years and have two children, a son in college and a daughter in high school.

John majored in biology at Augustana College in Sioux Falls, South Dakota, then attended grad school at Vanderbilt University in Nashville, where Laura taught high school math.

While the CMTA has a growing army of researchers trying to find treatment and cures for CMT, the three Dr. S’s are the generals.

John first became interested in CMT while training as a post-doctoral researcher at Washington University in St. Louis. He later joined the faculty at the University of Wisconsin in Madison and started working on STAR projects with Mike Shy. After being named to the CMTA Board of Directors, he went on to chair the CMTA’s Scientific Advisory Board, which provides input and oversight for research projects. He thinks the CMTA has done a great job of energizing and engaging the CMT community.

John says he is excited that CMT research has advanced to the point that clinical trials have been initiated and others are starting in the near term—and the fact that researchers have developed good biomarkers for those trials. While he originally thought that every CMT subtype would require a separate therapy, he now believes that significant crossover potential exists. Some of the more broadly applicable advances include gene therapy—such as the CMT4J trial taking place next year—and the potential to prevent the nerve degeneration that is the cause of progression in CMT.

A Star Trek fan, John enjoys biking, swimming and reading about history. —Clark Semmes



Dr. John Svaren

Detroit Patient/Family Conference Draws More Than 100

More than 100 people gathered in Detroit April 27 for the CMTA's latest Patient/Family Conference. Dr. Jun Li, PhD, clinical director of the CMTA Center of Excellence at Wayne State University, spoke on the "Clinical Presentation, Diagnosis and Management of CMT." He was followed by genetic counselor Karen Krajewski, who shared detailed information about genetics and genetic testing.

CMTA CEO Amy Gray presented a STAR research update detailing some of the exciting developments happening now and on the horizon. Pulmonologist Dr. Safwan Badr shared vital information about "Respiratory and Sleep Disorders in CMT," and neurologist Dr. Maher Fakhouri addressed the very important topic "Pain Management in CMT." Attendees also heard from therapeutic experts in the areas of physical therapy, orthotics and physical therapy research.

As usual, attendees also had the chance to meet, mingle with and question the presenters.

While their parents took part in the Patient/Family Conference, a group of teens ventured out for a day of adventure (and eating) with Youth Leader Jonah Berger. They started at the Aquarium at Belle Isle, an amazing collection of rare sea life on a beautiful island, then visited Bon Bon Bon, a downtown chocolate factory where they sampled delicious sweets. They lunched on Chinese food while sharing the ups and downs of life with CMT.



The National CMTA Patient/Family Conference is coming to St. Louis and Atlanta

St. Louis on September 7 • Atlanta on November 2

Please join us and learn about the many accelerated advances in CMT Research

The Charcot-Marie-Tooth Association is pleased to announce that The National CMTA Patient/Family Conference is coming to St. Louis on Saturday September 7 and to Atlanta on Saturday November 2.

Conference topics include neurology, genetics, breathing, surgery, physical therapy and much more!

More details and registration information to come.

Sponsored by



“FUN”ATHLON

LIVES UP TO ITS NAME

The Oxford “Fun”Athlon just keeps getting bigger and better. Now in its sixth year, this year’s event, held on Saturday, June 1, in beautiful Oxford, Maryland, included a one-mile open water swim, a 20-mile bike ride, a walk around historic Oxford and lunch at the waterside Doc’s Sunset Grille. For the first time, a separate golf tournament, Tee Off for CMT, was added on Monday, creating a full weekend of fun and fundraising.

To date, the events have raised over \$265,000 for the CMTA and CMT research with money still coming in. While event organizer Steve O’Donnell reported some tiredness following the three days of nonstop “fun”raising, overall he was energized and enthusiastic with the weekend’s success. Since Steve started swimming across the Chesapeake Bay in 2002, this year’s events bring Steve’s lifetime



The O’Donnell family—Steve, Sherri, Shawn and Jamie.

total fundraising to more than \$1.5 million.

A huge thank you to Steve’s friend, Steve O’Connell, for his individual contributions and to the golf tournament’s corporate donors—Bank of America Merrill Lynch, Barclays, Blackrock,

BNY Mellon, Credit Suisse, Goldman Sachs, J. P. Morgan, Morgan Lewis, RBC Capital Markets and Wells Fargo Bank—and everyone else who generously donated their time and money to support CMT research. ★



Some of the CMTA Family—DeAnn and Steve Scherer, Jeana Sweeney, Steve O’Donnell, Kim Magee, Andi Cosby, Marcia Semmes, Mark Scheidler and John Svaren.

NOT YOUR GRANDMA'S SHOES:

Five Trends People with Foot Problems Should Know

BY LAINIE ISHBIA

I've had a love/hate relationship with shoes my entire life. Finding the best shoes to wear with AFOs has never been easy. Long before I started wearing leg braces for my CMT, it was a struggle finding shoes that fit my high-arched, fused-ankle, drop-foot, callous-prone feet.

I remember the day I picked up my first pair of leg braces. When the orthotist brought them to me in a clear plastic bag, I instantly burst into tears like someone first viewing a loved one's corpse in an open casket. I was scared to touch them. When I caught my breath, my greatest concern came off as superficial: What kind of shoes do I have to wear with those things? That orthotist probably needed therapy for post-traumatic stress disorder after working with me that day. When he brought in a catalog with a pair of bulky, fake-leather clodhopper shoes on the cover, I literally started screaming at him, "Do you expect me to look like Fred Flintstone? No way am I wearing those!"

I left his office (AFOs still in the bag) and didn't take them out again for another six months, when cleaning bloody knees from sidewalk falls became a daily ritual. I moved out of denial and into typical solution-focused "Lainie mode." As my leg braces evolved through the years, so has my ability to find stylish shoes that I can wear confidently. Today, it is so much easier as the trendy shoe styles have features that people with hand and feet issues actually need. Here are five of my favorites:



Lainie Ishbia, AKA Ms. Trend-Able, with some of shoes she tried on her way to finding the best ones for people with CMT.

#1. Kicks Are Dope

Sneakers of all types are cool. They are no longer reserved for mall-walking, city-commuting or gyms. People can wear sneakers with anything and everywhere these days. Brands like Nike and New Balance have a ton more competition these days and it seems like every designer and brand has a line of sneakers. I mean, hello, even Ellen DeGeneres has her own line of sneakers. When worn confidently, sneakers can look just as great with a dress or skirt as they do with jeans. The key is to look like you planned to wear sneakers and not that you have to wear sneakers.

#2. Velcro Is Lit

For those of you without access to a teenager or slang decoder, "lit" means "hot" or "cool." When I first saw Velcro closures on shoes by popular designers, I wanted to jump for joy. I can't actually jump but I was REALLY excited. This trend has been life-changing. Velcro shoes are no longer just worn by nurses and small children. People without hand or foot problems

love the look and convenience of hook-and-loop closures. No more struggles with shoelaces for me.

#3. You Gotta Have Sole

People who have foot and balance issues are experts in flooring material. When I walk into a restaurant and see a shiny painted floor, my heart starts racing, and I go into survival mode. My eyes are always scanning the floor for spilled water, baby pacifiers or any other foreign object impeding my path and likely to cause a fall. I was so excited to see that rubber non-skid soles are now everywhere and on everything from sandals and booties to party shoes. As my 14-year-old son says, "Don't forget the rubber!"

#4. Elevate Your Style

One of the hardest things to accept when you have foot and balance problems is the inability to wear heels. Heels (or pumps as they called them in the 80s when my friends dyed them to match their prom dresses) are considered a necessity when dressing up, a no-brainer for able-bodied women. Heels allegedly make women look taller, thinner and sexier. Those of us who can't wear heels do not benefit from the illusion they give an outfit. My life was forever changed six years ago when I discovered the Wolky Jewel Sandal with a flat, platform sole. For the first time ever, I was walking comfortably in elevated

sandals that made my legs look longer too. Since then, I have found even cuter platforms (the completely level kind without a wedge) I can wear in any season.

#5. Zip It

When you wear leg braces or orthotics, putting shoes on is a high-intensity workout. Even when you remove the insoles and buy a size or two bigger than your real size, there's just not enough room in the opening to shove it in. It doesn't help that both of my ankles are fused with pins, preventing any movement. Like the struggle I have every summer trying to get my kid's sleeping bags shoved back inside the tiny nylon bags they came in, I eventually (sweat dripping) forfeit the fight cuz it ain't gonna happen. This year, I saw a ton of

cute hi-tops with zippers on each side. These hi-tops are not the Chuck Taylor style (I like those too) but rather, more grown-up, soft leather ankle boots that you can wear with anything. In general, hi-tops are awesome for women with foot issues. Not only do they provide built-in ankle support, but if you want to wear trendy ankle cropped jeans, hi-tops conceal your orthotic/AFO. Since laces are tough for me because of my fine motor issues and pull-on style shoes are totally out of the question, I was bouncing off my chair with excitement when I found a pair of hi-tops with removable insoles and two zippers. ★

Lainie is the creator of Trend-Able, a blog for women with invisible physical disabilities who want to look and feel their best (trend-able.com/blog).

Lainie showing how her AFOs work with boots.



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WHY WE GIVE:

THE CMTA is honored to announce a \$1 million challenge grant to support the organization's new Type 2 CMT research strategy. Starting September 1, long-time CMTA supporters Bob and Gail Buuck will match any donations to CMT2 research on a one-to-one basis. Donations will go to implementing the CMTA's exciting new strategy for Type 2, a multi-pronged approach that includes projects on gene therapy, axon degeneration pathways, mitofusin confirmation drugs and human stem cell studies. More details on the strategy will be announced soon—email Jeana Sweeney at Jeana@cmtausa.org with the subject line Type 2 to receive updates.



Bob and Gail Buuck

Bob and Gail Buuck are big believers in “impact giving,” the practice of strategically using time, talents and resources to make meaningful, measurable change.

The Buucks’ impact on the CMTA—and the impact their giving empowers the CMTA to make—are both enormous.

The relationship between the CMTA and the Buucks dates back to 1997, when Bob and Gail traveled from their home in Minneapolis to a Patient/Family Conference in Detroit, where Dr. Michael Shy then had his CMT clinic. The Buucks made their first grant to CMT research shortly afterward. In a story about the conference in the spring 1998 issue of *The CMTA Report*, Bob said, “It’s not enough to fund research. The CMTA must become a leading source for all relevant data pertaining to CMT. It’s a huge undertaking, but it must be done to properly guide research grant decision-making.”

Call Bob prescient: In the 20-plus years since that conference, the CMTA has done exactly that with its Strategy to Accelerate Research (STAR), bringing top researchers together with pharmaceutical and

2008 – 2019 CMTA 2A RESEA



Bob and Gail Buuck

biotechnology partners to accelerate scientific breakthroughs and develop therapies.

Bob and Gail entered the world of CMT when their son John, now 47, was born with a spontaneous mutation of CMT2A. He started showing evidence of symptoms while still a toddler and he was in a wheelchair by the time he was 12. A doctor at the Mayo Clinic finally diagnosed John at the age of 4 or 5, but at the time, Gail says, “There was nowhere to go.” There were no support groups and the CMTA wasn’t yet in existence.

The Buucks first connected with the Muscular Dystrophy Association, Gail said, but were basically told just to “live with it.” Instead, they found a rehab center in the Twin Cities that helped John and began making donations there.

That all changed in 1985 when Bob sold the medical device company that he had cofounded to Pfizer. With greater resources available, the Buucks made their first grant to STAR in 2011. In the early days, the CMTA’s grant application was simply directed toward CMT, Bob recounts, but it got more specific as the research progressed. The Buucks’ donations helped CMTA researchers develop

a rat model of CMT2A, Bob says with pride, adding, “We’re pleased with what we’ve seen over the past five or six years.”

The Buucks don’t designate particular projects for their donations beyond that they be for CMT2A. They entrust that call to the CMTA. “There’s been significant progress,” Bob says, “and I’ve always thought that the CMTA’s money was well-managed.” Gail agreed. “Our comfort level [with the CMTA] is very high,” she said, adding, “A lot of that comes from the fact that Dr. Shy is involved.”

“We understand the importance of a ‘lead gift’ to stimulate other people to give more,” Gail said of the couple’s matching donation. STAR’s success has also inspired the couple to increase their giving, Gail said. “We’ve seen results. This is successful so we’re more apt to give more,” and hope others do the same. Noting that their \$1 million gift is spread out over five years, Gail emphasized that donors “can do a larger amount over time.”

Today, their son John lives in California with his wife and their young son, who does not have CMT. John is very “self-adapting,” with a full exercise regimen of swimming and working out with a trainer. He is also very active, Gail said, with the Buuck Family

Foundation, which, in addition to giving to CMT research, focuses on issues of homelessness, poverty and children and families at risk.

Bob, 79, and Gail, 78, are retired now, but they’re not letting any grass grow under their

feet. The couple, who celebrated their 50-year anniversary in 2018, split their time between Minnesota and Arizona, where they are stewards at the McDowell Sonoran Conservancy. Gail, a former teacher, continues to volunteer with a number of causes in both states. In addition to working on his second novel, Bob is also very engaged at the University of Minnesota Foundation. The Buucks are a couple who make an impact wherever they are and whatever they’re doing and the CMTA is beyond grateful for their trust and support. ★

“We’ve seen results. This is successful so we’re more apt to give more.”

—Gail Buuck

RESEARCH PROGRAM MILESTONES

2015

2A Stem Cell Models Developed

2016

Buuck Family
2A Research Grant #3 Awarded

2017

Drug Screen on Cell Lines
PNS Meeting
Best Animal Models for 2A Available for Testing Drugs

2018

4 Gene Therapy Experts
Join STAR Advisory Board
Pharma Partners
5+ Companies
Type 2/Gene Therapy Strategy Developed

2019

Buuck Family
2A Research Grant #4 Awarded

GENETIC TESTING

(continued from page 5)

one gene, and a VUS in another gene, the most likely answer is that the person has only one type of CMT—the one that is caused by the disease-causing variant. The VUS is just a red herring.

There are also different types of inheritances in CMT. The most common inheritance is dominant, where a person needs to have a disease-causing variant in only one copy of the gene in order to be affected. Much less common (at least in the U.S.) is recessive inheritance, where both copies of the gene must have variants in order for a person to be affected. Each gene in CMT is known if it confers dominant or recessive inheritance (a few genes will do both). If a person has only

one variant in a recessive gene, this is not sufficient to cause CMT.

In a study looking at people with genetically defined CMT, it was found that 2.1 percent had more than one type of CMT. It is quite unlikely that any person with CMT has more than one form. Having multiple VUS on genetic testing does not mean that there is more than one form of CMT present.

If a person has genetic testing and is found to have a specific type of CMT, other family members will have the same type. The type of CMT does not change between generations. For example, if a person has CMT1A, and has an extra third copy of the *PMP22* gene, they can either pass down the chromosome that has two copies of the gene, and have a

child who also has CMT1A, or pass on the chromosome with only one copy of the gene, and that child will not be affected. There are exceedingly few exceptions to this. The only way to have different types of CMT in the family is if there is a new mutation that happens in either the egg or sperm. In my 12 years of working with people with CMT, I have seen this exactly once.

In other words, if you and another family member have CMT, you almost certainly have the same type. ★



Carly is a board-certified genetic counselor at Stanford University Hospital and Lucile Packard Children's Hospital. Specializing in CMT, she is also project manager of the Inherited Neuropathies Consortium.

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- Special mention in the CMTA Report
- A monthly "In-the-Know" email with news, updates and insider information
- One free pair of Aetrex shoes

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

Dear David,

Do you agree with Dr. Phil's position that one can be a caregiver to a disabled spouse OR a lover, but never both at the same time?

David replies:

For those who don't know, Dr. Phil is a psychologist with a successful TV show on which he gives advice and tries to solve issues that arise in individuals and families. Full disclosure: I am not a fan of his and find his style a bit heavy-handed.

Although it is often true that one member of an interabled couple does more of the physical work in the relationship, Dr. Phil is completely dismissing how much the disabled partner can contribute and give emotionally, psychologically and spiritually to create a loving and thriving union. All relationships have issues to work through just by the very nature of living with someone full time. Maintaining a balance of independence and healthy dependence is at the core of most relationships. Learning to accept help with love is equally as important as giving your spouse an opportunity to give love in a meaningful, purposeful way. Accepting an act of kindness is

being kind in itself. As a therapist who has worked with couples for many years, I can most assuredly state that without the emotional maturity of desiring your spouse's happiness, your relationship will not work. I was also put off by Dr. Phil's assumption that most interabled couples can afford to hire outside help. What world does he live in?

The challenge of any physical disability, or for that matter any emotional disability, can either shut both of you down emotionally or create a portal where the full range of emotions from sadness to joy can be experienced. Deepening your connection with open communication is the key. Helping your spouse with the most basic and personal activities can create a profound intimacy, giving you opportunities to deepen your love. Listening is an act of love and one doesn't need strong legs to master this ability. Listening and being supportive is probably one of the most important elements in a strong relationship. Being able to express your gratitude to one another is also important. Several months ago I was going through a rough time with my CMT and was particularly irritable and unpleasant

with my partner. I knew I was not being kind, but I wasn't quite ready to speak. I did manage to buy a card that simply said "I am so grateful for you." I gave it to him and it was a very intimate and loving moment for both of us.

Having a sense of humor about living with a disability is also crucial for a healthy marriage. Yes, even CMT can be funny at times. The other day I leaned against the wrong shelf in the refrigerator and down went a dozen eggs onto the kitchen floor. What a mess! I attempted to clean it up by getting down on all fours and my partner just looked at me and said, "Do us all a favor and just sit over there and stay out of trouble." I felt a little pathetic, but it was funny at the same time!

Yes, there are challenges in interabled relationships, but tell me what relationships don't include challenges. In Japanese pottery, artisans often repair cracks with gold, and the "brokenness" adds to the overall beauty of the piece. In relationships where one spouse has some obvious limitation, the challenges and the cracks are simply more visible. ★



Write to David at info@cmtausa.org.

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.

Corvette Restorers Rev Up for CMT

Members of the National Corvette Restorers Society, Heart of Ohio Chapter, got together at the Columbus Ohio Cars and Coffee event June 8 to host a fundraiser for Camp Footprint.

Sixteen chapter members provided "free" donuts and asked for a donation to support the camp. They brought in \$455 in cash plus a \$100 check. The club is kicking in the difference to make it \$1,000.

Thanks to the Heart of Ohio Chapter for helping fund Camp Footprint and giving kids with CMT a week they'll never forget.



REVIEW:

BILLY Shoes for Kids “Look Cool,” Hide AFOs

BY LIA AND TAU O’SULLIVAN



EDITOR’S NOTE: Last winter, we began hearing about a shoe for kids that went on easy and looked cool. BILLY Footwear, invented by a man left paralyzed after a three-story fall, incorporates zippers that go along the side of the shoes and around the toe, allowing the upper of each shoe to open and fold over. The wearer places his or her foot onto the foot bed, then pulls a zipper to close and secure the shoe. When we reached out to company co-founder Billy Price to ask for a review pair for Lia O’Sullivan’s new v-log, he couldn’t have been nicer. He asked that her mother, San Francisco Bay Area Branch Co-Leader Tau O’Sullivan, measure Lia’s feet to make sure the shoes would work for her. After comparing her measurements to the company’s sizing chart, he told us what shoes would work best and let her pick her favorite. She chose the Classic Lace High Kids Black Metallic Zip. Billy himself placed the order and soon the shoes were on their way to Lia, 9. She and “Moma Tau” report back.

LIA’S SHOE REPORT:

Hi I’m Lia and I have BILLY shoes. I LOVE THEM! I love them because instead of COMPLICATING laces they have ZIPPERS! Also, I love them because they look cool. Last I love them because they’re high tops, to hide my braces. There you have it. BILLY shoes are great!

MOMA TAU’S REPORT:

We all know shopping for shoes with a child who wears foot orthotics (FO) or ankle-foot orthoses (AFOs) isn’t easy. In fact, it’s probably the least enjoyable shopping experience ever. Not only do you get the “what is that” stares from fellow shoppers, there



aren’t many wide selections available to make room for the FO/AFOs. When shoes are wide enough, they are generally hideous looking, and difficult to put on, especially if your little precious one’s fine motor skills are not on point! That all changed recently for our Lia, who has CMT1B and has worn AFOs on both feet since she was diagnosed at the age of 2. BILLY shoes—specifically the Classic Lace High Kid’s cool looking selections—came into our lives.

As CMTA Advisory Board Member David Misener, an orthotist with CMT1B, says: “[F]ootwear is the base of the body, much like the foundation

BILLY Shoes Born of Necessity

Billy is himself a “differently abled warrior.” In 1996, he broke his neck in a three-story fall that left him paralyzed from the chest down. He lost the ability to move much of his body, including his fingers. Not only did he suddenly face mobility challenges, daily tasks like putting on clothes became much more difficult. Throughout the years he learned tricks for getting dressed more easily, but he never found an attractive pair of shoes that he could put on independently. So he and his business partner, Darin Donaldson, invented one, first for Billy’s personal use and then for others who want footwear that is functional, fashionable and accessible.

BILLY shoes are currently available only in kids’ sizes, but in August the company will be launching an expanded adult line that will include both high tops and low tops for men and women. The kids’ shoes are available at www.billyfootwear.com and other online sites, as well as select brick-and-mortar stores, including Nordstrom and Kid’s Foot Locker.



of a house: You need to have a solid footing beneath you to build a stable and supported body.” I can’t say it any better and I’m sure most if not all parents with a differently abled child would agree. In addition to Lia’s list of things she loves about her BILLY shoes, below are the features that make them such a good fit for her from my point of view:

- They have a wide base of support, providing room for FOs or AFOs.
- They have strong heel and ankle counters (those little plastic inserts used to reinforce the shoe) to resist sprains and instability.

- BILLY’s Classic Lace High Kid’s Black Metallic Zip are durable, light, soft, and flexible (but not rigid), which gives Lia freedom to play with the least restrictive environment.
- They are easy for Lia to put on and take off by herself, which encourages her independence.

Remember, what may work for us may not work for your precious one(s) but we hope you’ll find your best fit given its variety of cool selections. Lastly, BILLY Footwear was founded on the principle of inclusion and perseverance, which is exactly what we want for our differently abled warriors! ★



The Blue Denim Glitter BILLY Classic Lace High sells for \$55 at billyfootwear.com

THE CMTA GRATEFULLY ACKNOWLEDGES GIFTS...

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The Camp Footprint Effect: *Friends to Family in One Easy Meeting*

BY EMMILY STUFFLET

I remember May 31, 2018, like it was yesterday. I remember what I wore, the songs I listened to in the car, what I ate for breakfast and how I styled my hair. But most importantly, I remember who I finally met for the first time in person: my best friends Erin Black and Riley Williams. Erin, Riley and I all have CMT. I was diagnosed when I was 3 years old, meaning that I have been fighting my battle for most of my life. Growing up, I truly believed that nobody fully understood my situation and that I would never meet anyone I could relate to on a serious level.

That all changed on a September evening in 2017 when a girl named Erin asked to follow me on Instagram. I noticed that this mysterious girl had “CMT” written in her biography. Naturally, I accepted her request,

followed her back and messaged her. Erin immediately responded to my message and we instantly formed a connection. We talked about CMT, which eventually led to random topics such as our favorite musicians and clothing stores. In the mix of all those random topics, Erin told me about Camp Footprint, a weeklong camp held every summer in Pennsylvania for kids with CMT. She sent me the link to a video she created the first time she attended camp and I absolutely loved it, so much so that I desperately wanted to attend the next year. Erin and I concluded our conversation by exchanging phone numbers and we have talked almost every single day since that September evening.

Fast forward a few days to when I made another amazing friend. While I was looking through the comment section on Erin’s Camp Footprint post, I found Riley Williams, another girl close to my age who also has CMT. I messaged her on Instagram and, once again, we instantly formed a connection. I asked Riley numerous questions about Camp Footprint because she had attended with Erin in August 2017. She kindly responded to all my questions and made me feel so loved and appreciated. We gave each other our phone numbers and snapchat usernames to stay in contact.



Camp Footprint “reunion” in Disney World!
Left to right: Emmily Stufflet, Riley Williams,
Erin Black.



Emmily Stufflet, in the wheelchair,
Riley Williams and Erin Black.

A couple of weeks later, Erin created a group chat with the three of us to talk about Camp Footprint. After many long conversations regarding this amazing event, we became a trio. Since then, Erin, Riley and I have talked every single day. We all knew that we wanted to meet each other in person one day, but didn’t know how it could happen. The 2018 season of Camp Footprint didn’t work out for me, but my mother had the idea of inviting my two best friends to Florida over the summer. Immediately, I texted them and we started planning our trip.

For six months, Erin, Riley and I impatiently waited for May 31 to arrive. It was the longest wait any of us had ever experienced. Once the day arrived, we were all anxious and thrilled but ready to finally meet each other. Our morning at the airport con-

sisted of tears of joy, hugs and smiles. Then it was time to start the best five days of my life. The three of us explored all over Orlando, including the beach, Winter Park, Disney World, downtown Orlando and so much more. But, I can honestly say that the best part of our trip was spending time with each other. There was never a dull moment, only smiles and laughter. Within the first few hours of physically seeing one another, we had already shared numerous inside jokes and formed memories that we will never forget.

Saying goodbye was one of the hardest things I have ever done in my life. It sounds dramatic, but it's true. Within five days, Erin and Riley taught my family and me so much about strength, love and kindness, and for that we are forever thankful. Being able to experience something like this was truly life changing. I cannot stress enough how important it is to surround yourself with people who fully understand your situation. Not only do my best friends understand, but they introduced me to the CMT family, so I have numerous people that understand me and that I can talk to. It is insane how much my life has changed in the past year simply because of an Instagram direct message. Erin and Riley are true blessings and I cannot wait until the day we all reunite.

So, that's our story ... so far. Our friendship has just begun and we all are beyond thrilled for our future adventures together. Remember to stay humble, keep life in perspective and be kind to one another because you never know where you'll end up. ★

Emmily is 15 years old and will be a junior in the fall. She and her BFFs had a reunion in December and planned on meeting up again in June at the CMTA Youth Council retreat in Denver.



DREAM VACATION IN ITALY UP FOR AUCTION

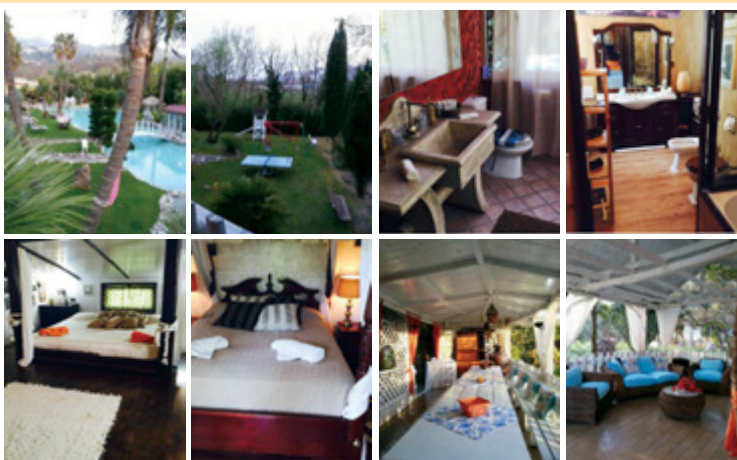
Win the Italian vacation of your dreams while supporting the Cycle and Walk 4 CMT and STAR, the CMTA's research initiative. Up for auction is a week-long stay at Villa Emilia, generously donated by long-time STAR supporters. The villa is located outside of the medieval village of Rocca d'Evandro, a spectacular mix of soft hills and countryside along the River Garigiliano.

This six-bedroom, five-bathroom villa sleeps up to 12 people. The main home has two king and three queen bedrooms. The chalet has one queen bedroom.

The property also has pools, a gym, horseback stables, a tennis court, a game room, sauna and steam bath and a children's play area. This package includes unlimited use of the wine cellar and one four-course meal prepared by a private chef.

Conveniently situated between Rome and Naples, Rocca d'Evandro is known for its vineyards and agricultural products, as well as its medieval fortress castle. The area's historical importance dates back to the Roman Empire and continues through World War II, when it was part of the "Gustav Line." Villa Emilia is just 20 miles away from the sea and the magnificent Pontine Islands, right across from the ports of Gaeta and Formia.

Bidding on this magnificent package starts at \$5,000, with a "Buy It Now" price of \$8,500. For more details, visit <https://cmta.rallyup.com/villacycle4cmt>. Good luck and arrivederci!



RAISING FUNDS

CMTER'S DAUGHTER GIVES BACK

Each spring my daughter Natalie's junior high school does a project they call "Giving Back." Financed by an anonymous donor and coordinated by Natalie's teacher, Mrs. Diane Majewski, students are given \$20 each to do projects that benefits someone other than themselves. Many of the students use their talents to make and sell things, turning their \$20 into even more money to donate, which was what Natalie decided to do.

She loves making clay figures so she created and baked almost 100 of them, including a shark! There wasn't a day that she didn't sell out and she even took special orders. There were so many special requests, she had a lot of late nights and sore fingers. I teach computer and art at her school and loved going in each morning and seeing the lines of students waiting for the new figures she

Natalie, Holly, and Audrey VanDyne at the Lakewood, Ohio, Walk 4 CMT



had to sell. In a week's time, Natalie was able to turn her original \$20 into \$75 that she donated to STAR.

Natalie doesn't have CMT, but I do and she sees how CMT affects my everyday life. She, her dad, and her older sister Audrey were my biggest helpers and supporters when I had my left foot completely rebuilt five years ago. She's always there to help me up when I fall, and last year we did our first CMTA Walk 4 CMT in Lakewood, Ohio, together. I love my girl and her big heart!

—Holly VanDyne

MURDER MYSTERY AFTERNOON RAISES \$1,002 FOR RESEARCH

The Seattle Branch asked the question "Who Dunit?" at its second annual Murder Mystery fundraiser April 28. The answer: the CMTers who raised \$1,002 for CMT research while enjoying an interactive fundraiser filled with intrigue and humor.

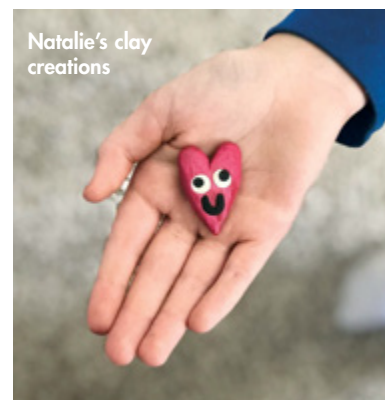
According to Branch Co-Leader Emily Osborne, the afternoon started when attendees arrived at a local brewery and enjoyed a light lunch, drinks



Seattle Branch Co-Leaders (and suspects) Denise Snow (left) and Emily Osborne (right).

and socializing. The guests were then divided into two groups, each with one "suspect." The two Murder Mystery actors led the show and eventually, a "murder" occurred. The guests gathered clues, used play money to bribe other guests for answers to their questions and tried to sleuth out "Who Dunit?" Afterward, participants each received a commemorative mug shot photo.

Emily said the fundraiser was an entertaining way to spend the afternoon and raise money for a great cause. It's one more example of the CMT community's inventive fundraising ideas. ★



Natalie's clay creations



AND AWARENESS

Walk This Way: San Diego Walk 4 CMT

Some 125 CMTA members, friends, family and volunteers gathered March 30 for the San Diego Branch's first-ever Walk 4 CMT, raising just shy of \$18,000 for CMT research.

Branch Co-Leader Kennie Trout said the walk met all criteria for a successful event: All metrics exceeded expectations and participants enjoyed themselves and made connections, hanging out and visiting long after the walk was over.

In the weeks leading up to the walk, Kennie invested a lot of time promoting it, getting sponsors, finding raffle items, and coordinating with the city of Carlsbad, CMTA staff and the property owners and managers. "After working through all the challenges, I was so happy that everything went as planned," he says.

That investment paid off in spades: The walk not only raised funds for research, but the committee's public grassroots advertising also raised awareness for CMT in the San Diego area—the walk was in a public area where their shirts, bandanas and banners stood out on that sunny March Saturday.

The walk committee had one cliff hanger when the owner of the original walk site cancelled on the group three days before the event. Luckily, Kennie said, the property manager (who has CMT!) where the walk was held convinced her property owner to let the branch hold the event there. Insurance and per-



Friends and Family at the San Diego Walk 4 CMT



missions weren't finalized until the day before the walk.

The walk committee has already started preliminary planning for next year's San Diego Walk 4 CMT 2020. Kennie says he would like to include a seated dance class as part of the event next year, like the one that Nashville, TN Branch Co-Leader Bridget Sarver hosts each morning at Camp Footprint.

Kennie called the walk a gratifying experience and says he got a lot of satisfaction from

watching everyone having a good time. "I would highly recommend that others plan a Walk 4 CMT in their hometown," he added.

The CMTA thanks Kennie and all the walk leaders across the country for stepping up to raise critical funds and awareness in their communities.

For more information on how to organize a local Walk 4 CMT contact National Events Manager Andi Cosby at andi@cmtausa.org, or visit www.Walk4CMT.org to find existing Walk 4 CMT locations.

Cycle (and Walk!) 4 CMT!

Sunday, August 25, 2019 • The Old Lantern, Charlotte, VT

CYCLE 4 CMT



www.cycle4cmt.com



EVENT DETAILS

CYCLE 4 CMT: Choose 6.5, 15, 25 or 40 mile routes. Fantastic views!

After - Party: For everyone - cyclists, walkers, supporters and sponsors. Live music, Vermont beer, cider and non-alcoholic drinks. Fully-catered meal. Silent Auction items from local sponsors, quality swag and more!

Can't make the event? Do your "OWN" Cycle or Walk 4 CMT - anywhere, anytime on or before August 25, 2019.

Whether you are a cyclist, walker or supporter, this event is for you! Register or donate at: cycle4cmt.com

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907-244-2100

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Little Rock
Candice Cargile
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Phoenix Area
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CALIFORNIA
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Alani Price
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Michael Huff
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Kendall Trout
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Emily Osborne
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Debi Weber
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Milwaukee Area
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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

JACKSONVILLE, FL

After a pop-up meeting last December, Tim Nightingale and Stephanie Burkhalter realized there was a group of people with CMT in north Florida looking to connect with others, but no branch. So they started one! Twenty people turned out for the Jacksonville Branch's first meeting, sharing their CMT stories and listening to a presentation on inherited neuropathy by Dr. Elliot Dimberg, from the neuromuscular group at Mayo Jacksonville. They plan to meet quarterly.

BOSTON, MA

More than 40 people came out to hear Dr. Reza Seyedsadjadi and Ken Cornell, CO, FAAOP, of Massachusetts General Hospital's CMTA Center of Excellence present to the Boston Branch on March 27. Many traveled from Connecticut, Rhode Island and New Hampshire to hear updates about the COE, which has seen well over 400 CMT patients since it opened. The COE is also involved in many research studies, and Dr. Seyedsadjadi said he is excited about promising drug development results and on-going clinical trials. Genetic counselor Trisha Multhaupt-Buell and research coordinator Natalie Grant have recently joined the center. Ken Cornell, CO, FAAOP, presented a full range of orthotics and bracing options for CMTers. Ken stressed the importance of support, stability and the ability to walk efficiently for people with CMT and that wearing the correct, properly fitted orthotic can be life changing. Special guest Jocelyn Duff, of Ipswich, MA, shared her family's CMT story and the importance of getting involved to support rare diseases. The Duff family's story has made headlines across Massachusetts and the country as they work towards the first gene therapy treatment for CMT4J.

NEWBURY, NH

The newly formed Newbury, NH Branch drew four members to its first meeting on April 20. They discussed a wide range of issues, includ-

ing the benefits of working out with an individual trainer and working with an orthotics specialists. They also shared contact information for doctors, physical therapists and occupational therapists. Group members were also assigned homework—to think of ideas for a future fundraiser, potentially during CMT Awareness Month in September.

CENTRAL NEW JERSEY

Branch Leader Mark Willis announced that the group's second Walk 4 CMT will be held in September, at a date to be announced. Last year's walk—held on a sunny day in a beautiful park—raised over \$14,000 for research. The amount entitles the branch to pick a guest speaker from the CMTA Advisory Board, and members discussed who that should be. The group also reviewed the new CMTA Physical Therapy and Occupational Therapy Guide, the newest neurotoxic



Sweet celebration in Central New Jersey

drug wallet card, and the new "What is CMT?" brochure. They shared their CMT stories, then enjoyed sandwiches and a special 10th anniversary cake.

NEW MEXICO

Eleven people attended the New Mexico Branch meeting on May 11. Advisory Board Member Clark Semmes talked via Skype about his participation in a clinical trial of ACE-083, a drug specifically to increase growth in a muscle related to foot drop. For the past seven months, he has gone to Philadelphia every three weeks to take part in the double-blind clinical trial. He indicated that the trial is still looking for participants and that there are a number of other locations around the country where a participant can go for injections. Members also asked about upcoming clinical trials and Clark advised them to go to www.clinicaltrials.gov.

SYRACUSE, NY

Fifteen people came out for the Syracuse Branch's first meeting on March 30. They

ranged in age from the early thirties to 80 and had all types of CMT, but they all agreed that a sense of humor is one of the best ways of coping with the disease. They also shared a sense of relief at meeting others who have CMT and knowing they are not alone. The group discussed future meetings, guest speakers and fundraising efforts. They also talked about fundraising and the possibility of holding a Walk 4 CMT. Branch Leader Michael Casey said he feels like "this could be the start of something big!"

CHARLOTTE, NC

Dr. Tyler Gonzalez, orthopedic foot and ankle surgeon from Wake Orthopedics, spoke to some 25 members of the Charlotte Branch May 4. Dr. Gonzalez focused on the things patients with CMT can do to help themselves, including physical therapy, strength maintenance and brain/body communication. He also discussed the benefits of bracing, the many different bracing options and who is a good candidate for bracing. Finally, he discussed surgical options for patients with CMT, clearly communicating that this should be the last option. Dr. Gonzalez encouraged members to be proactive in their care and to partner with their doctors to maintain the health of their muscles as long as possible



Dr. Tyler Gonzalez presents to the Charlotte branch

so that ideally they will not need surgery. The meeting was held at The Hanger Clinic, which set up a wonderful space with a projector and screen for Dr. Gonzalez. Hanger, a CMTA partner, also provided breakfast, an unexpected and much appreciated gesture.

LAS VEGAS, NV

The Las Vegas Branch had a great "get to know you" meeting on May 4, sharing CMT stories and talking about "best practices for living with CMT." The group planned out their meetings for the remainder of 2019 and discussed guest speaker options. They will meet again in August, October, November, and December.

SEATTLE, WA

Thirty people turned out to hear Dr. Thomas Bird speak at the Seattle Branch's April 13 meeting. He spoke about the CMT research he's seen while working at the University of Washington Medical Center over the last 40 years, then took questions. Group members introduced themselves to the first-timers in attendance, then talked about the branch's upcoming fundraisers—Wine & Chocolate, The Puzzle Project and the Second Annual Walk 4 CMT.



Dr. Thomas Bird speaks at the Seattle Branch meeting

NEW BOARD MEMBERS

KEVIN SAMI AND DAVID NORCOM
JOIN THE BOARD OF DIRECTORS

KEVIN SAMI brings extensive business and non-profit experience to the CMTA Board. He is currently an investment analyst in New York City, where he also serves as president of the Medhat F. Sami Foundation and as treasurer of Musical Chairs Chamber Ensemble, a Staten Island arts organization. Kevin previously worked as an investment analyst at Apis Capital Advisors and served on the steering committee of Families for Safe Streets, a New York traffic safety organization. Kevin holds a BBA in finance from the University of Wisconsin-Madison.



“I am personally affected by CMT,” Kevin said, “and am honored to join the board of the CMTA, an organization whose mission to develop treatments and support patients and families continues to have a tremendous impact on the lives of those affected.”

DAVID NORCOM has also joined the CMTA Board of Directors. David has more than 37 years of experience in investment analysis, asset allocation and capital markets research. He is the founder and chairman of Norcap Advisors, LLC, an investment manager in Dallas with over a decade of experience in the alternative asset space with risk management and capital preservation as the foundation of its investment strategy. Prior to founding Norcap in 2004, he spent 20 years as the director of consulting groups at Smith Barney and Morgan Keegan & Co. in Dallas. David served on the New York Stock Exchange Disciplinary Hearing Board for 10 years and was president of the Association for Professional Investment Consultants for two years. He earned a BBA in management economics from Texas A&M University and a Master of Divinity from Austin Presbyterian Theological Seminary. He is also a certified graduate of the Pension Fund Management Course at the University of Pennsylvania’s Wharton School of Business.

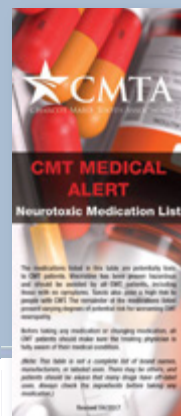
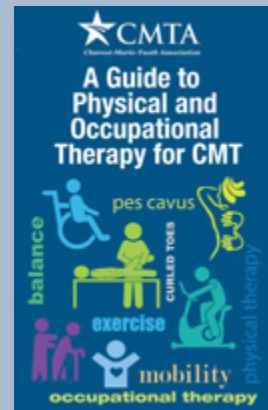


David said he firmly believes that “the time is ripe through the significant advancement in medical research to focus on a cure for CMT through gene therapy.” He is joining the board now, he added, because “I want to be present when that day comes and serve up a toast of celebration when we cure CMT.”

CMTA Board Chairman Gilles Bouchard said the new board members both bring “impressive business experience” and strong commitment to CMTA goals, “a huge help during this time of unprecedented growth and excitement around our STAR program.”

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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 (ddl, 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
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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.