

THE CMTA Report

JANUARY/FEBRUARY
2015

Information on Charcot-Marie-Tooth Disorders for Patients, Families, and the Scientific Community ★ www.cmtausa.org



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.

OUR VISION:

A world without CMT.

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Pat Dreibelbis: Celebrating the Life of a Mentor, Teacher and Friend

BY ELIZABETH OUELLETTE

When someone you love becomes a memory, the memory becomes a treasure. (1 Corinthians 13:7)

It is with the greatest sadness that we announce the untimely death of our beloved friend and colleague, Pat Dreibelbis, fondly known as Pat D. After nearly 24 years of unwavering dedication to the CMTA and the CMT community, Pat will be profoundly missed by the many whose lives she touched.

Patrick Livney, CEO of the CMTA, praised Pat's dedication to informing and teaching the global CMT community about CMT in order to better the quality of life for all. "Her two plus decades of devotion to the CMTA inspire me to succeed in delivering the first treatment to CMTers as quickly as possible," he said.

Pat's memory will live on through the numerous written treasures she left behind, including the CMTA's bimonthly newsletter, *The CMTA Report*, *The History of the CMTA*, and countless other publications, including her popular stories featuring Archy the Turtle.

For someone who was neither a doctor nor scientist, Pat's knowledge of CMT was second to none. Jeana Sweeney, the

CMTA's Director of Community Services, called Pat "a book of knowledge," adding, "If I did not know how to respond to a CMT-related question, I would always turn to Pat D. She'd have all the answers."

Pat was an integral part of CMTA's strategic planning team. Kim Magee, the CMTA's Director of Finance and a close friend, said, "Pat was always the voice of reason. We would all be discussing something, trying to work it out, and Pat would say one sentence and get at the heart of what we were trying to do."

Pat's love of the English language and schoolteacher background came into play daily, and no written materials ever went out from the CMTA without her blessing. Susan Ruediger, the CMTA's Director of Development, affectionately remembers Pat as a counselor and trusted advisor who helped her "understand the essence of



Pat Dreibelbis was a devoted CMTA staff member for almost 24 years.

the CMTA" and develop her writing skills.

For Pat, the highlight of her job was the opportunity to forge relationships with the people who called her asking for help, advice and feedback. Herb Beron, the CMTA Board Chairman, remembers: "Pat Dreibelbis was one of the first people I ever spoke with when I contacted the

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REMEMBERING PAT D.

(continued from page 1)

CMTA about my daughter Julia. We knew nothing about the disease, and she was a wealth of information to me and my family. Over the years as I came to know Pat better, we became good friends. I have always considered her to be the 'matriarch' of our organization, and her sudden passing leaves a void within our organization that we can only hope to fill with someone of Pat's character and kindness."

Bethany Meloche, the Director of Social Media and newest member of the CMTA team, pointed out that while it's not always easy to measure the impact of a life, in Pat's case, "The lives of over 2.8 million

people were made better because she was here. I have lost a colleague and supporter, the CMTA has lost a family member, and all of us with CMT have lost a friend."

For me, Pat was also my first point of contact with the CMTA. After I made a small donation, she took the time to call and thank me. From that point in time 12 years ago, Pat was an enthusiastic supporter of my work and involvement with the CMTA. Together, we worked together on many projects: CMTA Patient and Family Conferences, fundraisers, Support and Action Groups and a myriad of different publications. She inspired me and taught me more than I can say. I feel privileged to call

Pat a mentor and a close colleague.

Truly, the world has lost an amazingly compassionate, empathic and extraordinary human being. Everyone who ever had the privilege of working or speaking with Pat found it a special pleasure. Her quick wit and incredible sense of humor brought smiles and laughter to many. While Pat will be greatly missed, she will never be forgotten. The indelible mark she left on every layer of the CMTA and the treasures she left behind will continue to be a source of inspiration and information to all for many years to come. ★

To make a donation in memory of Pat, please go to www.cmtausa.org/pat or mail a check to CMTA, PO BOX 105, Glenolden, PA 19036.

NEW NAME, SAME GREAT PEOPLE!

CMTA Support and Action Groups Now CMTA Branches

Did you know that the entire CMTA grew from a single support group started in 1982? The members of that group formulated a plan for a larger organization, initially hoping to raise money for research, but also recognizing the need for a forum for patient self-help discussions and resources.

Over the past 30 years, much has changed within the CMTA, but one important thing we learned from that first group was that putting folks with CMT in a room together can lead to great things.

Today the CMTA has 68 groups in 36 states, plus four in Canada and one in Mexico! Throughout the year, folks from all over North America gather to share stories, listen to expert speakers and swap ideas on how to cope with CMT. These groups have also become important centers for fundraising and raising CMT awareness.

Because the groups and their leaders have become vital arms of the CMTA, we are renaming them CMTA Branches! With this name change, we want to emphasize the strong and growing connection between the CMTA and our local groups.

We look forward to working with each branch, searching for and serving every CMT patient and empowering them to make a difference! As we continue our focused STAR approach for a first treatment, we will strive to deliver a better quality of life for all who live with CMT!

To get involved with your local CMTA Branch, please visit www.cmtausa.org/branch. If there isn't a branch near you, please consider starting one and becoming a CMTA Branch Leader. For any questions about the CMTA Branches, please contact Jeana Sweeney at jeana@cmtausa.org.



ELIZABETH OUELLETTE NAMED NEW VICE CHAIR OF THE CMTA BOARD

BY HERB BERON, CMTA CHAIRMAN OF THE BOARD

Tireless, dedicated, passionate—these are just a few of the words that describe Elizabeth Ouellette and her efforts in helping CMT patients and their families and furthering the cause of the CMTA. As many of you know, it was Elizabeth's vision that helped revitalize our CMTA Support and Action Groups, which have grown from just a handful to 70 plus CMTA branches and counting! It was Elizabeth's desire to bring specialists directly to various parts of the country to meet with and educate patients and their families. Thus began an enhanced version of our hugely successful Patient and Family Conferences. It was Elizabeth who saw an increased role for dedicated specialists who could engage more directly with



the CMT community, which led to the creation and expansion of the CMTA Advisory Board. Elizabeth's unwavering passion and drive is essential to our organization, and I take great pride in naming her vice chair of the CMTA Board!

Much of Elizabeth's role will remain the same, but she will work closely with me to implement expansion of the Advisory Board and its services, increase the fre-

quency and content of Patient and Family Conferences and act as a liaison with the CMTA Board to facilitate better interaction with the greater CMT community.

Please join us in congratulating Elizabeth on her appointment and welcoming her to a new and even greater role within the CMTA!

NEW ADDITIONS TO THE CMTA ADVISORY BOARD

GLENN PFEFFER, MD, is Director of the Foot and Ankle Center at Cedars-Sinai Medical Center. He is also a Co-Director of the Hereditary Neuropathy Program and Co-Director of the Cedars-Sinai/USC Gloria Kaufman Dance Medicine Center. Dr. Pfeffer has written numerous scientific articles on orthopaedics and has edited seven academic textbooks on the foot and ankle. He has been treating foot and ankle problems in patients with Charcot-Marie-Tooth disease for 25 years. He is a past president of the American Orthopaedic Foot and Ankle Society and recently served as president of the California Orthopaedic Association. Dr. Pfeffer is frequently interviewed on foot and ankle topics, and has been featured on CNN, *Dancing with the Stars*, *Dateline NBC*, *Good Morning America*, and in *The New York Times*.



GREG STILWELL, DPM, is a board certified podiatrist diagnosed with CMT 1A, an inventor and the patent-holder of the Barefoot Orthotic™. He lectures internationally on foot and ankle topics, including CMT. He is fluent in both in English and Spanish. He is a published author on MRI results of calf muscle atrophy in CMT and has begun a philanthropic site called CMTHope, which will serve to fund those with CMT who need braces, shoes or orthotics and cannot afford them. Greg is dedicated to helping podiatrists in the US, as well as in Latin America, recognize CMT and become current on various diagnostic and treatment options.



CMTA Member Spotlight: Anthony Zahn

Anthony Zahn played soccer in kindergarten, but he wasn't great at it. As a freshman in high school, he tried to play football, but his knees hurt so badly he had to quit. Then, 15 years old and undeterred by his knee pain, Anthony took up cycling with the dream of riding in the Tour de France. It took a year and a half for a neurologist in Loma Linda, California, to realize the cause of his knee pain was CMT. Anthony brushed off the diagnosis and held onto his dream, deciding that he would be even more famous as the first man to ride in the Tour de France with a degenerative neuromuscular disease.

Anthony won the first race he ever entered, a relay triathlon in which he rode the bicycling portion of the race. He attributes that victory mostly to the other two people on his team. As time went on, Anthony began competing in bicycle road races, often finishing in the middle of the pack. Then one day, a fellow bicycle racer who had muscular dystrophy noticed Anthony's skinny legs, and they began talking. The disabled racer, a man named Ryan Levinson, recommended that Anthony get the level of his disability classified, and then race as a para-athlete. Anthony agreed, shifted his goals and kept reaching upwards.

The severity of Anthony's disability was officially classified (a requirement for a para-athlete) in July 2005, and he began competing as a disabled athlete. It took him two years to rise



Zahn (far right) receives a bronze medal in Beijing at the 2008 Paralympics.

through the ranks to the very highest level of competition, the USA National "A" Team. In 2008, he won a coveted spot on the US Paralympic team headed to Beijing. The Paralympics are held immediately after the able-bodied Olympics, in the same venues. Anthony describes them as a big, crazy circus. The athletes, according to Anthony, have a stunning array of body types, from tiny female gymnasts to enormous male bodybuilders. Throw in disabilities of every conceivable nature, and the people watching rises to a whole new level. With the nightclubs and other distractions in the Olympic village, the trick, according to Anthony, is to soak it all in while still staying focused on your races. Anthony must have achieved the right balance because he medaled in Beijing, bringing home the Bronze in the individual time trial.

Four years later, at a World Cup race in a small town in northeast Canada, a car somehow entered the course and collided with Anthony. The collision resulted in a concussion, two cracked ribs, a separated shoulder, and various other scrapes and bruises. Despite his injuries, Anthony traveled to London just six weeks later and was able to put together two eighth-place finishes in the 2012 Paralympics.

Anthony is no stranger to surgery. At 22, he had a procedure known as a "triple arthrodesis" (the fusion of three main joints) on his left foot, along with a tendon transfer. Post-surgery, Anthony was in a full leg cast for six months and a partial cast for another six months. It was more than a year before he could put a shoe on his left foot and 18 months before he could climb back on a bicycle. He has also undergone a

**IF YOU KNOW
SOMEONE**
who would be a good
subject for a CMTA
member spotlight, please
contact Clark Semmes
at 410-350-4812 or
clarksemmes@gmail.com.

carpal tunnel release and toe-straightening procedures.

Despite the injuries and surgery, Anthony does not take any pain medications, not even ibuprofen. No fan of drugs in the first place, his concern for being suspended from racing because of a drug test made the decision even easier. Anthony does not use any special equipment, either on his bicycle or his person. The bicycles he rides are expensive but not equipped in any special way for his disability. He once had a pair of custom cycling shoes with built-in AFOs, but the constant stress quickly broke them and now he wears standard clip-ins.

On July 6, 2014, at the Road Cycling National Championships in Madison, Wisconsin, Anthony retired from bicycle racing. After a 25-year career, retiring was bittersweet. He loved the racing and his fellow racers and had medaled at every level of competition, including three bronze medals at the world championships. But he no longer had the energy for the training required to compete at a world-class level. Anthony believes that while recreational exercise can be beneficial for people with CMT, the intense training he did probably exacerbated his condition.

Today, Anthony coaches other cyclists at all levels, from weekend athletes to Paralympic hopefuls. If you are interested in benefiting from Anthony's years of cycling experience, you can contact him on Facebook at www.Facebook.com/AtoZcycling.

Anthony Zahn has been a member of the CMTA since 2012. —Clark Semmes

HAPPY NEW YEAR!

This is a time to reflect on past successes and look forward to the accomplishments to come in 2015. Your past involvement has made a difference in our future. You helped us raise more than \$3 million in 2014, of which approximately \$2 million went directly to CMT research. With your help, we achieved significant breakthroughs in research for CMT drug delivery. Thank you!

As the CEO of the CMTA, the premier charitable organization fighting for those with CMT, I am looking forward to building on our success! Here are a few of my resolutions for 2015. The CMTA will:

- Share our resources with an additional 10,000 people worldwide.
- Host a national Patient and Family Conference in Boston.
- Grow our reach to 100 CMTA branches in North America.
- Build human cell lines for 1A, 1B, 2A, 2E, X, and 4 to expedite drug discovery.
- Succeed in building assays for CMT1B to be used to screen potential drugs.
- Begin the STAR strategy for type 4.
- Screen 1.7 million compounds at Genzyme for CMT1A.
- Grow the CMTA's Clinical Centers of Excellence worldwide to 19 from the current 17.
- Host 10 public webinars with CMT experts.
- Expand our outreach to Latin America, where CMT is prevalent.
- Host a national fundraising event in which everyone in every corner of the globe can get involved.
- Develop a guide to choosing the right orthotics for CMT.
- Develop a video series highlighting different physical therapy exercises to fight the symptoms of CMT.

Sticking to these resolutions will not only result in significant progress in our scientific research for drug delivery, it will improve the quality of life for those living with CMT.

How will you resolve to battle CMT? Are you going to tell someone you know about your CMT and all that the CMTA is doing to fight for you? Are you going to join one of our CMTA branches in North America to connect with others with CMT? Are you going to fund our groundbreaking research? Are you going to decide that this is the year to build a better future for everyone living with CMT?

Make 2015 the year you decide to be a part of the awareness and a part of the solution to CMT. This year, increase your connection and commitment to our success. Visit www.cmtausa.org or call 1-800-606-2682 to see how you can be a part of the end of CMT.

Together we will make our vision of a world without CMT a reality!
Warmly,



PATRICK LIVNEY, CEO



Beauty and the Beast: Genetic Heterogeneity

BY LAWRENCE WRABETZ, MD

Today I write about how the medical classification of CMT neuropathies both simplifies and yet confounds the development of treatments. The key phrase is genetic heterogeneity. OK, don't leave me just yet—the news is good on genetic heterogeneity and it involves the CMTA STAR initiative that you all know so well.

CMT neuropathies are genetically heterogeneous. Many of you will have heard your doctor mention this at a medical visit. What does it mean? Well, the idea is that even if many CMT patients have similar problems, their neuropathies are genetically different. OK, great, but what does that mean?

We are all familiar with the similarities of CMT neu-

ropathies: cramps, weakness and loss of muscle bulk in the feet and hands, foot deformities, difficulty keeping one's foot up with resulting loss of balance, clumsiness in the hands, slow reflexes and fatigue.

However, there are also differences—genetic heterogeneity. More than 1,000 different mutations (changes in the DNA) in more than 80 genes have been identified in CMT neuropathies. This number is changing so rapidly that within two weeks of writing this article, these numbers will have grown. This is an important result of both the human genome project, which produced an almost complete sequence of the DNA from several people, and the technological sequencing revolu-

tion, sometimes called Next-Generation or Next-Gen sequencing. These two advances have made it possible to sequence a patient's genomic DNA in only a few days, at a cost that is likely to soon fall below \$1,000, and then analyze the results quickly, using powerful computers to search for mutations in all genes. The result? New genes and mutations in CMT neuropathies are identified every couple of weeks.

The good news (the Beauty) relates to the similarities of CMT neuropathies. Many treatments aimed at helping with symptoms and disability are actually effective in many CMT neuropathies. For example, which mutation you have in which CMT gene is unlikely to alter the benefit of orthotics (foot braces and splints) for walking.

The bad news (the Beast) derives from the differences among CMT neuropathies. Because many different genes are affected by multiple mutations, there are probably many different mechanisms that produce disease. At first sight this may not seem very important, but it is for the development of new medicines, whose design is based upon trying to target specific disease mechanisms.

This is where the CMTA STAR (Strategy to Accelerate Research) initiative becomes important. STAR is based on developing rapid screens for new therapeutic leads—that is, new

KNOW YOUR NUMBERS!

How "common" is your form of CMT?

CMT Type	Percentage of All Types of CMT	Percentage of Specific CMT Type
CMT1A	50%	70% of CMT1
CMT1B	5%	10% of CMT1
CMT1X	15-20%	NA
CMT2A	4-5%	20-25% of CMT2
CMT2E	1-2%	NA
CMT4	<10% in North America and Northern Europe*	

* The prevalence gets up to 40 percent in North Africa, where consanguinity is more frequent. The prevalence is about 20 percent in Spain because CMT 4A (GAP1) has a founder effect there. According to the National Institutes of Health's Genetics Home Reference guide, a founder effect is the reduction in genetic variation that results when a small subset of a large population is used to establish a new colony. In Northern Europe and the US, CMT4C (SH3TC2) is the most frequent recessive form.

What Can You Buy with \$2 Million?

The progress made by CMTA's Strategy to Accelerate Research (STAR) in 2014 is really exciting! Here's what more than \$2 million of CMTA investments in research produced last year:

CMT Type 1A

- ★ We continued the screening of cells at the National Institutes of Health (which the CMTA pays for directly).
- ★ We continued the characterization of robust laboratory models at PsychoGenics.
- ★ We developed and managed the CMT1A Patient Database.
- ★ We developed a deeper understanding of the role of PMP22.
- ★ We initiated the screening of compounds at Sanofi-Genzyme. More than 70,000 compounds have already been screened!
- ★ We screened 400,000 compounds at the National Center for Advancing Translational Sciences (NCATS). Analyses of the hits are underway at University of Wisconsin.

- ★ We initiated the laboratory model testing of Addex compounds.

CMT Type 1B

- ★ We developed a cellular assay to screen compounds at University of Iowa and NCATS.
- ★ We developed laboratory models for eventual screening of compounds at the University of Buffalo.

CMT Type 1X

- ★ We tested multiple compounds to measure their impact on the CMTX laboratory model.
- ★ We explored the potential direct use of adult stem cells to treat the laboratory model.
- ★ We committed to testing a compound, made by a German pharmaceutical company, in the CMTX laboratory model.

CMT Type 2A

- ★ We developed genotypic and phenotypic laboratory models.
- ★ We continued our efforts on assay development.

CMT Type 2E

- ★ We procured a CMT2E laboratory model to be characterized at PsychoGenics.
- ★ We demonstrated detection of aggregation in cells.

CMT Type 4

- ★ We are developing a potential translational pathway for commonalities in the various subtypes.

In work applicable to all types of CMT, we launched a collaboration with the New York Stem Cell Foundation to produce adult stem cells for eventual use in drug screening!

The CMTA raised more than \$3 million in 2014, with 66 percent of it going to research and 21.5 percent going to programming.

If you want a treatment for CMT, be a part of the exciting progress and the solution by investing in your future today. Visit www.cmtausa.org/give2star and make your tax-deductible investment in STAR, or send your check to STAR, PO Box 105, Glenolden, PA 19036.

—Thank you!

chemicals that “flip the switch” in cultured cells that have been engineered to model a disease mechanism of neuropathy. Such chemicals provide the first step toward discovering new drugs for neuropathies. The new chemical can be thought of as a

key for a lock where the lock is the disease “target.” Once a key has been identified that flips the switch of the target, it can be made into a medicine by mounting the key onto known chemical supports. The supports help to get the medicine to

where it is needed (in nerves or the spinal cord for CMT) and make the medicine more stable.

Even if there are many mechanisms of CMT disease, one important STAR objective is to look for ways to group the

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CMTA Community Shares Tips for Helpful Products

When I started volunteering for the CMTA five years ago, I was immediately struck by the warmth and helpfulness of the CMTA community. Connected by a powerful common experience, our community is continually willing to listen, advise and share their personal stories.

Now, as Director of Social Media for the CMTA, I love seeing how the community has grown and developed into a tremendous force for good. With more than 20,000 people on our online pages and groups, all of us are reminded that we truly aren't alone in our CMT-related trials.

Recently, members of our Facebook group have helped us come up with helpful products



**CMTA Social Media Director
Bethany Noelle Meloche with her
new handwarmers**

for our new "Helpful Products" board on Pinterest. From key turners to button hooks to microwaveable slippers, these lit-

tle tools can make life with CMT a whole lot less frustrating, and the suggestions come from the people who know firsthand!

I received one of our helpful products as a gift this month from a CMTA friend who knew my hands and feet can get unbearably cold due to CMT. They are plush handwarmers, designed to look like smiling pieces of toast (really!), that heat up when plugged into the USB ports on my laptop. Adorable and pain-relieving, they are also a reminder of the good that social media and our community can do.

I guess you could say our community has added warmth to my life in more ways than one! —*Bethany Noelle Meloche*

GENETIC HETEROGENEITY

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mechanisms of different CMT neuropathies. To accelerate STAR and its objectives, CMTA CEO Pat Livney has assembled a diverse group of scientists from all over the world who work on neuropathies. They meet monthly on the telephone and twice per year in person to focus on STAR objectives, including discovering common CMT disease mechanisms, which, as explained above, means identifying common targets for treatments.

Therefore, my message is, be optimistic! Although genetic

heterogeneity represents a challenge for therapy, the genetic revolution is ever more rapidly cataloging new mutations and genes for CMT. The catalog makes it possible for medical scientists to organize the mutations into larger groups with common mechanisms of disease, hopefully making them accessible to similar treatments.

It is worth pointing out that some treatments may even cross traditional lines that separate diseases—lines that are defined by which area of the body is affected, i.e., nerves in the case of CMT. For example, we recently found a disease mechanism of some CMT neuropathies that

involves proteins not properly folded into their final shape. This disease mechanism is shared not only by CMT, but also by other more common degenerative diseases in other tissues, including some kinds of Alzheimer dementia and diabetes. The hope is that new medicines being developed for protein-folding problems could be successful in all of these diseases, including some CMT neuropathies. ★

Dr. Lawrence Wrabetz is a member of the CMTA Scientific Advisory Board, a professor of neurology and biochemistry and the director of the Hunter James Kelly Research Institute at the University at Buffalo.

TO SEE MORE HELPFUL PRODUCTS on the CMTA's Pinterest page, visit www.pinterest.com/CMTAssociation. If there is a product that has helped you live with CMT, please let us know by emailing Bethany at bethany@cmtausa.org. To connect with the CMTA community on Facebook, join our private group at www.facebook.com/groups/CMTAssociation.

Partnerships with Patient Groups Accelerate Therapeutic Development

BY CHRISTOPHER P. AUSTIN, MD, DIRECTOR,
NATIONAL CENTER FOR ADVANCING TRANSLATIONAL SCIENCES

Developing interventions for better human health differs from developing consumer goods in a fundamental way: Interventions to improve health generally are developed without direct input from the people they are meant to benefit. I believe this odd fact of history is responsible for much of the inefficiency and ineffectiveness of translational science, which “translates” findings from basic science into practical applications that enhance human health and well-being. Imagine for a moment that developers of a new snack food locked themselves away in a lab where the criteria for success were novelty and appeal to fellow developers, but they did not consult any consumers. The result could be an innovative snack food that would delight the developers but taste terrible; no one would buy it. This kind of scenario happens often in intervention development.

Patients—or more broadly, people, because most of us are or eventually will be patients—bring data, insights, connections, priorities and urgency to translational research projects. These cannot all come from professionally trained scientists. (I say “professionally trained” because many patients and families become experts in the diseases affecting them.)

To make the most of the enormous value patients can offer, I have challenged NCATS scientists to involve patients from the beginning in every project we do, as full members of the team. The NCATS-supported Rare Diseases Clinical Research Network is a leader in this approach and has had remarkable success in the pre-clinical arena enabled by a partnership with the patient-led Charcot-Marie-Tooth Association.

Patient groups like the CMTA bring funding, expertise in disease biology and advice on meaningful intervention approaches...

Charcot-Marie-Tooth disease (CMT) is the most commonly inherited disorder of the peripheral nervous system, affecting more than 2.8 million people worldwide. This incurable disease slowly damages the nerve cells leading to the arms, hands, legs and feet and results in pain as well as loss of muscle and sensation. A common form of CMT is caused by abnormally high production of a gene called PMP22; blocking this gene’s over-expression could potentially lead to new treatments. But researchers had never identified a small molecule drug with this sort of activity.

Several years ago, the CMTA proposed to NCATS a partnership to develop testing

systems that would help identify chemical compounds to transform into potential drugs to treat CMT. As part of the partnership, NCATS developed a new assay technique to screen for compounds that lower PMP22 expression. Because translation is a team sport, NCATS scientists accomplished this rapid work by collaborating with researchers at the National Human Genome Research Institute, the University of

Wisconsin and Sangamo BioSciences. To create the assay, the scientists used a new technique called genome editing to insert biological tools known as reporter genes into the DNA sequence of PMP22 in cells grown in culture. This technique is more specific than past methods, which inserted reporter genes at random locations into the cell’s DNA. The increased specificity led to discovery of an expanded number of potential treatment targets.

Collaborative relationships like this are truly synergistic: Patient groups like the CMTA bring funding, expertise in disease biology and advice on meaningful intervention approaches and NCATS brings expertise in therapeutic development. The result is a more patient-relevant and efficient route to new treatments—true translational innovation. ★

BULLYING: *What It Is, What It Is Not and How to Prevent It*

BY CMTA ADVISORY BOARD MEMBER SARAH KESTY



If your child has CMT, you probably worry about bullying. How can you protect your child

when he or she is at school and you're not?

First, you need to know what bullying is—and what it is not. Many of the student conflicts that get attention today are

not actually bullying. This is not to say that they are a lesser concern or not as hurtful and they certainly need to be addressed. But knowing the type of problem can reveal the best solution.

• **BULLYING** comprises repeated behaviors that intentionally cause physical and/or emotional harm to the victim and involve an imbalance of power between the bully and victim. Think of the movie-style bully who beats

about the problem are all ways to address conflicts.

• **MOMENTARY MEANNESS** is when one child is intentionally cruel to another, but the children are of equal status/power and it does not happen repeatedly over time. A child lashing out and calling another “stupid” would be an example of meanness. Meanness can be addressed with adult help and empathy lessons/activities.

• **TEASING** is when the parties involved are having fun, even though the content of the words could appear unkind. Think of two children laughing while calling each other “turkey” or “crazy heads.” It’s best to monitor teasing to be sure it isn’t masking or evolving into another issue, but it is generally harmless.

So, is it possible to make a child or school “bully proof?” Probably not, but there are ways to reduce the occurrence and impact of bullying. Here are two of my favorites:

• **CONFLICT** is a disagreement between two or more children of equal status, making all parties feel unpleasant, and usually having a solution. An example would be an argument over taking turns in a game. Peer mediation, teacher/counselor questioning or coaching, and opportunities to talk

• **EMPATHY:** Encourage staff to create a climate of empathy (ability to “walk in someone’s shoes”) at your child’s school. Students who can assume each other’s perspectives and experiences are far less likely to cause each other intentional harm and much more likely to speak up when they witness it. Focusing on the ways people can help one another (rather than the current anti-bully focus) embodies what we’re asking of students—that

CLASSROOM CONFLICTS



they adjust their “lenses” to see kindness and opportunities to support each other, rather than reacting out of fear and isolation.

• **PRELOAD THE COMEBACKS:**

Any child, with any ability level, will be the recipient of a mean phrase or two in school. Often, the child’s reaction to the meanness will impact whether or not it happens again; dramatic victims can draw attention and reinforce the potential bully’s experience. (I should know; I was a tear faucet growing up and wish I’d known how much the mean girls loved it.) Teach your child go-to phrases to use when someone is unkind. Something as simple as, “Really? Get a life!” or “It’s so sad you need to be mean all the time,” can catch a mean student off guard and take

away the fun for them. Having the comebacks preloaded and rehearsed will ensure your child can say them with very little emotion, potentially holding back the tears for when he or she is safe to let them out.

If your child comes home upset, listen to him or her. Be patient, open, and hear what he or she is saying—and do your best not to react as strongly as you’d like. Hearing your child’s suffering (maybe even revisiting your own) can be incredibly painful, but your anger or devastation may cause your child to leave out parts of his or her account for fear of upsetting you. Remember, some children will feel ashamed of bullying and will not report it, even to their parents. Look for signs of with-

drawal or changes in mood as possible indicators of social issues at school.

Most schools are appreciative of tips about students’ social interactions, so express your concerns to the school staff. Ask what their plan is to address the issue, then give it some time. You can check back with the school after a few days to be sure things have improved.

Bullying, mean moments, and conflicts will always be a part of growing up, but you can teach your child to handle them! Try to honor your child’s feelings, listen, and explore the lessons and growth hidden in the darkest of moments. You’re raising a wonderful child, and the CMTA community is always here to help! ★



WHAT’S ON YOUR MIND? ASK DAVID.

Dear David,

I am a 56-year-old woman who has had CMT for many years. I am grateful that although I have some mobility issues, I can still do and accomplish many things. My problem is that I am not sure how to handle good friends who are hesitant to call me because they know that I can’t keep up with them and don’t want to make me feel bad. I am feeling a little depressed and isolated, and I know how important it is for my overall health to have some kind of social life and be a part of a community.

David replies:

It takes work to maintain strong friendships, particularly as one gets older, whether or not one has CMT. All of us tend to waste time in thinking that

friends might not be calling as frequently. More often than not, they are simply involved with their own struggles. I like what noted self-help speaker and author Byron Katie says: “It’s not your job to like me; it’s mine.” Reach out to those friends and suggest getting together to do something that involves less physical strain. In other words, be proactive instead of feeling sorry for yourself and imagining reasons why they are not calling. When you begin to have negative thoughts concerning your friends not calling and think it’s because of your CMT, Byron Katie suggests asking yourself four questions: Is it true? Can you absolutely know that it’s true? What happens

when you believe that thought? Who would you be without that thought? So often our depressing thoughts stem from our own irrational thinking. I have found that most people are sensitive to my limitations. When they ask me how I am doing, I tell them directly and try not to dwell too long on the subject.

There is so much more to us than our physical health. Staying in touch by phone or computer is a great way to maintain connections. Thankfully, in 2015, there is no excuse to feel isolated. Don’t be shy about expressing your feelings to friends in a way that doesn’t provoke guilty feelings. Just say you miss them and would love to see them. ★



David Tannenbaum has an LCSW degree and has been a psychotherapist in New York City for the past 30 years. Write to David at info@cmtausa.org.

Visiting a CMT Center of Excellence

BY CMTA BOARD MEMBER STEVEN WEISS



Readers of *The CMTA Report* are well aware of the organization's work funding

promising research projects, supporting numerous branches for patients and their families, making information and resources available on its website and through social media, and so much more. When one considers everything the CMTA does on behalf of people with Charcot-Marie-Tooth disease, the CMT Centers of Excellence should also come to mind.

Thanks to the CMTA, there are now 14 Centers of Excellence in the United States, and three more in other countries. The centers, which are listed at www.cmtausa.org/coe, have been vetted by the CMTA's Scientific Advisory Board to ensure they offer comprehensive, high-quality care to people with CMT. The centers are led by some of the world's leading CMT experts, who are focused on finding treatments and cures for the disease.

A few months ago I had the opportunity to accompany a member of the Washington, DC Metro CMTA Branch to the Center of Excellence at Johns Hopkins University in Baltimore. Bella Eshuali is not your average person with CMT—she was born and raised in the African country of Cameroon. Until she moved to this country

a few years ago, she had never met anyone else with CMT or seen a doctor who knew anything about the disease. Desperate for help with her severely weakened hands and feet, she entered a lottery for a visa to come to the United States, and she was one of the few who were chosen.

Shortly after arriving in the Washington area, Bella was able to get a pair of leg braces. The braces helped her walk a little more easily than before, but they were bulky, heavy and painful to wear. She couldn't find shoes to fit comfortably over her braces, so even in the coldest months of winter she got by with loose-fitting sandals and a few

layers of socks. She also had a lot of trouble with her hands—tasks like opening bottles and picking up change were virtually impossible for her.

At Hopkins, Bella received a thorough evaluation, starting with Dr. Tom Lloyd, the clinical director. Tom gently asked Bella several questions, took careful measurements of her arm and leg strength, and closely evaluated her ability to walk and move. He also recorded her information for her file, and

with her permission he entered it into the large database that researchers are using to track the progression of CMT and learn about its impact on people's lives. This is one of the most wonderful things about visiting a Center of Excellence. We can contribute to CMT research by adding our information to the

Rare Disease Clinical Research Network database.

But Bella's visit was far from over. She spent most of the day in the clinic, as is standard practice at all CMT Centers of Excellence. In addition to Dr. Lloyd, she saw an occupational therapist, who talked about grips and other devices Bella could use to make things easier for her in the kitchen and around



Bella Eshuali, originally from Cameroon, recently visited the CMT Center of Excellence at Johns Hopkins.

the house. She also saw a physical therapist, who asked about her daily activities and recommended a type of leg brace that would work for her. Bella got a prescription from Dr. Lloyd that enabled her to get a better pair of braces, and the clinic accepted her health insurance, so Bella paid virtually nothing out of pocket. Life is still difficult for her, but now she better understands her condition and her limits, and she is leading as active a life as she can. ★

New Year, New Membership Benefits

With the ringing in of the New Year, the CMTA is excited to announce some exciting changes in membership benefits, as well as an entirely new and exciting membership option.

Here are the new CMTA memberships and benefits:

Registered Member of the CMTA Online Community

(Free)—Become a member of the CMTA Online Community at www.cmtausa.org/join, and you receive the following benefits:

- Online access to valuable information about living with CMT
- Access to the CMTA's Online Community, including access to CMTA Branches and Discussion, Circle of Friends and CMTAthletes groups
- A free information kit and a 10 percent discount at the CMTA store

Subscription Member (\$30 per year/\$80 for 3 years)

Subscription Members receive all benefits listed above, plus:

- *The CMTA Report*, the CMTA's bimonthly newsletter (electronic and/or hard copy)
- The *CMT Facts* series (electronic copy)

STAR Member (\$100 per year/ \$270 for 3 years)—

In addition to all the benefits listed above, STAR Members receive:

- One free pair of Aetrex Shoes in the style of their choice (new STAR members only)
- 50 percent off all Aetrex shoes (new and renewing STAR members)
- An expanded and updated copy of the CMT Survivor's Guide
- A 20 percent discount at the CMTA Store
- One *You're a STAR* auto decal

Wait! I'm currently a Premium Member. What happened to my membership?

1. Your current Premium Membership has been converted to a STAR Membership, and your expiration date will remain the same.
2. If your Premium Membership was set to auto-renew, the auto-renew feature has been cancelled due to the change in membership price.
3. You will receive a notice two weeks before your new STAR Membership expires, and at that time you can renew as a STAR Member or change to a Subscription Member. ★

JOIN THE CMTA TODAY to start engaging with others with CMT and to learn all you can about how to beat this disease!

REACH OUT TO YOUR "CIRCLE OF FRIENDS" TO RAISE FUNDS FOR CMT

Still trying to figure out how to help the CMTA find a treatment in 2015? How about a fundraiser that requires just a bit of your time, only takes a few minutes to set up, and doesn't require you to leave home?

The CMTA "Circle of Friends" (COF) provides an easy way for people to run a fundraising campaign to help the CMTA fund STAR Research. The Circle of Friends is a very personal appeal. Reaching out to those in your circle—friends, family, colleagues and acquaintances—and asking them to make a contribution to CMTA in your honor will greatly enlarge the number of people who support the CMTA. That, in turn, will enable us to fund more research and increase the chance of finding treatments. The CMTA website (www.cmtausa.org) offers a personal fundraising web page for each COF that's super easy to create, customize, and link to your own Facebook or Twitter account.

The COF fundraising web page can also be used to create a fundraising event, whether it's a walk, dinner or golf tourney. You'll be able to customize a page where folks can register for your event, make donations, and much more.

If you are interested in starting a COF with the CMTA, please contact Jeana Sweeney at jeana@cmtausa.org. TOGETHER, we can, and will, make a difference!



It's a Family Affair

BY ELIZABETH OUELLETTE

"The way you help heal the world is you start with your own family."

—MOTHER THERESA

It all started on one fateful day in Shelburne, Vermont, when I asked a few relatives to take part in the kick-off CMT Awareness month video of 2013. CMTA Director of Community Services Jeana Sweeney and I made up lyrics to a rap song and asked each participant to sing a line and get involved in spreading awareness of CMT. No recording contract came of the tune (no surprise there), but the time we spent together was precious from beginning to end. Despite our failed record debut, a lingering desire to somehow make a tangible difference in the life of my son Yohan and all those with CMT worldwide remained.

This adventure spurred conversation about the importance of fundraising to advance the CMTA's Strategy to Accelerate Research (STAR) initiative. Having recently taken up road biking, my brother Chris proposed a biking event to support Yohan and the CMTA's quest to

advance CMT research and Cycle 4 CMT™ was born.

Over the past 21 years, Chris's family and mine have stayed connected even though we live on opposite coasts. We spend summers in Vermont, and they come out to California for holidays. We even vacation together. Chris, his wife Mia and their two children, Warren and Lila, have become incredibly close to Yohan, forming tight bonds of friendship that will never fade.

I'm not sure Chris initially realized the time and effort involved in putting on a successful fundraiser, especially when planning started in June for a September event. Yet his determination and dedication to Yohan and the CMTA was unbelievably heartwarming and profoundly moving. Chris set out to support Yohan and make an impact in the community at large. And, when my brother Chris embarks on a meaningful journey, he puts his entire heart, soul and mind into making the most of the experience.

What started out as a small fundraiser turned out to be quite the event! Chris not only got friends, family and local business owners involved, but also random bikers, bike organizations and anyone who would listen to his emotional storyline. As Chris so eloquently put it, "Inspired by the positive and energetic attitude of my 21-year-old nephew, Yohan Bouchard, and the plight of the

2.8 million people affected by this disorder worldwide, our goal is to promote CMT awareness and support the ongoing research required to treat and eventually cure CMT." He readily connected with people by pointing out that "Simple activities that Vermonters enjoy and take for granted like hiking, biking, skiing or even walking are a challenge and might not even be possible for those with CMT." His plea did not fall on deaf ears.

With the invaluable support of relatives, and in particular, my sister-in-law Mia, who poured her creative energies into the event by making flyers, creating road signs and taking charge of the silent auction items, Chris's tenacity and commitment paid off big time. On Saturday, October 11, 2014, more than 100 people gathered for a day of biking, food, live music and heartfelt presentations. Thanks to Chris's relentless drive, more than \$50,000 was raised for the CMTA's STAR initiative.

With my family's active involvement, what I always thought of as "my cause" has now become "our cause." I feel blessed to have people in my life ready to join hands to make the world a better place for my son and all those in the CMT community.

And guess who is already in the beginning stages of planning for next year's second annual Cycle 4 CMT event? You got it—my brother, Chris. ★

From left to right:
Mia Ouellette,
Warren Ouellette,
Yohan Bouchard, Chris
Ouellette, Lila Ouellette





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Happy Hanukkah!

★ RAISING FUNDS AND

2nd Annual CMT
“We Got This” 5K
Walkathon

One year ago, I organized a fundraising walkathon along with my children—Jesse and Bethany Bertram. We all have CMT but are proud to say CMT does NOT have us!

Much has transpired in the year since then. There have been additional surgeries, hospitalizations and therapies, and Bethany was officially diagnosed with CMT, which we suspected she had. However, we still adamantly insist that CMT does NOT have us! The 2013 walkathon not only raised awareness of CMT, it also raised \$1,900 for the CMTA and was such a positive experience that we decided to do it again in 2014.

My personal goal this year was to raise awareness of this disease and to walk the course. I am happy to say I walked the entire 5K (3.1 miles) with my walker in front of me and my friends beside me.

We would like to thank our loved ones who participated in the 2014 “We Got This” Walkathon and generously donated \$1,000 to the CMTA. We are all in this together and get by with a little help from our friends, families and loved ones. We chose to name our walkathon “We Got This” because, yes, we do have CMT but we also like to say, “Keep calm, ‘cause we got this!”

—Angie Seames

CMT Walk and
Roll, Sarasota, FL

Braving winds of 25 mph, with gusts of more than 35 mph, scores of hardy souls walked and wheeled over the Ringling Bridge in Sarasota, Florida, on November 1, 2014, in the Sarasota, FL CMTA Branch’s CMT Walk and Roll. Members arrived early to festoon the entrance to Bird Key Park with banners, marking the registration point for the event. Banners in place, we waited to see who would come out on such a windy day. We watched the big whitecaps on the water and felt the sea spray at our registration table. Would the people who said that they would walk with us really come out when the wind threatened to blow them away? The answer was a resounding “Yes!”

At 10 am, we registered the first group of walkers and they started from the west end of the bridge. We had planned for everyone to hold up shiny blue stars emblazoned with the letters

CMTA in white, but the wind threatened to blow the stars away, so walkers clutched them to their chests. While the steepness of the bridge was a challenge, some members were able to go across the bridge and back, a distance of almost two miles. Members on motorized scooters were only able to go a short distance before the wind blew them back, but we applauded their fortitude for going as far as they were able.

The event drew support communitywide. The City of Sarasota Special Events Department not only obtained all the necessary approvals from the required city departments, but also arranged for us to have reserved parking spaces and a reserved pavilion to set up our registration area. Our 23 wonderfully generous sponsors came from all segments of the community: financial services, orthotics, certified public accountants, an insurance agency, pool service, wheelchair vans, doctors, an auto clinic, jewelry companies, seafood sup-



On a very windy day, Manuel Goldberg and Rachel Rivlin, left, and Sandy and Dick Dameron, right, hang on to the banner thanking the sponsors of the Walk and Roll.

AWARENESS FOR CMT ★

pliers, hairdressers, restaurants, and friends of our members. One of our biggest supporters was the party supply company that donated the blue stars that we carried, the letters CMTA to stick on the stars, and the banners. Participants also came from across the community. A fire chief brought his wife and four sons, carrying their baby in his arms while his wife pushed their 2-year-old in a stroller and their 5- and 7-year-olds walked across the bridge. A CPA came with his coworkers, an insurance agent with a friend and a physical therapist brought her sisters.

As we crossed the bridge, people passing in cars honked and waved to show their support. Walkers on the bridge stopped and asked us about CMT. We gave them the short version (we were braving strong winds!) of what CMT is, how many people have it, and how it affects us. It was a great opportunity to raise public awareness and educate people. That's what it is all about. If we can educate people and raise awareness, we hope that it will lead to raising funds to find a treatment and eventually a cure for CMT. In all, our CMT Walk and Roll raised \$3,390 to support the STAR research program. For a CMT branch that only came into existence in February 2014, we were very heartened by the support we received from our sponsors and friends. We all look forward to a world without CMT!

—Rachel Rivlin



Members of the winning team, left to right: Scott Wyte, Brandon Humpal, Jon Sheppard, and Gary Roberts.

6th Annual CMTA Golf Outing

Sixty-eight people from GE Aviation and Southern Industrial gathered together at The Crossings Golf Course in Durham, North Carolina, for a good cause. The CMTA focuses on helping to find a cure for CMT, a disease that impacts the nervous system. This disease is close to home for GE Aviation employees, as their friends, families and co-workers have been impacted by this disease. Every year they gather to raise funds to support the CMTA.

This year's golf tournament was a huge success. Eighteen foursomes teed off after a catered lunch from Danny's BBQ, sponsored by Alan Kelly. The day was filled with excitement, and a great time was had by all. Between mulligans, donations and golf entry fees, a total

of \$4,090 was raised for the CMTA. Special thanks to all of the sponsors (The Crossings, Carquest Auto Parts, Southern Industrial, Andrew Smarra and Lisa M. Angel, Rosen Law Firm, Mike Wagner, and James Bondeson) for supporting this event. Several prizes were awarded based on performance on the golf course. Congratulations to the Southern Industrial team on an outstanding finish!

—Kristen Neubauer

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CMTA Announces Collaboration with New York Stem Cell Foundation

The Charcot-Marie-Tooth Association announced in December that it will collaborate with the New York Stem Cell Foundation (NYSCF) Research Institute, a non-profit organization dedicated to accelerating cures through stem cell research, to develop a bank of induced pluripotent stem cell (iPSC) lines for a variety of neuropathies of known genetic causation. The ultimate aim of this research is to create a personalized medicine approach to rapid testing of human drug responsiveness in a dish.

As explained on the National Institutes of Health website, induced pluripotent stem cells are adult cells that have been genetically reprogrammed to an embryonic stem cell-like state, “de-differentiating” cells whose developmental fates had been previously assumed to be determined.

NYSCF will make stem cell lines from Charcot-Marie-Tooth patient materials that have been curated in a biobank assembled by Dr. Michael Shy at the University of Iowa, a member of the CMTA STAR consortium of sponsored investigators. Utilizing its automated technology, the NYSCF Global Stem Cell Array™, NYSCF will systematically generate iPSC lines from tissue samples obtained from patients representing a number of disease states.

These cell lines will then be used to develop methods for creating differentiated cells that

mimic the myelin-producing Schwann cells that are defective in Type 1 Charcot-Marie-Tooth (CMT) disorders of peripheral nerves, as well as the motor and sensory neurons that are defective in Type 2 disorders.

Members of the STAR consortium currently engaged in this CMTA-sponsored effort to differentiate iPSC lines include Dr. Robert Baloh, Cedars-Sinai Medical Center, and Dr. Gabsang Lee, Johns Hopkins University. The iPSC lines will also be expanded and banked by NYSCF and made available to the global scientific community to be used for research and the development of therapies.

Patrick Livney, CEO of the CMTA, called the NYSCF collaboration “an exciting opportunity for the CMTA to place research on therapies for Charcot-Marie-Tooth disorders in a personalized, patient context at a very early stage.” As Livney noted, “The Foundation has assembled the scientific and clinical key opinion leaders in CMT disorders, and the research tools necessary to validate therapeutic opportunities for their clinical potential. We have set out to engage drug makers to work together with the CMTA to advance new therapeutic approaches to our patients. Our STAR network

that combines this world-class research expertise with an operational capability has been highly enabling to the formation of collaborative alliances for this purpose.”

Susan L. Solomon, Co-Founder and CEO of NYSCF said the foundation is “very excited to partner with the Charcot-Marie-Tooth Association to develop resources that will enable the pursuit of new treatments and eventually cures for neuropathies. Partnering with the CMTA provides us with the necessary community of scientists, patients, disease experts, as well

“We believe that this type of interdisciplinary collaboration between various stakeholders is essential to move research forward in the pursuit of cures.”

as resources that allows us to move research forward. We believe that this type of interdisciplinary collaboration between various stakeholders is essential to move research forward in the pursuit of cures.”

NYSCF is an independent organization founded in 2005 to accelerate cures and better treatments for patients through stem cell research. It employs more than 45 researchers at the NYSCF Research Institute, located in New York, and is an acknowledged world leader in stem cell research and in developing pioneering stem cell technologies, including the NYSCF Global Stem Cell Array™. ★

For more information, visit www.nyscf.org.

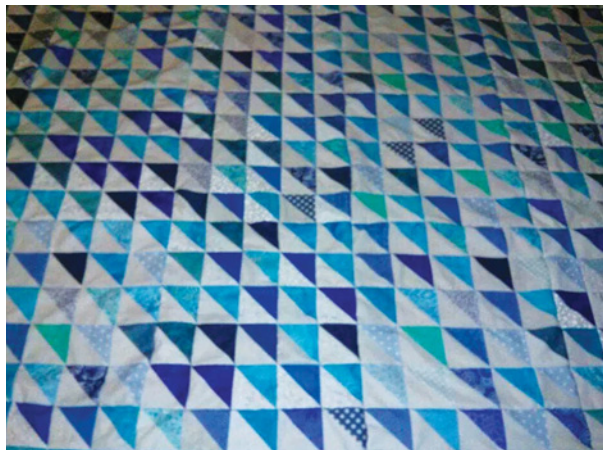
The Traveling Quilt

BY ANGELA BEAUMONT

CMT and making quilts are two constants in my life: CMT runs strong in my family and I've been quilting for about 20 years, though I've slowed down a lot since my disability began in 2010. I started the CMT Traveling Quilt Project because I wanted a new quilt for my bed, but when I finished the top in August, I happened to run across the idea of a signature quilt on TED Travels. I loved the idea so much I decided to put the two together. While I wondered how a signature quilt would do, when I put it in the CMT world on Facebook, the response was immediate. In one day the group grew by 75 members.

As the quilt travels, I'm asking for people who have CMT or a CMT connection to sign the quilt, make a quick entry in the accompanying journal, add a picture or post card and pay for the shipping to send it to the next person. I'm keeping all addresses confidential.

I never expected the quilt to be this popular. It's been hard to not get excited that it's wanted in so many corners of the world, though it can't seem to get off the East Coast: So far, it's been to Pennsylvania, West Virginia, New Jersey, Virginia and South Carolina. I have people patiently waiting their turns in Ohio,



CMT Traveling Quilt by Angela Beaumont

Georgia, Alabama, Louisiana, Nebraska, Washington, Oregon, California, Canada, the UK, Australia, Belgium, Romania, Scotland and South Africa.

Reactions to the quilt have warmed my heart more than any blanket. One Facebook friend said: "Right after I read this I broke down in tears thinking about how strange yet amazing it will be to touch something that so many people with CMT

have touched ... because I feel so alone with this disease ... other than my dad." Another said: "There are definitely some days where I just want to wrap myself up in a quilt and cover all the places that hurt. Then there are the days when my friends and family are the comfort I need. This project combines the two."

Because of the quilt, I'm becoming a CMTA branch leader in my area. The first meeting of the Vidor, TX CMTA Branch, serving southeast Texas and southwest Louisiana, is February 1, 2015. I never planned to get this involved, but here I am. The need is there.

You can follow the quilt on the Facebook page called "My CMT Traveling Quilt Project." ★

CMTA UPDATE COMING TO DALLAS!

The CMTA will hold a STAR Update on Saturday, March 21, 2015, at the Northwood Club, 6524 Alpha Road, Dallas, Texas 75240. CMTA CEO Patrick Livney will kick off the exciting program with an overview of STAR (Strategy to Accelerate Research), the CMTA's drug discovery program. He will be joined by speakers from the CMTA's STAR Scientific Advisory Board, Dr. John Svaren from the University of Wisconsin and Dr. Steven Scherer from the University of Pennsylvania.

This event provides a tremendous opportunity to hear and interact with these experts, as well as with staff and community members from the CMTA. Registration is just \$20 per person (non-refundable) and includes a seated lunch with the experts.

Don't delay. Sign up now for this informative morning conference. Register online at www.cmtausa.org/dallas or mail a check made payable to the CMTA with Dallas STAR Update in the memo line to PO Box 105, Glenolden, PA 19036. The deadline for registration is March 10, 2015. Seating is limited to the first 100 people to register.



CMTA BRANCH NEWS

• Arizona CMTA Branch

The group met on November 15, 2014, at the Burton Barr Central Library in Phoenix. Branch Leader Pam Palmer gave a summary of the Stanford Patient and Family Conference held in September 2014. Mary Almada from GeneDx (www.genedx.com) presented an overview of genetic testing and offered to answer questions about testing costs via email at malmada@genedx.com.

• Asheville, NC-Greenville, SC CMTA Branch

The combined Asheville-Greenville CMTA Branch met for the first time in Greenville

on November 18, 2014. Members spent the time getting to know each other, sharing stories and planning future meetings, speakers and events. Everyone also signed and took pictures of the Traveling CMT Quilt (*see related story p. 19*). The next meeting will be held in Asheville on Saturday, February 7, 2015, from 2-4 pm. Chad Dickenson, a podiatrist at the Orthopedic Appliance Co. in Asheville, will speak on braces.

• Hartford, CT CMTA Branch

Eighteen CMT patients and family members braved the cold and rain on December 9, 2014, to hear guest speaker Dr. Kevin Felice talk about the genetic factors causing different types of CMT and the resulting symptoms. Dr. Felice, a board certified neurologist and Chief of Neuromuscular Medicine at the Hospital for Special Care, discussed the facilities for CMT patients at his recently expanded department, including neurological, physical and occupational therapy evaluation. Several branch members are patients of Dr. Felice and recommend him highly. Branch leader Roy Behlke reviewed the status of CMT STAR research, including progress on CMT Types 1A, 2A, 2E, 1B and X, based on CMTA CEO Patrick Livney's presentation to all branch leaders. Behlke also discussed the year's fundraising efforts—the Hartford Branch Bowling Fundraiser, which resulted in a donation to STAR of \$2,380 and the Manchester Road Race, which raised \$1,500, for a com-

bined \$3,880 contribution to find a treatment. Roy also encouraged using IGIVE.com and Amazon Smile to benefit the CMTA when making online purchases.

• Naples, FL CMTA Branch

Branch Leader (and snowbird) Roy Behlke gave 13 members of the Naples FL CMTA Branch an update on CMTA news at their meeting at Naples Community Hospital on November 13, 2014. That news included the recently added partnerships with Genzyme and Addex, and the fact that both clinical and pre-clinical data by Addex show the potential of one of its drugs to address the needs of patients with CMT1. He also reviewed the events of CMT Awareness Month, including official proclamations by Florida Governor Scott and 19 other state governors. Roy reviewed the status of CMT STAR research, including progress on CMT Types 1A, 2A, 2E, 1B and X, based on CMTA CEO Patrick Livney's presentation to all branch leaders. Roy also presented charts from Katy Eichinger's webinar on the basics of balance since falls are a great risk to everyone with CMT. The next meeting will have a representative of GeneDx as guest speaker.

• Sarasota, FL CMTA Branch

Meeting in November for the last time in 2014, the group reviewed an impressive list of accomplishments for its first year of existence. Since the branch formed on February 22, 2014, members have heard speakers

(continued on page 23)

HOLIDAY LUNCHEON HELD IN HARTSDALE, NEW YORK

The Westchester, NY CMTA Branch's Holiday Luncheon/Fundraiser drew a capacity crowd of 72 enthusiastic participants to Pas-Tina's Restaurant in Hartsdale, New York, on November 1, 2014.

A delicious lunch was followed by a raffle that included 20 items and 18 gift baskets. Raffle items included a Pebble smart watch and Walmart and Sam's Club gift cards, many of which went for considerably more than face value. The generous winner of a 50/50 raffle donated her winnings back to the CMTA.

The process of getting all the gift cards, gift baskets and other donations took about a year but was well worth the effort, raising \$4,200 for STAR research. —Beverly Wurzel



From left to right: Luncheon workers Julia Wurzel, Kristyn Finelli, Beverly Wurzel and Mariah Wurzel

CMTA Branches

AL—North Florence

North Alabama CMTA Branch
Tina Smith
256-757-9250
Todd Oyen
256-810-6582

AZ—Phoenix Area

Arizona CMTA Branch
Pamela Palmer
ppalmeraz@gmail.com
480-236-3846
Jim Blum
480-272-3846

CA—Los Angeles Area

Los Angeles, CA CMTA Branch
Steve Fox
805-647-8225

CA—Los Angeles Area

Los Angeles, CA (South) CMTA Branch
Alani Price
310-710-2376

CA—Orange County

Orange County, CA CMTA Branch
Amber Biddle
714-343-3041
Myrna Andersen
310 987-3289

CA—South Bay Area

San Francisco Peninsula/South Bay CMTA Branch
Elizabeth Ouellette
800-606-2682 x107

CA—Sacramento

Sacramento, CA Support and Action Group
Holly Stevens
408-203-8804

CA—San Diego Area

San Diego, CA CMTA Branch
Jordan Thomas
619-549-0872

CA—Visalia Area

Fresno/Bakersfield, CA CMTA Branch
Melanie Pennebaker
559-972-3020

CO—Denver Area

Denver, CO CMTA Branch
Ron Plageman
303-929-9647
Dick Kutz
303-988-5581

CT—Hartford

Hartford, CT East CMTA Branch
Roy Behlke
239-682-6785

CT—North Haven

Southern CT CMTA Branch
Lynne Krupa
203-288-6673

DC—Washington, DC

Washington, DC CMTA Branch
Steven Weiss
Kimberly Hughes
301-962-8885

FL—Melbourne

Space Coast, FL CMTA Branch
Evelyn Quick
321-525-1336

FL—Naples

Naples, FL CMTA Branch
Roy Behlke
239-455-5571

FL—Sarasota Area

Sarasota, FL CMTA Branch
Rachel Rivlin
941-870-3326

FL—Tampa Bay Area

Tampa Bay, FL CMTA Branch
Vicki Pollyea
813-251-5512

FL—West Palm Beach

South Florida CMTA Branch
Phil Lewis
561-630-3931
Eileen Martinez
561-777-8471

GA—Atlanta Area

Atlanta, GA CMTA Branch
Susan Ruediger
678-595-2817
Katerina Marks
404-372-3158

IA—Iowa Area

Iowa City, IA CMTA Branch
Jeffrey Megown
319-981-0171

IL—Chicago Area

Chicago, IL (South) CMTA Branch
Dale Lopez
708-499-6274

IL—Norridge Area

Chicagoland (North) CMTA Branch
Charles Barrett
224-628-5642

IL—Springfield Area

Central Illinois CMTA Branch
Eileen Parn
217-787-8658

IN—Fort Wayne Area

Fort Wayne, IN CMTA Branch
Aimee Trammell
574-304-0968
Priscilla Creaven
260-925-1488

IN—Indianapolis Area

Indianapolis, IN CMTA Branch
Nancy Allen
317-459-8773
Patricia Wood
317-345-2254

KS—Wichita Area

Kansas Area CMTA Branch
Karen Smith
316-200-0543

MA—Boston

Boston, MA CMTA Branch
Mimi Works
617-913-4600
Jill Ricci
978-887-1014

MD—Baltimore

Baltimore, MD CMTA Branch
Clark Semmes
410-350-4812

MD—Easton

Easton, MD CMTA Branch
Missy Warfield
Seth Warfield
410-820-0576

ME—Portland Area

Portland, ME CMTA Branch
Mary Louie
207-450-5679

MI—Chesaning Area

Chesaning, MI CMTA Branch
Carolyn Koski
989-789-3656

MI—Kalamazoo Area

Southwest Michigan CMTA Branch
Jori Reijonen
269-341-4415

MS—Mississippi/Louisiana

Clinton, MS CMTA Branch
Flora Jones
601-825-2258
Cindy Chesteen
601-668-5439

NC—Ashville Area

Asheville NC - Greenville, SC CMTA Branch
Ruth Ann Carroll
(Asheville)
203-981-3738

NC—Durham Area

Research Triangle Area, NC CMTA Branch
Margaret Lee
919-359-6003

NC—Wilmington Area

Wilmington, NC CMTA Branch
Laurel Richardson
814-404-8046

NE—Lincoln Area

Lincoln, NE CMTA Branch
Karri Hood
402-641-0443

NJ—Central New Jersey

Central New Jersey CMTA Branch
Mark Willis
732-252-8299
Jacqueline Donahue
732-780-0857

NJ—Morris County

Morris County, NJ CMTA Branch
Alanna Huber
973-933-2635

NM—Albuquerque Area

New Mexico CMTA Branch
Gary Shepherd
505-296-1238

NV—Las Vegas Area

Las Vegas, NV CMTA Branch
Virgina Mamone
702-343-3572
Melissa Adams
702-339-0962

NY—Long Island Area

Long Island, NY CMTA Branch
Shari Loewenthal
631-254-8960

NY—Manhattan Area

New York, NY CMTA Branch
Amanda Imbriano
516-680-2981

NY—Upstate New York

The Upstate NY CMTA Branch
Elizabeth Misener
David Misener
518-527-0895

NY—Westchester Area

Westchester, NY CMTA Branch
Beverly Wurzel
Frank Wurzel
201-224-5795

OH—Cleveland Area

Cleveland, OH CMTA Branch
Heather Hawk Frank
440-479-5094

OH—Dayton Area

Dayton, OH CMTA Branch
Laura Wootan
937-859-6390

OR—Portland Area

Portland, OR CMTA Branch
Warren Beals
Joyce Beals
971-832-8272

PA—Bucks County Area

Bucks County, PA CMTA Branch
Linda Davis
Mitch Davis
215-943-0760

PA—Ephrata, PA

Ephrata, PA CMTA Branch
Rachel Weaver
717-984-8211

PA—Johnstown Area

Johnstown, PA CMTA Branch
J.D. Griffith
814-539-2341
Jeana Sweeney
814-269-1319

PA—Northwestern Area

Erie, PA CMTA Branch
Joyce Steinkamp
814-833-8495

RI—East Providence

Rhode Island CMTA Branch
Meredeth Souza
Raymond Souza
401-433-5500

SC—Greenville Area

Asheville-Greenville CMTA Branch
Rebecca Lauriault
(Greenville)
864-918-2437
Kitty Hodinka (Greenville)
610-761-6874

SD—Hartford Area

Hartford, SD CMTA Branch
Serena Clarkson
605-215-8853
Tom Clarkson
605-370-7595

TN—Nashville Area

Nashville, TN CMTA Branch
Bridget Sarver
615-390-0699
Gwen Redick
256-655-0391

TN—Savannah Area

Savannah, TN CMTA Branch
Reagan McGee
731-925-6204
Melinda White
731-925-5408

TX—Dallas Area

Dallas, TX CMTA Branch
Michelle Hayes
972-539-0905

TX—El Paso, TX

El Paso, TX CMTA Branch
Veronica Gallegos
915-203-4391

TX—Lubbock Area

Lubbock, TX CMTA Branch
Pam Downing
806-543-6647

TX—Vidor Area

Vidor, TX CMTA Branch
Angie Beaumont
281-229-2099

UT—Orem Area

Wasatch Front, UT CMTA Branch
Melissa Arakaki
801-494-3658

VA—Harrisonburg Area

Anne Long
540-568-8328

VA—Shenandoah Area

Northern Shenandoah Valley CMTA Branch
Teresa Frederick
540-336-4496

VA—Fredericksburg

Fredericksburg, VA CMTA Branch
Leigh Van Doren
540-370-1968

WA—Seattle Area

Seattle, WA CMTA Branch
Ruth Oskolkoff
ruth.oskolkoff@gmail.com

WI—Milwaukee Area

Southeastern Wisconsin CMTA Branch
Susan Moore
414-510-8736

WV—Vienna Area

Parkersburg/Vienna, WV CMTA Branch
Rebecca Knapp
304-834-1735

BRANCHES IN CANADA AND MEXICO

CAN—British Columbia

Victoria, BC CMTA Branch
Melanie Bolster
250-888-7713
Neville Tate
250-656-2547

CAN—Ontario

Eastern Ontario CMTA Branch
Robin Schock
613-389-1181

Northern Ontario CMTA Branch

Brenda Spencer
705-788-0408

Southern Ontario CMTA Branch

Kelly Hall
519-843-6119

Mexico

(This branch will be in Spanish.)

México CMTA Rama
Gina Salazar
Gina_oviedo@hotmail.com

Most CMTA Branches can be accessed via the CMTA Online Community at www.cmtausa.org.

In memory of Rick Alber, the family requests that donations be made to the Charcot-Marie-Tooth Association, PO Box 105, Glenolden, PA 19036, or online at www.cmtausa.org/donate2star. Please indicate that the donation is in memory of Rick Alber.

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BRANCH LEADER SPOTLIGHT

Heather Hawk Frank, Cleveland, Ohio

When Heather Hawk Frank was a child, her dad noticed she wasn't progressing like other kids. Her parents took her to a neurologist at the Cleveland Clinic who diagnosed her with neuropathy, but she wasn't officially diagnosed with Charcot-Marie-Tooth Type 1A until she was in high school.

Heather has never let CMT stop her from achieving her dreams. In kindergarten, she told her parents she wanted to become a teacher, and in 2006, she graduated from Ursuline College in Pepper Pike, Ohio, with degrees in education and history. She currently teaches seventh and eighth grade at Saint Mary of the Assumption School in Mentor, Ohio. Heather has a master's degree in education and is currently working on her principal's license.

While dating with CMT was an adventure, Heather met her husband Dave in 2005, and they were married in 2009. Heather counts her husband, along with her parents and sister, among her biggest supporters. This past year, Heather once again triumphed over CMT, losing more than 100 pounds and completing



Heather Hawk Frank with husband Dave Frank at the end of the Chase Your Tail 5k to benefit One of a Kind Pet Rescue

her first 5K (sporting her CMT awareness gear throughout). In her free time, Heather enjoys reading, traveling (especially to study history), and spending time with her nieces and nephew.

Heather knows of no other family members with CMT and met no one else with the disease while growing up. One night, after a difficult appointment at the neurologist, she was looking for information about CMT and found the CMTA. Although Heather was excited to discover a group working to raise awareness and find a treatment for CMT, she was disappointed to see that there were no support groups in the Cleveland area. A month later, she began to organize the Cleveland Support and Action Group. The group held its first meeting in May 2012.

Heather treasures the friendships she has made in the group. "It's great to be around people who truly understand my struggles and who are there for each other," she says. Each month the group's meetings include the good, the bad and the ugly, but they always include laughter. The Cleveland Support

and Action Group (now the Cleveland, OH CMTA Branch) meets every other month at Panera Bread in Brooklyn, Ohio.

Before starting the group, Heather never told anyone about her CMT or showed anyone her AFOs. Meeting others and starting the group gave her new confidence, though, and she now proudly wears her CMTA gear. She even met one of her good friends when she walked into a doctor's appointment wearing a CMTA shirt. The girl checking her in said, "I have CMT too." Soon after that, the two became close friends.

Heather is so grateful to have the CMTA in her life as well as all of the friends she has made in by being a part of this wonderful community. In the future she hopes for a world without CMT! ★

CMTA BRANCH NEWS

(continued from page 20)

with important information about living with CMT. They also raised \$1,305 for the CMTA by selling \$5 passes to Macy's Charity Shopping Day and another \$3,390 with a CMT Walk and Roll over the Ringling Bridge across Sarasota Bay (*see related article, p. 16*). It has been a busy, productive year for this new Sarasota, FL CMTA Branch, and they look forward to 2015.

• Southern CT CMTA Branch

Meeting on December 10, 2014, group members listened to and asked numerous questions of Kevin Felice, MD, Chief of the Department of Neuromuscular Medicine at the Hospital for Special Care in New Britain. Dr. Felice outlined the services available in the recently expanded department and shared how his department/hospital is positioned for upcoming clinical trials. He also stressed the importance of knowing what type of CMT one has and registering with the Rare Diseases Clinical Research Network at the National Institutes of Health (www.rarediseasesnetwork.org). Dr. Felice invited the nine members to visit the hospital and/or call to make an appointment.

• Washington, DC Metro CMTA Branch

Meeting on December 28, 2014, the group heard from Alex Davis of GeneDx (www.genedx.com), which specializes in genetic testing for rare hereditary disorders, including several types of CMT. The test must be prescribed by a doctor, and GeneDx works with patients and their insurance companies to make the test as affordable as possible. If you have questions, contact Alex at adavis@genedx.com. The group also recognized the passing of long-time CMTA employee Pat Dreibelbis. ★

CMTA MEMBERSHIP, PUBLICATIONS & ACCESSORIES ORDER FORM

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If you are a STAR member or are joining as a STAR member now, you may purchase publications and accessories at discounted prices. (Some exclusions may apply.) To check your membership status, please call 1-800-606-2682, ext. 105.

Subscription Membership Benefits:

- Online access to valuable information about living with CMT
- Access to the CMTA's Online Community, including access to CMTA Branches and Discussion, Circle of Friends and CMTAthletes groups
- An information kit and a 10% discount at the CMTA store
- Bimonthly delivery of *The CMTA Report*, (electronic and/or hard copy)
- The *CMT Facts* series (electronic copy)

STAR Membership Benefits:

- All of the benefits of Subscription Membership
- PLUS**
- One free pair of Aetrex Shoes in the style of their choice (new STAR members only)
- 50% off all Aetrex shoes (new and renewing STAR members)
- An expanded and updated copy of the CMT Survivor's Guide
- A 20% discount at the CMTA Store
- One *You're a STAR* auto decal

JOIN THE CMTA:

Subscription Membership <input type="checkbox"/> \$30/year <input type="checkbox"/> \$80/3 years STAR Membership <input type="checkbox"/> \$100/year <input type="checkbox"/> \$270/3 years	Choose your newsletter format (check one or both) <input type="checkbox"/> PDF <input type="checkbox"/> Print		
QUANTITY	COST		TOTAL
	Regular Price	STAR Member Price	
The Patients' Guide to Charcot-Marie-Tooth Disorders [CD Format]	\$10	\$8	
CMT Facts IV	\$10	\$8	
CMT Facts V	\$15	\$12	
CMT Facts VI	\$15	\$12	
My Child Has CMT	\$5	\$5	
Teaching Kids about CMT...A Classroom Presentation (DVD/1 hour)	\$10	\$8	
CMTA Titleist Hats Quantity and Color: Blue___ Lt. Blue___ Black___ White___	\$25	\$20	
CMTA T-Shirts <input type="checkbox"/> Navy Blue with white logo) <input type="checkbox"/> White with blue logo) Quantity and Size: S___ M___ L___ XL___ 2XL___ 3XL___	\$15	\$12	
CMTA Shark-O T-Shirts Quantity and Size: S___ M___ L___ XL___ 2XL___ 3XL___	\$20		
Be a STAR Wristbands	\$1.50 each*		
Be a STAR Necklaces (Includes battery)	\$5 each*		
Washable CMTA Tattoo (Pack of 5)	\$1 per pack		
CMTA Pin	\$3 each		
CMTA Brochure & Neurotoxic Drug Card	FREE		
Donation to the CMTA (100% Tax-deductible)			
Shipping & Handling (Orders under \$10, add \$3.50; orders \$10 and over, add \$7.50)			
*Quantity discounts for these items available online			
ORDER TOTAL			

☐ Check payable to the CMTA (US residents only; non-US residents, please use credit card or international money order.)

☐ Money Order ☐ American Express ☐ MasterCard ☐ VISA

Card Number: _____ Expiration Date: _____

Mail to: CMTA, P.O. Box 105, Glenolden, PA 19036; or fax to 610-499-9267

A copy of the official registration and financial information may be obtained from the Pennsylvania Department of State by calling, toll-free within Pennsylvania, 1-800-732-0999. Registration does not imply endorsement.

CMT PATIENT MEDICATION ALERT:

**Definite high risk
(including asymptomatic CMT):**
Vinca alkaloids (Vincristine)

**Moderate to
significant risk:**
Amiodarone (Cordarone)
Bortezomib (Velcade)
Cisplatin and Oxaliplatin
Colchicine (extended use)
Dapsone
Didanosine (ddI, Videx)
Dichloroacetate
Disulfiram (Antabuse)
Eribulin (Halaven)
Gold salts
Ixabepilone (Ixempra)
Leflunomide (Arava)
Metronidazole/Misonidazole
(extended use)
Nitrofurantoin (Macrochantin,
Furadantin, Macrobid)
Nitrous oxide (inhalation abuse)
Perhexiline (not used in US)
Pyridoxine (mega dose of
Vitamin B6)
Stavudine (d4T, Zerit)
Suramin
Taxols (paclitaxel, docetaxel)
Thalidomide
Zalcitabine (ddC, Hivid)

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

Negligible or doubtful risk:
Allopurinol
Amitriptyline
Chloramphenicol
Chlorprothixene
Cimetidine
Clioquinol
Clofibrate
Cyclosporin A
Enalapril
Glutethimide
Lithium
Phenelzine
Propafenone
Sulfonamides
Sulfasalazine

THE CMTAReport

The Charcot-Marie-Tooth Association
P.O. Box 105
Glenolden, PA 19036
1-800-606-CMTA (2682) FAX (610) 499-9267
www.cmtausa.org

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WHAT IS CMT?

- ▶ CMT is the most commonly inherited peripheral neuropathy, affecting approximately 150,000 Americans.
- ▶ CMT may become worse if certain neurotoxic drugs are taken.
- ▶ CMT can vary greatly in severity, even within the same family.
- ▶ CMT can, in rare instances, cause severe disability.
- ▶ CMT is also known as peroneal muscular atrophy and hereditary motor sensory neuropathy.
- ▶ CMT is slowly progressive, causing deterioration of peripheral nerves that control sensory information and muscle function of the foot/lower leg and hand/forearm.
- ▶ CMT causes degeneration of peroneal muscles (located on the front of the leg below the knee).
- ▶ CMT does not affect life expectancy.
- ▶ CMT is sometimes surgically treated.
- ▶ CMT causes foot-drop walking gait, foot bone abnormalities, high arches and hammer toes, problems with balance, problems with hand function, occasional lower leg and forearm muscle cramping, loss of some normal reflexes, and scoliosis (curvature of the spine).
- ▶ CMT has no effective treatment, although physical therapy, occupational therapy, and moderate physical activity are beneficial.
- ▶ CMT is usually inherited in an autosomal dominant pattern, which means if one parent has CMT, there is a 50% chance of passing it on to each child.
- ▶ CMT Types that can now be diagnosed by a blood test include 1A, 1B, 1C, 1D (EGR2), 1E, 1F, 1X, 2A, 2B, 2E, 2F, 2I, 2J, 2K, 4A, 4C, 4E, 4F, 4J, HNPP, CHN, and DSN.
- ▶ CMT is the focus of significant genetic research, bringing us closer to solving the CMT enigma.