

THE CMTA Report

SEPTEMBER/OCTOBER
2013

Information on Charcot-Marie-Tooth Disorders for patients, families, and the scientific community ★ www.cmtausa.org

REPORTS FROM ATTENDEES AT THE CMT CONSORTIUM IN BELGIUM

CMT in Malaysia

*Tey Shelisa, Faculty of Medicine,
University of Malaya*

On the 27th of July, I travelled 9,000 miles from Kuala Lumpur to Antwerp to attend a meeting that would leave a deep impact on me. I had been selected to present my findings in front of experts in the field, some of whom also had traveled across the globe to be there! As I stepped onto the stage, I pushed myself to breathe and I began to tell the story of my work on CMT in Malaysia. Let me share some of that story here with you.

I work in a team together with Azlina Ahmad-Annuar, Sarimah Samulong, Nortina Shahrizaila and Goh Khean Jin, and we have been working on a Malaysian family with CMT. The parents are of Indian ethnicity and are first cousins. Two sons are affected with CMT, while a daughter and another son are normal. When their eldest son first came to the hospital, he was only 13 years old. Clinical examination revealed that he had CMT, but he also presented with some extra phenotypes which are not typically seen in CMT patients. He has been a regular visitor to the neurology and physiotherapy clinic since then. Seven years later, his youngest brother was also diag-



CELEBRATING AWARENESS, Southern Ontario Support and Action Group members Ron Detta, Kelly Detta-Hall and Julie Detta pose with Michael Chong (Member of Parliament of Wellington-Halton Hills) to show off the proclamation recognizing September as CMT Awareness month in Canada.

nosed with CMT at 13 years old and, in addition, he also has mild intellectual disability. We performed genetic screening of the commonly associated CMT genes on both brothers, and the tests were negative. Upon discussions with our collaborator Marina Kennerson in Sydney, we sent DNA from the brothers for exome sequencing to identify the causal gene.

Sixty-thousand DNA variants later, I am down to a few promising candidates, but there are still many others that need to be screened, and we are still refining the method to analyze the data. This is partly because the brothers are from a consanguineous marriage, so we are taking various approaches. The inheritance pattern in the family is suggestive of an autosomal

recessive disorder, but since only the brothers are affected, it could also be X-linked. The progress of analyzing the data is long. I need to validate each potential variant from the exome data and check to see if it is only present in the two affected brothers and not in their normal siblings and other normal individuals. I have tested almost 50 and there's still some way to go. We are excited about

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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.

OUR VISION:

A world without CMT.

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How to Organize a Fundraiser

BY HERB BERON, CHAIRMAN OF THE CMTA BOARD OF DIRECTORS

Back in May of 2007, my wife Rachael and I were talking about our family's charitable giving. Our son Josh was 10 years old and our daughter Julia was 6. When Josh was a newborn, he was diagnosed with a heart defect known as Tetralogy of Fallot. (He subsequently underwent open heart surgery when he was seven months old.) Our charitable donations at that point went primarily to the children's hospital where his surgery was performed. It was at that point—we had learned of Julia's CMT within the prior year—that we decided that we needed to start giving to CMT-related causes. During Internet searches, my wife found that there was someone swimming in the Chesapeake Bay in Maryland to raise money for the Charcot-Marie-Tooth Association. Since she was from Baltimore, my wife felt an immediate connection and somehow got in touch with Board Member Steve O'Donnell. The rest is history. I am one of those people who know what he's good at (and more importantly, know what he's *not*!) For those who know me, swimming is clearly not my forte, but I knew that I was organized and could put together a plan to start a fundraiser.

Having been solicited by so many charities over the years, I felt that I had a decent idea of what is successful and what is not. The first step was to create a compelling reason for people to donate. I composed a well-

worded (but not too long) email, which I sent to my entire personal and business email directories. After speaking with the CMTA, we realized that we'd need our own specific fundraising website (to make it easy for donors to contribute).

Once that was done, our initial email was sent. I followed up with personal thank-you emails, swimming status updates, and a final picture. That first year, we really didn't know what we were doing. My wife and her sister were the only swimmers for Julia, yet we were able to raise a significant amount of money just through our personal and business contacts. The following year, many of our friends wanted to swim as well, and seven cars caravanned from New Jersey to Maryland. I asked all of the other swimmers to forward that email (or make their own) to all of *their* email directories as well. When a donation came in from someone whose name I didn't recognize, I would forward an email to all of the swimmers and ask them to identify the donor. They could then send the appropriate thank-you.

The year after that, we decided to bring the swim to our home state of New Jersey. We belong to a local lake club and approached the manager about hosting the swim. He gladly provided lifeguards as well as boats to accompany the

swimmers (in case somebody started swimming like *me!*). Looking back, that was an important year for our swim. As this had been entirely about raising money, it now also became equally about raising awareness

The first step is to create a compelling reason for people to donate.

in our community. We actively recruited swimmers of all ages to join us (and only required them to spread the word). We started a silent auction—originally we asked local businesses (i.e. restaurants, retail

stores, gas stations, etc.) to provide gift baskets or gift certificates. We would package several restaurant certificates together in one basket and call it "Taste of Montville" or "Caldwell Eats." We also asked the swimmers to solicit businesses or obtain any items that we could place in our silent auction. We now regularly get donations of sports and entertainment tickets, memorabilia, and even several "experiences" (set visits to "Modern Family," "NFL on CBS," "Wipe-Out," and the "Rachael Ray Show," just to name a few). It turns out that *everybody knows someone* who can offer something unique that can be auctioned. That year, we also started having custom tee-shirts donated (that we sell at the event). Three years ago, we started our own TeamJulia Facebook page (which we use to spread the word to an even larger audience). Our swim has

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become a Labor Day tradition in our town—and now as Chairman of the Board, it makes it even sweeter for me that it happens at the beginning of Awareness Month. At the time of this writing, TeamJulia has raised \$538,226.73 (and counting!) since its inception!

The bottom line—fundraising is hard ... but my primary suggestions are:

- **Make your fundraising “ask” personal.** Almost everyone you know gives to some form of charity, and especially to causes of friends (or friends of friends). Don't be scared to open yourself up. It's difficult, but it works

(and the correspondence that you'll receive from so many people is often *very* therapeutic as well).

- **Get creative.** We never thought about having a silent auction before. Now it has turned into a lot of fun and is very profitable. We have 100-150 people attend our event each year, and everyone looks forward to seeing which auction items we get each year.
- **Keep up the correspondence with your donors.** Send personal thank-you emails (or make calls) to let them know how much their gifts mean to you.

Send updates on your event—we send our first email three months before the actual event, then follow up with status emails each month until the day of the event, and then a final email with pictures, etc. On your subsequent emails, keep them short and funny. (I'll often joke about the swimmers working on their stamina, etc.)

Best of luck always—if you ever have any questions about the process, feel free to reach out to me at hberon@optonline.net. ★

SPREADING AWARENESS...

Many of the CMTA's Support and Action Groups and their members requested proclamations from their Governors or Members of Parliament recognizing September as CMT Awareness Month. Along with Canada (pictured on page 1), the states of CT, NM, NJ, NV, MD and TN all sent proclamations.



REPORTS FROM ATTENDEES AT THE CMT CONSORTIUM IN BELGIUM

MALAYSIA

(continued from page 1)

our promising candidate genes, because, to our knowledge, these genes have not been linked to CMT before. To be sure about which gene is the culprit, I will need to examine the effect of the mutation on the gene and analyze how this could lead to CMT.

This family is one of several that we have recruited from the University of Malaya Medical Centre (UMMC). Our team pioneered research into CMT in Malaysia, and, in fact, literature on CMT in the rest of Southeast Asia is lacking. Recently, we published a paper in *Muscle Nerve* on 25 Malaysian CMT cases. Around 60 percent of the cases had mutations in the commonly associated genes, PMP22 and GJB1. Based on this cohort,

we found that X-linked CMT was the predominant type in Malaysia, which differs from other studies reported in Korea, Japan and China, where CMT1A (PMP22 duplication) is the most common subtype.

The meeting in Antwerp really energized me and my research into CMT, and the advice that I received from the other researchers has been invaluable. I was impressed with how freely knowledge and experience were shared, and the imaginative approaches of the researchers inspired me to be more creative in my scientific approach. I would like to express my gratitude to the CMTA for the travel grant support and also to the families participating in this study. Nine thousand miles was a long way to go, but it was worth it!

these mice were studied well into their adult life. Nerve conduction and structure appeared normal, even in older mice. In addition, most behavior and physical ability was unimpaired by either mutation. However, the R94W mutation caused an age-dependent, progressive decline in unprovoked movement, so that by two years of age, R94W mice covered only 75 percent of the distance that their wild type littermates voluntarily explored. While these mice do not display the severe phenotype seen in humans, they can provide us with cells that can be used to screen potential drug therapies.

Aminoacyl-tRNA Molecules

Na Wei, Post-Doctoral Researcher, Scripps Research Institute, CA

I'm a postdoctoral researcher at The Scripps Research Institute in California. Studies in our lab are mainly focused on aminoacyl-tRNA synthetases. Over the last decade, tRNA synthetases have emerged as a new class of regulatory molecules with widespread functions beyond their classic role in protein synthesis. At the same time, they have become one of the largest protein families associated with CMT diseases.

In my research work, I found that tyrosine tRNA synthetase (YARS or TyrRS), whose mutations are causally linked to a dominant intermediate type of CMT (DI-CMTC), enters into the nucleus and activates a transcription factor E2F1 when cells

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PEDIATRIC CLINIC OPENS FOR CMT CHILDREN IN FLORIDA

Dr. Richard Finkel has started a new multidisciplinary neuromuscular clinic in Orlando at the Nemours Children's Hospital. This new facility opened in October, 2012, and is already seeing many infants, children and adolescents with both common and rare disorders of nerve and muscle, with a special focus on CMT. Dr. Finkel has participated in the Inherited Neuropathy Consortium and helped develop the "CMT Peds Scale," which is now the standard means of evaluating children with CMT. Team members include Dr. Finkel (Neurology), Dr. Stephanie Reid (Pediatric Physical Medicine and Rehabilitation), Dr. Steven Frick (Orthopedics), Carrie Crain, MS (Genetic counselor), Sissy O'Reardon, ARNP (Neurology Nurse Practitioner), Matt Civitello, MPT, and Jenna Turner, MPT (Physical Therapy), Betsy Burgos, OTR (Occupational Therapy and Assistive Technology), Aimee Molineaux (Medical Nutrition) and social work support. Dr. Finkel also performs EMG and nerve-conduction testing on children. To inquire about this CMT clinic call 407-650-7250 or email Heather.Franceschini@nemours.org.

Characterization of Two Mitofusin 2 Knock-in Mouse Models

Alleene Strickland, Neuroscience PhD Candidate, University of Miami Miller School of Medicine

These mice contain the common, dominant R94W and R94Q mutations in Mfn2, a gene associated with many severe cases of CMT. In mice, inheritance of two copies of either gene is lethal to newborns, but we were able to study cells harvested from these mice, which showed deficits in energy production and mitochondrial function and structure.

Inheritance of a single copy of either gene was not lethal, so

The Seven Stages of Drug Development

As you probably know, the CMTA is aggressively fighting to find a pharmaceutical treatment, and, ultimately, a cure for all types of CMT. We have gathered a robust Scientific Advisory Board, found experts in the industry, and forged partnerships to accomplish our goals. Through these collaborations, we have the right knowledge base, technology, and expertise to deliver success. In fact, we have developed protocols and partnerships for 90 percent of all types of CMT—1A, 1B, 2A, and X.

In our journey toward success, there are seven stages for drug development we must accomplish:

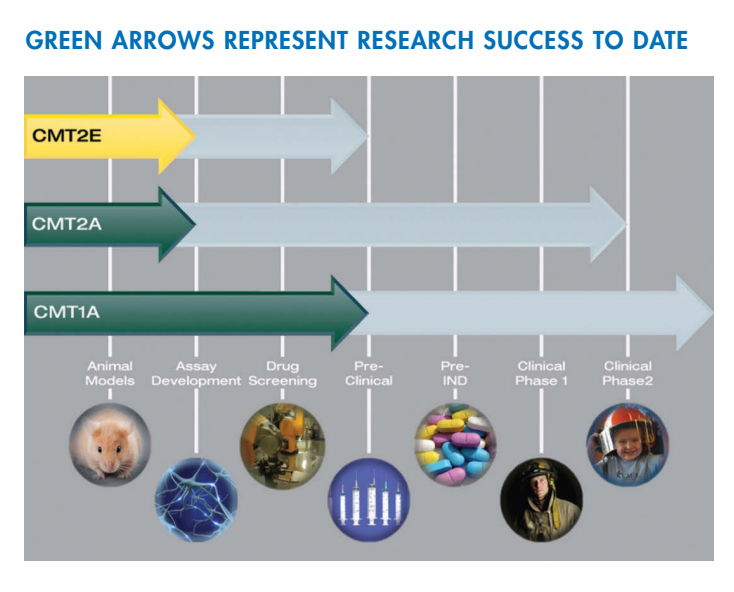
1 Create laboratory disease models that replicate each type of CMT. Once we have a robust population with offspring that exhibit symptoms, we can use those models to test candidate pharmaceuticals.

2 Build cellular assays. Create and reproduce cells with each type of CMT that we can use for large testing of many different pharmaceuticals.

3 Screen the cellular assays with different pharmaceuticals. This has been done for type 1A at the National Institutes of Health, with over 350,000 pharmaceuticals tested. Success has been achieved with one class of drug compounds, leading us to stage 4.

4 Test the candidate drug compounds in the laboratory models. Give the symptomatic models doses of candidate compounds to measure their improvement. Testing is currently underway for type 1A.

5 Test the compounds for dosage, toxicity and viability. This is done in private and academic laboratories to find



the exact dosing which will ameliorate the root cause of CMT.

6 Conduct pre-clinical Phase 1 human trials. A small group of strong, healthy people without CMT will take the dose of the compounds to test for human side effects and toxicity.

7 Conduct pre-clinical Phase 2 trials. Through the CMT Centers of Excellence, small groups of people with CMT (type specific) will engage in pharmaceutical trials to understand the positive impact the drug will have on CMT. Once these trials are concluded and the FDA has approved the use of the drug, it can be delivered to the market, accessible to

everyone with the type-specific CMT.

The graphic above outlines the seven stages with the green arrows demonstrating our current successes. The yellow arrow represents the protocols which have been developed. Since this graphic was created, protocols have been developed and financial support has been given for types 1B and X.

The CMTA is proud of our research achievements. We expect significant success in each of the stages in the coming months.

If you have any questions about our research, or if you want to support our research programs, please contact us at info@cmtausa.org.

As part of Awareness Month activities, people submit letters recommending their friends, family and others to be recipients of the "I'm a Shooting Star" (for children) or "I'm a Star" (for adults) awards. The awards celebrate the energy and enthusiasm with which people with CMT live life. This year the Awards committee chose Allison Stanley as the Shooting Star and Michele Kekac as the Star. Here are their winning letter entries.

Two 'Stars' Receive Awards

A Shooting Star: Allison Stanley

*Nominated by her parents
Mark and Christine Stanley*

We would like to introduce you to our ten-year-old daughter, Allison. She was diagnosed with Charcot-Marie-Tooth just over two years ago. She has CMT Type 4. We were very surprised because there is no history of CMT in the family. The doctors say that she is a rare case. She had surgery on both legs to lengthen her Achilles tendons shortly after her diagnosis. She spent eight weeks in a wheelchair while she healed. She now wears braces on her legs most of the time. She does physical therapy and horseback riding therapy to keep her strong.

She just started fifth grade. She is on the safety patrol. She is going to run for student council. She is also a nearly straight-A student. Her school is very supportive. They have even done a couple of fundraisers for her.

We have been involved with the Muscular Dystrophy Association since her diagnosis. She is currently the Michigan Ambassador for the MDA. We have attended many Lock-Up fundraising events to support the MDA. Our biggest accomplishment this year was raising over \$5,200 for the Muscle Walk. All of the money went to the MDA summer camp, which she attended for the second time this summer.

She is a very determined young lady. Recently, she had to make one of the hardest deci-

sions in her life. She had to give up her dancing. Her legs just were not strong enough anymore. Instead of getting mad or sad, she found something else that she loves. She took up horseback riding. She is amazing at it. She recently did a horse show and won a first, third and fourth place.

One of the biggest things that everyone notices about her is her smile. She has a smile that warms everyone's heart. She is also one of the most caring and loving young ladies that you will ever meet. She has never let this disease define her.

There is something she said that I will always remember. Shortly after giving up dance, she said to me, "Mom, I know my legs don't work right anymore, but I know that God gave me this disease for a reason. It was so I could help other people know about it."

We recently had to get a kit to turn her bike into a three-wheeled bike because she doesn't have the muscle tone to support a two-wheeled bike. Allison was just so excited because she finally had a bike to ride. We now incorporate this into her physical therapy program. She rides three miles at least three times a week.

We are so blessed to be a part of her journey with CMT as



Ten year-old Allison Stanley, diagnosed with Type 4 CMT, shows off her newly created three-wheeled bike which she uses in her physical therapy regimen.

she gets older. We know that with the love and encouragement of her family and friends, Allison will do amazing things and never let her CMT keep her from trying new things and loving life.

A CMT Star: Michele Kekac

*Nominated by her sister
Kalila Dias Brandao*

Michele was very young when the pain started. We lived in a small town in Brazil at the time, and all the doctors told us it was growing pains. She was very clumsy and fell all the time; her legs seemed too weak for her body. We never thought anything of it. In 2005, after years of suffering and seeing

countless doctors, Michele was diagnosed with CMT Type 1A. By that time, the disease had advanced considerably; her feet were full of calluses, and the pain had intensified.

We went through all the stages: first came denial, then acceptance, research, awareness, surgeries, disability, and, finally faith and an unbending will to make a difference. I cannot say how I'd react to this situation if I were in her shoes. I am certain that I would not be as brave, or as strong. As CMT played havoc with Michele's body by changing her life and limiting her abilities, it also changed her spirit and gave her purpose.

Michele is a CMT Star because of her kindness, strength and persistence. In January 2010, she started a blog, "This is my journey: My life with CMT." At first it was a way to communicate and update family and friends. Soon, it became an escape valve where she poured out her heart, anguish, hopes and fears. Then, other patients with CMT were drawn by her words and found hope in her strength. Most, like her, had never talked with or met another CMT patient. The blog became a way to connect to people who shared the same reality.

Fueled by her new experience, Michele decided to be more active in the CMT community. Through some of the

friendships she created online, Michele helped to create a group blog, "Clicks for CMT: Bloggers Raising Awareness." This group of motivated women helped raise much awareness during last year's CMT Awareness Month by chronicling their stories and experiences.

One of the hardest decisions my sister has had to make was to retire at the ripe old age of 31 from a job she loved due to chronic pain and chronic fatigue. Her firm was just as sad to see her go and honored Michele by selecting the CMTA as the organization spotlighted at one of their largest events. The Women's Tea in 2012 was a huge success and exposed many professionals

to CMT who had never heard of it. This event helped raise \$1,800 for the CMTA.

After Michele experienced the positive reaction from this event, she decided to make an even bigger impact. She is now planning her first fundraising and awareness event in her town of Shelton, CT.

She is hoping to sell over 100 tickets, and the donations have started coming in. With a month to go, she has already helped to raise over 30 percent of the \$5,000 goal. She is also spreading the word in the Connecticut media by giving interviews to local papers and is even taping her first on-air segment—all to raise awareness of this wonderful cause. ★



Michele Kekac is, indeed, a star because she is kind, strong and persistent and is currently working on a fundraiser for the CMTA in CT.

A Special Thanks to All Federal Employees

Thank you very much for your past support of the CMTA through the Combined Federal Campaign. In total, 2012's contributions were over \$20,000, allowing us to continue to meet our mission every day. As you know, the CMTA has three focus areas: improving the quality of life for those with CMT by providing resources and information, increasing awareness about CMT and funding research for the development of treatments for all types of CMT. Our progress includes:

- Creating 55 Support and Action Groups throughout North America, providing local resources for those with CMT.
- Building our social media presence and interacting with over 10,000 people through Facebook, Twitter, LinkedIn and Pinterest.
- Spreading the word about CMT to physicians and clinicians through trade magazines and partnerships.
- Gathering a robust Scientific Advisory Board, identifying experts in the industry, and forging partnerships to develop protocols and pathways for 90 percent of all types of CMT—1A, 1B, 2A, and X.

Your generosity has helped us achieve these major milestones. Thank you!

In the coming weeks, the CFC will open again for the 2014 giving year. As promised, the CMTA has been named as one of the Best in America as certified by Independent Charities of America. The CMTA is also approved as a national charity for the 2014 giving year. Our CFC number is 10597.

Thank you for all of your support! We are very grateful to you!



Parents: Do You Want to Do More? Donate to Research.

Many of us with children with CMT face difficult obstacles in dealing with the disorder, both practical and emotional. On the practical side, we try to address the problems as they develop. These include issues like when and how to use orthotics/AFOs, when and how much bracing should you insist upon, how often is physical therapy needed, and at what point do you turn to surgical options.

Along with each decision comes an emotional response. In addition, as parents, we face other practical issues as children move through their youth. These include involvement with sports as well as non-athletic activities such as theater and music. All of these activities can be more difficult for a child with CMT, particularly if he or she has siblings that do not suffer from CMT.

Perhaps harder are the emotional issues of wanting to do whatever you can to make life easier for your child. In addition to trying to provide emotional support for your child as he/she makes his/her way through life, there is always the feeling of “I could do more!” Many times this feeling is suppressed because you do not know what else you could do to help.

There is something you can do, and it is easy—donate to research to find a treatment for CMT.

There has never been a better time to use any resources available to move the research

ball just a little bit closer to the goal of finding a treatment and cure for CMT.

With its world-class Medical Advisory Board and strong leadership, the CMTA has made tremendous strides in leveraging any donations to research to have the maximum possible impact. Anyone who has read these newsletters or looked at the brochures or other materials involving the “STAR Program,” the “Time is Now” campaign, or any other research-based initiatives knows that there are plenty of opportunities to assist in finding the treatment sooner rather than later.

Moreover, there are so many ways to contribute. In addition to standard cash donations, bequests, or planned giving programs, there are opportunities to fund specific projects or researchers, where you can see your dollars at work. Along these lines, I would share a story of how I was able to assist (through my donation and the generosity of a close friend) in funding a three-year post-doctoral fellow at the National Institutes of Health (NIH) who was dedicated specifically to a project involving CMT1A research. Not only did this add one more specialist to the CMT medical world, but it focused projects and work at the NIH on CMT-specific research.

As the STAR Program advances, many similar specific funding opportunities will present themselves. These include funding certain milestones that will exist in collaborations with pharmaceutical and biotech companies, as well as targeted research projects.

Do you want to know what needs funding? Just ask Pat Livney, the CEO of the CMTA.

The main goal of the CMTA is a world without CMT. The goal inherently involves research efforts that go beyond supporting the status quo. It is only through research discoveries that we can find drug treatments and therapies that will help everyone with CMT.

When I first discovered my son had CMT1A, I did not know what I could, or should, do to help. I got involved in the organization and contributed to its efforts. These actions were well worthwhile and gave me a sense of involvement in doing something to help my son.

So if you ever feel “I Could Do More,” you can, by simply contributing to the CMTA research efforts. I urge you to make a contribution, no matter what size, for CMT research and, particularly, the STAR program. We will only get treatments through your help.

—Gary Gasper,
Board of Directors

The CMTA has made tremendous strides in leveraging donations to research for maximum impact.

Physical Activity

A Physical Therapist's Perspective

BY KATY EICHINGER

A common barrier to engaging in regular physical activity or an exercise program is lack of knowledge of where to begin or what to do. A visit with a physical therapist may be able to help get you started. Physical therapists are health care professionals who hold a college degree in physical therapy and are licensed by the state in which they practice.

CMT affects individuals differently, and, therefore, recommendations regarding exercise and physical activity often need to be individualized. The Physical Activity Guidelines were established by the U.S. Department of Health and Human Services in 2008 (<http://www.health.gov/paguidelines/>). In brief, it is recommended that adults participate in 150 minutes of moderate intensity exercise per week and in resistance training involving all major muscle groups two times per week. For individuals with disabilities, the

recommendations are to engage in regular physical activity and avoid inactivity.

Physical therapists are well equipped to be able to guide individuals who are interested in increasing their physical activity. An evaluation by a physical therapist involves taking a history to better understand your individual experience with CMT symptoms as well as learning about how other conditions you may have affect your health. The physical therapist will also perform a thorough assessment of your strength and range of motion, sensory involvement, your functional abilities, and will often inquire about pain and fatigue. Additionally, the physical therapist will inquire about your current physical activity including your recreational interests as well as your familial and occupational demands.

The information acquired can then be used to develop a physical activity plan and set

goals. This plan can include the types of exercises and activities that meet your individual needs. In addition, recommendations regarding the intensity and frequency of these activities can be addressed. It is essential, however, that you learn to recognize your own body's response to exercise and activity as well as how to balance physical activity with other physical demands related to your occupation or family.

In making recommendations regarding exercise, a physical therapist will often work with you on a consultative basis. Physical therapists work in a variety of settings and have differing levels of knowledge regarding CMT. Many physical therapists will be interested in learning about your condition and how it has affected you. Providing them with this information will facilitate a partnership between you and your care provider that will enable you to reach your fullest potential. ★

"SMALL STEPS WALK" HELD IN ONTARIO, CANADA

On July 14, the First Annual Small Steps Walk for CMT was held in London, Ontario. It was a huge success! With over 100 participants, the five-kilometer walk raised over \$5500 for STAR and a whole lot of awareness for CMT! The walk was in support of all people with CMT and their families; however, it was inspired by Evan Hedges. Evan is three years old and was diagnosed with CMT last year. With frequent rests in his wagon, Evan completed the walk ... AFO's and all! As he came down the home stretch, the crowd erupted with cheers!



Celebrating My First Year with CMT

I celebrated 365 days under the diagnosis of Charcot-Marie-Tooth at a fundraiser in Gloucester, MA. My friends and I helped unload and set up everything for the event. As my family arrived, everyone happily pitched in—cooking food; setting out fresh salads, pasta and desserts; clearing plates and tables; and selling t-shirts and raffle tickets. It was amazing how hard everyone worked. My parents were busy greeting guests as everything moved forward.

Our surprise guests were Senate Leader Bruce Tarr and State Representative Ann Marie Ferrante. Ms. Ferrante gave a short speech congratulating us

on what we all had accomplished. Later in the night, my mom and I also spoke about the past year. At the end of a long, but great, day we realized we had 150 people show up and raised \$15,000 for the CMTA.

In August of last year, my mom asked me how I felt about talking about my diagnosis and CMT. She wanted to do something but would only move forward if I was okay with the idea. I didn't have to think long; I wanted to get involved, too.



Vittorio Ricci has joined CMTAthletes and, along with biking, he is getting ready for cross country this school year.

So, on August 25, 2012, my family and I joined the CMTA and started “Vittorio’s Circle of Friends”. It feels good to be a part of the CMTA; we receive support and answers that we don’t get elsewhere.

I was the patient profile in the November/December

2012 CMTA newsletter. I remember how happy and emotional my family was when it came out. My mom had extra copies printed. I did my first CMT presentation to my old

TAKE ON A NEW CHALLENGE!

Help the STAR Pathways Campaign

You have been hearing about the Warfield Family Challenge for the past six months! We asked for your support to raise much-needed funds for the CMTA STAR Pathways Campaign and you responded generously. The campaign went well beyond our challenge and raised a total of \$338, 516!

Now we have another “challenge.” Since this is only the beginning of the \$25 million STAR Pathways Campaign, we have a long way to go. Our family is hoping that, following our lead, another family living with CMT will jump on the bandwagon and make its own challenge while the momentum is so good. The CMTA needs your help!

We all have friends who know what we deal with daily. Those friends would probably love to help fund the research with gifts small and large. They simply need to be asked. That’s how we launched the Warfield Family Challenge. You can, too!

The challenge to be matched could be any amount with which the challenging family is comfortable. Then, you would need to go through your address book and send a packet of information to everyone on your list. It can be done by “snail mail” with actual CMTA brochures and a note from you, as we did. Or, it could be done by e-mail with a note and links to those very same brochures.

We had help from the CMTA, and our friends were very impressed by the science that backs up the information provided by the CMTA. We were proud to have such professional background material to share. Anyone can take on this project! We’d be happy to talk to you about our experience.

Finding a family willing to take up the next phase of fundraising is our new Warfield Family Challenge. We all know the need. We’ve heard how close the researchers are to finding the first treatment for CMT1A. Please step up to help us move closer to a “world without CMT.”

Take our new “challenge!”

—Seth & Missy Warfield

Elementary School. The third-through- sixth graders were fascinated with my leg braces. It was my first speech and lasted five minutes, and the school raised \$700. A few months later I spoke to my freshman class and added more detail. My classmates wanted to know more about the CMTA than my diagnosis. My freshman class raised \$900. Then, over the summer, Brooks Summer Camp held their annual Swim-A-Thon. I spoke to camp counselors without a prepared speech at hand, and campers raised \$3,750.

I joined CMTAthletes and Donna DeWick's "Got Nerve" for CMT Awareness month on Facebook. I enjoy sharing challenges, advice, and motivation with other athletes like myself. I have gone through five pair of Allard leg braces in one year and was recently fitted for custom "Noodle" AFO's through Kinetic. I will have them just in time for cross-country. I can't wait to get them. I have met a few people with CMT through the Boston Support and Action Group, but the majority of my contact with others who have CMT is through the internet.

The best, and most important, thing I learned this year about having CMT is that doing anything is helpful. Each opportunity has led to another, and I'm thankful to have had so many doors open.

With the help of my family, friends and the CMTA, my Circle of Friends raised \$37,000 this year. There is more to be done, and I look forward to my second year of new opportunities.

—Vittorio Ricci

PATIENT AND FAMILY CONFERENCE TO BE HELD IN NOVEMBER

The CMTA is thrilled to announce a CMT Patient and Family Conference to be held on Saturday, November 9, 2013, in Lake Buena Vista, Florida! The location for the Patient Family Conference will be the beautiful Holiday Inn, a Downtown Disney Resort Area Hotel (*please note change in location), and there will be a CMT Youth Outing at the Magic Kingdom. (For more details about the youth outing visit www.cmtausa.org/url/youthouting.) The Central Florida Area has never hosted an event like this one! You can't afford to miss it!



Patrick Livney, CEO of the CMTA, will kick off the program with some research news, which will be followed by presentations from some of the prominent minds in the fields of CMT research and treatments. We are excited to present Michael Shy, MD, Richard Finkel, MD, Stephan Züchner, MD, PhD, and Sean McKale, CO, LO among our featured speakers.

This event provides a tremendous opportunity to not only hear, but to interact personally, with these experts. Registration is just \$50 per person (non-refundable) and includes "Lunch with the Experts," during which you will have time to talk with the presenters and other leading CMT authorities, including Support and Action Group Facilitators and CMTA Board Members, while enjoying delicious food. Don't delay. Register now for this informative and interactive conference. You can do it online at www.cmtausa.org/url/orlando-pfc, or you can mail a check made payable to the CMTA and designate Orlando PFC on the memo line. The deadline for registration is November 1st.

The Patient Family Conference will also feature exhibitors offering valuable information and a variety of suggestions and products to assist with mobility, alternative pain management, and exercise for people affected by CMT.

Need added incentive? In addition to the PFC's exciting agenda with outstanding speakers, we are pleased to offer special rates made available to us at the Walt Disney World Theme Parks for those attending the conference. Please visit www.mydisneymeetings.com/cmta13t to view special pricing and even order tickets directly.

Register your family and friends now. It's a conference you don't want to miss! At the time of this printing, there were still spots available. Don't delay, and register *now* if you plan to attend.

We look forward to seeing you there!



GIFTS WERE MADE TO THE CMTA

IN MEMORY OF:

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TO THESE CMT CIRCLES OF FRIENDS:

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SPECIAL THANKS
to old and new
friends whose
thoughtfulness and
generosity helped
the Warfield Chal-
lenge to exceed
expectations and
resulted in a total
of \$328,516 for
the STAR Pathways
Campaign.

Wheelchair to Walking: Maybe I Really Am My Mother's Daughter

BY BETHANY MELOCHE, DIRECTOR OF SOCIAL MEDIA

I come from a line of athletes. My mother is a long-distance runner. Her father was a born sprinter.

But that is my maternal line—the line without the gene for CMT.

I grew up admiring Mom's marathon-finisher medals. I'd run my tiny fingers over their bronze finishes, reading the inscriptions, knowing that I would never get one. I had CMT.

Children are told that they can be whatever they want to be, if they just set their minds to it. But no one ever entertained the possibility of me being an athlete: I had CMT.

By the time I turned 18, I required a wheelchair to get around. The muscular imbalances in my feet and calves had led to severe bone deformities. My feet were contorted and curled, and the shortest walks left me with painful blisters and sores from the rigid plastic of my leg braces.

It was then that, for the first time, I considered my last resort: surgery. The doctors recommended 20 procedures over two different surgical events. They described it as “destroying and rebuilding [my] feet.” In the words of one consulting surgeon: “For a little while you will hate the bastard who does this to you.” He also told me it would be worth it.

It was.

The surgeries were an astounding success. After six months of operations and recovery, I could walk without pain. Against the advice of my family, I sold my wheelchair and scooter and insisted on walking. I could walk, and that was what I was going to do!

Each day I walked farther. With every step I grew stronger, and I began to see my body for what it could accomplish, and

not just how it could hurt me.

I learned through a “for-fun” genetic testing service that I have fast-twitch nerve fibers. It classified me as a “likely sprinter.” At

first I laughed. But then I felt good. I had some of those athlete genes, too, and why should CMT negate all of them? I thought I would put them to the test.

I decided to do my own Walk-a-Thon. My goal: Seven miles. I had never walked that far in my life.

The walk was to be a celebration. The girl who was in a wheelchair would now walk seven miles. With the support of my local CMTA Support & Action Group, I planned to use the walk to raise money for the CMTA, to help research find treatments and a cure. I set another outrageous goal: I

wanted to raise \$1000 for every mile I walked.

On August 25th, with family and members of the CMTA community, I walked 7.06 miles in 3 hours 23 minutes 47 seconds. A new Bethany world record!

At the end, my Mom placed a custom-engraved medal around my neck.

I have CMT.

And I win medals. ★

Postscript: We raised over \$9300 for the CMTA, \$1328.57 for every mile. Reporters covered the event, spreading awareness of CMT. Best of all, we had a lot of fun on a beautiful Mountain View summer day doing something nobody with CMT ever takes for granted: walking. Bethany plans to walk 13.1 miles next year. Want to join her? Email her at bethany@cmtausa.org.

No one
ever thought
I could be
an athlete:
I had CMT.

Renee Meloche placed a custom-engraved medal around Bethany's neck to celebrate her “win.”





WHAT'S ON YOUR MIND? ASK DAVID.

Dear David,

I'm wondering if there is anything that a parent can do preemptively to keep a child with CMT from being picked on or bullied at school. With the school year getting started and my daughter in a new school, I'm quite worried.

David Tannenbaum answers:

Having worked for years with parents of children with special needs, I know how much you want to protect your children from bullying. It hurts to feel your child's pain with this issue. I can't say enough about how important it is to get involved with the school when your child is diagnosed with CMT. At the same time, talk a lot to your child about CMT as well, and build up his or her self-esteem.

Being different is just being different, no matter what others say. This will need to be repeated many times. I would think it is time to begin these discussions when your child begins to notice the differences between herself and others, or when you think she is ready. In the meantime play up your child's strengths and love her with everything you've got to give her a good foundation of security to meet future challenges. She will have the emotional equipment to deal with these challenges and stand up and advocate for herself when the need arises. Keep in mind that love is a verb!

Encourage your child to be active in the areas that she can participate in, even in a modified



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

way. I remember as a child I was really awful at sports, but I was able to be in the school play and on the debating team. It was a little dorky, but it really built up my self-esteem. I would have loved a teacher who understood my challenges. I didn't need pity or coddling, just some understanding.

Maybe, if possible, your child could participate in educating other kids about CMT with the guidance of an understanding teacher or other professional. This is such a great opportunity to teach kids about inclusiveness. Teachers need to be educated about CMT. Your child needs to be seen as unique, not handicapped or inferior. This is simply another opportunity for diversity training. When other children don't understand something, they tend to react negatively. The more they know, the more they often like the opportunity to help and be kind. By the way, kids are fascinated with technology, like braces! I know that many kids want a certain amount of anonymity, but the sooner we can accept our uniqueness, the stronger we will become. It is important for your child to truly understand that, despite her differences, she is just as beautiful and lovable as anyone else. Kids can be so cruel. It has been

shown in studies that kids as young as three months reject others who are different. It is in our DNA to choose others who are most like us. This isn't their fault. They just have to be taught differently. It is the teachers' and parent's responsibility to be aware of this and know how to intervene when a child is bullied or is the object of teasing. It might build character for the child to advocate for herself, but at 8 or 9 or 10 or 11, we need some help.

Parents need to be aware of their own feelings of self-acceptance if they, too, have CMT, before they teach their children about loving their uniqueness. Parents often struggle with their own feelings of imperfection. In truly loving yourself you are modeling something precious for your child to learn. I don't think this is just about CMT because I know that many parents put pressure on themselves and their kids to be perfect with or without CMT. I remember working at a summer camp for emotionally and physically challenged children many years ago, and I remember that the counselors created such a loving and accepting atmosphere that these kids thought that anything was possible. There was a joyous spirit throughout that affected them for the rest of their lives. ★

In this column, David discusses bullying. This is such an important topic that Archy also wrote about it in this issue (see page 18).

SAG FACILITATORS SPOTLIGHT:

Steve Fox, Ventura, California

Steve Fox decided to start the Ventura/Santa Barbara SAG after noticing there wasn't an active group in his area. Now the one-year-old group is relocating closer to Los Angeles to make it easier for more people to attend.

"We've had a good response already, but Ventura is about 70 miles north of Los Angeles, and a more central location will be more convenient for CMTA members," says Fox, who at age 68 has dealt with his CMT for more than 50 years.

"I've been fortunate," he says. "My sister, who is six years younger than I, has had a tougher time since her CMT symptoms are more severe. I had triple arthrodesis surgeries on each ankle, one when I was 16 and the other at 17, and I think that made a real difference in my case."

Fox has had an active career as a journalist, working for *The New York Daily News*, *Los Ange-*

les Times, *Associated Press* and *Investor's Business Daily*, and also teaching journalism at the University of Southern California. He now runs his own corporate communications business.

"I decided early on that I wasn't going to let CMT hold me back from the things I wanted to do, and although I'm always slower and clumsier than others, I get there eventually," he says. "Attitude really matters with a progressive disease, and while I wouldn't have chosen to have it, my CMT has made me more patient, more tolerant and more resourceful than I think I would have been otherwise."

Born in New York City, Fox moved to Southern California in 1968 so he could enjoy his primary recreational passion—sailing—year-round. He and his wife, Carol, have owned a number of boats and now cruise California's beautiful Channel Islands, a marine preserve, in a 29-foot sailboat.



Steve Fox enjoys his primary recreational passion, sailing, along California's Channel Islands.

"I've really enjoyed being a SAG facilitator, and I look forward to meeting more folks in the future," he says. "I think we will see enormous medical advances for CMT patients in the years ahead, and, with the great work the CMTA is doing with the STAR Initiative, I believe it's entirely possible that there will be a medication to prevent CMT and, perhaps, even a cure within my lifetime." ★

CMTA REMEMBRANCES

Your gift to the CMTA can honor a living person or the memory of a friend or loved one. Acknowledgment cards will be mailed by the CMTA on your behalf. Donations are listed in the newsletter and are a wonderful way to keep someone's memory alive or to commemorate happy occasions like birthdays and anniversaries. They also make thoughtful thank-you gifts. You can participate in the memorial and honorary gift program of the CMTA by completing the form below and faxing it with your credit card number and signature or mailing it with your check to: CMTA, P.O. Box 105, Glenolden, PA 19036.

Honorary Gift:

In honor of (person you wish to honor)

Send acknowledgment to:

Name: _____

Address: _____

Occasion (if desired):

- Birthday Holiday Wedding
 Thank You Anniversary Other

Memorial Gift:

In memory of (name of deceased)

Send acknowledgment to:

Name: _____

Address: _____

Amount Enclosed: _____ Check Enclosed

VISA MasterCard American Express

Card # _____

Exp. Date _____

Signature _____

Gift Given By:

Name: _____

Address: _____





SUPPORT AND ACTION GROUP NEWS

• CA—South Bay Area

The meeting was held on June 29th with 30 people in attendance, including new members to the group. Many thanks to Ashlee for presenting Aetrex's line of shoes and insoles. She indicated which shoes would work best for those with CMT. She also introduced iStep technology and how the process works to fit clients with the right orthotics. Thanks also to Ashlee for the pizza! Thanks to Harriet and Frank for providing dessert. Thanks to the members for all the valuable feedback and suggestions.

• CT—North Haven

Nine members attended the July 10th meeting. Thanks to Bob Festa, group member, and his Farmington Rotary Club for donating \$500 to the CMTA. Thanks to Bob for hosting Jeana Sweeney and Lynne at his March meeting. Also, thank you to Doug Sutherland for all his efforts working on a proclamation from the Governor declaring September CMT Awareness Month in Connecticut! Michele Kekac discussed all she and her husband, John, have done to get the September Awareness Month and fundraiser event, "A Slice of Hope for CMT," up and running.

• IN—Lafayette

The group held their meeting June 12th. Doug Mullin, CPO of MidWest Brace and Limb, Inc., was the key speaker. He focused on different types of bracing available, the process of

making a brace, and how the brace is fitted to the needs of a patient. Thanks to Lawane Lord for donating a wonderful array of bakery cookies, utensils, plates, tablecloth, and other snacks. Also, a huge thank you to K. Dees Coffee and Roasting Company for the donation of Columbian Coffee. A reporter from the Journal and Courier newspaper also attended and took notes.

• MA—Boston

The Boston Support Group met on June 26th. They discussed beginning a group list of email addresses, CMT research, genetic testing options, and CMT's effect on the family. Members wanted more information on clinical trials, genetic testing options and costs, groups available for different age groups, and resources in the Boston area. To participate and learn more about CMT research, Jeana suggested joining the contact registry.

• MI—Kalamazoo Area

The group met on July 18th and had six in attendance, including one new member. A lawyer who specializes in Social Security Disability claims presented an overview on SS Disability and answered questions.

• NM—Albuquerque Area

The CMTNM group met on August 10th. Two of the eight in attendance were new to the group. This meeting focused on videos from the November CMTA facilitator meeting in

Chicago. We viewed the three-part "Science of CMT." Everyone found the information to be of value, and the group had a lot of lively discussion. Everyone was encouraged to register with the CMT patient registry database.

• NY—Upstate New York

Sandra Varno MS, RD, CDN, and Resource Educator at the Cornell Cooperative Extension, Albany County, served as guest speaker for the July meeting. Sandra discussed "Eating and a Healthier You." She shared several tips on exercise and weight management. Deacon Hook shared information on the upcoming Christine A. Hook Memorial Concert in October. Proceeds of the concert are to benefit the CMTA.

• OH—Cleveland Area

There were 11 people in attendance at the July 10th meeting. Topics discussed included the tee-shirt fundraiser, which raised over \$1,400 for the CMTA, the upcoming Awareness Month picnic, and guest speakers at future meetings.

• WV—Vienna Area

The group met August 10th for a picnic. There were 24 in attendance and all went well. Thanks to everyone for preparing the great food. Entertainment for the day included members baring it all—feet! One member even shared a zipper secret that everyone found very useful. Overall, it was a good time of food, fellowship and lots of fun. ★

CMT Support and Action Groups in Your Community

AL—Birmingham

No group currently meeting. Will accept calls.
Dice Lineberry
205-870-4755

*AL—North Florence

North Florence CMT Support and Action Group
Tina Smith
256-757-9250

AZ—Phoenix Area

Arizona CMT Support and Action Group
Pamela Palmer
ppalmeraz@gmail.com
Jim Blum
480-272-3846

CA—Santa Barbara

Ventura-Santa Barbara, CA CMT Support and Action Group
Steve Fox
805-647-8225

CA—Stockton

Stockton, CA CMT Support and Action Group
Nina Anselmo
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San Diego, CA CMT Support and Action Group
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Santa Rosa, CA CMT Support and Action Group
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CA—Visalia Area

Visalia, CA CMT Support and Action Group
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CO—Denver Area

Denver Area CMT Support and Action Group
Ron Plageman
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Dick Kutz
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CT—Hartford East

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CT—North Haven

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Iowa Great Lakes and SW MN Regional Virtual CMT Support and Action Group
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IA—Iowa Area

Iowa City, IA CMT Support and Action Group
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IN—Fort Wayne

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Priscilla Creaven
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IN—Lafayette

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Karen Smith
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LA—Baton Rouge

Louisiana CMT Support and Action Group
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MI—Kalamazoo

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No group currently meeting. Will accept calls.
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MS—Mississippi/Louisiana

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Eastern Ontario CMT Support and Action Group
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Southern Ontario CMT Action and Support Group
Kelly Hall
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Mexico

(This group will be in Spanish.)
México CMTA Grupo de Apoyo y Acción
Gina Salazar
Gina_oviedo@hotmail.com

Most Support and Action Groups can be accessed at www.cmtausa.org. They can be found in the CMTA Online Community under Support and Action Groups.

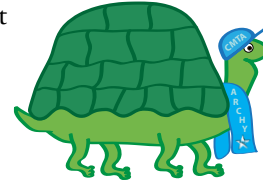
***NEW SUPPORT GROUPS**

Back to School

I usually look forward to the start of school each fall. I'm a pretty good student, and I get a lot of praise for the work I do. Besides, after a summer away from them, I am anxious to meet up with some of my friends. This year, however, is quite different.

I'm going to a different school where all the kids and teachers won't know or understand about my CMT. I've grown up with most of the kids from Greenwood Elementary

School. But now I'm off to the merged middle school called Greenleaf Middle School. I'm just not looking forward to being in the lowest level grade and being different. As much as I think I've come to accept my CMT, and as well as I do at most activities in spite of it, I'm really dreading the first time someone makes fun of how I walk or how slowly



Our mascot "Archy" writes about his experiences as a turtle with CMT.

I complete physical tasks.

My mother has been reading about the "meanness" of middle-school-aged kids. It has something to do with not really knowing who they are or how they fit into the scheme of life. But, if you are on the other end of the comments and the teasing, it doesn't really matter why they do it, just that they do.

AMINOACYL-TRNA MOLECULES

(continued from page 4)

are challenged by stress. Importantly, the DI-CMTC causing mutant YARS over-activates this transcriptional factor. As over-activation of E2F1 causes abnormality in cell cycle progression and apoptosis in neurons, and has been linked to various neurological disorders, my work raised the possibility that E2F1 over-activation is also linked to DI-CMTC.

With the help of the travel fellowship from the Charcot-Marie-Tooth Association (CMTA), I was able to present my work at the 5th International CMT Consortium Meeting in June, 2013. The meeting assembled the world's greatest minds in CMT research, from whom I obtained valuable feedback. For example, generating the knock-in mice model can help me to analyze the pathway that may be changed in the

animal. Also, we could design specific inhibitors of the pathway that we are particularly interested in and test them in vivo. These suggestions provided guidance for my future studies.

Although this was my first time attending the CMT meeting, from my conversations with other attendees, it was obvious that the area of CMT research is in fast development. Ample latest-research achievements, such as the identification of new genetic mutations in different CMT families and the sharing of the latest knowledge among investigators coming from all over the world, were inspiring. I believe that the more genetic mutations we are aware of, the clearer is our understanding of the disease-causing networks and mechanisms. It may be possible that one day researchers can find out an overarching scenario that explains the CMT disorder based on a well understood disease-causing mechanism of these

genetic mutations. The state of the research methodology used by investigators in the field (e.g., whole exome sequencing, various animal models and the high-throughput drug screening systems) also made a strong impression. I have no doubt that these technologies will accelerate the drug discovery process for finding a cure for CMT.

As an experimental biomedical researcher spending most of my days behind the bench, the greatest gain for me from the CMT meeting was the opportunity to meet with the most experienced CMT physicians in the world. They are the people who know best the suffering of the patients and their needs. Although tremendous progress has been made in disease diagnosis, there is still no cure for CMT. Working alongside all the researchers and clinicians, I am hopeful that I can contribute to making the first drug for CMT a reality. ★

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I'm a pretty self-assured kid. Having parents who understand my disorder and who don't cut me too much slack because of the CMT has made me a fairly strong kid. But, let's be totally honest. None of that takes away the sting of being picked on. I do have my really good friends to support me and help me if problems arise, but in the middle school, we each have unique schedules, and I might not really be with my good friends except at lunchtime.

So, I'm nervous. I don't want my mom to know because she already feels guilty that she's the reason I have CMT. My dad is a great father, but he has trouble remembering what school can be like and how mean some kids are. He tells me that bullies are just insecure kids who get a sense of power from hurting kids who are smaller or weaker in some way.

My plan (if you can call it that) is to beat the bullies to the "joke" that they think I am by using what my dad calls self-deprecating humor. Look up the word. What it means in the practical sense is that I am going to make fun of myself and the way I walk and run. That way, the bullies lose their chance to pick on my lack of skill. If they decide to beat me up, I'm guessing the humor won't be very successful, but that's where the teachers and my friends come in. I'm not going to take any bullying. I'm going to do what my dad suggested ... strongly. I'm going to report it before it gets out of hand.

I'm a strong little guy. I know all the "right" things to do. The test this year will be whether I can put all my sense and knowledge into play. I sure hope I can ... no, I'm confident that I can! ★

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 CMTA Report

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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.