

THE CMTA Report

MAY/JUNE
2010

Information on Charcot-Marie-Tooth Disorders for patients, families, and the scientific community * www.charcot-marie-tooth.org



**Charcot-Marie-Tooth
Association**

OUR MISSION:

To generate the resources to find a cure, to create awareness, and to improve the quality of life for those affected by Charcot-Marie-Tooth.

OUR VISION:

A world without CMT.

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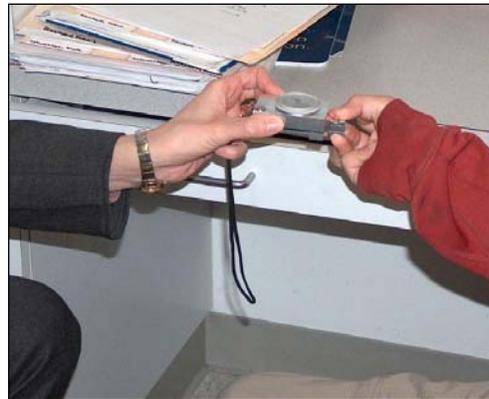
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CMT Findings Reported at the American Academy of Neurology

In April, the AAN met in Toronto and heard presentations on several new research findings related to CMT. Such emphasis on CMT research is encouraging. The summaries of the reports follow:

- Mice with a genetic mutation in the myelin protein zero (MPZ) gene, develop a disease which is similar to CMT1B. The mice in the study benefited from receiving curcumin, a component of the spice turmeric. The curcumin appears to help mice perform better than their untreated littermates on a test of their ability to hold on to a rotating rod and on their grip strength. They did not improve on other tests of motor function. This study was reported by Agnes Patzko, a research associate at Wayne State University.
- Zarife Sahenk, professor of pediatrics, neurology, and pathology at Ohio State, presented findings on NT3 gene therapy for CMT1A. Mice that were treated with an intramuscular injection of neurotrophin 3 (NT3) showed improvement in grip strength, the ability to stay on a rotating rod, and strength in their nerve signals.



As part of the new pediatric scale, tests of pinch strength are measured.

The study showed that NT3 gene therapy delivered intramuscularly has some potential for treating people with CMT1A and possibly other forms of CMT because NT3 is considered to be good for nerve fibers in general.

- Richard Finkel, director of the CMT Center of Excellence at Children's Hospital of Philadelphia, reported on the CMT pediatric scale developed by Dr. Michael Shy, Wayne State, and other CMT specialists around the world. The pediatric scale will measure foot posture, hand dexterity, sensory function, ankle positioning, hand and foot strength, balance, and endurance. A second scale called the Pediatric CMT Quality of Life

instrument has been developed in the United States and Australia. The instrument includes a questionnaire which covers topics such as physical symptoms (pain, cramps and fatigue). Also included are social issues such as playing with peers, how others perceive their cognitive functioning,

emotional distress, and emotional bonding. The availability of scales that measure physical functioning and overall quality of life in children with CMT is an important step in developing treatments for the disorder and measuring the success of such treatments. *

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Join the New NIH/CMT Contact Registry!
See box on page 3

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PARTNERING WITH THE NIH

Since the commencement of the CMTA's STAR research program, we have consistently referenced one of our key scientific partners, namely the NIH National Chemical Genomics Center (NCGC). As you know, the NCGC has been instrumental in advancing our research agenda not only through their high-throughput screening program, but also by supporting our cellular assay development project.

Under the leadership of Dr. James Inglese, the NCGC affords the CMTA cutting-edge technology and world-class scientific leadership. As such, I thought it appropriate to give you a sneak peek at a feature article that will soon appear on the NCGC website (<http://www.ncgc.nih.gov/>). I am sure that the article below will give you a better appreciation for our enthusiasm and optimism for the success of the STAR program.

—David M. Hall, Chief Executive Officer

The NIH Chemical Genomics Center and the Scientific Community

The NIH Chemical Genomics Center (NCGC), housed within the National Human Genome Research Institute at the NIH, is a member of the Molecular Libraries Probe Production Centers Network (MLPCN). As such, the NCGC investigates biological and chemical space by screening biochemical and cellular assays against libraries of chemical compounds to determine activity, and then optimizes chemical compounds of interest for use as biological probes or potential leads for early-stage therapeutic development.

In the past year, the NCGC has initiated collaborations with nearly 300 scientists in academia, industry, and the government. These collaborations extend throughout the U.S. and abroad. In addition to its work with the MLPCN and its assay providers, the NCGC brings

projects into its pipeline through novel mechanisms; these include interagency agreements with the U.S. E.P.A. and other NIH institutes, and collaborations with disease foundations.

The NCGC's focus on rare and uncommon diseases has led to numerous unique collaborations in many areas. One such disease collaboration involves Charcot-Marie-Tooth disease (CMT). Amongst inherited neurological disorders, CMT disease is the most common. The disease causes nerve degeneration and muscle weakening in patients' extremities, sometimes accompanied by sensory nerve function loss. Over the past two years, the NCGC has worked with the Charcot-Marie-Tooth Association (CMTA) to develop and screen an assay based on PMP22, the major gene involved in the genesis of CMT1A, which is the

most prevalent type of the disease. The aim of the assay is to find compounds that may inhibit PMP22 expression or cellular accumulation.

The NCGC-CMTA collaboration is a model for future collaborations between NCGC and the scientific community. The collaborative process was carefully planned from the beginning to design an experimental program with the best chance of success. The collaboration was initiated when CMTA scientists visited the NCGC, after learning about its assay development, screening, and probe optimization expertise and capabilities. A kick-off symposium was held at the NCGC and attended by CMTA-funded investigators and advocates; this symposium brought the participants up-to-date with research in the field and catalyzed thinking about how to move forward with the

current available knowledge and cell lines. A series of meetings were held between CMT experts and NCGC scientists to map out a plan for developing a high-throughput assay using these cells. The assay based on these cell lines was transferred from the outside laboratories to NCGC for optimization and validation.

A successful key to this collaboration has been the hiring of a dedicated post-doctoral fellow for the project at NCGC. Sung-Wook Jang, Ph.D. joined the NCGC in July 2009 from an external CMTA-funded laboratory at the University of Wisconsin to work on assay development and screening. Since his arrival, he has worked successfully with scientists across the CMT community to develop a robust assay for screening for inhibitors of the PMP22 gene. Dr. Jang validated the assay at NCGC using man-

ual and robotic assay technology to determine its validity. After a successful validation, he used the NCGC's Kalypsys robotic system to perform quantitative high-throughput screening on over 300,000 compounds with the goal of looking for small molecules that may be active in the assay. Along with other scientists at NCGC, he is currently verifying active compounds from the screen. After potentially active compounds from the initial screen have been verified, Dr. Jang will work with outside CMTA expert scientists to test these compounds in secondary assays, to both confirm activity and determine a plan for chemical optimization.

The NCGC provides a unique training environment for post-doctoral fellows such as Dr. Jang. Industry expertise is melded with academic freedom and the ability to work openly

with disease foundations, delving deeply into chemical biology space and relating it to human health and disease. The CMT-related research at NCGC can continue far beyond the current high-throughput screen; little is known about targets that modulate CMT1A, and these targets could be the subject of future screening efforts. It is hoped that leads from the primary screening effort will be developed into strong chemical probes, which could eventually lead to therapeutic development in the area of CMT. Since the scientific methods underlying this research plan were carefully laid out, and continuous contact is maintained between the different laboratories involved in the project, scientific findings will effectively piece together the puzzle that is Charcot-Marie-Tooth disease. *

A New Place for Information on CMT— Join the Contact Registry!

CMT is being researched as part of the National Institutes of Health-sponsored Rare Disease Clinical Research Network (RDCRN), in a group called the Inherited Neuropathies Consortium.

Researchers from Wayne State University, University of Pennsylvania, Children's Hospital of Philadelphia, University of Rochester, NY, National Hospital for Neurology and Neuroscience, Queens Square, London, UK, University of Sydney, Australia, and

C. Besta Neurological Institute, Milan, Italy are teaming up to create the largest repository of information on people with CMT.

All information is sent through a secure website using a patient code to the University of South Florida, which houses this information for all of the disorders in the RDCRN. All protected health information is kept confidential.

You can be entered into this research by seeing a participating physician at one of the

sites listed above, or you can join the Contact Registry at www.cmtausa.org/registry. You'll be among the first to know about any new information from this research and about clinical trials that become available for your type of CMT.

To find out more about the RDCRN, the Inherited Neuropathies Consortium, and the contact registry, visit the RDCRN website at <http://rare diseasesnetwork.epi.usf.edu/>. *

CMTA-NCGC Partnership Supports Additional CMT Studies

A new CMT type 2E study recently began at Columbia University under the direction of Dr. Ronald Liem, Professor of Pathology and Anatomy and Cell Biology—who received a 2-year/\$5 million grant from the NIH to identify compounds to treat CMT type 2E.

This exciting project was similar to an ongoing CMTA-funded study at the University of Missouri under the direction of Dr. Michael Garcia. CMTA CEO David Hall and CMTA Board Member Herb Beron coordinated a meeting between Dr. Liem and his team at Columbia and Dr. Michael Garcia, and the CMTA STAR team at the NCGC. The purpose of the meeting was to introduce the NCGC, their technologies, and expertise to Drs. Liem and Garcia in the hope that the scientists could find beneficial areas of collaboration. The meeting was a great success as the three

Three teams of researchers have come together to find beneficial areas of collaboration on 2E research.

groups have established a strong dialogue and will continue to share information to ensure they are 1) getting access to the best technology; 2) leveraging each other's work to prevent duplication of efforts, and 3) pursuing future areas of collaboration that can generate additional funding opportunities to advance their research.

Please stay tuned to future issues of *The CMTA Report* for continued updates on the CMT type 2E project.

Below is a summary of the CMT type 2E project at Columbia University

SPECIFIC AIMS

Mutations in NEFL, the gene encoding neurofilament-L (NFL), cause CMT2E. Dr. Liem has studied these mutations and found that in both neuronal and nonneuronal cells, mutant NFL resulted in misassembly of the intermediate filament network. In neuronal cells, Dr. Liem found that the mutant proteins caused defects in axonal transport leading to

degeneration of neurites. His studies showed a perfect correlation between pathogenic mutant NFL and the misassembly of filaments in the cultured cells. He hypothesized that inhibitors of neurofilament misassembly will lead to therapies for CMT2E. Therefore, the goals of this project are to identify small molecules that inhibit misassembly and to test their effects in a mouse model of CMT2E.

SPECIFIC AIMS:

- **Aim 1: Screen for small molecules that inhibit neurofilament misassembly.**

Dr. Liem will perform high-throughput visual screens for compounds that inhibit the abnormal assembly of neurofilaments caused by expressing NFL mutants in cultured cells.

- **Aim 2: Generate a mouse model of CMT2E caused by NEFL mutation.**

Dr. Liem will generate knock in mouse models of CMT2E with mutations in the NEFL gene.

Mice will be characterized phenotypically with the goal of using them to test compounds that are identified in Aim 1 for their ability to improve peripheral neuropathy.

The proposed experiments should lead to potential small molecules that could be useful in the treatment of CMT2E and will provide a mouse model to test these molecules. *

CLINICAL STUDY AT JOHN P. MURTHA NEUROSCIENCE AND PAIN INSTITUTE

If you experience weakness, fatigue and pain with your CMT, you might be eligible for a study that will last one year and require 12 visits to Johnstown, PA. Participants will receive a free physical exam, free laboratory testing, the study drug and a small travel stipend. You must be at least 18 years of age. For more information or to determine if you are eligible for the study, call Janet Goodard, RN, Study Nurse Coordinator at 814-259-5288 or 1-866-707-8424. The study is sponsored by the Memorial Medical Center and funded through the Department of Defense. *

The CMTA “Circle of Friends”

Since we began the CMTA Circle of Friends program, our members have found many new and creative ways to support the work of the CMTA. People who initially began with a simple letter to their friends and family have built upon their success and now hold annual dinners, walks, and tournaments.

Others have met the challenge by finding ways to get their employers and other companies involved through sponsored events and matching gift programs.

If you would like to take up the challenge to get involved and start your own Circle of Friends, call us today at 1-800-606-2682, email us at cof@charcot-marie-tooth.org, or visit us on the web at www.charcot-marie-tooth.org/cof.

Working together, can create a world without CMT!



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Eating doughnuts for CMT research!

Dear CMTA,

I found out that I had CMT when I was 16. From my days in elementary school, I always knew there was something different about the way I walked and so I was both happy and scared to find out why that was. When I was 19, having difficulty walking around my college campus, I was fitted with braces and continue to wear them now at age 40. What a godsend they have been—just like the CMTA.

The CMTA has always been a reliable and valuable source of information and support for me, and I recently realized it was time to give back to the CMTA. Last September, I had the privilege of co-starting a new support group in Williamsburg, VA, for others, like me, who live with

CMT every day. In Las Vegas this past March, I had the most supportive experience yet, meeting members of the CMTA staff and Board, CMT researchers and clinicians, other CMT support group leaders from around the country and, most importantly, a large roomful of people with CMT. That experience in Vegas made me feel like I wasn’t alone and that my challenges were understood, a feeling I had not quite ever had before. It also energized me to enact my idea for a fundraiser to help the CMTA raise money for the STAR program in the hope of finding a treatment and cure for those like me with CMT.

My fundraiser involved the purchase of 100 boxes of Krispy Kreme doughnuts. When you purchase that many, you get

them for \$3 a box. I sold each box for a “minimum donation” of \$6. While my goal was \$500, my Circle of Friends exceeded it and together we raised \$937 for the STAR initiative. From doing this fundraiser, I did more than just raise money for the CMTA. I helped to increase awareness of CMT and the CMTA throughout my community. I received significant emotional support from my friends and family, and I got to eat doughnuts!

Thanks to Pat and Dana for supplying me with CMT pamphlets to give out with the doughnuts. And thanks to the CMTA for all of their support and continuing effort to find a treatment and a cure for CMT.

Sincerely,
Nancy Mollner
Williamsburg, VA

Our CMT Experience... *and the Plan We Have to Help Stop It*

BY BROOKE ERWIN

In October of 2006, my daughter, Gretchen, was diagnosed with CMT. After the initial shock and concern wore off, we kind of ignored the symptoms, diagnosis, and thoughts of CMT altogether for about the next 6 months. By that time, Gretchen began to have more complaints and problems, and we knew we needed to pay attention. In the beginning we noticed the shape of her foot changing rapidly, and her physical ability didn't seem to be quite up to par when compared to her peers. She had difficulty running "the dreaded mile" required by the Presidential fitness test, and she occasionally complained about her feet and legs hurting. Once the complaints became more frequent and her foot steadily arched higher, we knew it was time to re-visit the CMT diagnosis.

We met with several different specialists to find the best matches for our family, and once we found the right doctors, we were all evaluated. As it turned out Evan, our youngest son, also had some classic signs and early symptoms of CMT, and later we discovered it came from me. Evan was a late walker—not walking on his own until around 17 months, and within a year of that he began "toe-walking." He spent most of his active time on his tip-toes and we constantly had to remind him to "walk on flat feet." To this day, he doesn't

seem to display any additional symptoms, but we have worked with him from the time of his diagnosis. He had physical therapy along with his big sister, continues stretch routines at home, and wears night splints to help maintain his heel cord flexibility as long as he can.

Now, almost 4 years later, we deal with the effects of CMT everyday. Since Evan was so young when we discovered our family's CMT, he doesn't see himself as really being any different from anyone else. How-

thrilled she has such a great group of friends. On the other hand, she is a teenager and she is struggling with all the normal adolescent pressures on top of learning to deal with her CMT. We have the same parent/teenage conflicts most families have, like squabbles over the sloppy stacking job she did during dish duty, her meltdowns over losing her favorite t-shirt, or not getting an A on the project she poured herself into. But throw in the physical therapy she loathes, talk of an upcoming



The "Bad Dreams Go Away Spray" kit contains a small book, a spray bottle, and a page of stickers.

ever, Gretchen struggles both personally and physically. She has been very secretive with her problems and her AFO leg braces. She has only told her best friends she wears them. Everyone she has chosen to share her secret with has been very supportive, and we are

surgery, or even my daily reminders to put on her AFOs and you'll have child that's about as warm and fuzzy as a rattlesnake.

Our biggest hurdle right now is getting her ready for the reconstructive surgery she needs to loosen the tendons and mus-

cles and straighten her badly deformed feet. Some days I think she hates me for scheduling this procedure. Most of the time she accepts and handles her CMT very well, but when it comes to doctor's appointments,

It is our responsibility as her parents to make the tough decisions (surgery) for her right now.

physical therapy, braces, or surgery, she morphs into a bitter and sometimes mean little bugger. It breaks my heart. There are days when I honestly want to say, "Fine, you win. I will not stay on you about your braces or make you have your surgery. Do what you want. You're the only one who is going to suffer in the long run." But that's right. She will suffer, and it is our responsibility as her parents to make these tough decisions for her right now. After having a little pity party for myself, I pull myself up and put my big, mean momma face back on and make her do those things she doesn't want to do because I know it is in her best interest. I just hope she will appreciate it one day and realize we were not trying to hurt her.

When Gretchen is really feeling low or grumpy, we remind her about all the great things going on with the CMTA and all the research efforts tak-

ing place now—even as you read this. It gives us great hope that we can stop or maybe even reverse these painful and sometimes debilitating symptoms, but better yet we may be able to stop CMT for good.

My family is so motivated to help support those research efforts that we have been brainstorming for new fundraising ideas. Recently, I released a children's book that's included in a fun and creative activity kit for children and parents. To help raise awareness in our community, I have written several articles for area magazines and newspapers, and I have contacted and informed our local news stations about Charcot-Marie-Tooth and our fundraiser. Though my book and product have nothing to do with CMT directly, they are relevant to many young children, and most of you adults know at least one child. This creative children's product would be a perfect gift for any child between the ages 3 and 8. It's also a great way to help support the CMTA and their STAR initiative. A portion of every purchase of my book and activity kit will be donated to the CMTA quarterly.

The book, titled *Bad Dreams Go Away Spray*, is the first in a series published by my "mom-entrepreneur company," called KidLogic. My books approach common childhood issues all parents face such as: bad dreams, undesirable table manners, and even not-so-happy nappers in very simple "kid logic" ways. Every "Bad Dreams Go Away Spray" parent/child activity kit includes: a cute, colorful and distinctively illustrated

bedtime storybook, a "chubby" kid-sized spray bottle—perfect for little hands, recipes for your own protective potions, and a silly monster sticker sheet. The kit is available online for \$14.55 plus \$5.95 shipping/handling. To order please see my website: www.baddreamsgoawayspray.com and remember: \$1 of your purchase will go to the CMTA! *

Brooke has already had one article published in the *TowneLaker Magazine*.

Help Find a Cure for CMT

by Brooke Erwin

When my daughter Gretchen was seven, she complained of foot and leg pain. Her feet started developing high arches and her gait became clumsier. She had trouble keeping up physically, and she started falling regularly. Gretchen was tired and frustrated.

At age 11, Gretchen was diagnosed with Charcot-Marie-Tooth or CMT, the most commonly inherited neuropathy affecting one in 2,500 people; 300,000 in the United States. Soon after her diagnosis, the rest of the family was tested. Tests revealed her little brother, Evan, also has CMT and it was inherited from me. CMT affects the peripheral nerves in the body. The nerves begin to lose function, which causes the muscles to lose stimulation and action. It starts in the feet and hands. Toes curl, the foot arches and ankles and calf muscles weaken.

For some, CMT can be painful - for others, simply frustrating. Imagine once being a top athlete in school and in a short time, tripping over your feet as you walk across a room. Those affected by CMT do not have a physical awareness of where their body is in space, often bumping hips and knees on the corners of tables. Loss of balance and foot drop cause people to trip and fall.



Years ago, people living with CMT would have only one fate - eventually living in a wheelchair and relying on others for help. But today, there is hope for people with CMT. The CMTA (Charcot-Marie-Tooth Association, www.charcot-marie-tooth.org) has united top scientists, neurologists and geneticists from around the world to find treatments and a cure. With the help of the U.S. government and the National Institutes of Health, the CMTA is months away from a pharmaceutical treatment that may stop nerve degeneration. They are hopeful that they will find a cure soon, reversing the nerve deterioration that would allow Gretchen and Evan to physically function better.

Our family attends the bi-monthly CMTA Support and Action Group of Atlanta, which serves the greater Atlanta CMT community by providing resources, medical updates and research information. Because of the collaborative research and the support and action group, we have a reason to believe in a world without CMT.

I am a children's book author and illustrator, and I recently committed to donate \$1 of each sale of my recently released book, *Bad Dreams Go Away Spray*, to the CMTA to fund research. Please visit www.baddreamsgoawayspray.com for more information on the book and help us find a cure!

Overcoming Obstacles in Europe

BY YOHAN BOUCHARD

Over spring break, I went to Europe with my father and about 20 students from my high school, Palo Alto Prep. From the time we landed in Vienna, I knew that we would be exploring three large cities on foot: Vienna, Budapest, and Prague. Having CMT, I was concerned about walking for hours without much rest. However, I have been learning methods to cope with the pain and fatigue, and I employed them to deal with the constant hindrance to my ability to walk for long periods.

Upon our arrival in Vienna, we first took a bus to our hotel and decided to recover from the 11-hour plane trip. During the second day, we visited the Hapsburg Palace, the former home of the Austrian Emperors. The palace was extremely large, and it

took a good hour to complete the tour, but I focused on all the interesting artwork and architecture within the palace to distract me from my pain. Although it was under renovation, the palace still looked majestic. The following morning, we thankfully we

I knew I wasn't the only one who was exhausted. That night we spent around 30 minutes figuring out where the restaurant was, and when we got there, we learned that it was expensive, but the food was decent. By the time I went to bed that night,

I overcame the physical challenge of walking in Europe with a variety of coping techniques.

took a bus to the train station and a train for the four-hour trip to Budapest.

When we arrived in Budapest, the entire area seemed to be run down or under construction. We had to walk to our hotel, where all the students just crashed after the long journey, so

my feet were in such pain after pounding the pavement all day that I had to distract myself by reading *A Clash of Kings* by George R.R. Martin.

As we all gathered in the lobby of the hotel the next morning, I groaned as I learned that we were to take a walk to the downtown area. Luckily, the walk took less than an hour, and when we arrived, we stopped to eat lunch, allowing me some time to rest. That afternoon, we spent the next four grueling hours walking around an Easter Festival and looking at shops with handmade apparel (not my favorite activity). As a way of resting my tired body, I was in favor of stopping at a coffee house before returning to the hotel. I knew I had to get a good night's sleep because the next day our group had planned to visit several monuments and a large 150-year-old church. The train trip to Prague lasted about

(continued on page 18)

Gilles Bouchard and his son, Yohan, pose during their school trip to Vienna, Budapest, and Prague.



Archy Anticipates Summer Vacation

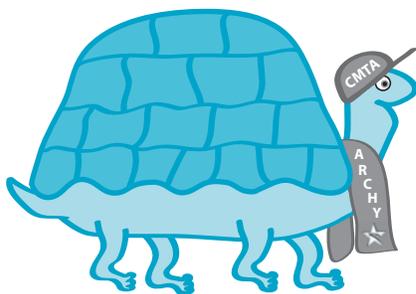
“Archy,” our mascot, writes for the CMTA newsletter about his experiences as a turtle with CMT.

One of the big decisions that our family always starts making in the spring involves where we will go for summer vacation. We always go somewhere that the whole family agrees on. As my sister and I get older, I guess that might get harder and harder to do, but right now, we still like to go places with our parents. My sister, Tara, is thinking about going to Adventure Girl camp for a week, but she’ll also go with the rest of us wherever we choose.

Right now, the top contender seems to be a water park in Wildwood, New Jersey, called Morey’s Pier. Even though the park is right next to the Atlantic Ocean, the water they use in the park is fresh water, so it’s safe for us turtles. The reason the water park seems like such a good choice is that I can still swim really well and that gives me a chance to enjoy all the same activities as my sister. Besides, this year my grandparents, Ralph and Edna Turtlebaum, are coming along and my Pop Pop, who also has CMT, can still swim even though he’s pretty old. In fact, he and I often have races to see if I’m getting faster—and I am, although I sometimes wonder if maybe he is really getting slower instead.

One year, we went to famous

battlefields in our area. My parents thought my sister and I should know about the parts of American history that relate to the people in our lives. I really liked the old Civil War battlefields, but my sister was sort of bored. I think she would have liked to go to a place called a



“mall” more. The one thing that my parents and we kids discussed was how the Civil War really tore this country apart and how, in some cases, brothers wound up fighting against brothers. We agreed that we didn’t believe in ever letting our different opinions tear our family apart. Nothing would be worth that.

Another favorite past vacation was a trip to Disney World. I know that people always talk about how accommodating the guides are to people who are handicapped. Well, you should see how they work to make the visit safe and pleasant for us animals. Of course, our time in the park is different from the time when the people are all there so it’s much safer. We go in really early in the morning, and the dew on the rides and on the walkways makes it much more pleasant for our little turtle feet.

We can even ride Dumbo and the swirling teacups because they are wet in the early morning. Once it gets close to the regular opening time for the park, we quietly make our exit to go back to our motel and swim or just enjoy the quiet time.

Thinking back on our previous vacations and looking ahead to this year’s, it comes to my attention that where we go really isn’t the point. The fact that we all go together and enjoy each other’s company is what it’s really about. I’m sure we’ll have a good time again this year, and the only bad thing about summer vacation is that my teacher will make me write an essay next September called “what I did on my summer vacation.”

Yuck. *

ARCHY TO ATTEND “COOTERFEST”

In October, Archy will be attending Cooterfest in Inverness, Florida. Cooterfest is a festival that celebrates the legend of a little turtle with a big heart. This year the festival will be held from October 29 to 31.

The CMTA will have an informational booth at the event and will also have the Archy Train, created by Jerry Cross, there for kids to ride. Several staff, board members, and support group leaders will be in attendance, and some, it’s reputed, will be dressed as fellow turtles in support of Archy and his campaign to bring awareness of CMT to the country.

You can visit www.cooterfest.com to learn more about the festival and, if you live nearby, consider visiting the CMTA booth and Archy. *

Board Member Holds Successful Fundraiser for Research

In January the San Francisco Giants held a fantasy baseball camp in Arizona. Having attended the baseball camp for several years, new Board Member, Vasi Vangelos, thought that he could use the week at camp as a fun and somewhat simple way to raise money for CMT research.

He sent letters to a large number of friends and professional associates asking them to pledge a sum of money for the number of hits that Vasi might be able to get in his week of camp. Of course, people were also offered the option of giving just a set sum of money for his efforts at camp. Vasi spent an entire week



Vasi Vangelos, CMTA Board Member, and "part-time" San Francisco Giant outfielder in his official baseball card.

from January 24 to 31 with his son, Will, the batboy and his brother, Steve. His position was the outfield, which he jokingly

said was "because of his speed." (Vasi has CMT, although he remarked in his letter that he considers himself fortunate to live with a less debilitating form of CMT.)

Vasi's fundraiser was an amazing success, with the total amount being donated to the research program. For the donors who chose to "bet" on Vasi not being a fabulous hitter, they were probably quite disappointed to find out that Vasi hit an amazing 10 hits in 21 at bats. *

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, 2700 Chestnut Parkway, Chester, PA 19013.

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: _____



GIFTS WERE MADE TO THE CMTA

IN MEMORY OF:**Irene Ashworth**

Mr. and Mrs. David R. Beach

Anton Betschart

A.L. Gilbert Co
Mr. and Mrs. Frank Aufdermaur
Ms. Mary Brand
Mr. and Mrs. Don Brink
Mr. Gerald Burch
Mr. and Mrs. Gerry Dieringer
Mr. and Mrs. Joe Duarte
Mr. Louie Durrer, Sr. and Family
Mr. and Mrs. Gregory Elder
Mr. and Mrs. Conrad Grass
Mr. Hansueli Muhlethaler and Family
Mrs. Marie Ott
Mrs. JoAnne Plooy
Mr. Hank Plooy, Jr.
Mrs. Rita Sigrist
Mr. and Mrs. Frank Silva
Mr. and Mrs. Lenard Suter
Mrs. Joey Vogel

Ronald Beyer

Ms. Aurora L. Costa-Esnard
Mr. and Mrs. F. Robert Kobbe
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Mr. Robert Stephens

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Mr. and Mrs. Norm Winter

John Gomez

Ms. Florence Fiscella

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Mr. Michael K. Grife

William C. Hadaway

Mr. Michael Hadaway

Harold Hoppe

Dr. and Mrs. Alan F. Frigy

Charles T. Lynch, Ph.D.

Mr. Tom Lynch

May F. Meloche

Ms. Diane P. Justusson

Irene Minter

Mr. Robert R. Minter

Marvin Napier

Ms. Rhonda R. Belknap
Ms. Mary Ann Bishop and Family
Mr. and Mrs. Gerald D. Carlisle
Ms. Betty Corman
Mr. and Mrs. Brad Cutter
Ms. Ellen Eckert
Mr. and Mrs. Keith Elliott
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Mr. Tony Harrell
Ms. Marie Jenkins
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Mrs. Marjorie C. Orwig

Donald K. Remy

Mr. and Mrs. Mike Barry
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Mr. and Mrs. Rich Fischer
Ms. Geraldine Kurk
Mr. and Mrs. Skip Rueter
Ms. Charlotte Stroot
Mr. and Mrs. Arthur Vahlkamp
Mr. and Mrs. Steve Vahlkamp and Anna
Mr. and Mrs. Mike Vanmeter and Family
Ms. Sharon Zehnle and the Neighbors

Alan Sohnen

Mrs. Belle Sohnen

Vivian Westerfer

Ms. Diane L. Westerfer

IN HONOR OF:**Riley Ashe**

Mrs. June McCarthy
Mr. John McCarthy

The Atlanta Support and Action Group

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Joshua Beron

Mr. and Mrs. Ira Geller
Mr. and Mrs. Alan Goldstein

Robbie Campbell

Mr. Jordan Grable

Tina Cox

Ms. Querlyn E. Marques

Billy Kennerly

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Pam Kleinman

Mr. and Mrs. Howard Fuchs

Zachary Korowitz

Mrs. Julie Altman
Mrs. Erica L. Axelrod
Ms. Hope G. Becker
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Mr. and Mrs. Irwin Scharf
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Ms. Sharon Steinberg
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Mr. Alan M. Weissman
Mrs. Barbara Wineburg
Mr. Eric Wollman
Mr. and Mrs. Hyman Wollman

Melinda Lang

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Tyler Ray Lopez

Ms. Jean A. Moore

Lauren Roskopf

Mrs. Holly Halsted-Smirl

Phyllis Sanders

The Erika and Kenneth Witover
Family Foundation

Betty Williams

Ms. Judy W. Goyer

Thank you to all of you who have contributed to the Board Challenge. To date, we have received over \$252K, but we still have \$98K left to reach the \$350K Board Challenge. If you have not yet contributed, please do so today. We do not want to lose our matching funds.



Genetics Questions

ANSWERED BY CARLY SISKIND AND SHAWNA FEELEY, WAYNE STATE UNIVERSITY

Editor's note: These questions were posed by support group members and answered by the genetics counselors during the meeting of support group leaders in Las Vegas.

Can one person have 2 or more types of CMT?

Yes, but only rarely, as they would need mutations in two or more genes that each cause CMT.

Why and how does CMT skip generations?

It doesn't. For some people, symptoms may be so mild that they go unnoticed. For other families, the type of CMT may be one that presents differently in different sexes. For example, in CMT1X, females often have less severe symptoms than the males in the family, and in some cases, have no symptoms. Therefore, it may seem that the CMT is skipping a generation, but it is not.

Why is there so much variation in the presentation of the same type of CMT in two different individuals?

A good question—one that researchers are trying to figure out. The truth is that no one really knows at this time.

Why does it cost so much to determine the different types of CMT through genetic testing?

Genetic testing is expensive and this is due, in part, to the complex task of sequencing a number of genes in order to identify mutations. To help reduce cost, we encourage people to test in an appropriate way by narrowing the number of genes that you

test for to those that are most likely to yield a positive result. As technology changes and new methods for genetic testing become available, the tests will be performed at lower costs.

Is it true that the DNA test for CMT uses white blood cells, not red, and is this a reason why the test is expensive since more work is involved?

The cost of genetic testing is not associated with the type of blood cells being evaluated. When genetic testing is being performed on a blood sample, they do not use red blood cells because there is no nucleus in red blood cells. Since the nucleus houses the DNA, there is no DNA in red blood cells. Cells that do have a nucleus, such as skin, muscle, and cheek cells, can be used for genetic testing, but extracting DNA from white blood cells is the easiest way to get the most DNA to work with.

I read an article by Linda Crabtree, and it sounds as though she has vocal cord involvement. How common is that?

The main symptoms of CMT are length-dependent motor and sensory neuropathy. This includes weakness and sensory loss beginning in the toes and traveling toward the body. Some people have more involvement with their hands than their feet. There are different degrees of severity. Sometimes these are predictable based on the type of

CMT, but other times it varies significantly. Some people do have vocal cord involvement. It is, as far as we know, a portion of people who have CMT2A and a portion of people with CMT4A. It is usually seen in people who have more severe cases of CMT. Another thing that may be seen in people with CMT is hearing loss. This is hearing loss that is caused by damage to the nerves and is not helped by hearing aids.

How do you suggest parents with CMT explain the inheritance issue to their young teens?

That is a difficult question. It is hard to know the right time to explain a genetic condition to a child, both from the standpoint of inheritance and what the condition really is. The general guideline, when it comes to the right time, is typically when kids start asking questions. All questions should be answered as honestly as possible. Many teens take biology in their first or second year of high school. The teaching of genetics in class may be an appropriate time to bring up the subject of inheritance.

For some kids and parents, the best thing may be to sit down and have a frank conversation with the child. Ask your kid, "Do you know what CMT is?" "Do you have any questions about it?" "Are you concerned about the future?" "Did you know this is something that can be passed on, and your children could have it?" Kids are extraor-

dinarily capable of finding information on the Internet. You may find that they know everything you want to tell them. You may find that they have stumbled on information that scares them. I had one patient who came to see us at 19 years of age who told me that she was sure she would wind up in a wheelchair. It was the first time her mother had heard of this fear (and for the type of CMT she has, type 1A, 95% of people never end up in a wheelchair). Kids may have heard or found

information that is true for some people, but does not apply to the type of CMT that the child actually has. Having a conversation with a genetic counselor can also be useful because genetic counselors are trained in breaking bad news and restating information so that people can understand it more easily.

The bottom line is that we believe that information can be a powerful tool in terms of helping people cope with a disorder such as CMT. This is not only true for adults, but children, too.

Helping them to understand and articulate why they may need to wear braces or do things a little differently than their peers empowers them to be able to explain it, for example, on the playground when a peer is asking questions. This ability to talk about issues can lessen the confusion or fear about them. Avoiding these topics or not discussing them with children can lead to a feeling that there is a stigma associated with CMT, which might make the child feel worse about himself. *

CMT IN THE NEWS

- Abbey Umali was reappointed as the National Goodwill Ambassador for the Muscular Dystrophy Association. She is only the third ambassador to be chosen three times to represent the association. Abbey turned 11 in March and is diagnosed with CMT. She is a top fifth-grade student and loves animals of all kinds. She hopes to be a veterinarian one day. She is from Redlands, CA, and has been touted for her spirit and dedication.

- A link has been found between CMT 2C and two rare forms of spinal muscular atrophy (SMA). Three teams of researchers in the United States and Europe identified mutations in chromosome-12 for the TRPV4 protein that tie together the origins of the two diseases. Generally it has been assumed that SMA and CMT arise from very different causes. SMA is a

disease of motor neurons and CMT is a disease of peripheral nerve fibers. The TRVP4 protein is a cellular channel present in a variety of tissues, including nerve fibers, that opens and closes to allow calcium and other positively charged particles to move in and out of cells. The mutations in TRVP4 cause abnormalities of calcium concentration (either too high or too low) in the nerve fibers. The unexpected findings indicate that the rare forms of SMA result from abnormalities in the nerve fibers of the periphery rather than cell bodies in the spinal cord.

- CMTA member Lisa Eichlin has written a book for younger children which can either be read by the child or to the child. When you purchase the book, you get a code which enables you to download an audio file of the book being read that you

can then use in an iPod or burn to a CD. The book is entitled *The Bravest Boy I Ever Knew* and is told from the vantage point of a young boy whose best friend is in a wheelchair. He talks about how similar they are and what special things his "brave" friend can do. The book is available from Amazon.com for \$8.99.

- Dr. Rebecca Ionasescu, 86, and her husband, Dr. Victor Ionasescu, 83, both passed away in February. Victor Ionasescu was a well known scientist and educator and was long affiliated with the CMTA. Both Rebecca (Gabi) and Victor began working on CMT in 1982 and spent 15 years searching for genes that cause the disease. At one point, Victor had the largest database of CMT patients in the United States. Both doctors worked at the University of Iowa hospitals into their seventies. *

CMTA Support and Action Groups: Our Facilitators are on the Move!

BY ELIZABETH OUELLETTE

Over the past several years, I've spoken with many people who would positively like to attend a CMTA support group meeting and those who inquire enthusiastically about starting a group in their communities. However, I have also been in conversation with a great number of people who experience overwhelming trepidation and disquiet when the term *support group* is used. For a plethora of reasons, the phrase gives some individuals goosebumps, as it conjures up various images of sad, despondent and desperate-looking people, sitting around in a circle, commiserating and complaining about the horrible deck of cards life has dealt them. Others imagine that the attendees are all over 85, sitting with walkers, canes, and wheelchairs as their only friends, staring lifelessly into space, waiting for the inevitable. Others imagine a reflection of themselves in 20 to 30 years, in some imaginary world that has yet to be. Still others may think that these groups have a cultish quality, and fear getting sucked into some mystical and ritualistic meeting, clad with bells and heavy with wisps of pungent incense floating around in mid air.

Because of some of the above negative and erroneous connotations, the term *support group* was modified at the Las Vegas conference. After some

debate, we unilaterally decided that *Support and Action Groups* (SAGs) renders a much more accurate picture of what these meetings are all about (the acronym SAG was not the first choice, but it brought out so much laughter, we just had to keep it). In essence, these self-help groups provide a place where members can share stories, experiences, and ideas for coping with CMT, in an environment of acceptance, empathy, and understanding. These are not static groups, but dynamic and vibrant entities, whose members are actively changing the world of CMT, one day at a time. Some of these activities include spreading awareness about CMT and STAR to health professionals, providing education to CMT members, friends, and family, creating synergy to improve the lives of everyone with CMT, and fundraising for a much-needed cure.

Upon our return from the Las Vegas conference, most of our support group leaders, who are now called "facilitators," to underline the organizational and collaborative nature of their function (see "The Goose Story") have been working hard to promote our cause. Here are just a few examples: Nancy Mollner, from Williamsburg, VA has just raised approximately \$1000 by selling Krispy Kreme donuts to her members and

friends. Suzanne Ruediger, from Atlanta, Georgia is gearing up to do a Circle of Friends on behalf of the CMTA. Beverly Wurzel, from Fort Lee, New Jersey is getting her community involved in raising money for Ken Gomez' cross-country ride. All monies donated to Ken's ride will be matched by the Paul Flynn Charitable Trust (<https://secure.charcot-marie-tooth.org/kensride.php>). Beverly has already brought in close to \$1000 for the CMTA. Among the many activities that Jeana Sweeney from Johnstown, PA has undertaken was persuading Pepsi-Cola Bottling Co. in Johnstown to sponsor her annual softball tournament for the CMTA. Donna Rennie, from Walnut Creek, CA, is organizing a "Fundraising 101" conference in the Bay Area for all those who are interested in learning more about raising money for our growing nonprofit. Gretchen Glick, who created and currently manages the CMTUS (www.CMTUS.yahoo.com) from Solvang, CA constantly updates her members about STAR and continually promotes ways to support the CMTA (igive.com, fundraising ideas, updated CMT news, etc). Mark Willis from Freehold, NJ and Ruth Korowitz, from Plainview, NY along with Jeana Sweeney are organizing a CMT Kids Day in the fall. Dale Lopez, from Chicago, Illinois, is expanding

As our support and action group facilitators build grass-roots communities across the country and spread awareness of CMT, they will inevitably need as many hands as are willing to help. When the universal dynamics of team building are understood, we realize that no one person can, or should, do or be everything. True success really depends on creating a network of individuals, committed to a common goal, who share the overall workload from beginning to end. The story at right exemplifies this tenet.

her annual Tyler's Walk to include all her support and action group members. Ronnie Plagman, from Ocala, Florida, has invited us to join him at Cooterfest (www.cooterfest.com), where the CMTA staff will man a booth and support Jerry Cross and his famous Archy Train.

So as you see, all across the nation, our facilitators have been working very hard since the month of March to provide support and tangible results that will benefit each and every one of you who has CMT. Come and join our ever-expanding CMTA community: join a group, start a group, or contact the CMTA to discover new ways in which you can make a difference. To the many individuals who have made a unique difference in the livelihood of our Association and in the lives of others, we send deep appreciation and heartfelt thanks. Keep up the great work SAGs!!! *

THE GOOSE STORY

Next
fall, when
you see geese
heading South for
the Winter, flying along
in V formation, you might
consider what science has discovered as to why they fly that way:
as each bird flaps its wings, it creates an
uplift for the bird immediately following. By
flying in V formation the whole flock adds at least
71% greater flying range than if each bird flew on its own.

***People who share a common direction and sense of community
can get where they are going more quickly and easily
because they are traveling on the thrust of one another.***

When
a goose falls
out of formation,
it suddenly feels the drag
and resistance of trying to go it alone
and quickly gets back into formation to take
advantage of the lifting power of the bird in front.

***If we have as much sense as a goose,
we will stay in formation
with those who are headed the same way we are.***

When
the head goose
gets tired, it rotates back
in the wing and another goose flies point.

***It is sensible to take turns doing demanding jobs
with people or with geese flying South.***

Geese
honk from behind to
encourage those up front to keep up their speed.

What do we say when we honk from behind?

Finally,
and this is important,
when a goose gets sick, or is
wounded by gunshots and falls out
of formation, two other geese fall out with that
goose and follow it down to lend help and protection.
They stay with the fallen goose until it is able to fly, or until
it dies. Only then do they launch out on their own, or with another
formation
to catch up with their group.

**IF WE HAVE THE SENSE OF A GOOSE,
WE WILL STAND BY EACH OTHER
LIKE THAT.**

*by Dr. Harry Clarke Noyes
ARCS NEWS, Vol. 7, No. 1, January 1992*



SUPPORT AND ACTION GROUP NEWS

California – East Bay

At the May meeting, Donnie Rennie set out to turn a passive group into an active one. The Las Vegas conference was discussed and people were asked to take on various responsibilities such as resources, fundraising, and support group concepts. Someone took on the fundraising chores and the group has already committed to two nights at Applebees and a spaghetti dinner in October. The next meeting will be in June with a presentation by Hangers Orthotics.

California – Sacramento Area

The group met for the first time on March 27th. The room held 48 people and by 1:05, the room was packed. Later, the facility opened up the dividing wall and ultimately, we had about 60 people in attendance. The meeting started with an ice-breaker, allowing us to meet and

understand a little about the people in attendance. Nathan Sherman played the video “A World without CMT” and then walked everyone through the STAR program slide show. Following lunch, the group had an open discussion on topics such as AFOs, genetics, hip displacement, the work place, and respiratory issues.

California – Santa Barbara

The group met on April 25th for lunch and a discussion of the use of vitamins and supplements. Prior to the meeting, Gretchen met with seven new members, one, an Hispanic family, who had found her through Craigslist. She gave them the brochure in Spanish and helped them understand the basics of CMT, including fatigue and balance problems. After lunch, the group learned about Dr. Sahenk’s mouse study of injectible NT3 (neurotrophic factor).

California – San Francisco Peninsula/South Bay

The last meeting was held on April 25th at the Los Altos Library. Rick Alber will be the new co-facilitator of the group. Many members of the group stepped up to take on jobs such as treasurer, snacks coordinator, official greeter, and resource manager. A fundraising committee is looking at the possibility of holding a Poker/Texas Hold’Em tournament in the fall. The next meeting will be in June, with the annual picnic scheduled for August.

Florida – Tampa Bay

David Hall, CEO of the CMTA, flew down to Florida to discuss how to get CMT “out of the closet” by sharing our story with family and friends. He discussed how the CDC and NIH are now aggressively working on CMT because the CMTA spoke up and finally asked for their help. Dave also explained how STAR is progressing. The fact that “treatments” are finally something we can believe in brought tears to many members in attendance. The group was thrilled at how quickly the knowledge and understanding of all types of CMT has progressed.

Georgia – Atlanta

The March meeting had the largest turnout to date...38 people, with five new members. The speaker was Nancy Duncan from the Disability Resource Group. She discussed her disability—blindness—and the Americans with Disabilities Act. After Nancy’s presentation, group facilitator Susan Ruediger discussed what she had learned at the Las Vegas support group meeting. Of all the presentations, the most significant was the report from Dr. Michael Shy about the success of the STAR initiative. They are on the brink of a treatment for type 1A and after that is accomplished, they will begin to build a cell line for type 2A.

New Hampshire/Vermont

The group met on March 27th at the Dartmouth-Hitchcock

(continued on page 18)

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We Need Your Best Thinking!

Please take a quick survey to help design our new web strategy...

CMT may be classified as a rare disease, but there’s no reason why it should remain obscure. To take advantage of social media and other digital strategies, the CMTA is designing a comprehensive web strategy.

Our goals are to serve our members better, to build a national and international registry for clinical trials, and to put CMT on the radar of the significant leaders in Washington and around the globe. Ultimately, the plan is designed to achieve our goal of finding a cure.

Go to www.cmtausa.org/mythoughts and take a five-minute survey to join us in our efforts to improve the lives of those with CMT.

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**SUPPORT AND ACTION
GROUP NEWS***(continued from page 16)*

Medical Center in Lebanon, NH. There were 15 people in attendance. The topic for the meeting was pain management. We learned about various interventions for the management of pain and where to go for help. We also learned that chronic pain can produce depression and that the Wong-Baker FACES Pain Rating Scale is often used to define the level of pain being experienced. The next meeting will be in June.

New York – Westchester Area

At the May 1st meeting, Ken Gomez and Nancy Homiak were guest presenters. Ken will be riding 4,200 miles across the United States to raise money for research. Every dollar he

raises up to \$22,000 will be matched by a memorial fund established by his cousin, Paul Flynn, who was the first Executive Director of the CMTA. Nancy, another member of Paul's family, talked about fundraising ideas that the group might try. There was a large turnout and everyone enjoyed a potluck brunch.

North Carolina – Triangle

The group met on April 10th. Fifteen people were in attendance, with people coming from Charlotte to Greenville. Susan Salzberg presented information from the Las Vegas support group leaders' conference. The group watched the slides produced by Ken Wysocki on the robotics at NIH and the study of compounds which may provide treatment for CMT1A. The importance of fundraising was

stressed. The expense of running animal and human trials for just one compound is around \$2 million.

Pennsylvania – Johnstown

The group met on April 24th with 14 members, including one new member in attendance. Jeana Sweeney's four-year-old daughter, Rylee was also in attendance. Tony Franke, RN, did a presentation on foot care. Following her presentation, the group watched a modified PowerPoint presentation on STAR. JD handled the scientific issues and Jeana talked about fundraising. By the time Jeana was done, the members were fired up about fundraising and JD managed to get the scientific results of the high-throughput screening explained so that members were equally excited about that aspect of things. *

EUROPE*(continued from page 8)*

six hours and I used those hours to get some much-needed R&R.

Upon our arrival in Prague, we took the subway to our hotel. The ride took about 30 minutes and was a good opportunity to move around after sitting for six hours in the train. We left the hotel to find a restaurant at about 6 PM. Our party split into five groups, each of which chose a separate restaurant. My group chose a restaurant hastily, based on its appearance. Little did we know that they would charge extra for converting our payment from

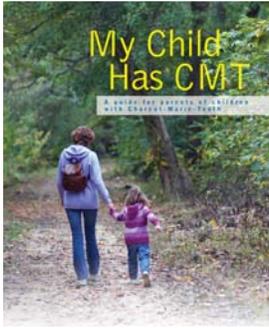
their currency into Euros. This experience left a bitter taste in our mouth for the remainder of our trip!

The next day, we visited several synagogues that survived the Second World War. One of them had been converted into a Holocaust memorial with thousands of victims' names inscribed on all the walls. This experience was a solemn reminder of the Holocaust. After visiting this memorial for several hours, I began to notice the pain in my feet so I sat down and rested for a short time. We then returned to the hotel. The following day we visited an extremely large cathedral dating

back several hundred years. That night at the hotel, our party prepared to say our farewells to Europe. The next morning we departed for San Francisco via Munich.

My journey to Europe presented me with physical challenges that I overcame with a variety of coping techniques. Through the use of brief resting periods, a distracting novel, and perseverance, I was able to cope with the constant difficulties that arose from my CMT symptoms. Also, all my friends, parents, and teachers supported me in any and all ways possible, which made for a very memorable and fun trip. *

MEMBERSHIP APPLICATION/PUBLICATIONS ORDER FORM



Pediatric Publication Now Available

My Child Has CMT is a 32-page booklet with full-color photos and topics such as "When We Found Out Our Daughter Had CMT," questions and answers from Dr. Mena Scavina, A.I. DuPont Institute in Wilmington, DE, "How to Keep a Medical History for Your Child," and a dictionary of common medical terminology. A list of resources and organizations helpful to families with CMT is the final chapter. The booklet is only \$5 for CMTA members and \$7 for non-members.

ALSO AVAILABLE:



Circle of Friends T-Shirts



Be a STAR Wristbands

NAME: _____ / _____ / _____
First MI Last

ADDRESS: _____

CITY: _____ STATE: _____ ZIP: _____

COUNTRY/POSTAL CODE (IF NOT US): _____

DAYTIME PHONE: _____ EVENING PHONE: _____

EMAIL: _____ USER NAME: _____
Required for PDF Newsletter/Website Access *Required for Website Access*

Note: If you are joining now, you may purchase publications at active member prices.

	QTY	COST	TOTAL
MEMBERSHIP Members have the option of receiving <i>The CMTA Report</i> in print, PDF via email, or both. Receive newsletter as: <input type="checkbox"/> Print or <input type="checkbox"/> PDF via email Receive both Print <i>and</i> PDF Newsletters		\$25 \$30	
The Patients' Guide to Charcot-Marie-Tooth Disorders [Print Format]		active members \$10 nonmembers \$15	
[CD Format]		active members \$7.50 nonmembers \$10	
CMT Facts I <input type="checkbox"/> English <input type="checkbox"/> Spanish		active members \$3 nonmembers \$5	
CMT Facts II <input type="checkbox"/> English <input type="checkbox"/> Spanish		active members \$5 nonmembers \$7	
CMT Facts III		active members \$5 nonmembers \$7	
CMT Facts IV		active members \$8 nonmembers \$10	
CMT Facts V		active members \$12 nonmembers \$15	
CMT Facts VI		active members \$12 nonmembers \$15	
A Guide About Genetics for CMT Patients <i>(No shipping and handling on this item only)</i>		active members \$4 nonmembers \$5	
Teaching Kids about CMT...A Classroom Presentation <i>(1 hour DVD)</i>		active members \$7.50 nonmembers \$10	
NEW! My Child Has CMT, A Guide for Parents		active members \$5 nonmembers \$7	
Be a Star Wristbands (Pack of 5)		\$5	
Women's Circle of Friends V-Neck T-Shirt Quantity and Size: ___M ___L ___XL		active members \$9 nonmembers \$10	
Men's Circle of Friends V-Neck T-Shirt Quantity and Size: ___M ___L ___XL ___2XL ___3XL		active members \$9 nonmembers \$10	
West Coast Patient-Family Conference <i>(5 hours—2-DVD set)</i>		active members \$15 nonmembers \$20	
CMT Informational Brochure, Neurotoxic Drug Card, and Letter to Medical Professional with Drug List		FREE	
Physician Referral List: States: _____		FREE	
Donation to the CMTA (100% Tax-deductible)			
Shipping & Handling <i>(Orders under \$10, add \$3.50; orders \$10 and over, add \$7.50)</i>			
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, 2700 Chestnut Parkway, Chester, PA 19013; 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 (ddl, Videx)
Dichloroacetate
Disulfiram (Antabuse)
Gold salts
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)
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
Fluoroquinolones
Glutethimide
Lithium
Phenelzine
Propafenone
Sulfonamides
Sulfasalazine

THE CMTA Report



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What is CMT?

- ◆ is the most common inherited neuropathy, affecting approximately 150,000 Americans.
- ◆ may become worse if certain neurotoxic drugs are taken.
- ◆ can vary greatly in severity, even within the same family.
- ◆ can, in rare instances, cause severe disability.
- ◆ is also known as peroneal muscular atrophy and hereditary motor sensory neuropathy.
- ◆ is slowly progressive, causing deterioration of peripheral nerves that control sensory information and muscle function of the foot/lower leg and hand/forearm.
- ◆ causes degeneration of peroneal muscles (located on the front of the leg below the knee).
- ◆ does not affect life expectancy.
- ◆ is sometimes surgically treated.
- ◆ 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).
- ◆ has no effective treatment, although physical therapy, occupational therapy, and moderate physical activity are beneficial.
- ◆ 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.
- ◆ Types 1A, 1B, 1C, 1D (EGR2), 1E, 1F, 1X, 2A, 2B, 2E, 2F, 2I, 2J, 2K, 4A, 4C, 4E, 4F, 4J, HNPP, CHN, and DSN can now be diagnosed by a blood test.
- ◆ is the focus of significant genetic research, bringing us closer to solving the CMT enigma.