THE CMTA Report

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

JULY/AUGUST
2010

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.

EMAIL CMTA AT:
info@charcot-marie-tooth.org

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Research Information Revealed at PNS Satellite Meeting

BY PAT DREIBELBIS

Peripheral nerve researchers from around the globe listen to the opening lecture of the Satellite Meeting in Sydney, Australia.

The satellite meeting of the Peripheral Nerve Society met from July 5-7 in Sydney, Australia. The first presenter of the combined meeting was Dr. Michael Shy, Detroit, Michigan. He talked about the advancements in CMT research, including STAR, and the need to make all of the research efforts global ones. His talk was particularly significant because of the high numbers of attendees from Asian countries with whom there has not been as strong a relationship as we would like going forward.

Several researchers reported on a new form of X-linked CMT that is caused by a mutation in the copper transporter gene ATP7A resulting in a distal hereditary motor neuropathy. Another report focused on CMT type 2C, which is caused by TRPV4 mutations and results in a form of CMT with diaphragmatic and vocal cord involvement. Carly Siskind,

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INSIDE:

Tonight the "End" of CMT Begins!

Patrick Livney, President and Chairman of the CMTA Board, introduced the first honoree of the night—his brother Roland.

That slogan was the theme of this year's "Honor a STAR. Be a STAR" gala. As David Hall said in his opening remarks, the focus of the event was the power of one and the impact that each person can have in the fight against CMT. A combination of scientific innovations and advancements, along with great leaders who are working on behalf of the CMTA, will allow the CMTA to continue to push toward a cure.

Sadly, the wonderful event was lessened slightly by the absence of Penn State Coach Joe Paterno, who was ill and hospitalized in State College. Despite that, his wife Sue and a former assistant offensive coordinator at Penn State, Fran Gantner, came and spoke eloquently of Joe's commitment to excellence and his refusal to ever accept defeat. His attitude was likened to the approach the CMTA is taking in finding a cure for CMT—striving for excellence in choosing the researchers who are leading the charge and never accepting defeat even though, at times, the task seems overwhelming.

Chairman of the Board Patrick Livney mentioned the advances that have been made since last year's event and promised that he would continue his personal mission of guiding the CMTA toward its vision of a world without CMT. To that end, he quoted Vince Lombardi when he said, "I firmly believe that any man's finest hour, the greatest fulfillment of all that he holds dear, is that moment when he has worked his heart out in a good cause and lies exhausted on the field of battle—victorious."

The honorees at this year's event were Roland Livney, who is Patrick Livney's fraternal twin brother. Although Patrick is diagnosed with CMT, his brother does not have the disorder. He has, however, been a champion for the CMTA for more than five years, establishing the Livney Foundation, which has given more than $1 million to CMT research. Roland demonstrates that the bond between brothers is profound and the will to find a cure drives both of them.

The second honoree was Amy De Silva, who at age 14 has been a youth ambassador for the CMTA and has used her vocal talents to spread the message of hope and encouragement to oth-

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Herb Beron and his daughter Julia proudly noted the reasons that Amy De Silva was an honoree of this year's gala.

CMT Continues to Be Recognized in Washington, DC

On June 15, 2010 the U.S. House of Representatives Committee on Energy and Commerce, Subcommittee on Health held a hearing titled "NIH in the 21st Century: The Director's Perspective." Testifying before the Congressional Subcommittee was NIH Director Dr. Francis Collins. During the hearing, Dr. Collins lauded the CMTA's research collaboration with the NIH's National Chemical Genomics Center (NCGC) and acknowledged that the CMTA-NCGC relationship can serve as a model for future research and drug development programs for rare and neglected diseases.

Responding to a specific question by Representative Eliot Engel (D-NY), Dr. Collins also suggested that the CMTA-NCGC program represents the type of partnership that could entice companies to take an active role in future drug development efforts.

To that end, the CMTA thanks Representative Engel and the Subcommittee for their continued support of CMT research. The CMTA is honored to have the NCGC as a partner in our quest to find therapies and cures for CMT, and is greatly appreciative of Dr. Collins and his colleagues throughout the NIH for their commitment to rare disease research and drug development. The success of STAR can only be sustained through the continued support of the national CMTA community, along with our scientific, academic, and public sector partners.

We remain eternally grateful for everyone's support and look forward to updating you on the latest advancements within the STAR program.

Below is a transcript of the exchange between Representative Engel (D-NY) and Dr. Collins during the Congressional Hearing.

—Dave Hall,
Chief Executive Officer

United States House of Representatives, Committee on Energy and Commerce, Subcommittee on Health

"NIH in the 21st Century: The Director's Perspective"

ENGEL: I'd like to ask you about Charcot-Marie-Tooth Disease. There is an innovative partnership between the NIH's National Chemical Genomic Center and the Charcot-Marie-Tooth Association. I've worked with that association, and I think that this can serve as a model of future rare disease research and drug development, and so I'm wondering if you could describe the program and offer some thoughts on it.

DR. COLLINS: I appreciate the question because I do agree this is a very exciting program. My father-in-law has Charcot-Marie-Tooth disease and so this is a disorder that is not only something from my clinic but also from my family's experience, and this is an interesting disorder which causes a weakness of the legs particularly, but also the hands over the course of time and can be quite debilitating, but it is well-understand now what the cause of that is…the genetic abnormality has been now laid out in great clarity, but what could you do about it? Working with the NIH Genomic Center, which is this remarkable facility that has been mentioned already at least once in this hearing. It is…an effort is being made to identify a small molecule, which is sort of a drug that would basically compensate for the genetic problems that's found in individuals with this type of Charcot-Marie-Tooth disease, so-called CMT1a, and that is an early stage effort but it's a good example of this therapeutics for rare and neglected diseases effort that NIH is putting an increasing effort into. Charcot-Marie-Tooth is too rare for companies to generally see this as a good investment for them in terms of developing a therapy. But with the chemical genomic center working with academic investigators who know a lot about the disease…if they can push this forward to the point of identifying a promising compound, then you can imagine a company getting pretty interested in licensing it out and carrying it all the way through to a clinical trial.

Additional information about the hearing, including a transcript of Dr. Collins testimony can be found at the website below:
http://energycommerce.house.gov/index.php?option=com_content&view=article&id=2042:hearing-on-nih-in-the-21st-century-the-directors-perspective&catid=132:subcommittee-on-health&Itemid=72

CMT GALA

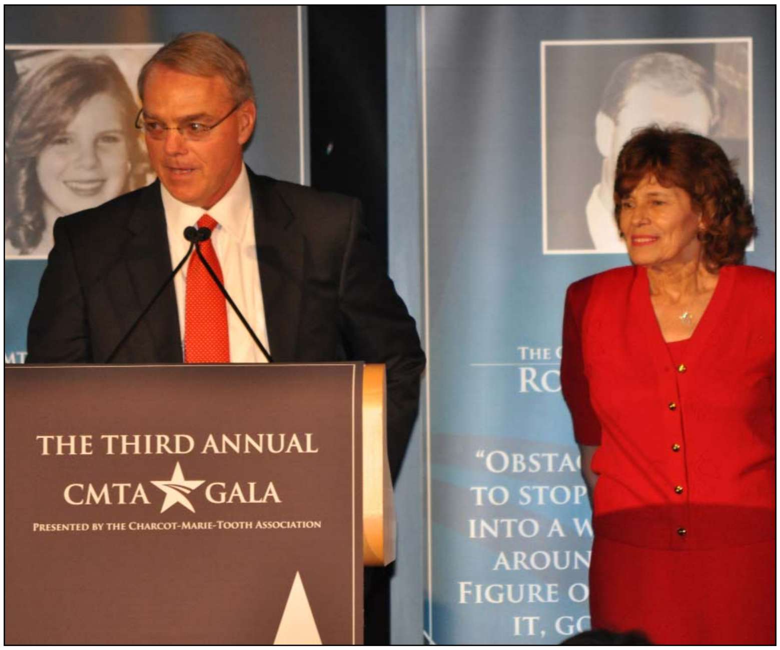
(continued from page 2)

ers. Amy has performed the National Anthem at Boston's Fenway Park and Kansas City's Kauffman Stadium as a representative of the CMTA. On her Facebook page, Amy's STAR Voyage, she shares her personal insights into her life and the challenges of living with CMT. Her goal has always been to help other children and teenagers living with CMT, and her personal journey has inspired thousands outside of the CMT community.

The final recipient of this year's award was Kurt Coleman and the Ohio State University for their work in making the CMTA the recipient of their fundraising efforts in the 2009 football season. Senior Kurt Coleman urged Terrelle Pryor, whose father has CMT, to join him in hosting a video game challenge that pitted the Ohio State offense against the defense with fans paying to watch the competition. The gaming event was just one effort by "Uplifting Athletes" that benefitted a rare disease. Public service announcements aired on the Big Ten Network throughout the 2009 football season and brought additional awareness of CMT to the thousands who follow Big Ten football.

After the awards ceremony, the audience participated in a live auction, bidding on such items as four box seat tickets, food, and parking for the Penn State-Michigan home game in the fall of 2010. This auction item was a gift from the Paternos and brought in $4,000. There was also a golfing weekend in North Carolina given by Robert Kleinman; a golf outing at the Congressional Country Club in Washington, DC, given by Gary Gasper, and a cast-signed poster from the first *Twilight* movie, four premium seats to a Dodgers/Yankees baseball game in Los Angeles, and four house seats to the Billy Elliott Show on Broadway, all donated by Board member Vasi Vangelos. During the pre-dinner cocktail hour and the dinner itself, a silent auction was also conducted which featured items from history-making sports moments, like a famous Willie Mays' catch, political moments, like the election of JFK, and show business legends like a poster and autographs of the original Rat Pack. All proceeds will benefit the STAR initiative.

CEO David Hall ended the night's festivities by thanking everyone who "individually" has had an impact on the fight to find a cure for CMT and who has helped "this night be where the end of CMT begins."



Fran Gantner speaks on behalf of Joe Paterno as Joe's wife Sue listens and looks on.

A "STAR" AT THE STAR GALA

While Pat Dreibelbis, Dana Schwertfeger, and Jeana Sweeney were waiting for the elevator down from the ballroom, they realized they were waiting with Ricky Martin of "Livin' La Vida Loca" fame. It was definitely the high point of the evening for Jeana, who managed to have her picture taken with Ricky once they reached the ground floor.

Stepping It Up for CMT

BY JEANA SWEENEY, CO-LEADER, JOHNSTOWN SUPPORT GROUP

Children in the Alleghenies of Pennsylvania are raising money for the Charcot-Marie-Tooth Association. To this date, three elementary schools—Richland Elementary, Black Lick Valley Elementary, and Westmont Hilltop Elementary—have participated in a program called "Stepping It Up for CMT," raising an amazing amount of $13,600. All in all, these three schools, comprising 1900 children and the children's parents, have been educated about CMT. So Stepping It Up for CMT not only raises money for research, but also brings much awareness to CMT!

How the program works: I go to each grade with the goal of educating the kids about CMT by teaching them what CMT is and reading a story about Archy the turtle. This is then followed up by the children wearing pedometers during the school day to "Step it up for CMT." After wearing the pedometers, the children take a sponsorship sheet home with the number of steps they took. They then ask parents, grandparents, friends, and neighbors to be their sponsors. The sponsor has the choice on what they want to give, as there is no designated or mandatory amount of contribution—it could be $1 or $20. The children have about two weeks to get as many sponsors as they can, and then they bring the money back to the school and it is collected by a CMTA representative.

"Stepping It Up for CMT" not only raises money for research but it also raises awareness of CMT.

The children are excited to raise money for CMT, but they have another motivating reason to participate. Every child that brings in $1 or more will receive a participating gift (a CMTA bracelet). The child that raises the most money will be rewarded with lunch for them and a friend, from anywhere they choose (this does not seem like a lot, but it's a really big deal to them) and tickets to somewhere fun in their area.

I personally have been doing this program, and I must say it brings tears to my eyes hearing the children talk about CMT and how excited they get about raising money for the CMTA. Every school has its own special story, but there is one particular story that stands out the most. A little girl from one of the schools gave 10¢. To most people this is not a lot of money, but to me it is, because no matter how much pocket money you have, it's about giving what you can and every penny counts.

I would love to bring this program to your school and I am willing to help in every aspect I can. If you would like to learn more about "Stepping It Up for CMT," e-mail me at sagcf_johnstown@cmtausa.org

If you know anyone who might donate some pedometers to this project, please let me know. They will receive a thank you which designates the value of their contribution for tax purposes. ✻

CMT Awareness Week in September

The week of September 19-26th will be CMT Awareness Week. One of our plans to make the week special (and fun) will be to provide posterboard copies of Archy, the CMTA's spokesturtle, to anyone who requests it. Then, the task will be for each person who is participating to get Archy's picture taken with a famous landmark in their home town or with a famous person in their area (a mayor, a sports legend, an author, or anyone who matters to your part of the world.) The picture will then be sent to the office in Chester, electronically, info@charcot-marie-tooth.org, and it will be entered into a contest with the other photos. The picture which is the most imaginative or the most awe-inspiring will win a prize.

The CMT "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!

Fruit of the Vine

Wine Tasting with a Circle of Friends

It began with a Facebook friend on May 16 and ended four weeks and nine days later. It was a wine tasting fundraiser for the CMTA. This tasting "quaternity" was held over four consecutive weekends, beginning with the Memorial Day holiday and ending on Father's Day.

Bella Cavalli Farms in Solvang, California, hosted the tastings, giving 10% of the total sales over four weekends to the CMTA. Over 500 people came out to taste and support CMTA's research efforts.

One morning while sitting at my computer, I noticed Jeff Lockwood on my Facebook "suggested friends" list. I read his profile, and saw he lived in Solvang, so I added him to my friends' list. The next day I saw a Facebook wall post from him saying he was preparing for the weekend American Cancer Society fundraiser. I thought "I wonder if he'd do anything for CMT"? So, I contacted him via Facebook. He responded right away with his phone number and said to call. In that one call I learned he could have a wine tasting for CMT! We chatted briefly about the event and met the next day.

Bella Cavalli Farms is both an equestrian rehabilitation facility and a vineyard.

To my complete surprise, Bella Cavalli Farms is about two miles from my home, I had driven by the front gate nearly every day for 8 years, and never knew it was there! The property is also an equestrian rehabilitation center. It is an undiscovered unique and precious gem of an estate, well hidden from the main highway and city lights.

In our meeting, Jeff explained how his tastings work and how 10% of the sales would be donated to the CMTA. He requested a copy of the association's 501(c)(3) letter, and a refundable day use permit fee for the State Alcohol Beverage Control Department. (California law). Dana, from the main office, immediately sent me the 501(c)(3) letter and that was quickly followed by the permit fee from Kim.

With those two pieces of paper, I met with Jeff once more, and we completed one form for the ABC and sent the check. Jeff taught me more about the wine industry and explained how the CMTA label would be on wine bottles. Because of his commitment to non-profits, two weeks later, I had a wine tasting event scheduled for Memorial Day Weekend. Three days! I took Jeff some brochures and newsletters to place in the "Boots and Bottles" tasting room. Then I sent the news to area media, my college and grad school friends, my "electronic Circle of Friends," personal friends, and members of the professional community.

I was ecstatic and spent the weekend speaking to guests about the CMTA and STAR as they tasted four wines (Pinot Noir, Pinot Grigio, Chardonnay, and Syrah) along with "Doc Jack's" barbecue sauce and olive oil before touring the immense grounds and meeting the beautiful horses in residence. This tasting was held outside on the spacious south lawn under shady oak trees with plenty of tables and chairs for comfort. Fresh-baked bread was on hand for dipping into the barbeque sauce and olive oil. Joanne, Jeff's wife, loves to bake, and they both enjoy hosting guests on an intimate, individual basis. This contributed to a unique atmosphere for wine tasting. I took many photos.

I was tired by the end of the weekend, but still wildly happy the event was so well received. A few days later, I phoned Jeff regarding pickup of any leftover newsletters or brochures and to thank him, his wife Joanne, and father "Doc" Jack. Jeff was giving a riding lesson, so I left a message on his machine.

The next day, Jeff returned my call and then we chatted about the successful weekend event. Then came another surprise! Jeff asked me if I wanted to have the following three weekends as additional fundraisers! Of course I said yes! This time all I needed was to sign the State SBC form and Jeff faxed it to Sacramento. I had two days to prepare. Not much time to get to the news media so I made some flyers and placed them at area libraries, medical offices, gyms, and even City Hall! I was out of brochures and didn't have business cards yet. However, I still wore my CMTA T-shirt!


"Bella Cavalli" is Italian for "beautiful horses."

The second weekend went even better. By now, I was familiar with the winery and property, so I spent more time taking photos of individual guests and learning they came from as far away as parts of Europe and the US East coast. This time we were set up on an even more private north lawn near the tasting room. Many more people learned about CMTA and STAR. Rave reviews of Bella Cavalli were given by all. Guests were genuinely very interested in CMT. In addition, several bachelorette parties dropped by, giving the tastings a celebratory mood! Joanne, was so generous in providing chocolate-covered strawberries for guests to nibble.

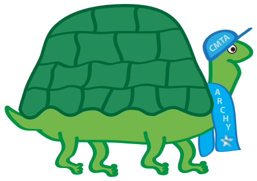
By the third and fourth weekends, I was well prepared with additional brochures, business cards, and CMTA T-shirts for Jeff, Joanne, and Jack. By this time I felt like part of the family! I still greeted guests and spoke about the CMTA with brochures and business cards in hand. I also felt like a pro when guiding guests around the grounds. Jeff added a special wine auction to these two weekends, including four bottles of wine, a jar of barbecue sauce, and a bottle of olive oil. The minimum bid was $118. Joanne provided attractive plates of fruit, cheese, and crackers and explained equine rehabilitation methods to all. As the last guest drove out the gate, Jeff and Joanne invited me to stay and watch the filming of a video of their young thoroughbreds. As a long-time horse lover, I was in heaven! It was a perfect conclusion to the four weekends of raising awareness and funds for the CMTA.

Additional fundraisers at Bella Cavalli are planned.

—Gretchen Glick

Wine sporting the CMTA label is available for purchase at http://www.bellacavallifarms.com/winery/order.php When you order, in the "comment box" state that you want the Charcot-Marie-Tooth Association label.

The Country Turtle and the City Turtle



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

One of the strange ideas that my mother came up with this summer was to invite a turtle who lives in the city to come and live with us for a few weeks to get the experience of living in our woods. It's part of some national program called "Fresh Air" and, normally, I guess it applies to kids. It sounded like an "okay" idea, but I knew if we had a boy visiting, the job of entertaining him was going to fall on me, and I just wanted to have my own private fun this summer. My mother gave me the inevitable lecture about thinking about others and not just myself and reminding me that I'm very lucky to live in such a nice place. Not everyone has that advantage, she said. Okay. So, she shamed me into saying I would go along with the plan. Privately, I prayed that the visitor would be a girl turtle my sister's age.

Of course, that's not what happened! The turtle chosen for us was a boy named Vinnie and he lived in a aquarium in New York City. Yuck. I'm sorry, but I don't make friends easily and having one thrust upon me is not my idea of a fun adventure. When Vinnie was "delivered" to us by bus, the awkwardness began. I had no idea what to say to him. Of course, my mother was all warm and gushy and he seemed to like her right away. I was another story. He looked at me, noticed the braces on my legs (my really cool camouflage ones) and seemed to totally withdraw. I couldn't decide if he thought I was crippled and wouldn't be able to do anything with him or if he was embarrassed for me because I was different. Frankly, it didn't matter what he thought about me, but it did put a definite freeze on our "friendship."

Another adventure this summer was going with CMTA Director of Program Services Pat Dreibelbis to Australia and meeting an actual koala bear.

The first day went by quickly. He got settled in *my* room, and he ate my mother's cooking. We barely spoke. On the second day, it was pretty obvious that I was going to have to make the first move. So, I invited him to go swimming with me in our pond. He said "yes," but I could tell he wasn't really sure of himself. It turned out that he has lived his whole life in a tank or aquarium, so his swimming hasn't really been perfected like mine has. He was actually timid even though he was bigger and stronger than I was.

I'm a star when it comes to swimming, and Vinnie hasn't really ever had to swim in open water, so he was slapping the water and generally making a lot of noise. I didn't think he was in any trouble, though, because how could a turtle drown? I mean, really, turtles are meant to be in the open water, aren't they? You can probably tell where this is going. He panicked and started flailing around. Only when he yelled did I realize that he couldn't get upright and was taking in a lot of water. I swam over and righted him so that he could catch his breath. I don't think he

Editor's note: Archy now has a Facebook page and would love for all of you to visit. You can visit his Facebook page at http://www.facebook.com/pages/Archie-the-CMTA-Turtle/132200940139421?v=info to read what people are saying to him and to comment on his observations.

was in any real danger, but apparently he thought I had "saved his life." He told every single animal in the woods and everyone in my family about my miraculous "save."

That night, when we were in my room, he finally asked about my legs and the braces. I told him what CMT is and how it affects me. He was really interested and was quite impressed that in spite of CMT I could swim amazingly. Once we started talking, I realized that he was a pretty cool turtle. His experiences weren't anything like mine, but he does have a sister, so we could talk about being bossed around by an older sibling. We shared some laughs about things we have done to our sisters (like hiding favorite pieces of clothing and toys) and we even shared some grudging comments about times that our sisters had been nice to us and come to our aid. As we talked, it became apparent to me that Vinnie and I were going to have a lot of fun while he was living with us in Greenwoods. I promised to help him with his swimming, and he decided he would help me learn to dance the way the turtles who live in New York City do.

I'm going to be sooooo cool! I think this "summer in the country" is a great idea. As it turns out, I am learning as much as Vinnie is, and we are already talking about my coming to visit him in the city next summer. I might have found a "forever" friend. You don't have to be like someone to like someone. That's my philosophy for today. ✻

Contemplations

BY NICOLE BURGGRAAFF

When I was five, I knew what I was going to do with my life. I was going to live in a house made of candy, travel in a spaceship to Mars, and be a Mom.

When I was eleven, I knew what I was going to do with my life. I was going to move to Boston, marry Joe McIntyre of the pop group New Kids on the Block, and be a Mom.

When I was sixteen, I knew what I was going to do with my life. I was going to convince Kurt Cobain to let me ghost write his autobiography, buy a loft apartment in Manhattan, and be a Mom.

Personally, CMT has not prohibited me from much. I could say it stopped me from becoming a surgeon because my hands lack dexterity—but I think failing grade 11 chemistry is what really killed that career path. CMT also kept me from joining a running group with other young Moms in my neighborhood —but I don't like to sweat, so I doubt a CMT-less me would be running alongside them anyways.

CMT did not stop me from having a group of ridiculously fun friends to venture through adolescence with. It did not prevent me from moving out of my parents' house to "the big city" to earn a University degree. It did not hinder me from finding the most awesomely loving (and good looking) man to become my husband.

But what kind of Mom gives her kid a 50% chance of inheriting a progressive neuromuscular disease? Especially a Mom who knows first-hand what it's like to be stuck in a body that is constantly tired, slow, and clumsy. A Mom's job is to protect her child—not give him a permanent disability.

My husband Jason and I considered adoption. Adoption gave us the opportunity to have a child without playing CMT roulette. It also meant missing out on experiencing pregnancy and seeing Jason's pretty blue eyes or my "Cabbage Patch Kid" nose on a smaller person.

We also had a conversation about being "childless by choice," a chat that lasted less than 5 minutes. We wanted to be dirty nose wipers, school bus good-bye wavers, and long division explainers.

Parenthood was our ambition.

When I was twenty eight, I knew exactly what I was going to do with my life. I was going to take our newborn son home, marinate him in love, and show him that CMT is a speed bump, not a stop sign. I had arrived. I was a Mom. ✻

GIFTS WERE MADE TO THE CMTA

IN MEMORY OF:

Sammy W. Abernathy
Mr. and Mrs. Robert Haynie

Shirley Amador
Ms. Sandra Briguori
Spa Lady Group

Debbie Baxter
Mr. Doug E. Kahle

Ronald Beyer
Mr. and Mrs. Laurence Beyer

John W. Brown
Ms. Marie T. Brier

Myrtle Browne
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Sabra Drohan
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Ms. Virginia Stahle

Malvina Labeskis
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Mrs. Gracia Kather

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Mr. Steven C. Sygerych

Jack Walfish
Mr. and Mrs. Jerry Walfish
Shirley Wegener
Mr. and Mrs. Michael J. Coslo

Darlene Weston
Mr. and Mrs. Jeffrey Mattox
Ms. Vicki Pollyea
Mrs. Theresa Risch
Mrs. Jean Meyers Ryan

IN HONOR OF:

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Mr. Jason Angell
Ms. Susan L. Bloom
Mrs. Laura Hoffman
Mr. Jack D. Little

Dudley C. Bowlby
Mrs. Dawn M. Orr

Wil Brady
Mrs. Sarah Smith-Brady

Esther Bunis
Mr. Michael S. Weiner

Betty Darrenkamp
Ms. Jenna Darrenkamp

Stephen Gasper
Mr. Stephen Dembitzer

Esther Haber-Kranitz
Happy 90th Birthday!
Ms. Ruth Cohan
Dr. and Mrs. Gary Cohen

Elizabeth Heiles
Ms. Karen B. McCormick

Robert and Pam Kleinman
Mr. and Mrs. Raymond Greenberger

Zachary Korowitz
The Stein Family

Robert Kramer
Mr. Andrew R. Kramer

Julie Leonard
Happy 87th Birthday!
Mrs. Shirley Weiner

Dan Martens
Mr. William Martens

Elizabeth Ouellette
Mr. and Mrs. Webb McKinney

Phyllis Sanders
Mr. and Mrs. Howard Maier
Mr. and Mrs. C. Glenn Schor

Gary Shepherd
Ms. Ruth A. Loyd

Judy Sieg
Mr. and Mrs. Bruce Burke

David Tannenbaum
Ms. Amy Wachtel

Jerry Walfish
Ms. Leslie S. Walfish

Lisa Weiner
Ms. Mary Elliott

Eugene Zacharias
Baltimore Heritage Quilters Guild

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: ______________________________
____

Rolling Through Airport Security

My First Time Wearing AFO Bracing

BY JORI REIJONEN

Shortly before my trip, I had an unusual dream. I was trying to get ready to leave for the airport, and my husband was trying to hurry me.

When I went to put on my Ankle Foot Orthosis (AFOs; custom-made orthotics for my feet with braces that go up to my knees), I found that instead of having easily fastened Velcro straps, they fastened with chains and a lock, looking much like handcuffs. After some fumbling, I finally got them on and tried to stand up from the floor. My shoes, now roller skates, slid underneath me and I could not get my balance.

"Well, this would help me get across the airport faster," I thought in my dream, "if I could just figure out how to stand up in them. And what am I going to do at the security checkpoint?"

When I woke from this memorable dream, I knew immediately why I had it. This would be my first time flying since I was diagnosed with Charcot-Marie-Tooth Disorder (CMT), my first time flying since I had begun wearing the AFOs. I had been wondering what to do at the security checkpoint at the airport when it was time to take off my shoes.

Should I take off my shoes and AFOs? My balance has been affected, and I did not know whether I could take off the AFOs or put them back on again standing up without the aid of a chair. And how would I be able to stand, wait, and walk through the line without my shoes and AFOs? I have significant foot problems, including, as the physician so kindly put it, "visible deformities," so standing or walking barefoot quickly becomes a problem.

I also considered simply not wearing my AFOs to make taking off my shoes easier. This might appear to make sense, but I would be doing a lot of walking and standing during my travels, through airports and then through the hotel. I really needed to wear the AFOs to avoid fatigue, numbness, weakness, and tripping over my own feet (called "foot drop" by my physician: until diagnosis with CMT, I always thought I was just clumsy). While I was looking forward to my trip, anxiety regarding the security screening process had worked its way into my dreams.

Some research helped to reduce my fears. Through an online search, I found out that the Transportation Security Administration (TSA), the agency responsible for airport security screening in U.S. airports, has anticipated the needs of travelers with mobility issues. The TSA also has provisions for many other types of disabilities and health issues. The TSA website contains detailed information for special needs travelers at http://www.tsa.gov/travelers/airtravel/specialneeds/index.shtm.

I would not be required to remove my AFOs and stand and walk through security without them. Instead, I would be hand-screened by a same-sex screener upon my request. If I wished to be privately screened, that could be arranged.

My trip through the security checkpoint went surprisingly smoothly. I let the TSA staff know that I wear orthotic braces that help me to walk and cannot easily be removed. When it came to my turn, a TSA security officer led me to a small open booth next to the regular screening lane. The female security officer was friendly and respectful.

"I'm going to be giving you a little back rub now," she said as she patted me down.

"A little to the left," I replied.

The security officer also used a small handheld device to check me for explosive residue. She then told me that we were finished. Altogether, it took about 5 minutes—a little longer than walking through the metal detector.

The next time that I fly, I won't have to worry about how to get through airport security. And, hopefully, before I travel, I'll dream about sandy beaches and relaxation, rather than AFOs on roller skates. ✻

SUPPORT AND ACTION GROUP NEWS

WRITE TO US!

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California – South Bay Area
The group met on June 6th and learned about the STAR research project. Members also discussed CMT issues while enjoying a luncheon. In addition to solving problems, the group discussed easy fundraising activities. Elizabeth will make jars that can be placed in local stores and eateries, someone else is holding a garage sale to benefit the CMTA. The annual picnic will be held on September 15th at Elizabeth Ouellette's home in Los Altos. The following meeting in November will feature Dr. Steven Scherer from the University of Pennsylvania. Dr. Scherer has more than 150 CMT patients in his care and has authored more than 90 research papers.

Michigan – Chesaning Area
The group had its third meeting in June. It was a small gathering, but one new member was welcomed. The group has about 14 members and several are children, a 9-year-old boy who was just diagnosed, and two girls, 9 and 14, who are cousins. At the June meeting, a speaker came from True Touch Theapeutic and discussed how to help the body by eating right and exercising. The next meeting will be on August 8th and will be a pot luck lunch in the park.

New Jersey – Central Area
The group met on June 6th with 15 people in attendance. The meeting was a time to share "My CMT Story" and one first-time attendee who was a bit overwhelmed, said "I have never been in a room with anyone else that has CMT before." The leader, Mark Willis, shared information from the Las Vegas conference and news from the monthly facilitator conference calls with Dave Hall, CEO. The group discussed doing a fundraiser to coincide with CMT awareness week in September.

New Mexico – Albuquerque
The group met on June 12th with a small attendance of five. The meeting was successful as the members discussed fundraising and noted the success of the Solvang, California, wine tastings. There are a number of wineries in New Mexico, so the group may pursue that same concept. Some attendees also expressed an interest in doing a Circle of Friends fundraiser. The group raised the issue of videotaping some support group meetings that might be of interest to all groups, such as ones featuring noted researchers who would not be available to travel to remote locations.

Oregon – Portland
The Portland group met on June 8th with ten people in attendance. Since no speaker was scheduled, everyone had a chance to share their "CMT story" with the group. It turned out to be a great meeting. The next meeting is scheduled for August 17th, where we plan to discuss fundraising ideas.

Pennsylvania – Pittsburgh
The group met on June 5th to hear orthopedic surgeon Dr. Steven Conti. He brought his associates Michael Rees and Frank Erdeljac with him. The eleven support group members saw Dr. Conti's slide show presentation on how foot and ankle problems can be solved through surgery. His associates discussed how braces and other orthotics can be used to help people avoid surgery. The group will take a summer break and be back in the fall. ✻

IN MEMORY:
DARLENE WESTON

Darlene Weston led a CMT support group in Portland, Oregon, for over a decade. She passed away on June 6, 2010.

Darlene's husband Dean, as well as three of their adult children, had CMT, but it was Darlene who was the voice of CMT support for many people. In addition to the support group, Darlene facilitated a chat list on the Internet. She had a unique ability to share a wealth of information about CMT, practical advice on nearly everything, and a shoulder for support. She was very empathetic and was extremely gracious, never wanting to cause anyone to worry. She had the unique talent for turning negative feelings into positive results.

After the loss of her husband and her own failing health, she remained a positive, loving voice. She always sought to take care of others. She is survived by her loving children, grandchildren and great-grandchildren and by her many world-wide friends with CMT.

—Vicki Pollyea,
Tampa Bay Area Support Group Facilitator

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AUSTRALIA

(continued from page 1)

genetic counselor from Wayne State, reported on the distribution of genetic subtypes of CMT, by analyzing the results of DNA tests ordered by Wayne State University. The overwhelming number of positive CMT results were for the types 1A, 1X, HNPP, 1B, and 2A. Another study conducted in Belgium looked at the hereditary neuropathies with early childhood onset and found that most patients displayed either early neonatal hypotonia and breathing difficulties or a phenotype consisting of early foot deformities and delay in motor milestones.

> Across a person's life, quality of life issues do not change much because patients adjust their expectations as their condition worsens.

A presentation by Shawna Feeley, a genetic counselor from Wayne State University, revealed some interesting data about the most common form of type 2 CMT, which is 2A. CMT2A is caused by mutations in the mitofusin 2 gene (MFN2) and represents about 25% of all type 2 axonal neuropathies. Some cases of CMT2A are quite severe, and it is believed that the severity depends on the specific causal MFN2 mutation. When the patients have GTPase domain or the R3 domain, the mutations which disrupt those regions are particularly likely to cause severe neuropathy.

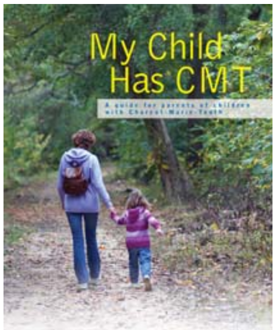
Running parallel to the inherited neuropathies' session was a series of meetings on the inflammatory neuropathies. In all, about 165 researchers and clinicians attended the three-day event.

Joshua Burns, Children's Hospital at Westmead, University of Sydney and Dr. Sindu Ramchandren, Wayne State University, Detroit, discussed the development and validity of the CMT pediatric scale. The pediatric scale measures symptoms such as foot and ankle involvement, hand dexterity, hand strength, foot strength, pinprick and vibration sensation, sensory symptoms, balance, and motor function. The scale was developed to complement the already existing CMTNS (CMT neuropathy scale). Dr. Ramchandren discussed the issue of quality of life in addition to the basic CMTPedS. She found that health-related quality of life in children with CMT is significantly reduced compared with population norms. The quality-of-life instrument will include such things as pain, cramps and fatigue, social function such as playing with peers, perceived cognitive function, emotional distress, and emotional bonding. It has been found that across a person's life, quality-of-life issues do not change much because patients adjust their expectations as their condition worsens.

One of the most fascinating talks of the sessions was given by Dr. Kiyoshi Hayasaka of the Yamagata University School of Medicine, Yamagata, Japan. He discussed the clinical and molecular diagnosis of CMT in Japan. Interestingly, he found that the PMP22 duplication is not so prevalent among Japanese CMT patients and that there are few de novo cases in Japan. He estimated that, contrary to the number that is commonly used (1/2500), the incidence of CMT in Japan is only 1/10,000. He attributes this to the fact that fewer CMT patients seek medical help and that fewer of them actually realize that they have a problem because the symptoms are less severe in the Japanese population.

In addition to the array of speakers, there were also poster presentations to look at and absorb. But the three days were not without their "fun" times as well. On Monday night, there was a reception held at the Sydney Opera House which featured Aboriginal men playing native instruments on the steps of the Opera. Following that musical entertainment, we walked up hundreds of steps to the glassed room where we enjoyed drinks and assorted finger foods that were brought around by employees of the Opera House. On the final evening, dinner was held at the University of Sydney in an old building which houses a spectacular antiquities exhibition. (Note: Neither of the events featured kangaroo meat.)

All in all, the trip was both mind-boggling and eye-popping. The science was deep and difficult, but the views of all the famous landmarks in Sydney, Australia, were truly amazing. ✻



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:

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

CMT PATIENT MEDICATION ALERT:

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

Moderate to significant risk:
Amiodarone (Cordarone)
Bortezomib (Velcade)
Cisplatin and Oxaliplatin
Colchicine (extended use)
Dapsone
Didanosine (ddI, Videx)
Dichloroacetate
Disulfiram (Antabuse)
Gold salts
Lefluonamide (Arava)
Metronidazole/Misonidazole (extended use)
Nitrofurantoin (Macrodantin, 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-Flouuracil
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.