

THE

CMTA Report

SPRING 2017

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SPRING FOOT ISSUE

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OUR MISSION: To support the development of drugs to treat CMT, to improve the quality of life for people with CMT and, ultimately, to find a cure.
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The Ins and Outs of Foot Surgery

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THE **CMTA** Report

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NEW CEO TAKES HELM OF THE CMTA

The CMTA Board of Directors announced May 15 the appointment of Amy J. Gray as the new chief executive officer of the Charcot-Marie-Tooth Association.

“We are delighted to bring Amy on board as our new CEO,” said CMTA Board Chair Gilles Bouchard. “With her extensive experience, proven leadership and passion for our mission, Amy will help us accelerate the momentum in our fight against CMT.”

Gray told *The CMTA Report*: “I am honored to have been selected by the board as the next CEO of the CMTA. I firmly believe there has never been a more exciting time in the organization’s history than now. The CMTA has seen tremendous progress the last five years and I look forward to building on this success in the years to come. My most important priority will be to aggressively pursue potential treatments, and ultimately a cure for CMT, with the Strategy to Accelerate Research (STAR). I firmly believe the CMTA is on the brink of advancing game-changing breakthroughs for the entire CMT community and I intend to tirelessly dedicate myself to this effort every day with the highest sense of urgency.”

Gray brings to her new position more than 17 years of management experience in national voluntary health agencies, combined with a strong understanding of mission-driven



organizations. Her experiences working for three major non-profit health care organizations have shaped her expertise in fundraising, staff development, financial and operational management, strategic planning and community engagement.

Gray was most recently the senior vice president of constituent and community relations for the National Parkinson Foundation (NPF), responsible for providing the overall leadership, strategy and vision for the foundation’s fundraising efforts, planned giving legacy society and network of chapters. She joined the NPF in 2013 in the role of vice president of chapter relations and community partnerships, providing guidance to its nationwide network of affiliates in organizational, mission, board and fund development activities. In 2014, she led the foundation and its affiliate chapters through a process of unification, integrating the chapters and foundation into one organization.

Before joining the NPF, Gray served in executive roles at the Crohn’s and Colitis Foundation of America and the Muscular Dystrophy Association. In these roles, she successfully built strong chapters and robust relationships with donors, volunteer leaders and key stakeholders.

Gray is a graduate of St. Cloud State University and lives in South Florida with her husband and children.

CMTA/MDA Co-Fund Grant to Study Gene Therapy in CMT1X

The Muscular Dystrophy Association (MDA) and the Charcot-Marie-Tooth Association (CMTA) in March announced a research grant totaling \$119,999 to Kleopas Kleopa, MD, for a study on the effectiveness of a gene therapy approach in CMT1X. In an earlier study, Kleopa and his colleagues showed that a single injection of the gene that is mutated in CMT1X was associated with production of normal protein in nerves and improvement of peripheral nerve health and motor performance.

The MDA and the CMTA are co-funding the grant under a partnership formed in 2016 that

aims to advance CMT research, therapy development and clinical care, and increase understanding about the disease by improving education for kids and adults affected by CMT, medical professionals and the public.

CMTA Board Chair Gilles Bouchard said, “Partnerships are at the core of the CMTA’s STAR (Strategy to Accelerate Research) approach to finding treatments for various types of CMT, so we are very pleased to announce the first research project jointly funded with the MDA.”

“MDA is pleased to collaborate with the CMTA to fund this exciting research,” MDA Scien-

tific Program Officer Amanda Haidet-Phillips said. “Working together allows us to have a greater impact as we pursue our common goals to help individuals and families with CMT.”

Kleopa is a professor and senior consulting neurologist at the Cyprus Institute of Neurology and Genetics, Cyprus School of Molecular Medicine, in Nicosia, Cyprus. The two-year grant, which became effective February 1, 2017, will fund research on whether gene therapy treatment after disease onset leads to functional improvements in CMT1X.

Kleopa’s new work will

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CMTA-Funded Study on 3D Printing Technique for Comparing CMT Heel Surgeries Named “Game Changer” at AAOS Meeting

A pioneering new surgical technique using 3D prints to compare different surgical approaches in the same patient was named one of six “Game Changers” of the year by the American Academy of Orthopaedic Surgeons (AAOS) on March 17. The technique was developed by Dr. Glenn B. Pfeffer, director of the Foot and Ankle Surgery Program at Cedars-Sinai Medical Center, with funding from the Charcot-Marie-Tooth Association.

“Ultimately our findings offer hope for better techniques to help patients with Charcot-Marie-Tooth disease have a better quality of life,” Dr. Pfeffer said.

The abstract for the study was one of six selected from more than 900 for the “Game Changers” session at the Academy’s annual meeting. The designation goes to techniques most likely to change the practice in the next three years. The study, “The Use of 3D Prints to Compare the Efficacy of Three Different Calcaneal Osteotomies for the Correction of Heel Varus,” was written by Dr. Pfeffer and Cedars-Sinai investigators Max P. Michalski, MD, Tina Basak, MD, and Joseph Giaconi, MD. Their



One of 18 identical models of a malaligned foot produced using a 3-D printer

technique has application for other types of deformity correction research in orthopaedics.

The study focused on the deformity of the heel bone, which is frequently twisted inward in patients with Charcot-Marie-Tooth disease, causing them to have an extremely unstable gait. Researchers began with a CAT scan of a patient who was unable to walk even a few feet without assistance because of foot deformity caused by CMT. They then used a 3D copier to print 18 identical models of the malaligned foot so they could compare the correction obtained by three different operations. A specialized jig accurate to within 1/10,000th of an inch enabled highly precise and reproducible cuts in the models, which replicated the procedures done in the operating room. After the bone cuts, a repeat CAT scan was performed on each model to measure the correction.

The researchers concluded that none of the uniplanar bone cuts provided adequate correction of the complex three-dimensional heel deformity. The “Z” bone cut

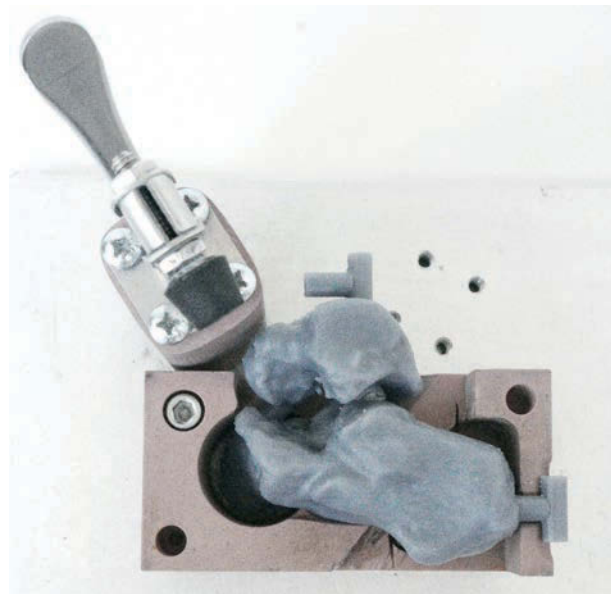
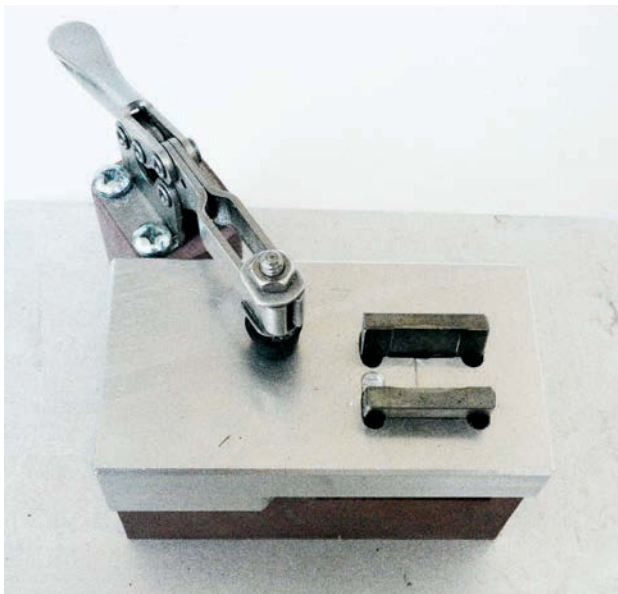
Study authors are currently in the second phase of the research to see what type of reconstructive procedure is best.

provided significantly more correction in the coronal plane (varus/valgus), with no significant shortening of the calcaneus compared to the “Dwyer” and “Oblique” cuts. The Z cut however, produced much less correction than anticipated, with only 3 degrees of final heel valgus. None of the osteotomies provided more than 6 mm lateral translation of the tuberosity. The study authors are currently in the second phase of the research to see what type of reconstructive procedure is best. That research should be completed in six months.

CMT kills the long nerves to the hands and feet, causing the muscles around them to atrophy. Cavovarus foot deformity is caused by an imbalance in opposing muscles, often resulting in a foot that has a high arch (cavus), clawed toes, a heel that turns inward (varus), and a foot drop. ★



Glenn B. Pfeffer, MD, is the co-director of the CMTA Center of Excellence at Cedars-Sinai Medical Center and co-director of the Cedars-Sinai/USC Gloria Kaufman Dance Medicine Center. He is also a member of the CMTA Advisory Board.



The micro-sagittal saw used to make “bone” cuts, enabling researchers to compare different surgical alternatives

Falling in Love with My AFOs

BY LAUNA BARNETT

“Do you trip or fall a lot?”

asked the neurologist.

“Not a lot,” I replied.

“Do you spend a lot of time and energy making sure you don’t trip or fall?”

“Yes,” I sighed.

Ugh, yes ... yes, I do. I’m always looking down, taking note of the terrain, constantly scanning for pinecones, gravel, sticks or anything else that might lead to a rolled ankle or cause me to catch my toes and trip. I avoid walking on the beach (darn you, soft sand).

I cringe when I see that I must tread across bark chips. I cross to the other side of a street so that the sidewalk slants in a direction that supports my soft ankle. I make sure my daughter, husband or a trusted friend is nearby when I’m walking on uneven surfaces so that I have a hand to hold if needed. These precautions have just become my reality. I can’t quite pinpoint when it got to this point, but here I am.

“I need braces,” I told myself at this doctor’s visit in August 2016. “I know it. That’s why I’m at this appointment. But I don’t want them. I really don’t want them.”

I’ve always known I had CMT and have accessed the medical community for support very sparingly over the years—but that’s a story for another day. After having inserts made about 15 years ago in my early 20s, I had managed my life with CMT relatively smoothly. The progression of my CMT has been slow and my muscles have weakened over the years without my really taking note. I couldn’t really see what I was missing out on and avoiding until the doctor asked me that single question.

The doctor encouraged me to get AFOs. He answered my questions about how they would affect my life. He told me that I would have more endurance and less fatigue, and that I would probably love them. My prescription for bilateral ankle-foot orthoses sat on my desk for at least a month before I called and made the appointment for an initial consultation. I was worried and fearful about the next stage. Needing braces seemed to mark a stage of my disease that I wasn’t ready for. It felt like the beginning of the end of my independence. It felt quite similar to grief—I was griev-



Author Launa Barnett with Ota and her beloved AFOs

ing the loss of an independent life. I spent hours contemplating how braces could impact my work. I was angry. I was scared and anxious. I was worried.

I had my initial consultation and fitting appointment in mid-October. I entered the office with apprehension and anxiety about what was to come.

Fortunately I was matched with an orthotist who could handle my honesty and directness. I explained my resistance to the process and hesitancy about how braces might negatively impact my ability to do my job as a speech pathologist working with young children, which often required me to get on the floor with them—and get back up. I stressed the importance of functionality. We settled on a plan—customized Arizona AFOs.

It took a while to get the braces fabricated and they weren't ready to be used until early December. I wore them inconsistently throughout the first month because they were uncomfortable. I experienced a fair amount of rubbing on my left ankle and took them back in for another troubleshooting session. They were sent back to Arizona for another adjustment. I finally had them back and ready to wear in early February. I started wearing them to work most of the time. I found that I was still having a little discomfort, but I was seeing the benefits. But I still wasn't in love with them ... yet.

I can pinpoint the exact moment when I fell in love with my AFOs. It was March 5, 2017, and my husband and I were walking our dog with a friend. We were at 1000 Acre Dog Park near Portland, Oregon (a place I had

previously avoided). I found myself walking “alone.” The dogs were running ahead and the guys were lagging a bit behind. I looked

to my right and gazed at the river; I looked to my left and took note of the trees. I inhaled that wonderful forest scent. I thought about the 3.5 miles we had covered—the

sand, mud, gravel, sticks and other unstable ground I had walked on. Bam! I was overcome with an unexpected moment of strong emotion. My heart fluttered and my eyes filled with tears. For the first time in years, I was out in nature and didn't need a hand to hold. I wasn't staring so intently at the ground worrying about trip-

For the first time in years, I was out in nature and didn't need a hand to hold.

ping or rolling my ankle that I missed the beauty all around me. I wasn't concerned that my legs would fatigue to the point that they wouldn't cooperate. I wasn't spending all of my energy making sure I didn't fall. The realization was extraordinarily powerful.

All along I was worried that this chapter of my life, the one with AFOs, marked the end of my independence, but I was so wrong. It marked a renewal of my independence, something I hadn't even realized was slowly slipping away.

When my husband caught up with me, he asked me a question, one that's his way of checking in to see if I need a break or a hand. “You good?” he asked. I'm sure I was beaming when I answered, “Yeah, I'm good.” And I meant it. I'm SO good! ★

CMTA CENTERS OF EXCELLENCE (www.cmtausa.org/coe)

CMTA Centers of Excellence are patient-centric, multidisciplinary CMT clinics where children, adults and families affected by CMT can be assured of receiving comprehensive care by a team of CMT experts. The Centers roughly correspond to the 21 international sites that make up the NIH Inherited Neuropathies Consortium (INC)—a group of academic medical centers, patient support organizations and clinical research resources sponsored in part by the CMTA. The centers will become even more important as the CMTA begins clinical trials, which will depend on how much we know about the “natural history” of CMT—how different types of CMT progress over time and whether novel medications are slowing the course of the disease. Much of that information will be supplied by the Centers of Excellence.

CMTA CENTER OF EXCELLENCE

Cedars-Sinai Medical Center (Los Angeles)	Drs. Robert Baloh and Richard Lewis
Children's Hospital of Philadelphia (Philadelphia)	Dr. Sabrina Yum
Connecticut Children's Medical Center (Farmington)	Dr. Gyula Acsadi
Massachusetts General Hospital (Boston)	Drs. William David, Florian Eichler, Vera Fridman & David Chad
Johns Hopkins University (Baltimore)	Dr. Thomas Lloyd
Lucile Packard Children's Hospital at Stanford (Palo Alto)	Dr. John Day
Nemours Children's Hospital (Orlando)	Dr. Richard Finkel
Stanford Neuroscience Health Center (Palo Alto)	Dr. John Day
University of Iowa (Iowa City)	Drs. Michael Shy, Laurie Guttman and Nivedita Jerath
University of Miami (Miami)	Dr. Mario Saporta
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University of Minnesota (Maple Grove)	Dr. David Walk
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University of Rochester (Rochester, NY)	Dr. David Herrmann
University of Texas Southwestern (Dallas)*	Drs. Susan Iannaccone and Diana Castro
University of Utah (Salt Lake City)	Dr. Nicholas Johnson
University of Washington (Seattle)	Drs. Michael Weiss and Leo Wang
Vanderbilt University (Nashville)	Dr. Jun Li

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INTERNATIONAL

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

MAGIC FEET:

The Ins and Outs of Foot Surgery

BY BRANDON LEDERER, DAVIDA HAWKES, ANNROJ FERNANDEZ AND KAREN BROWN

Six to eight months ago, the authors—two women and two men ranging in age from 34 to 48—each had a foot osteotomy/tendon transfer surgery. Author Karen Brown created Karen’s Foot Group before her surgery to share and document the experience. She also posted in the CMTA Emotional Support Group and the CMTAthletes group. The authors started out as strangers, but as they posted and texted each other throughout their long recoveries, they became what Karen calls “CMT friends.” They share their collective experiences with the aim of helping others in the CMT community who may be considering foot surgery.

“THINK OF THE MAGIC OF THE FOOT, comparatively small, upon which your whole weight rests. It’s a miracle ...”

—Martha Graham

The choice to have foot surgery is a big one. The recovery time is six to 12 months, which is a lot to ask of ourselves, our families and our employers. We each ultimately decided that trying to reset the arc of our individual CMT’s progression was worth the time, energy and expense.

We recommend getting at least two opinions before deciding on surgery. Discuss the pros/cons of every option with every doctor you’re considering for the surgery, including why the doctor recommends one approach over another, what will be done, and how confident s/he is about a positive outcome. Also ask about in-home physical therapy (with the cast on) during your initial recovery, which will help maintain some strength during this time.



Annroj, 36, lives in California. He doesn’t know his type and had surgery in September 2016.



Davida, 48, lives in Cardiff, Wales, has CMT1A and had surgery in August 2016.

Figure out the mobility solutions that will work for you before surgery, when you have two good feet. Can you hop on one leg? If not, then you will need a wheelchair or a knee scooter. Be cautious about using crutches. With CMTers’ impaired balance and weakened hands/arms, it is perilously easy to topple and end up slamming that post-op foot down to keep from falling. Hitting your cast against cement is very painful and sends a shock all the way to your brain.

In the United States, equipment must be authorized by your

insurance company or purchased independently; in the United Kingdom, necessary equipment is loaned to you while needed, although you may be given only crutches unless you push for a walker/frame instead. A 4-post solid walker/frame is an option (use RUBBER cane tips, not standard plastic walker feet), but is difficult in tight areas. Walkers with two wheels and two feet worked well for us. Some of us used a wheelchair for a while (insurance often covers the rental). Measure your doorways before you get your wheelchair, and



Brandon, 35, lives in Nebraska, has CMT1X and had surgery in August 2016.



Karen, 45, lives in Arizona. She has CMTA1A and had surgery in October 2016.

remove your doors if necessary to gain about two extra inches of clearance. Knee scooters are another option, but are difficult in tight spaces and are generally not covered by insurance. If you are covered for only ONE device (walker/crutches/wheelchair), use your insurance for the most costly aid and buy or borrow any cheaper one(s) yourself. Good places to look for loans include churches, community centers and senior centers. Craigslist is a good place to look for low-cost options.

The surgery typically lasts a few hours. In our collective experience, outpatient surgery took about eight hours from check-in to discharge, and hospital stays were two to three days. After the surgery, have someone with you 24/7 for at least the first three days, and ideally for the entire first week. In addition to needing help so that you can stay in bed with your foot up as much as possible, you can quickly go septic from an infection, which can cause severe confusion for someone on pain medication. After the first week, you can be alone during the day but you will still need significant help.

Pain management in the first two weeks is about rest, elevation, ice and pills. Have your caregiver place everything you might need within arm's reach of your bed. The only reason you should get up is to use the bathroom. Your foot **MUST** be elevated above your heart and iced (20 minutes on/20 minutes off) as much as possible to keep the swelling down. Rest it on pillows and/or a wedge (cover with a towel; bleeding through your splint is likely). Be mindful that when using a wheelchair or knee scooter, your foot is well below your heart. The more it's down,

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PRE-SURGERY PLANNING

This list is not exhaustive, but the following items are essential to consider:

- Does your surgeon/hospital/surgery center/radiologist/anesthesiologist/etc. take your insurance?** Don't assume because your surgeon does that everyone else does as well. Find out who your team will be, and contact all of them before the surgery to discuss coverage.
- What medical equipment will your insurance cover?**
- Line up your disability paperwork** if you are taking time off work for this. Six to eight weeks of recovery time is not uncommon; doctors will generally write what you ask for. It is harder to go back a second time for more, so ask for a little more than what you think you'll need. You don't have to use it all, but if you need it, it's there.
- In the UK, get a sick note for three months**, and extend it if you need to, depending on how much leg work your job entails and how much paid time off you have.
- Talk to your employer about time off and modifications you may need** once back to work: closer parking, shorter hours for the first few weeks, changing duties so you are not on your feet for long periods of time, telecommuting, etc.
- If you plan to use crutches or a walker/frame, practice with them in advance.** You do not want to lose your balance and come down on that foot. Remember that you will also be fatigued and medicated in the weeks after the surgery. Are these aids realistic for you?
- If you choose a knee scooter, you can buy one online for a lot less** than you can rent one from medical supply, but the cushion quickly gets uncomfortable on the knee. Trim down a garden kneeling mat as an extra cushion.
- Stock your freezer with as many meals as you can manage to cook or buy in advance.** Choose healthier meal options, to avoid post-op weight gain during your months of reduced activity.
- Talk to friends, family, and neighbors to line up your support team** for school runs, grocery shopping, drug store runs, bringing meals, light house-keeping, taking you to doctor appointments, etc.
- If you have children at home, let them know what to expect.** Keep it light and results-focused with younger children (I'll be slow and need your help for a while, but after I'm better, I'll be able to do so much more with you!) so they don't feel afraid.
- If your house is more than one story, rearrange your space for first-floor living.** Practice getting into your house (and into bed) with one leg never touching the ground.
- If you have cats or dogs, think about how to keep them away from your foot** those first two weeks while you are in a soft cast.
- Driving: If your surgery is on your driving foot, you won't be able to drive** until you are cleared by your physician. If it's your other foot and your car is automatic, you may be able to drive in a month. Remember, though, that your feet are way below your heart while driving and keep the trips short at first.
- If you intend to drive before you can bear weight on your new foot** (after you put your wheelchair/ scooter in the back of your vehicle), how will you get in the driver's seat? Practice this in advance.
- What pain meds will you be given and what do you need to expect in terms of side effects or withdrawal symptoms?** Percocet, for instance, causes constipation when you're on it and flu-like symptoms when you go off it. You might need some emotional support withdrawing, which can be as hard.

FOOT SURGERY

(continued from page 9)

the more it swells, and the more it hurts. This gradually improves but you'll have swelling for at least six months.

After about two weeks in splints, each of us moved to hard casts. The time spent in hard casts varied from four to 10 weeks, depending on the specific surgery and orders. Three of us had the foot cast at 90 degrees the whole time; one had a neutral position (~120 degrees) cast for the first three weeks, then 90 degrees for the next cast. When we were ready to bear weight on the new feet, we all progressed to walking boots for six to eight weeks and finally back to shoes, which had to be wide enough to accommodate our still-swollen feet. We used walkers or canes for stability.

Physical therapy (PT) outside the home starts when the hard cast comes off. Be aware that starting PT can be emotionally difficult, because your new foot hasn't learned how to work yet. The more consistently you do your home exercises, the faster your rehab will progress. Our PT orders varied quite a lot ... from about eight hours to 125 hours!

Expect it to take six months before your foot is functional and a year for full recovery. Training your new foot and rebuilding its strength are slow processes. At six months, we still experienced swelling, fatigue, aching, some pain, stiffness and limited range of motion.

Three of us gained weight during our recovery. The one who did not ate meals heavy in protein and vegetables with snacks

of fruit and no "empty calories."

Two of us have had unexpected complications from scar tissue: One has continued limited range of ankle motion; the other has two toes that now catch the ground more easily and sharp pain at the base of those toes. We all experienced secondary pain/discomfort in the joints above the reconstructed foot, as well as the "other" foot. It is common for knee and/or hip pain to increase after the surgery, but hopefully that is temporary. Physical therapy comes at the perfect time to help strengthen the knees and hips while rehabilitating the new foot. Some of us believe our unreconstructed foot worsened more quickly because it was our only foot for a time. Be sure to elevate and ice that foot as well whenever you're able.

My Allard ToeOFF
makes me...



FIERCE

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OH, THE EMOTIONS!

The emotional challenges are hard to anticipate and prepare for. These are some of the moments we felt emotionally overwhelmed and/or caught off guard:

The first time we saw our new feet.

You're likely to tear up or maybe full-on cry; just let it all out. It's HUGE. It's a mixture of gratitude and wonder (at having a foot that is straight for the first time in your life), concern (over all those cuts, scabs and staples) and maybe even some grief over no longer having that curved foot you didn't realize you'd become attached to seeing down there. It looks like somebody else's foot is on your leg. That takes some getting used to.

Missing moments with our families and friends. Special occasions, school events, date nights and playing Legos on the floor. It sometimes feels like you're missing EVERYTHING that truly matters. It's normal to feel that way, and it's temporary, but that doesn't make it easy.

Feeling like you're falling short in every arena of your life. Your house is a mess, your hair is unwashed, your desk is an avalanche waiting to happen, you can't remember if you paid that bill or not, your child is lonely for you (again), you haven't had sex in too long and you're not yet where you thought you'd be in your recovery.

Wondering if it was worth it or if you put everyone through all of this for nothing.

Contemplating the effects of a degenerative disability. Wondering if the surgery will help, if the other foot will need it, if other major surgeries may be necessary and if one day no surgery will help. This one is huge.

HOWEVER... if you open yourself up—to your partner, your family, your friends and your com-

munity—and ask/allow them in to help you through this, you will also experience feeling loved on an entirely new level. People will show up, they will help you, feed you and your family, clean your home, run your errands, bring your child home from school or take him to the zoo when you need a break. If you let them, the people in your life will fill your heart with love and gratitude. This is a time for you to accept help and love from others.

WOULD YOU DO IT AGAIN?

Brandon: Absolutely 1000 percent yes. I was at, "Wow this is pretty good" about three days into the boot. After six months I walked on gravel and grass/uneven terrain, which used to be painful and scary. If I had known it could be this good, I wouldn't have waited so long. Every day continues to amaze me. It is still getting better and improving. Six months out I have little to no pain in my reconstructed foot, and my other foot has pain before the one that got the surgery does. This is incredible to me. I expected it would at least be a year before I reached this milestone.

David: Generally, I think yes, although I need to see where I get to with this one. I'm probably 80-90 percent sure it was the right thing. At eight months out, I've just lately begun to feel like I'm almost there, but I'm not "there" yet. My ankle is straight. It doesn't roll and my balance is improving too. The strength in my foot is so much better that I'm moving pretty well. I think I am now able to do things I couldn't do pre-op. I'm not able to run yet. I do a sort of skittle and the "bounce" is poor but I've started back doing some step aerobics and I feel great. I wouldn't do the second foot straight away; I'd need to wait one

USEFUL AIDS & EQUIPMENT

- **A walker/frame, wheelchair, and/or knee scooter**
- **Toilet safety bars**—these can be wall-mounted, toilet-mounted, or a freestanding frame that stands around the toilet. Do NOT use a sink as a support unless it is mounted in a full cabinet; it can rip off the wall if you lean on it.
- **A shower bench** or stool for bathing
- **Extra pillows/wedge** to elevate your foot
- **A cast cover**—not essential but it does relieve some of the concern about getting your cast wet in the shower.
- **A cooler** by the bed to hold ice packs



to two years to make sure I was truly happy with the first!!

Anroj: I do feel more stable when standing, but my range of motion is still poor; thus I can't fully say it was the right choice. My PT said I have so much scar tissue that it hinders range of motion as well. I think I have recovered 50 percent and I can drive now. I'm moving a little bit better but still no brisk walking and jumping, and it's still painful when I walk sometimes.

Karen: The hope for my surgery was that it would stabilize my foot (done), correct the deformity (done), prevent future deformity (only time will tell), enable me to pick my foot up myself (not there yet and may not get there) and leave my house without AFOs (unlikely). My new foot is amazing, and I may yet be able to pick it up myself. A month ago, I was very discouraged with my recovery, and chose to work harder to get where I want to be with this new foot. The progress is slower than I expected, but it is progress. I am happy that I took the leap of faith to do it. I plan to do my other foot in 2018. ★

STAR Explained: *Turning Science*



Editor's Note: CMTA Board Chairman Gilles Bouchard (pictured above) delivered the clearest and most concise explanation of STAR to date at the Miami Patient/Family Conference. We reproduce it here.

In 2008, the CMTA's Board of Directors launched its Strategy to Accelerate Research, or STAR. It was based on two important ideas:

IDEA #1: THE GENES THAT CAUSE CMT ARE KNOWN.

We know the causes of many types of CMT. The big breakthrough came in 1991 when PMP22, the gene that causes CMT1A, was discovered. Today 90 different genes that cause CMT have been identified and more are being discovered each year. Once we identify a gene that causes CMT, we can duplicate it in the laboratory. This is the foundation of the STAR strategy. As a famous inventor put it:

"A problem that is well stated is half resolved." This is true of CMT, unlike many other diseases whose causes are either unknown or very complex.

IDEA #2: MANAGE RESEARCH ACCORDING TO SOUND BUSINESS PRINCIPLES.

STAR IS BASED ON FIVE CORE BUSINESS PRINCIPLES:

- 1. Develop a strategy** based on knowing the cause of the disease and where to focus.
- 2. Find the best researchers** in the world and ask them to implement projects that support the strategy.
- 3. Create accountability**—Hold those researchers accountable for achieving their goals. We take your money very seriously. Our researchers are not fully paid until they fully deliver.

4. Demand collaboration—We bring the researchers out of their silos to collaborate with each other. We are now seeing more and more technologies and therapies emanating from many different fields of study.

5. Encourage partnerships—It costs between \$400 million and \$1 billion to bring a new drug to market. The CMTA does not have this kind of money. We have to work with big, strong pharmaceutical companies that have the money to develop drugs. In the end, they will carry the ball over the line for us.

OUR STRATEGY HAS FIVE KEY ELEMENTS:

1. Assays—Assays are tests. We recreate CMT in Petri dishes and then use high-throughput screening (HTS) to test hundreds of

Steps on the Path to Drug Discovery for CMT

Laboratory Models



For each type of CMT, develop a robust population with offspring that exhibit symptoms and can be used to test candidate pharmaceuticals.

Assay Development



Build cellular assays, reproducing cells with each type of CMT that can be used for the rapid testing of many different chemical compounds.

Drug Screening



Screen the cellular assays using the chemical libraries at the National Institutes of Health and major pharmaceutical companies.

Pre-Clinical



Test the candidate drug compounds by giving doses to the symptomatic laboratory models and measuring their improvement.

Pre-IND



Test the compounds for dosage, toxicity and viability in private and academic laboratories to determine the exact dosage that will improve the root cause of CMT.

Clinical Phase 1



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

Clinical Phase 2



Conduct pre-clinical phase 2 trials. Recruited through the CMT Centers of Excellence, small groups of people with type-specific CMT will participate in pharmaceutical trials to determine the effect the drug will have on CMT.

nce into Therapies

thousands of drugs to see if they affect it. We are looking for hits—drugs that correct the defect of a particular type of CMT.

2. Animal Models—We take the promising hits and test them on laboratory animals, narrowing the millions of potential compounds down to a few of the most promising.

3. Stem Cells—We take human skin samples and put them through a stem cell process to create neurons (nerve cells) and Schwann cells (which make myelin). This way, we create assays that better represent human biology. We have developed models for CMT1A and CMT Type 2.

4. Partners—We've tested millions of compounds to see if they affect CMT1A. With the help of a major pharmaceutical company, we've found several promising ones that need to be fine-tuned for humans. We've created a "toolbox" with the assays, animals and tests so that companies with new therapies for CMT can work with us to test them. This includes new technologies from other domains. We can get solutions from the entire medical field. For example, four different drug companies that work on many different diseases recently reached out to the CMTA to discuss potential therapies.

5. Clinical Trials—We are working to get ready to conduct clinical trials, including developing outcome measures, which measure a drug's effectiveness against CMT.

HOW DO WE WORK?

We created an advisory board with top-notch researchers. The Scientific Advisory Board has 14 world-class scientists. Because the work of STAR involves translational research—turning science into therapies—we created a Therapy Expert Board (TEB)—a group of experts who tell us how good the science is in terms of turning it into therapies for those with CMT. More recently, we realized we had to get ready for clinical trials so we created the Clinical Expert Board (CEB), a group of experts who are helping us and our partners think about how to design clinical trials.

CLINICAL TRIALS—HOW YOU CAN HELP

Everyone with CMT has an important role to play. There are currently 22 Centers of Excellence in the United States and abroad (see page 7). Clinicians need as much data on as many patients as possible to help drug companies conduct successful trials. You can help by joining our Patient Registry. We are also developing "outcome measures" to see if drugs are effective as soon as possible so that we can keep the trials short and inexpensive. The traditional CMT test scores require too much time to show if a drug is working or not, so we are looking at various "biomarkers" such as fat content in calf muscles or certain chemicals in the blood. ★

TO PARTICIPATE IN CMT RESEARCH STUDIES, please join the Patient Registry (www.rarediseasesnetwork.org/cms/inc/registry). You do NOT need to know the exact type of CMT you have to join this registry. And everyone in the world who has CMT can join.

CMTA RESEARCH UPDATES BY DISEASE:

CMT1A Over the past eight years, we've done animal studies, performed HTS, identified hits and worked with a company called Genzyme. Today, we're focused on two families of compounds that are being fine-tuned in the lab. Genzyme, a traditional pharmaceutical company, takes a small molecule approach, which utilizes chemical formulas. In parallel, we are also working with biotech companies, which create biological living proteins, or large molecules. For example, a company came to us with a very interesting approach—RNA interference—which inserts little pieces of DNA into nerves to affect the way the cells create the protein that overexpresses PMP22. We've seen promising results in rat testing. This technology is currently used in two approved drugs on the market.



CMT1X is the second most common type of CMT. Researchers have identified a relationship between CMT1X and inflammation. We've identified the source of this inflammation and we are going after therapies to target this source. The approach comes from cancer research. Another approach is gene therapy, meaning that a virus sends DNA to the nerves that replaces the defective gene. We are also investigating gene therapy for CMT4.



CMT1B We have good assays and mouse models. We've also had several hits and potential compounds. As in CMT1X, inflammation may play a role in CMT1B, so CMT1X research might help CMT1B.



CMT2A We've patented a rat model and have seen promising results using stem cells. We will also complete a small screening of FDA-approved drugs this year.



CMT2E We have stem cell assays and good animal models. Testing will commence soon.



Miami Patient/Family Conference Features Speakers from CMTA's Newest Center of Excellence

BY RACHEL RIVLIN

Speakers at the Miami Patient/Family Conference on March 18 delivered relevant, exciting and thought-provoking information to 145 people with CMT, their families and others who care about them.

Mario Saporta, MD, PhD, MBA, answered the genetic testing question asked by many in the CMT community: "Why bother?"

Saporta, director of the CMTA Center of Excellence at the University of Miami Miller School of Medicine, gave two reasons: One is to add to the CMTA's library of CMT genes and mutations and the second is to confirm a diagnosis and thus avoid unnecessary or harmful treatments and procedures.

Saporta said patients in his clinic also want to know about their estimated risks, estimated

prognosis and progression, how to modify medical management of other diseases, whether CMT will be passed to their children and their eligibility for clinical trials.

Stephan Züchner, MD, PhD, talked about gene discovery. Züchner is the chairman of the Dr. John T. Macdonald Foundation Department of Human Genetics at the University of Miami Miller School

of Medicine and a member of the CMTA's Scientific Expert Board. More than 90 percent of CMT patients receive a diagnosis of CMT1, he said, and while researchers have found 90 genes that cause CMT, fewer than half of patients with CMT2 know which gene caused their disease.

Researchers hope that gene discovery will guide the way to gene therapy, Züchner said, and because CMT is part of a larger group of genetic diseases that overlap, collaboration and inclusion of other rare diseases in research is important. Currently, gene discovery is at an all-time high and is proceeding at great speed, providing hope that a treatment will eventually be found, he added.

Züchner revealed that researchers found the gene that caused Julianna Snow's rare and aggressive form of CMT, which affected her breathing. Julianna, the 5-year-old girl whose choice of "heaven over hospital" triggered a debate about a child's right to choose, died in June 2016.

Dr. Michael Shy, head of the CMT Clinic at the University of Iowa and co-chair of the CMTA's newly formed Clinical Expert Board (CEB), wrapped up the conference. The CEB is tasked

with providing the natural history and clinical expertise to design, develop and enable clinical trials, collaborating with scientists in the development of clinical biomarkers and

ensuring the adequate recruitment of carefully evaluated patients and experienced investigators to conduct these trials.

Measuring the effectiveness of a potential therapeutic is a key part of clinical trials. Shy explained that one such measure is the CMT Disability Progression Score, which detects changes that occur over time in adults with CMT, but needs the participation of approximately 1,000 people to be reliable.

Another measure is the MRI biomarker test, which can pick up CMT before symptoms are evident, but measures only leg and calf muscles.

For those patients who want to participate in clinical trials, Dr. Shy stressed the importance of registering with the Rare Disease Clinical Research Network (RDCRN) (www.rarediseasesnetwork.org/cms/inc/registry). Patients seen at a CMTA Center of Excellence like the one in Miami are also encouraged to enroll. ★

Rachel is the Sarasota, FL Branch Leader.

Researchers hope that gene discovery will guide the way to gene therapy...



NEUROTOXIC MEDICATION LIST UPDATE

Dr. Louis Weimer of Columbia University recently updated the CMTA's neurotoxic medication list as part of his review for an upcoming issue of *Current Neurology and Neuroscience Reports*. The following medications have been added to the **moderate-to-significant risk** category:

Arsenic Trioxide (Trisenox)

Immune Checkpoint Inhibitors

Ipilimumab (Yervoy)
Nivolumab (Opdivo)
Pembrolizumab (Keytruda)

Immunomodulatory Drugs (IMiDs)

Lenalidomide (Revlimid)
Pomalidomide (Pomalyst)

Monoclonal Antibodies

Cetuximab (Erbix)
Brentuximab Vedotin (Adcetris)

Swimming with Dolphins, Finding New Friends

BY ERIN BLACK

Editor's note: During the Miami Patient/Family Conference, a lucky group of CMTeens had a day they'll never forget swimming with the dolphins at the Miami Seaquarium. A big-thank you to the wonderful folks at the Dralla Foundation, which funded the adventure.

Before March 18, I had never met anyone else like me. Meaning, I had never met anyone who wears big chunky braces, falls down and faces the other terrible and unique challenges of CMT. Swimming with the dolphins at the Miami Seaquarium was great, but meeting other people who share my disease was even better.

My adventure began in January. I was scrolling through my social media feed when I stumbled across a beautiful picture of palm trees posted by the Charcot-Marie-Tooth Association (CMTA). I was immediately interested, warm weather and the CMTA being two of my favorite things. The post was about a CMTA Patient/Family Conference in Miami. Reading through all the information, I soon realized how badly I wanted to be there. Unfortunately, I am only 15 years old with not much of a budget. My wonderful family, including my amazing Aunt Peggy, agreed to help me get there.

On the day of the Dolphin Odyssey, my family dropped me off in the hotel lobby, where I met a group of 15 kids and chaperones. We started off as strangers. The only thing I really knew about these kids was that they were around my age, lived in many different places and, most significantly, had CMT. We hopped (or should I say slowly helped each other) into a big van and headed toward the Miami Seaquarium. We were

warmly welcomed and ate lunch while learning about each other. The conversation flowed surprisingly well: When I mentioned physical therapy to the table, everyone moaned and groaned. That is when I realized, "These are my people."

After seeing several exhibits and a whale show, it was time to meet the dolphins. We changed into wetsuits for the swim. Taking off our braces and getting into the cold pool water was hard for almost all of us. That is the amazing part: I was not alone for once in my life. It was okay to struggle because everyone else was struggling too, so we just helped each other and had fun doing it. After we changed, we got in the water with the dolphins. Swimming



CMTeens at the Miami Seaquarium

with these crazy adorable, yet extremely intimidating, creatures was incredible. We kissed them, shook their flippers, splashed and played. Just being in the presence of the 400-pound dolphins was wonderful.

After spending time with the dolphins, we changed into dry



clothes and headed for a luau-themed beach party. On our way, we talked about the perks and struggles of living with CMT. One of the chaperones asked us each to talk about the best thing about having CMT. To be honest, I had never thought about it before. I said that overall, CMT makes me stronger, makes me the person I am and makes me realize how blessed I am to have such amazing friends and family. Finishing the night off at our luau was the perfect end to a perfect day. We drank mocktails (non-alcoholic cocktails) by the ocean and ate what I like to call "fancy kid food."

It is hard for me to put into words what the day meant. We all started as awkward acquaintances and ended the night as unique friends. I even met my online friend Julia for the first time. She wasn't the only friend I made.

In just 10 hours, we all became friends. To see and hear how CMT affects all these amazing kids made me feel grateful and appreciate what I have. The chaperones with CMT inspired me to get involved and reminded me that this disease should not and will not define me. ★

Erin, 15, lives in Cleveland, Ohio.

FUNDING A CURE

BAILEY'S BEAT THE BITE 5K RAISES MORE THAN \$10K FOR STAR

An impressive 76 walkers turned out for the first annual Bailey's Beat the Bite 5K Walk for CMT on March 25, in Tucson, Arizona. The walk was inspired by 7-year-old Bailey Roestenburg, a Tucson native who has CMT. Bailey was diagnosed with CMT when she was just 3 years old. The Oaxaca/Roestenburg family—including her mom Vanessa, aunts

and grandparents—decided to bring awareness about CMT to the Tucson community and to raise funds to help find a cure. The first-ever Bailey's Beat the Bite was a huge success, raising a grand total of \$10,623 for STAR, more than twice the \$5,000 goal. Many local businesses—including QMA Patios, Mobile Maintenance and Towing and Prestige Pool Plumbing—got involved by sponsoring the walk, while others, including Texas Roadhouse, Reid Park Zoo and Roadhouse Cinemas, donated gift cards/passes to their businesses.



Local businesses donated all materials, food, raffle items and prizes. The Dauphin County Anglers and Conservationists (DCAC) generously donated space along Clark's Creek at the E. J. Stackpole Memorial Trout Nursery and stocked it with several hundred trout. The DCAC is run entirely by volunteers and funded by donations. The site where the fundraiser was held is designated for children 12 and under and people with disabilities. It has a paved walkway that makes the clubhouse and surrounding area accessible for those with mobility issues.

Jordan Mermelstein, 12, Harrisburg's youngest branch member, set his own personal fundraising goal this year. "Jordan's Trout Hunters" raised more than \$2,000, making him the highest individual fundraiser of the group! **WAY TO GO JORDAN! ★**

FISHING FOR A FIX NETS MORE THAN \$5,000

The Harrisburg, PA CMTA Branch raised more than \$5,000 at its 2nd Annual Fishing for a Fix Youth Trout Derby on April 23. Twenty-seven kids ages 12 and under competed for prizes for the "first fish caught," "first palomino" and top three trout by length, while their parents bought tickets for seven different raffle baskets and a kayak.



SOUTH CAROLINA MIDDLE SCHOOL RAISES \$1,072 FOR CMT RESEARCH



HELLO, I'm Joseph Matthews from the Northwood Middle School in Taylors, South Carolina. I decided to raise Spirit Week money for CMT this school year because my younger brother, Jeffrey, has it and I watched him struggle with certain things as he was growing up.

So for Spirit Week, my fellow Student Council members and I created two games and sold water bottles to raise money. The first game was called "The Wishing Well" and involved dropping a little pebble from above down into a cup. The second game involved guessing how many pieces of candy two filled jars contained. The one who guessed the closest won the candy inside. We raised a total of \$1,072 and donated it to our local CMTA branch. They were so happy with what we accomplished and so proud to see that people truly do care about CMT.

The CMTA Gratefully Acknowledges Gifts...

IN MEMORY OF:

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Mr. Jason Angell

MR. AND MRS. MARTY BERGER - Happy 50th Anniversary!

Mr. and Mrs. Kenneth Handel

BRANDON BURCHARD

Mrs. Adriana Burchard

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Mr. Dillon Flatt

SHEILA DEANNUNTIS - Happy Birthday!

Mr. Michael Chonoles
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CHLOE LOVELLO

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DOROTHY MAMINSKI - Happy 100th Birthday!

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SUSAN RUEDIGER

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Ms. Christiane Palpant

ERIN SHELLEN

Chicago Ridge Elementary
Schools

JENN STENANDER

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TYLER SZADAJ

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Mr. Brian Edwards

LAURA TERRY

Ms. Maureen Carleton

SUSAN VAHER

Ms. Angela Calendine

ANN VAN DOREN

Fredericksburg City
Public Schools

HARRIET WEISS - Happy Birthday!

Wendy, Sue, Linda,
and Nurit

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, PO Box 105, Glenolden, PA 19036.

Honorary Gift:

In honor of (person you wish to honor)

Send acknowledgment to:

Name: _____

Address: _____

Occasion (if desired):

- Birthday
- Holiday
- Wedding
- Thank You
- Anniversary
- Other

Memorial Gift:

In memory of (name of deceased)

Send acknowledgment to:

Name: _____

Address: _____

Amount Enclosed: _____ Check Enclosed

VISA MasterCard American Express

Card # _____

Exp. Date _____

Signature _____

Gift Given By:

Name: _____

Address: _____



CMT CLINIC AT VANDERBILT MEDICAL CENTER: SERVING THE CMT COMMUNITY SINCE 2009

The CMT Clinic at Vanderbilt Medical Center, Nashville, Tennessee, is a designated CMTA Center of Excellence that serves patients mainly from Tennessee and the surrounding states of Alabama, Georgia, Kentucky, Arkansas and Mississippi. Clinic director Dr. Jun Li and the neuromuscular team have dedicated their practice to research and treatment of CMT since 2009.

To be seen at the clinic, patients must have their primary care physician or local neurologist make a referral to the Neuromuscular Division of the Neurology Department at Vanderbilt, specifying that it is for CMT.

In addition to a physical examination, clinical evaluations include review of medical records, lab results, EMG and nerve conduction studies, and follow-up with family members for a genetic history. The clinic provides consultation and referrals for physical therapy, occupational therapy, orthotics and other medical



THE CMT CLINIC AT VANDERBILT (pictured, left to right): Valerie Hanks, OTR/L, CPAM, C/NDT; Robin Yawn, RN, Neuromuscular Case Manager; Jun Li, MD, PhD, Professor of Neurology, Director of Vanderbilt CMT Clinic; and Penny Powers, PT, MS, ATP

services providers as needed.

On the research side, Dr. Li's laboratory is investigating a therapeutic approach for the mouse model of hereditary neuropathy with liability to pressure palsies (HNPP). They have discovered a novel genetic cause for CMT2P and produced an animal model they are using to explore the pathogenesis of this disease. In col-

laboration with Vanderbilt Imaging Center, the team is also studying how a variety of MRI techniques can be used to measure outcomes for clinical trials. The Vanderbilt CMT clinic has also participated in the Inherited Neuropathy Consortium's clinical studies, including a natural history study of CMT, a genetic modifier of CMT1A and discovery of novel genetic mutations for CMT.

Dr. Li and the clinic's physical therapists have also investigated the benefits of Kinesio® Taping as a therapeutic intervention. The Kinesio Taping® Method is a rehabilitative taping technique designed to facilitate the body's natural healing process while providing support and stability to muscles and joints.

For more information, contact Robin Yawn, RN Case Manager, at 615-936-0060 or fax information to 615-936-7147. Patients should send the clinic their medical records before the appointment and bring a copy with them to the appointment. ★

EACH ONE, TEACH ONE: SPREADING CMT AWARENESS

Bill Severn, a member of the Easton, MD Branch, teaches a high school biology course in Worcester County, Maryland, for students considering medical careers.

For a recent segment about genetically transmitted medical conditions, Bill assigned students to investigate a particular medical disease and then be interviewed by the entire class about the symptoms, problems and prognosis as if they had that disease. Because Bill had 15 students, but only 14 diseases in his lesson plan, he assigned CMT to his best student, then worked with her to investigate the condition.

Bill interviewed that student during a subsequent class. After the interview, another student said he had never heard of CMT, and didn't know anybody who had it. Bill replied, "Well, now you do, because I have it and that is why I have difficulty walking and standing."

The bottom line is that 15 young people, possible future medical professionals, now know something about CMT. And Bill is including CMT in his lesson plan for next year. Thank you, Bill, for doing your part to spread CMT awareness!



CMTA BRANCH NEWS

NAPLES, FL

Eight people attended the branch meeting on March 23, including two new members. Current members gave the new members tips for coping with CMT and recommendations for CMT-knowledgeable local clinics, neurologists and orthotists. Branch Leader Roy Behlke reported that the Naples branch raised \$491 for STAR research last year. He also raised \$275 this year at his Annual Glen Eagle Golf Club Art and Craft Show and encouraged members to sponsor their own fundraisers.

★

INDIANAPOLIS, IN

Five people watched a webinar titled “Brace Yourself” at the branch’s first meeting of the year on January 28. The webinar by CMTA Advisory Board Member David Misener, BS, CPO, MBA, can be found at www.cmtausa.org/webinars. In it, Misener talks about how AFOs can prevent or slow some of the symptoms of CMT. Misener shared that AFOs should be re-padded about every six months because the padding gets soft and loses its ability to protect. While most insurance policies only allow AFOs to be replaced every five years, they may need to be replaced more often because CMT is progressive. To make the case to the insurance company, the CMTer’s doctor should document body changes.

★

IOWA CITY, IA

Nine people turned out for the branch’s March 26 meeting. They discussed how often to schedule visits to the CMTA Center of Excellence at the University of Iowa, bracing options, the helpfulness of rollers, physical/occupational/aquatic therapy, the idea of having a collection of items like canes and exercise equipment for members to try, the importance of stretching, the pain experienced by people with CMT and how illness in general makes CMT worse. Post-meeting, the group discussed on Facebook the idea of impromptu get-togethers to walk local trails and the possibility of having board/card game nights.

★

BALTIMORE, MD

The group welcomed six new participants and two guest speakers to its April 9 meeting. Harrisburg, PA Branch Leader Erin Gaul talked about her CMT journey, and yoga instructor Cindy Prudhomme demonstrated a variety of chair yoga poses. The group also discussed the upcoming Oxford Biathlon, Daniel Fry’s heroic journey along the Appalachian Trail, PEOPLE magazine’s

coverage of CMT kids swimming with dolphins, and the CMTA’s partnership with the MDA on gene therapy for CMT1X.

★

ANN ARBOR, MI

More than 55 people attended the Michigan branch meeting on March 25. Fifteen drove more than 50 miles to get to the meeting, and one even traveled five hours. The agenda was as packed as the meeting. The branch welcomed guest speakers Dr. Sindhu Ramchandren, the head of the CMT Clinic at the University of Michigan; orthotist Alicia Foster; Rachel Melvin, who gave a great walking pole demonstration; and



CMTA Social Media Director Bethany Meloche, who read from her new book, “How Should a Body Be?”

★

WESTCHESTER, NY

Ted Drygas CPO, FAAOP, from Care Crafters Prosthetics & Orthotics in Nanuet, New York, spoke at the branch’s March 11 meeting. Ted brought and demonstrated many custom and off-the-shelf bracing options for CMT patients. Eleven people turned out even though it was an exceptionally cold late-winter day. Most were not aware of the many bracing possibilities Ted presented so it was a very informative and very interesting meeting.

★

WILMINGTON, NC

Representatives from Paws 4 People Foundation—and special guest speaker “Watson”—shared information about this amazing organization at the Wilmington Branch’s meeting on March 18. The founda-



tion has 500 highly trained assistance dogs providing support in areas including mobility and psychiatric service, educational and rehabilitative assistance and therapeutic visitation.

★

PORTLAND, OR

Inspired by “101 Tips for Dealing with Charcot-Marie-Tooth Disease,” members at the March meeting of the Portland, OR branch shared their own suggestions for CMTers. One member shared a tip about buying shoes on the Nordstrom website, which allows split sizing: Order two pairs, two different sizes, and return the shoes that don’t fit, regardless of whether or not they match in size. Another member emailed a great idea for exercise, especially helpful for those confined to wheelchairs: Before rising in the morning, jog in bed. For devices to help with strength and dexterity issues in the hands, check out www.WrightStuff.biz. Another CMTer offered some very useful tips for feet, including toe socks for protecting hammer-toes and bunions, rolling feet over tennis balls to decrease pain and stiffness, and relieving cramping by getting on an exercise bike and peddling.

★

PITTSBURGH, PA

Three new people came out for the branch’s March meeting. They heard from Patty Yerina from the ACHIEVA Family Trust, who discussed future planning for individuals with special needs. Members also discussed plans for a one-day tour of Camp Footprint, which is held 40 miles north of Pittsburgh at Camp Kon-O-Kwee. The branch raised \$2,325 in 2016, including all memberships, subscriptions, donations, purchases from the CMTA store and a chocolate-covered pretzel fundraiser.

★

SOUTHEAST WISCONSIN

The branch heard from Hanger area director Dave DiTorrice at its March 4 meeting. Members discussed AFOs, AFO materials, and how AFOs can help with balance, fatigue and other CMT-related issues. The group also talked about its Third Annual Wisconsin/Milwaukee Walk 4 CMT, which will be held on July 29. Like last year, participants will walk on the rubberized running track that surrounds the huge ice oval at the Pettit National Ice Center.

At its April 22 meeting, the branch heard from Rick Slama from the Northwest Milwaukee Social Security Office. He talked about the ins and outs of applying for Social Security Disability and Supplemental Security Income (SSI), which is intended to help disabled people who have little or no income. Rick encouraged people to be proactive in their applications, to reapply if necessary and to seek assistance from an attorney if needed. He also encouraged attendees to create a “MY Social Security Account” by visiting www.socialsecurity.gov/myaccount and noted that people can file an appeal online if they recently applied and were denied for medical reasons, something that often happens with CMTers.



CMTA Branches

Most CMTA Branches can be accessed
online at www.cmtausa.org/branches

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Toronto Area
Linda Scott Barber
416-997-5084

Interested in starting a branch in your area?

Contact CMTA Director of Community Services Jeana Sweeney at Jeana@cmtausa.org.



ASK AN EXPERT

I would be very interested in hearing how a specialist thinks the gene editing technology CRISPR might eventually help prevent CMT—or even correct the disease. To me it seems a lot more promising than drugs, Schwann cells, stem cells, etc.

John Svaren, PhD, chair of the CMTA's Scientific Advisory Board, replies:

CRISPR (clustered regularly-interspaced short palindromic repeats) is a gene editing technology that can be programmed to target specific stretches of genetic code and to edit DNA at precise locations.

The CMTA has used CRISPR extensively in many CMT projects. For example, CRISPR, and an earlier version of genome editing technology were used several years ago to create cell

lines for screening compound libraries for CMT1A at both the National Institutes of Health and Sanofi-Genzyme. CRISPR has been used to create the CMT2A rat, which has turned out to be the best rodent model of CMT2A.

CRISPR is also being used with iPSC (induced pluripotent stem cells) lines in the lab of Robert Baloh, MD, PhD (director of the Neuromuscular Medicine Department of Neurology at Cedars-Sinai Medical Center in Los Angeles) to create comparison cell lines to identify the precise defects that are associated with motor neurons made from CMT2A iPSCs, and other stem cell projects are using similar approaches.

This brings us to the possibility of gene therapy using CRISPR,

which has triggered a lot of popular interest. To “correct” the mutations in every Schwann cell (or neuron for CMT2), we will have to be able to deliver CRISPR to every such cell. A lot of progress has been made on the gene therapy front with lentivirus (specifically, the lab of CMTA-sponsored investigator Kleopas Kleopa, MD, at the Cyprus Institute of Neurology and Genetics) to deliver genes to Schwann cells in order to correct defects in a mouse model of CMT1X, and this, combined with CRISPR, may eventually become a therapy. However, there are a number of safety issues that would have to be addressed before this could be tried in humans, so it is likely that any such trials will be more than five years down the road. ★

OXFORD BIATHLON ORGANIZERS DOUBLE FUNDRAISING GOAL

Steve O'Donnell and Clark Semmes, organizers of the highly successful Oxford Biathlon, have doubled their fundraising goal for 2017. Last year, the Biathlon, a family-friendly event with something for everyone in beautiful Oxford, Maryland, brought in more \$100,000 for the CMTA and CMT research. With that success in mind, Steve and Clark are reaching for \$200,000 this year.

This year's Oxford Biathlon will take place on Saturday, June 10, and will once again offer participants a choice of a one-mile open water swim, a 20-mile bike ride, walks of various distances or simply lunch on the gorgeous Oxford waterfront.

For the first time, the 2017 event will be entirely run by the CMTA, including the swim, which previously piggybacked on the SharkFest swim. While the Masthead Restaurant remains the starting and ending point for this year's event, it is under new ownership, and has been renamed Doc's Sunset Grill. The new owners, Bo and Chooch, are looking forward to the Biathlon and offering a great lunch to all participants.

Anyone interested in registering for or donating to the 4th Annual Oxford Biathlon can visit www.cmtausa.org/oxbi for more information.



Swim, Bike & Walk for the Cure
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WHAT'S ON YOUR MIND? *Ask David.*



Write to David at info@cmtausa.org.

Dear David,
I can't believe the comments I get from people I don't even know, who come right up to me and ask if I was in an accident. Sometimes in my mind I feel like saying, "If you don't get out of my face, there really will be an accident!" I usually try to be a gentle, understanding person, but sometimes the comments are just too much for me to take. Am I overreacting?

David replies:

I can absolutely understand your internal response to this kind of insensitivity. More than once I have thought of my cane as more than just a cane, if you know what I mean. There is such a difference in my response to questions about my CMT when I feel that someone is asking in a truly empathetic way. A year ago, I was on vacation trying to get in the pool when the

woman in the next chair noticed me, looked right in my eyes and asked me if I needed some help. I hesitantly said I did, and she held the chair I was using as a support so it wouldn't slip. She began talking to me about her son who has a congenital weakness in his hands, but is doing well. There was something about her questions about my CMT that did not feel at all intrusive. Here was someone who was truly interested in making a connection over our shared experience. I felt very warm towards her and actually talked to her and her husband several times during my stay.

I remember many years ago someone actually stopped me on the street and said, "What a cool way of walking!" I was dumbfounded and could not come up with a response. She had boldly

pointed out the thing I was trying to hide. I will give her the benefit of the doubt and chalk it up to her discomfort. Even today when I try to think of a response, the only thing that comes to mind is "Thank you." Now that I'm a bit more grown up, I can handle those comments more easily, but it still amazes me how many people are uncomfortable when they are confronted with a disability. The worst is when they look at you with pity and comment on how brave you are to deal with being so limited. I am not defined by my limitations; I am defined by what I can do despite them. I truly believe that most people are uncomfortable with their own imperfections and their discomfort is projected onto anyone who is different. I can feel in my gut the difference between someone who asks me a question

Tennessee CMter Tackles Appalachian Trail Thru-Hike

Only one in four people who attempt a thru-hike of the 2,190 mile Appalachian Trail succeeds. Tennessean Dan Fry is determined to be one of them, despite the fact that his CMT forced him to have both hips replaced by the age of 19. Dan, now 24, set off from the Georgia Trailhead April 6 on a modern-day hero's journey to bring attention to the rare disease and to raise funds for research.

Dan knows some people might write him off as crazy for undertaking such a massive physical challenge. But in the last year, he got interested in learning more about CMT, how it works and the people it affects. He went online and found Facebook, forums, and blogs full of CMT patients in pain who found it hard to imagine a happy future. "I heard them," Dan says, adding, "People deserve to know about this disease. Patients deserve to be hopeful that they can still accomplish things bigger than CMT. Parents deserve to believe that their kids with CMT will be all right. For these reasons, I'm going walking."

On a typical hero's journey, the hero often encounters helpers along the way who provide comfort and support. On his journey, Dan will connect with members of the Charcot-Marie-Tooth Association, which has branches up and down the East Coast. Dan's helpers will deliver Ibuprofen and trail mix, share stories and laughs and walk along the trail with him for part of his journey. Harrisburg, PA Branch Leader Erin Gaul plans on picking him up and taking him to her home for a hot meal and a hot shower, not necessarily in that order.

The Appalachian Trail is the longest hiking-only footpath in the world, ranging through 14 states from Georgia to Maine. Dan sold all his belongings and took a six-month leave of absence from his job as a finance manager at a car dealership to complete the trail.

Donations to Dan's fundraising campaign can be made at www.cmtausa.org/donate/daniel/.





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about my CMT in a caring way, as opposed to someone who is just curious but has no interest in connecting or seeing who I am apart from my weird way of walking. With people who are detached and unable to relate to me in an engaging way, I give them a brief and rather dismissive response and move on. Sometimes I admit I will be a little outrageous and say "Yea, I was skiing and was hit by a snowmobile." Hey, I need a little fun!

Lastly, I would advise moms and dads who want to prepare their kids for this kind of insensitivity to tell them that most people are kind-hearted but can say dumb things. The better your kids feel about themselves, CMT and all, the less these comments will affect them. ★

David Tannenbaum has an LCSW degree and has been a therapist in New York City for the past 30 years. He has specialized in helping others with the task of growing emotionally and spiritually through physical challenges. "My CMT has been my greatest challenge and my best teacher in life," says David.

GENE THERAPY GRANT

(continued from page 3)

advance and expand on his previous work as his team examines whether repeated injections lead to increased protein levels, and whether treatment at later stages of the disease leads to improvements similar to those seen for treatment in the early stages.

The CMTA funds and promotes targeted activities in its STAR research network with the aim of advancing therapies via alliance partnerships, with 40 current research projects and a funding total of just over \$5 million over the last three years.

The MDA and CMTA Boards of Directors approved the grant after careful analysis and deliberation by the MDA's Research Advisory Committee and the CMTA's STAR Advisory Board, peer review processes overseen by leading clinicians and scientists in volunteer roles. ★

CMT PATIENT MEDICATION ALERT:

Definite high risk (including asymptomatic CMT):

Taxols (paclitaxel, docetaxel, cabazitaxel)
Vinca alkaloids (Vincristine)

Moderate to significant risk:

Amiodarone (Cordarone)
Arsenic Trioxide (Trisenox)
Bortezomib (Velcade)
Brentuximab Vedotin (Adcetris)
Cetuximab (Erbix)
Cisplatin and Oxaliplatin
Colchicine (extended use)
Dapsone
Didanosine (ddI, Videx)
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Nivolumab (Opdivo)
Pembrolizumab (Keytruda)
Perhexiline (not used in US)
Pomalidomide (Pomalyst)
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WHAT IS CMT?

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