



cmtupdate



The Landmark “The Voice of the Patient” Meeting:

Patient-Focused Drug Development Meeting for Charcot-Marie-Tooth to be Held in 2018

The Hereditary Neuropathy Foundation (HNF) will be holding a landmark externally-led Patient-Focused Drug Development (PFDD) Meeting for Charcot-Marie-Tooth (CMT) and related inherited neuropathies (INs) in Washington, D.C. on Friday, September 28, 2018. The meeting will be held in conjunction with September National CMT Awareness Month. HNF was accepted by the U.S. Food and Drug Administration (FDA) under the agency’s externally-led PFDD initiative to host this meeting on behalf of the hereditary neuropathy community. Patients, treatment developers, healthcare providers, and regulatory professionals from the FDA will be in attendance to focus on the needs of this rare neuromuscular disease community, which impacts 1 in 2,500 (adults and children) in the U.S. and 2.6 million people worldwide.

PFDD meetings integrate patient insights into the drug development process. Learnings will help inform clinical trials, and could potentially be used to assess other therapies to evaluate the benefits of seeking additional indications. This groundbreaking meeting will include facilitated panel discussions designed to provide the FDA with perspectives from people with hereditary neuropathies, advocates and caregivers. Following the successful model that the FDA developed to host similar meetings, the day will focus primarily on a range of patient viewpoints on CMT/INs, covering the symptoms and impacts to daily life that are most important to patients and patients’ perspectives on existing and future treatments. This input can help inform the FDA’s

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1-855-HELPCMT (435-7268)

www.hnf-cure.org

The Hereditary Neuropathy Foundation’s mission is to increase awareness and accurate diagnosis of Charcot-Marie-Tooth (CMT) and related inherited neuropathies, support patients and families with critical information to improve quality of life, and fund research that will lead to treatments and cures.

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PATIENT-FOCUSED DRUG DEVELOPMENT – CONTINUED

(continued from page 1)

decisions and oversight during drug development and the review of marketing applications for new drugs.

Allison Moore, HNF Founder/CEO, stated, “This is an unprecedented time for our community: with the first-ever CMT clinical trials already in late stages and more clinical trials on the horizon in 2018, we must take action now to educate the FDA and other stakeholders in the drug development process on the impact that CMT/INs have on patients’ and their families’ lives and quality of life to ensure their perspectives are considered throughout the drug development continuum.” Moore continued, “It is also critical that we document the impact new therapies could have on mortality rates and disease severity among those patients with the fatal disease phenotypes.”

HNF is inviting all advocacy and stakeholders in the CMT community to join this meeting to advocate for the needs of the CMT/IN patient community. Patient voices are critical in understanding how to develop and approve therapies that provide clinical and meaningful benefit to those living with CMT. Bringing the voices of patients, caregivers, and advocates to the conversation will produce the data and documentation needed to publish a summary of the day’s proceedings in the form of a “Voice of the Patient” report that can be used by FDA leadership as well as other stakeholders in the drug development process. This meeting also serves to address industry’s concerns regarding committing resources to drug therapy development related to the gaps in patient-reported data, clinical natural history studies,

risk-benefit assessment, and validated clinical outcome measures that are sensitive to capture disease progression and/or improvement. The learnings of a patient-led meeting will help to document CMT/INs patients’ experiences to further define the devastating effects of living with this debilitating disease.

HNF has been active in pursuing stakeholders (industry) that agree there is a commercialization opportunity for drug development; however, data gaps are blocking translation. Furthermore, finding additional disease mechanisms, targets, and biomarkers are important for many of the rarer types impacting young children and adolescents. An improved understanding of patients’ impaired function presents a critical path to the future of therapy development.

With these learnings, it will be possible to implement outcome measures that will capture what matters most to patients. Disease manifestations are not always equal among patients, so patients’ perspective on the significant impact on activities of daily living and quality of life (QoL) helps to develop guidelines and reporting for patient-focused research initiatives.

FOR MORE INFORMATION ABOUT THE PFDD MEETING, TO GET INVOLVED, AND/OR TO BECOME A MEETING SPONSOR, PLEASE CONTACT ALLISON MOORE AT ALLISON@HNF-CURE.ORG

OR VISIT [HTTPS://WWW.HNF-CURE.ORG/CMT-PATIENT-FOCUSED-DRUG-DEVELOPMENT-MEETING/](https://www.hnf-cure.org/cmt-patient-focused-drug-development-meeting/)

10 Reasons to Join the CMT Inspire Community TODAY!

1. Get real life answers from other patients and caregivers that are experiencing CMT firsthand.
2. Control what you share on a user-friendly platform with extensive privacy settings.
3. Share stories with patients and caregivers in an online community that can help educate, inspire and empower others.
4. Join multiple Inspire communities (i.e. Invisible Disease Alliance) to connect with more people.
5. Receive regular updates about CMT and the health topics you care about.
6. Connect to explore and share your feelings with others who are dealing with the same health concerns.
7. Learn about treatments and therapies for CMT.
8. Network with individuals that live in your area for face-to-face support.
9. Connect with others from anywhere at anytime for support and comfort.
10. Discover opportunities to participate in clinical trials and other research.

Landmark “Voice of the Patient” Meeting Needs All Hands on Deck!

What a year we have had! Let’s do it again in 2018 by making CMT the biggest rare disease EVERYONE has heard of! But we need your help to get the job done! As we prepare for the externally-led Patient Focused Drug Development (PFDD) meeting accepted by FDA and hosted by HNF, we need to raise the funds to reach our goals!

We will have our voices be heard at that meeting that day by key FDA officials. We will demonstrate the impact CMT is having on so many lives. **Most importantly, the FDA will hear that early intervention with pediatric treatments will result in quality of life improvements and will essentially halt disease progression for so many people.** The outcome of this meeting will accelerate possible therapies, drug trials, and approvals.

The top five reasons to DONATE to this groundbreaking effort:

- Your generosity will underwrite travel expenses for patients and their family members who will give live testimonies to the FDA on the impact of living with CMT.
- We require financing to support the planning and development of the agenda, finalize all of the meeting venue details, and publish the final “Voice of the Patient” summary report to the FDA website.
- We need to produce a live webcast of the meeting so thousands across the U.S. can join in and be a part of this monumental day.
- We need help to attract more attention for CMT from the pharma and biotech industries by funding improved patient-reported outcomes research studies with multiple surveys and polling leading up to this meeting.
- Your donation will let you give thanks and honor to your loved ones suffering with CMT by ensuring the meeting day is a huge success.

Please donate in honor or memory of a loved one and help support this ground-breaking event!

CMT Champion.....	\$1,000
CMT Promoter.....	\$500
CMT Advocate.....	\$250
CMT Supporter.....	\$100
CMT Friend.....	\$50

On the donation form, there is a place to write your personal sentiments related to CMT—these sentiments will be displayed at the meeting—another way to show FDA officials and other key officials the impact of CMT on so many lives.

With your permission, we will also acknowledge your stewardship of this landmark event in the meeting program booklet and the 2018 year-end *CMT Update* newsletter.

Be a part of the change we all want: help us ensure CMT is the biggest rare disease everyone has heard of!

CLICK HERE FOR SPONSORSHIP:
[HTTP://WEBLINK.DONORPERFECT.COM/PFDDSPONSORSHIPPAGE](http://weblink.donorperfect.com/pfddsponsorship)

New Executive Director for HNF!

BY JOY ANDAL KAYE, BOARD CHAIRMAN, HNF

The Board of Directors of the Hereditary Neuropathy Foundation is pleased to announce that Courtney Hollett has been named Executive Director effective December 1, 2017. Courtney joined HNF in February 2012 to assist with fundraising, database management, and financial reporting. In addition, she provided support to our many fundraising event coordinators and to the growing membership of Team CMT. Finally, she managed social media outreach for HNF as well as managed all administrative aspects of HNF as a whole. Courtney earned her B.S. from Salve Regina University in Rhode Island.

Familiar with the effects of CMT on members of her own family, Courtney has seen first-hand the importance of raising awareness and funds to cure CMT. Needless to say, she has been an integral part of the growth of HNF, and we are excited to have her lead the organization into 2018!



Please join me in congratulating Courtney!

HNF JOINS CHRONIC PAIN ADVOCACY GROUPS TO PROTECT ACCESS TO PAIN MEDICATIONS

BY TINA TOCKARSHEWSKY, DIRECTOR, MEDICAL AND PUBLIC AFFAIRS, HNF

HNF recently joined over 30 patient advocacy groups and health care professional societies in signing a letter to members of Congress regarding the Ensuring Patient Access and Effective Drug Enforcement Act that unanimously passed both the House and Senate and was signed into law on April 19, 2016. The groups are advocating to maintain the law in its current state and are opposing its repeal.

Due to a concern that thousands of people with pain conditions were unable to obtain their necessary prescription medications as a result of arbitrary closures of controlled substances distributors by the DEA, this bi-partisan bill was sponsored by Senator Orrin Hatch, among others, in strong support of the pain community. Senator Hatch helped to ensure that the 2011 Institute of Medicine pain report and the Interagency Pain Research Coordinating Committee were inserted into the ACA, and he has also been a leading voice trying to advance the National Pain Strategy. The letter is in support of Senators Hatch and Whitehouse's efforts to ensure that patients do not suddenly lose access to their pain medications as had been happening a few years ago.

Recently, CBS aired a one-sided "60 Minutes" segment on October 15, 2017, in which Joe Rannazzisi, an ex-DEA assistant administrator who now consults for lawyers suing the opioid industry, accused the bill sponsors—and Congress as a whole—of caving in to the "big pharma" opioid lobby and tying DEA's hands so they could no longer shut down rogue distributors. This is simply untrue. As a result of the "60 Minutes" segment, no fewer than five legislative bills were introduced to repeal the law after the program aired.

The language for the new law was written by Obama Administration officials at the Justice Department and DEA—and both supported the final bill which requires DEA to: (1) inform DEA registrants that they are under investigation and what measures they are suspected of violating; and (2) provide them 30 days to correct the situation. The law additionally authorizes the DEA to bypass this notification process and immediately suspend a distributor's registration if there is substantial likelihood of death, bodily harm, or abuse of controlled substances.

Since one distributor could service scores of pharmacies, each with hundreds of customers, this leads (and did lead) to widespread shortages of prescription opioid drugs, which consequently left innocent Americans struggling with debilitating chronic conditions suddenly cut off from their treatments.

The signers of this letter want distribution centers who have engaged in illegal activities to be dealt with appropriately—and even shut down in cases where there is imminent danger to the public. The advocates do not believe this fair and balanced law should be repealed. This may embolden DEA to resume and increase distributor shut downs with no warnings, again leading to pharmacy shortages.

HNF proudly stands with the chronic pain community in protecting access to therapies and treatments to help manage the debilitating effects pain has on quality of life. We will continue to monitor this situation on behalf of our patients and advise you of future advocacy efforts where patient advocates' voices may be needed to represent the needs of our community.





HNF developed the Therapeutic Research in Accelerated Discovery (TRIAD) as a collaborative effort with academia, government and industry, to develop treatments for CMT. Currently, TRIAD involves many groups that span the drug discovery, drug development, and diagnostics continuum.

Hope on the Horizon

BY JOY ALDRICH, DIRECTOR OF ADVOCACY, HNF

The Hereditary Neuropathy Foundation (HNF) is a staunch champion for accurate and early Charcot-Marie-Tooth (CMT) disease diagnosis, recognizing its critical importance for patient care, research, and treatments. For those with CMT and suffering with its progressive muscle atrophy and weakness in their legs, feet, arms, and hands, there are currently no cures nor approved curative or symptomatic medications.

A change is on the horizon, though!

In December 2016, Pharnext, a French biopharmaceutical company, completed patient enrollment for the international Phase 3 clinical trial of PXT3003, Pharnext's lead PLEODRUG[®], for treatment of CMT1A (the most common form of CMT). The trial's primary endpoint is the change in the Overall Neuropathy Limitation Score (ONLS) to determine improvement of patient's disability after 12 and 15 months of treatment. The trial results are expected in the second half of 2018.

HNF has been partnering with Pharnext to encourage this clinical trial's development, and we want to thank all who are involved with the study, from the researchers and developers at Pharnext, the clinicians in the field and at our HNF-designated Centers of Excellence, to the patient-participants; all contribute and play a critical role.

HNF has been pleased to contribute to this groundbreaking research by providing U.S. clinical site identification and patient recruitment support for this pivotal Phase 3 trial of PXT3003. HNF resources such as the Global Registry for Inherited Neuropathies (GRIN), our online Inspire Patient Community and our CMT-Connect local patient empowerment programs have been instrumental. We are enthusiastic that our joint effort with Pharnext could result in the first-ever therapy for CMT, and we look forward to continuing our collaboration to tackle the more rare forms of CMT in the future.

We can't rest on our laurels!

It will take many, many clinical trials to build the pipeline of potential therapeutics necessary to serve the diverse needs of the entire CMT community. That is why we have created the [Clinical Trial Support Fund](#), specifically to raise the money for the clinical trials desperately needed by our patients. Your tax-deductible donation will ensure that HNF can continue to support clinical trials for years to come.

NOW AIRING ON TELEVISION:

WWW.HNF-CURE.ORG/PHARNEXT-PLEODRUG-CMT1A/

ACE-083 PHASE 2 CMT STUDY UPDATE

ACE-083 is an investigational drug that is based on a naturally-occurring protein and works to enhance the body's own promoters of muscle growth. ACE-083 can be customized to patients' needs by administering the drug into target muscle(s) with the aim of increasing muscle size and strength. Accelaron is developing ACE-083 for neuromuscular diseases in which patients experience weakness and atrophy in specific muscles, such as CMT patients with foot drop.

The Phase 2 clinical trial consists of two parts designed to evaluate ACE-083 in CMT patients with muscle weakness in the tibialis anterior (TA), a muscle in the lower leg involved in foot dorsiflexion (raising the foot at the ankle). Part 1 is an open-label, dose-escalation study, to evaluate safety and efficacy in a range of dose levels. Part 2 is a randomized, double-blind, placebo-controlled study using the optimal dose level selected in Part 1.

Enrollment and treatment are ongoing in Part 1 of the Phase 2 trial of ACE-083 in patients with CMT disease.

FOR ADDITIONAL INFORMATION ABOUT THIS CLINICAL TRIAL, PLEASE VISIT CLINICALTRIALS.GOV, IDENTIFIER NCT03124459.





Team CMT

MEMBERS CAPTURE NATIONAL TITLES!

BY CHRIS WODKE, TEAM CMT FOUNDER

On November 12, Team CMT members Joy DeCaro Perry and Chris Wodke competed in the Miami Man Triathlon in Miami, Florida.

The race was a National Championship for Aqua Bike and Long Course Triathlon. Joy competed in the Aqua Bike race, which was a 1.2 mile swim and a 56 mile bike ride. Chris competed in the Long Course Triathlon, which added a 13.1 mile run to the Aqua Bike swim and ride.

It was a challenging day, with 20 mph winds and 88 degree heat. These types of conditions are tough enough for any athlete, much less one with the challenges of CMT. Just finishing a race with such conditions is a victory. Both athletes raced in the Physically Challenged division, each taking first place. So, Team CMT now has two National Champions! The race is also a qualifier for the World Championship to be held next July 2018 in Denmark. Chris has already qualified for the World Championship in Denmark in Aquathlon—Aquathlon consists of a swim and run.

The race was also a fundraiser for CMT programs and research.

“We were both proud to be racing in Miami and raising awareness of CMT. I am also so proud of Joy. She beat a tandem team, which is really hard to do. I took 1st place in my triathlon, but there were only two of us.”

– Chris Wodke

YOU CAN STILL MAKE A CONTRIBUTION IN HONOR OF THESE CHAMPIONS AT:

[HTTPS://HNF.DONORPAGES.COM/TEAMCMT/CHRISWODKE/](https://hnf.donorpages.com/teamcmt/chriswodke/)

Thank you, Chris and Joy, for taking on this challenge to help others with CMT.



Gearing Up for the TD Bank Five Boro Bike Tour

BY COURTNEY HOLLETT, EXECUTIVE DIRECTOR, HNF

The TD Bank Five Boro Bike Tour will take place on May 6, 2018. HNF has been fortunate to participate in the TD Bank Five Boro Bike Tour as a charity rider for America's largest cycling event. This means more participants riding to increase awareness of Charcot-Marie-Tooth, and thousands more dollars raised to fund research into treatments and a cure.

HNF Riders get VIP treatment, which includes breakfast and lunch as well as priority start, special bike parking at the start, and a Team CMT singlet. The event provides participants the unique and fun experience to bike through all five boroughs—a 40 mile, traffic-free ride for 32,000 cyclists. Starting just north of Battery Park, the tour runs up Manhattan, through Central Park, around a brief loop in the Bronx and down to the Queensboro Bridge passing countless New York City icons on the way. After a ride over the Pulaski bridge passing through Brooklyn, riders enjoy an incredible view from the lower deck of the Verrazano Bridge.

Join the HNF Team and help raise funds for CMT research!

“We love coming up to NYC and being on Team CMT! It's one of our favorite events. Great energy.”

– Jeff W, CMT Patient

“Nothing is more inspiring than riding in the 42 mile TD Bank Five Boro Bike Tour next to my inspiring and relentless friend and founder of HNF, Allison Moore. Although I ride for her and Team CMT, she would be the one I would want on my team biking through the beautiful boroughs increasing awareness for CMT. GO TEAM CMT!”

– Gabby G, Friend

JOIN THE HNF TEAM IN THE TD BANK FIVE BORO BIKE TOUR
[HTTPS://WWW.HNF-CURE.ORG/TD-BANK-FIVE-BORO-BIKE-TOUR/](https://www.hnf-cure.org/td-bank-five-boro-bike-tour/)

Join Team CMT – Be a part of finding a cure!

Team CMT is a grassroots community fundraising program founded by Chris Wodke, a CMT athlete who partnered with the Hereditary Neuropathy Foundation (HNF) to raise awareness for Charcot-Marie-Tooth disease.

Building awareness helps us to create a better understanding of the challenges faced by those living with CMT and assists us in increasing support, research, and the likelihood of finding a cure. By joining us, you can truly make a difference. Register today to become a Team CMT member and you'll be a part of an international effort to change the future for those living with CMT.

When you join the team and make a minimum donation of \$25, we will send you a Team CMT shirt that you can proudly wear to help promote awareness. You can wear your Team CMT

shirt to the gym, physical therapy, or to an athletic event—anywhere you want to engage and encourage a conversation about CMT.

If you want to do your own fundraising campaign, we can help you with a dedicated fundraising page to track your efforts: <http://www.hnf-cure.org/team-cmt/> Team CMT is not just for adults, and we want to encourage everyone to get involved!

Check out our special Team CMT Kids webpage to learn how to support children living with CMT by registering your school or creating a team: <http://bit.ly/2pSDXS9>

We are our own best advocates for managing our health and promoting greater awareness of the challenges we face while living with CMT. Together, as a team, we can increase CMT awareness—please consider joining Team CMT TODAY!



Introducing the New Helios GX with Monica Tullar

Q: Can you tell us about your experience with bracing for CMT?

A: When I was diagnosed with CMT in the 1980s, there were very limited options in bracing, and I was less than enthusiastic about any of them. There was a buzz about a new spiral brace, and I had huge expectations! Here was a brace that looked cool and was going to give me balance and stability while still being slick enough to give me shoe options...after all, I was a teenager being braced for the first time.

Unfortunately, manufacturing was discontinued, and I was heartbroken. As a matter of fact, I was so disappointed, it was years before I attempted any bracing at all. Eventually, I progressed and began the LONG, BUMPY, UNEVEN journey from AFO styles made of different materials to the Helios brace. I worked with everyone from Duke University Hospital in North Carolina to the Mayo Clinic in San Diego; somehow I was blessed to find Ortho Rehab Designs here in Las Vegas. I've worked with many orthotists, but Mitchell Warner is the only one who has not only met my expectations, but also exceeded them with each new design. Until now, every brace had to step up to me—With the Helios GX, I have increased my overall activity level! I have yet to find a limitation for this brace.

Q: What are the benefits of the Helios GX?

A: Within minutes of putting on this brace, I was HOPPING. Now, this wasn't the first time that I was hopped up with enthusiasm after putting on one of Mitchell's new designs; however, this is the first time in 30 years that I was actually literally hopping! The energy return from this brace is far beyond anything I expected.

The Helios E/I has energy return, and it helped me so much with walking stability. The Helios GX literally had me walking faster, with stability, comfort, and that extra spring to support me. I found myself walking so well around Mitchell's office, and I was so excited and inspired by the Helios GX, that I began hopping for everyone: friends, neighbors, patrons at my local pub, and, finally, for my physical therapist. I walked into Comprehensive Physical Therapy with a hop in my step! I was greeted by Traci Cassidy, and I eagerly performed my bunny interpretation. Within days I was jumping! I know that may sound like nothing; however, when you've been told "YOU WILL NEVER DO THAT AGAIN"—this was unexpected to say the least!

Q: Can you describe what balance means to you when wearing an AFO? Do different designs affect your balance?

A: The issue of balance and stability, in my opinion, is addressed in all of the different Helios designs. Ortho Rehab Designs' continuous diligence in addressing foot deformities and proper alignment give them a superior foundation to build on. Mitchell's casting technique and custom fit literally builds from the ground up. You just don't get that from an off-the-shelf brace. I truly believe that if it was not for the triplanar correction and alignment, I would be in a wheelchair today.

At 14, doctors predicted I wouldn't walk past the age of thirty. If I had continued the wear on my joints, ligaments, and tendons, I think I would have proved them right. I can't stress enough the importance of early bracing. It's because of proper bracing for twenty years that I am surgery-free at forty-five, and I can't imagine what I would have done with this brace if I had it when I was younger. Hop... Hop... JUMP!

Q: Can you tell the difference with the new and improved design?

A: I feel like the Helios E/I and the GX both have their place and their respective patients. Bracing is so individual and unique to each person. This is where the designer is imperative. What Mitchell is doing is totally custom designed. Each foot, each leg, each patient: there is not one that is better than another, only what is best for you! The E/I is great for uneven surfaces or if you're in a situation where you do need to crouch or get up and down off of your knees. However, for us maximum performance/high-impact patients, it's the GX. There's no questioning the balance of the E/I and no comparison to the performance of the GX.

Q: How are you doing with different types of shoes?

A: As far as shoes go, I am wearing EVERYTHING in the Helios GX. I only went up a half size. I found that with a little creativity, there is a plethora of shoes out there. I have found both of the Helios designs to be compatible with a cobbler (not the cherry pie kind), and there are lots of possibilities. Through the years, I have accumulated several shoe tips and will be sharing them on Ortho Rehab Designs' YouTube channel, along with feedback on my continued efforts to find the limitations of this new Helios GX. Stay Tuned!

HELPING CMT PATIENTS GET AND STAY PHYSICALLY ACTIVE

For many patients with CMT, walking the dog, carrying the groceries, or putting them away at home can be fatiguing, painful, scary, and sometimes insurmountable tasks. These activities of daily living and others, are activities which may be improved with focused and progressive therapeutic exercise. The general population, young and old, and certainly those with medical issues are encouraged to exercise by doctors, family, governmental health agencies, and even the media to improve health and quality of life.

A consistent exercise program is an essential part of attaining and maintaining a healthy lifestyle. Regular physical activity helps to maintain a healthy body composition, strengthen joints and muscles, improve balance and cardiovascular function. For those living with CMT, progressive exercise has the potential to improve functional ability, independence, and quality-of-life. An ever-increasing body of clinical and scientific information indicates that most patients with CMT, when not contraindicated, should regularly participate in exercise and physical activity under the guidance of a qualified professional.

Starting an exercise program can be intimidating to many patients with CMT, when routine tasks such as buttoning a shirt, navigating stairs, or opening a car door are daily challenges. Foot deformities, balance deficits, foot drop, and increased risk of falling also may prevent those living with CMT from participating in regular forms of physical activity and exercise. Choosing the “right exercise” or the “best activity” is often a factor steering those with CMT away from enjoying the benefits of regular exercise and physical activity.

Exercise regimens for patients with CMT should be developed under the guidance and with the advice of informed professionals, such as physical therapists, occupational therapists, psychiatrists, and exercise physiologists familiar with CMT.

HNF created “Band Together for CMT” to give patients with CMT some suggestions and a starting point to begin an exercise program. Each individual should seek professional guidance as to their individual program in terms of intensity, volume and frequency of exercise. Patients with CMT may have unique strengths and weakness, limitations and potential precautions to be considered when starting a program. Every exercise and activity displayed has potential modifications in position, action, and more to enable everybody to participate. The general principles herein are more important than the specific exercises displayed. These are some basic movements which may be of benefit to those with CMT. There are many choices of appropriate exercises and activities, including balance activities, cardiovascular exercise, and stretching (all important), which are not included in this edition. A comprehensive, individualized program is always the best way to begin. If you haven’t exercised in sometime, start VERY slowly, progress carefully, if you have been exercising, listen to your body as you progress.



HEREDITARY NEUROPATHY FOUNDATION

BAND TOGETHER

FOR CHARCOT-MARIE-TOOTH (CMT)

*Start moving, start exercising!
Everyone can do something: speak with your doctor or physical therapist.*

BY DR. JAMES NUSSBAUM, PT, PHD, SCS, EMT

ALWAYS, speak with your doctor and/or physical or occupational therapist prior to beginning any new exercise or activities. Discuss the intensity (how hard you should work), the frequency (how many times a week), mode (what type of exercise or activity to do), and duration (how long you should exercise each day) with a professional.

THANK YOU TO ACCELERON PHARMA FOR SPONSORING!



ORDER YOUR “BAND TOGETHER FOR CMT” BOOKLET HERE:
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More Gain, Less Pain —

WHEN CMT PATIENT TURNS TEACHER



BY CHRISTOPHER DITO

My name is Christopher Dito, and I'm a 27 year old male living with Charcot-Marie-Tooth 1A. I was diagnosed with CMT at the age of 13 and used a wheelchair by the age of 25. Although doctors argued that it was strictly CMT-related, I humbly beg to differ. I now like to think of this disease as a kicker that I must live with on top of the accumulation of bad decisions and living habits I adapted to growing up. These habits left me unhappy, anxious, hopeless, and—last but not least—in pain.

I was constantly patching up my life with dozens of different band-aids, never taking the time to consider or address the root cause of my wounds.

It was after almost 25 years of ignoring signals (including spending weeks in St. Francis Hospital for heart complications, shoulder injuries, ankle injuries, a blown ACL, a severe neck injury, lower back injuries etc), that my body had enough. In October 2014, I was walking through the supermarket when my feet suddenly gave out on me. Every tendon and muscle below both knees felt like rubber bands about to snap. It was from this point forward, and for the next year, that I would have to use a wheelchair with no real definitive answer of what had happened to me.

After many struggles (including two complete foot reconstructions requiring 26 operations), I now stand here today as a physical trainer specializing in human biomechanics. My love for the human body dates back to when I was just a kid, unable to fulfill a childhood that most people would deem “normal.” Today, my interest has become my profession and, most importantly, my passion.

My self-education of human physiology truly began while I was confined to a wheelchair; but, once mobile, it quickly turned into something more serious. I began traveling the country and partaking in continuous educational courses in the hopes that I could find the answers to my own problems as well as the problems of others. After attending courses and countless hours of research, there still seemed to be so much missing. I began to think outside the box, which led me down a few roads I'll briefly share with you today.

I was initially overwhelmed with this influx of information, as many CMT sufferers may also experience reading this article. Some might be thinking, “If I have an incurable nervous system disease, how the hell does all this help me?” Well, it may not cure us, but becoming more in touch with our body can help us understand that we have more control over our nervous systems and pain than we perceive we have.

What would happen in a world where we focused on wellness instead of illness?

This was one of the most important questions I asked myself during my recovery process. If our environment influences our health, why not work on changing our environment—or, at the very least, work on changing how our bodies respond to the environment we are in. The more I explored and researched areas such as diet, stress control, circadian rhythm, sun exposure, alternative holistic medicine and, of course, my primary scope of practice human biomechanics, the more I found out how closely they are all related.

While browsing the internet, I began finding people and methodologies that were innovative and operated outside the “ordinary.” I discovered the practice of Functional Patterns (which I am currently labeled a human biomechanics specialist under), I learned about our fascial system by reading books like *Anatomy Trains* by Thomas W. Myers and I explored the work of Dr. Jack Kruse (a neurosurgeon and biohacker). After delving down these roads and adapting them to my own body and circumstances, I’ve concluded that it all boils down to this: whether we are born with an incurable disease or performing at the highest levels of athletic ability, we are all human beings.

After this moment of realization, I truly began to see mental and physical progress. I was forced to let go of my ego and abandon all of the previous dogmas that I had held onto so desperately. I slowly began implementing all of these new elements into my recovery process, and I started listening to the signals from my body that I had once ignored.

My approach to fitness and therapy no longer replicates that of a traditional standpoint. I train myself and others to focus on the primary functions humans evolved to do: breathing, walking, running, and throwing. Who would have thought? Exercising and rehabbing our bodies in relation to our biological blueprint is key. Of course, it is more complicated than it sounds; yet, the reasoning is so simple and often overlooked.

During an educational trip to Austin, Texas, I spent exorbitant amounts of time absorbing sunlight, eating high quality seafood, and exercising with others who were determined to educate themselves on optimizing human functionality. It didn’t take long to reap the benefits of what I was doing. I was able to feel the all of the inflammation, aches, and pains slowly escaping my body... something I had previously only experienced on a much smaller scale while dieting.

As someone who has dealt with a lifetime of chronic pain and injuries and who now also helps others experiencing these issues, I think that it is important to first ask two questions: what is pain and what causes pain?

Unfortunately, what many people fail to recognize is that this subject, as a whole, is still somewhat of a mystery. Although scientific studies continue to be conducted and definitive conclusions remain unknown, I can assure you that pain is more of an output of information rather than an input. In fact,



pain is a byproduct of our environmental context. Pain can be a response of the body to cope with our own personal environment (response to stress/non-stressful environment, past trauma, stimulation of nociceptors, danger signals, etc.). All of these environmental factors play a very significant role in the state of two divisions of the autonomic nervous system called the sympathetic and parasympathetic nervous

systems. They also play a tremendous role in our bodies’ fascial system, another subject often understudied and overlooked.

While in Texas, I decided to go on a hike using minimalist shoes, something I would have never attempted to do in the past. I was able to feel my feet connected to the ground, rather than the usual “floaty” and unstable feeling. My body was being propelled forward using muscles I had never quite felt work (or at least not in that context). The usual extreme tightness I would experience in my calves never quite came, and I was able to complete the entire hike.

It’s a wonderful feeling to have this heightened sense of awareness and connectivity to your body when you’ve gone your whole life without it.

Unfortunately, most people don’t know they are feeling bad until they are feeling good—a common theme, even among my most highest of functioning clients.

Evidence-based Research? Public Research Findings? These are two of the most sought out pieces of information among health practitioners and scientists all over the world. Unfortunately, the hard truth is that both of these pieces of information are the result of our boundaries of understanding. Some amazing discoveries have come from research findings, and my intent is not to bash the achievements of these phenomenal breakthroughs. It is simply to open the eyes of the listener.

My own personal journey continues with its good and bad days, as does the journey of those I help; but, I am slowly seeing light at the end of the tunnel. We are complex organisms, but we are also merely human...so we must treat ourselves as such. I will always stand by and have an extra sense of connection to the CMT community because this condition has helped make me become the man I am today. Growing up with a disease, you are often given limitations and told that your standards shouldn’t be as high as others. I, myself, refuse to believe that there should be a lower standard to my quality of life or that I should accept chronic pain and limitations. One day, I wish to see proper treatment provided to sufferers of all physically-debilitating conditions and to the human race as an entirety. Until then, it’s up to us to do all we can on our own to combat this disease and to pursue a life without limitations.

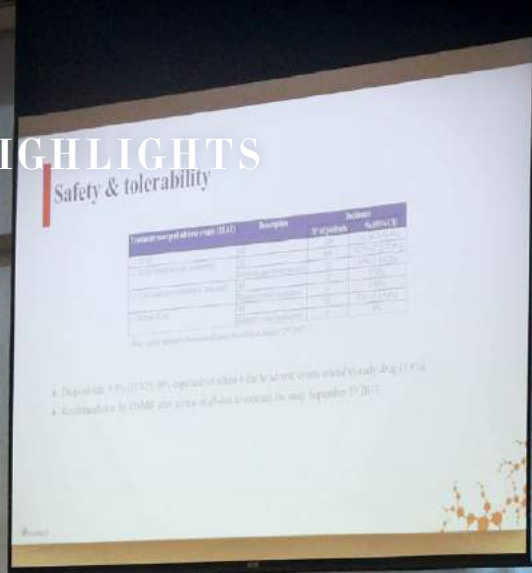
TO LEARN MORE ABOUT CHRIS DITO AND THE METHODS HE EMPLOYS, VISIT:

WWW.MADEFORMOTUS.COM

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Summit Overview 2017



“Well worth the trip. Helped re-frame my thoughts on adaptive equipment and ‘pain.’ Loved meeting another active person my age, and inspired by what others are doing.”

— Marian C

René Goedkoop, Pharnext

BY JOY ALDRICH, DIRECTOR OF ADVOCACY, HNF

On November 3, 2017, nearly 100 participants gathered in Boston for HNF’s Patient-Centered CMT/HNPP Pain Summit. It was an incredible meeting of people with hereditary neuropathies and their family members, caregivers, clinicians, researchers, funding agencies, payors, leading pain experts and pharma industry coming together to get an understanding of chronic pain in the CMT/HNPP community, including its impact on quality of life.

The event began with a welcome from HNF Founder and CEO, Allison Moore, and a report from Robert Moore (Allison’s husband) on a voice survey about pain that was conducted by using TrueReply phone polling. Data regarding descriptions of pain symptoms (stabbing, shooting, aching, pressure), descriptions of how the pain impacts quality of life (“My pain keeps me from doing the things I love”), and currently used methods of pain management (fentanyl patch, gabapentin, marijuana, yoga, mindfulness) was recorded. The data collected in the phone survey will help inform academia, government, and industry in developing effective pain management programs and therapeutics specifically for the CMT/HNPP community’s needs.

Pamela Katz Ressler, RN, MS, HNB-BC and founder of Stress Resources in Concord, MA, then eloquently presented the Keynote Address, “Chronic Pain and the Importance of Social Networks.” She described how pain can cause isolation and loneliness, and that online communities (including HNF’s Inspire community with 4,900 members) help to facilitate gathering and sharing information, as well as provide the opportunity to mentor others, which reduces suffering and isolation.

John Novack, Director of Communications at Inspire, then conducted a live-time text poll with Summit participants to further explore chronic pain within the CMT/HNPP community. We learned:

- 64% of audience respondents stated they had chronic pain that was a combination of neuropathic, muscle and skeletal/ bone pain
- 61% of respondents stated they had a combo of achy/sore, sharp/ stabbing, and tingling/electrical pain
- The majority of respondents said their chronic pain rated 4-6 on a pain scale (with 10 being worst/completely debilitating pain)
- Most reported moderate to severe interference with their general activity, mood, and stress levels
- Most reported moderate to severe impacts on sleep

As a result, HNF now has some initial data to support further research and, hopefully, publication that documents the correlation between CMT/HNPP and pain!

We then heard from Clifford J. Woolf, MD, PhD, a leading, internationally-renown pain researcher as well as a professor at Harvard and researcher at Children’s Hospital Boston. Dr. Woolf gave us a preview on his new, potentially groundbreaking, research approach for studying painful chemotherapy-induced peripheral neuropathies and the implications his learnings might have for other types of painful neuropathies including CMT/HNPP.



Xavier Paoli, Pharnext, Allison Moore, HNF, and René Goedkoop, Pharnext



Workshop: The CMT/HNPP Patient Experience Workshop

The rest of the morning session was focused on methods to treat CMT/HNPP pain.

- Dr. Florian Thomas, director of HNF’s Center of Excellence at Hackensack University Medical Center, presented a much appreciated overview: “CMT/HNPP 101” including discussion in “simple” language of the anatomy of a nerve, demyelinating vs. axonal types of CMT, inheritance patterns, symptoms and their variability even within families, prevalence, and diagnostic methods.
- Dr. Jafar Kafaie, director of HNF’s Center of Excellence at St. Louis University, gave an overview of the types of chronic pain and the current standards of care for treating neuropathic/nerve and musculoskeletal pain.
- Dr. James Nussbaum, clinical and research director of ProHealth & Fitness PT OT, presented his case for the importance of physical and occupational therapy for CMT/HNPP patients. He recommended his “Gravity Supported Program.” The program consists of three phases over twelve weeks, with a goal of increased repetitions of four core exercises. He also discussed the HNF-supported clinical trial of the AlterG® anti-gravity treadmill.
- Mitch Warner, CPO, founder of Ortho Rehab Designs, presented his case for how ankle-foot-orthosis can treat pain by providing better alignment and a more natural gait. Warner created the Helios® brace, but his presentation shared an important list of corrections that patients should require of any/all AFOs: balance restoration, triplanar correction, corrected alignment, prevention of further deformity, more functional gait, and energy return.
- A panel including Bob Twillman, PhD (executive director of the Academy of Integrative Pain Management–AIPM), Pamela Katz Ressler (our keynote speaker) and Kristin Gelzinis (HNF patient advocate) discussed the current national focus on the “Opioid Crisis,” its effect on chronic pain patients, and the possible misunderstanding and misrepresentation of the cause of the “crisis.” While new prescribing guidelines have been issued, some physicians are opting out of prescribing pain medications and some insurers are limiting access to safer forms of opioids because they are more expensive. Meanwhile, there is little movement for insurers to cover non-pharmacological treatments for pain. This led to a conversation about the practice of Mindfulness-Based Stress Reduction and how powerful it has been in helping some CMT patients reduce the amount of pain medication they require.

During lunch, we heard from personal trainer and CMT-patient, Chris Dito. His inspiring story of getting himself out of a wheelchair and into the gym training others was powerful. Dito is the owner of Made for Motus and is a physical trainer specializing in human biomechanics with a focus is on posture and breathing.

The National Organization for Rare Disorders (NORD) representative, Allison Seebald, presented an overview of the robust natural history study to support the second most common inherited neuropathy, HNPP, as well as the rarer types for which there are major gaps of information. HNF is one of twenty rare disease communities chosen to develop this study with funding from NORD and the FDA.

Dr. Mamatha Pasnoor, from HNF’s Center of Excellence at the University of Kansas Medical Center, then presented her study of four common drugs used to treat pain in idiopathic neuropathy patients and how the study incorporated patient-reported outcome measures. Preliminary results show that there was no clear winner, but overall nortriptyline (75mg day) and duloxetine (60mg day) outperformed pregabalin (300mg day) and mexiletine (600mg day). This study is being funded by a grant from the Patient-Centered Research Outcomes Institute (PCORI), who also supported HNF’s 2016 and 2017 Summits as well.

After these presentations, the Summit participants then broke out into two groups to participate in one of two highly interactive workshops:

- “The CMT/HNPP Patient Experience Workshop,” where patients developed case studies to present to medical school students about CMT/HNPP with the help of facilitator Libby Bradshaw, DO, MS-academic director of the Pain Research Education and Policy Program at Tufts Medical School.
- “Communicating Your Pain Workshop,” where Gwenn Herman, LCSW-C, DCSW, clinical director and founder of Pain Connection (a U.S. Pain Foundation program), taught how to communicate with their healthcare providers by using a “Tree Plan” with seven branches (representing aspects of daily living): physical, cognitive/emotional, spiritual, social, career, leisure/hobbies and opportunities.

The Summit then wrapped up with several research updates. Allison Moore reported on current HNF initiatives, while Rene Goedkoop, MD (Pharnext), Kenneth Attie, MD (Acceleron Pharma), Thomas Wessel, MD, PhD (Flex Pharma), and Sean Ekins, MSc, PhD, DSc (Collaborations Pharma) presented their respective work on clinical trails for CMT and research for CMT/HNPP.

A big thank you to all of the attendees, presenters, sponsors, and exhibitors who came together to make this year’s Summit another great success!

It starts in infancy

and lays dormant in the body until one day...
the symptoms start to appear.

Everything seems okay until your...

<p>Feet Start to Look Different</p> 	<p> Balance Becomes Unstable</p>	<p>Toes Start to Curl</p>	<p>Neuropathic Pain</p> 
<p>Ability to Hold, Grasp & Turn Things is Lost</p> 	<p>Muscles are Wasting</p>	<p> Shoes Don't Fit Well</p>	

Even if you are lucky enough to get a proper diagnosis of this genetic disease—Charcot-Marie-Tooth—there is no cure, and it is progressive.

But the Hereditary Neuropathy Foundation is on track to finding treatments and cures! **Learn more at www.hnf-cure.org**

HNF'S CMT CENTERS OF EXCELLENCE

The national network of HNF-designated Centers of Excellence (COE) provides patients with resources to find hubs of expertise in caring for and treating CMT, as well as locations where CMT research is being conducted. Our primary goal for the program is to ensure that access to care results in positive outcomes for each individual patient's clinical experience. We are honored to have these premier Centers and their leading experts in partnership with us to improve the future for people with inherited neuropathies.

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Events

UPCOMING 2018 EVENTS

Save the date!

5/6/18

TD Bank Five Boro
Bike Tour
New York, NY

9/28/18

Externally-led
Patient-Focused
Drug Development
(PFDD) Meeting
Washington, DC

9/29/18

Patient-Centered
CMT Summit
Washington, DC



Chrome Divas Host Another Successful CMT6 Event

On September 9th, The Rochester Chrome Divas hosted a Scavenger Hunt in the Finger Lakes Region of New York State to raise money for CMT6 research. Mother Nature provided a sunny, cool Fall day for the ride!

Ninety-eight participants enjoyed spending the day with Zach Houliares (who has CMT6) and the Divas. Grown-up “bikers” played duck duck goose, hugged a stranger, and folded laundry for a stranger, all while spreading the word for CMT6. Chrome Divas came in from Xenia, OH, Watertown, NY and Cortland, NY to help support such a worthy cause. Other bike clubs joined the day not only to show the Divas support, but to learn about “Our Man” Zach.

Each person had a chance to meet Zach, have their picture taken with him, and see where he worked this past summer. The day was a great SUCCESS! Everyone returned safely from the hunt and LAUGHING because they had so much fun. Many asked to be alerted about the next event so they can join in again. HNF is happy to report that The Rochester Chrome Divas raised over \$11,000 for CMT6 research! Thank you, Divas, and cheers to “Our Man” Zach!

LEARN MORE ABOUT ZACH'S TEAM: WWW.CURECMT.ORG

Grace's 2017 Courage Crusade

We had another amazing turnout for our 9th annual Grace's Courage Crusade fundraiser and raised over \$18,000 for CMT2. This year we decided to try something new. Our venue at the Charlestown Rathskeller Tavern included a delicious variety of food, a signature drink, and the competitive corn hole tournament.

Guests enjoyed sitting under the tent or relaxing outside by the fire pit. Our raffle included many diverse items including our ever-popular flat screen TV! Friends and family joined together to make our event one of our best yet. Grace's friends took photos, set up a henna station, sold raffle tickets, T-shirts, pop sockets, and, of course, our “Arlene on the Scene” books (available on <http://bit.ly/2fjtyiq>). Grace's family members also pitched in: even her big brother and his friend scooped ice cream...and Poppy too!

We would like to thank Grace's Nonna for her unending support and organizational skills, the Charlestown Rathskeller Tavern staff, and, of course, our partner in crime—Tonia Hassell.

Until next year...!

Thank you everyone for all of your support,

Marybeth and Chris Caldarone



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Hereditary Neuropathy Foundation

@CMTNeuropathy



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