



cmtupdate



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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.

Letter from the CEO HNF’s Roadmap to Clinical Trial Development

BY ALLISON MOORE, CEO/FOUNDER, HNF

Dear Friends:

As we embark on the New Year, I am humbled by the support of our stakeholder community and the dedication to accelerate therapies to you and your families in the next few years. We are so close to the first treatment for CMT and we are doing our best to accelerate more treatments in the next few years. 2018 will be “the year” for our community as we prepare for the externally-led Patient-Focused Drug Development Meeting.

This meeting will take place in Washington, DC, on Friday, September 28. Our annual Patient-Centered Summit will follow on Saturday, September 29.

FOR MORE INFORMATION: WWW.CMT-PFDD.ORG AND WWW.CMTSUMMIT.ORG

HNF is inviting all advocates 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 the “Voice of the

(continued on page 2)



HNF'S ROADMAP TO CLINICAL TRIAL DEVELOPMENT

(continued from page 1)

Patient” report that can be used by FDA leadership as well as other stakeholders in the drug development process. This input can help inform the FDA’s decisions and oversight during drug development and the review of marketing applications for new drugs.

It is important for us, as patients to understand the years it can take to reach commercialization of a therapy approval by the FDA. The typical timeline from “bench to bedside”, which in layman’s terms means from disease models to successfully treating the disease, takes many years. The process of testing possible therapies in assays (cell and animal models), Biomarker Development is 12-15 years minimum. That means from the time of Basic Research (1-5 years) and Discovery to PreClinical (5-10 years) to Clinical to Distribution and Access (2-3 years). For people like us living with CMT, this is way too long!

HNF Stands for Acceleration of Therapies to Patients TODAY!

What’s next? HNF will be relaunching the new and improved Patient Registry — Global Registry of Inherited Neuropathies (GRIN) — and now, more than ever, we need your help! These patient reported outcome (PRO) research studies will be analyzed by CMT experts in the field and presented at the PFDD meeting. These patient outcomes will enhance the therapy development of our industry partners, inform all CMT scientists that have had break-throughs that may lead to treatments and most importantly, give us more insight into what matters most to you as CMT patients.

It is our time to let our voices be heard by FDA officials, legislators, the National Institute of Health and all stakeholders who have committed to joining us on this monumental day, September 28, 2018.

JOIN GRIN: WWW.NEUROPATHYREG.ORG



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.

Muscle Cramp Study Underway

Flex Pharma, Inc. is a biotechnology company developing innovative treatments for cramps and spasticity associated with severe neurological diseases, such as Amyotrophic Lateral Sclerosis (ALS), Multiple Sclerosis (MS) and peripheral neuropathies such as Charcot-Marie-Tooth (CMT). In October 2017, Flex initiated a Phase 2 study of CMT, referred to as the **COMMIT** study. The study will evaluate FLX-787, the Company’s co-activator of TRPA1 and TRPV1, in patients with CMT who suffer from muscle cramps. Approximately 120 subjects at 20 study centers across the United States are expected to take part. Participants will be in the study for approximately three months and will visit the study clinic three times. Enrollment is currently ongoing.

“Many of our patients with CMT neuropathy suffer from frequent and severe muscle cramping which can limit their activities of daily living and have a profound impact on their quality of life,” reported CMT clinical trial lead investigator, Dr. Nicholas Johnson, Assistant Professor of Neurology, Pediatrics and Pathology at the University of Utah. “We are hopeful FLX-787 will provide significant clinical advantages because of its safety profile and the lack of systemic exposure.”

Dr. William McVicar, Flex Pharma President and CEO, added “An important goal of the Hereditary Neuropathy Foundation (HNF) is to raise awareness of inherited neuropathies, such as CMT. Flex Pharma and the HNF both support research aimed at addressing the needs of patients, so we are excited to be partnering with the HNF in this effort to find a treatment for CMT patients suffering from painful cramps that impact their quality of life.”

FOR ADDITIONAL INFORMATION ABOUT THIS CLINICAL STUDY, PLEASE VISIT CLINICALTRIALS.GOV, IDENTIFIER NCT03254199 OR [HTTP://COMMITSTUDY.COM](http://COMMITSTUDY.COM)





What is an externally-led Patient-Focused Drug Development (PFDD) Meeting?

PFDD meetings integrate patient insights into the drug development process. Following the successful model that the FDA developed to host similar meetings, the day will focus primarily on a range of patient viewpoints on Charcot-Marie-Tooth (CMT), covering the symptoms and impacts to daily life that are most important to patients and their perspectives on existing and future treatments. This input can help inform the FDA's decisions and oversight during drug development and the review of marketing applications for new drugs.

Q: What is the format of the meeting?

A: The agenda will consist of speakers who will educate the FDA about CMT. In addition, there will be patient testimonials throughout the day to give the FDA an insider glimpse of the challenges patients face daily. This groundbreaking meeting will include facilitated discussions designed to provide the FDA with perspectives from people with CMT. It will also include caregivers, advocates, and all stakeholders who are part of the drug development continuum.

Q: Why is it important to educate the FDA?

A: 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***. Our goal is to educate the FDA and other stakeholders in the drug development process on the impact that CMT and other related inherited neuropathies have on quality of life for patients and families. We also seek to, as well as highlight what matters most to patients. This will impact clinical trial design, biomarkers, and improved outcome measures. It is also critical that we document the impact new therapies may have on mortality rates, comorbidities and disease severity.

Q: What are biomarkers?

A: Short for biological marker, a biomarker is something that can be measured to advance specific applications for diagnosing and/or predicting therapeutic response to a treatment. The biomarker helps to determine the presence of progression or improvement of a disease or disorder in the response to a treatment.

Q: What are outcome measures?

A: An outcome measure is the result of a test or tests that are objectively used to determine the baseline function of a patient at the beginning of a treatment. Throughout the clinical trials process, the same tests can be used to determine progress and treatment efficacy.

Q: What are comorbidities?

A: Comorbidities are the simultaneous presence of two or more diseases that typically are a result of, or strongly related to the primary disease which in our community is CMT.

Q: Who should attend the PFDD Meeting?

A: All CMT community members should join us either in person or by webcast to strengthen our impact on the FDA. This opportunity offers patients a birdseye view of HNFs research initiatives, and the impact and importance of bringing the patient's voice to the forefront of therapy development.

Q: How can I contribute to the meeting and mission?

- You can network and speak with fellow members of the CMT community through our interactive sessions to share your challenges to FDA officials and other stakeholders.
- You can submit your story for consideration to present at the meeting. A powerful and personal testimony will help us reach our goals. Travel and hotel expenses will be provided.

EMAIL COURTNEY@HNF-CURE.ORG

- You can contribute by sponsoring our Patient Focused Drug Development Fund. Your contribution will be tax deductible.

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

- We are developing a "Sentiment Wall", and we want to hear from you! These sentiments will be displayed at the meeting (First name and last initial only)— another way to show FDA officials and other key officials the impact of CMT on so many lives.

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



HEREDITARY
NEUROPATHY
FOUNDATION

Our Voices Will Be Heard

The Hereditary Neuropathy Foundation will be hosting two ground-breaking patient-centered outcome meetings to capture the "Voice of the Patient" so we can improve and implement protocols, treatments and the development of new therapeutics to meet our Charcot-Marie-Tooth (CMT) patients' needs.

If you are a health care provider, researcher or industry stakeholder with an interest in improving outcomes for CMT patients, this important two day event should not be missed.

Day 1

Friday, September 28, 2018

Public Meeting for Charcot-Marie-Tooth (CMT)
Externally-led Patient Focused Drug Development
(PFDD) Meeting
cmt-pfdd.org

Day 2

Saturday, September 29, 2018

Patient-Centered Charcot-Marie-Tooth (CMT)
Behavioral Health Summit
cmtsummit.org



For more information, email: info@hnf-cure.org or call: 212-722-8396

HNF'S HEALTH CARE PROVIDER DIRECTORY!

Join HCP

BY ESTELA LUGO, HNF MANAGER, MEDICAL OUTREACH

One of the most frustrating experiences for CMT patients is having to explain the disease to their own doctors and health care providers who have no experience in treating the patient community. Patients are often met with blank stares or misinformation. This climate creates feelings of hopelessness and mistrust for patients. HCPs have limited knowledge of CMT, in fact some have never even heard of it. HNF has made a stance to improve health care providers knowledge of CMT, help development standard-of-care-guidelines, and offer extended referral resources for providers included in our HCP Directory.

As a CMT patient and the Manager of Medical Outreach, I'm dedicated to offering patients a reliable resource for connecting to medical specialists with unique expertise and familiarity in caring for people with CMT. The specialists included in our directory will help patients manage their CMT and improve their quality of life across the spectrum of needs they may have. The directory includes neurologists, physical therapists, orthotists, and many others.

The HCP Directory aims to improve the process of finding local care resources of professionals with CMT-related experience, a list of HNF Centers of Excellence and HNF approved facilities that offer physiologic assessment as well as behavioral care. Our mission is to continue to grow the directory and connect patients to qualified healthcare professionals in the United States.

Dr. James Nussbaum, PT, PhD, EMT is a proud directory member, as well as the clinical Research Director of ProHealth & Fitness. His dedicated team of physical and occupational therapists have firsthand experience working with CMT patients.

"I joined the HNF Provider Directory because I am passionate about caring for people with CMT, and I know that the HNF is just as passionate in helping the patient community in so many ways. I'm confident my team can help people with CMT improve the quality of their lives through skilled physical and occupational therapy. Together with the HNF, we can connect like-minded expert providers, including orthotists, neurologists and others in creating specialized care teams to be a tremendous resource to patients and their families."

I am working with the HNF to help educate providers, specifically neurologists, so that each patient (with CMT) who sees them, is appropriately referred for a PT and OT evaluation, an orthotist assessment, and possible genetic counseling for help getting educated and genetically tested.

Such referrals by the neurologist should be the gold standard, not the exception. Each neurologist and her/his staff should have a clear and structured pathway for referral to expert providers who will all be part of the team of healthcare providers caring for the individual diagnosed with CMT."

We encourage Health Care Providers to join in HNFs Medical Educational Program.

By joining the directory, HCPs will not only make it easier for patients to find them, but they also will become a part of the network. Health Care Providers will also gain access to the following:

- Access to the Assessment Test in collaboration with AANEM
- Free CME course
- Inclusion in our patient HCP Directory
- Referral resources for your patients
- Registration discount to future CMT/IN meetings sponsored by HNF
- Access to affordable genetic testing
- Printed copy of the Quarterly CMT Update newsletter
- Immediate notification of ground-breaking research and clinical trials

Please share this opportunity with your HCPs to become part of our growing network!

FOR HCPs: [HTTPS://WWW.HNF-CURE.ORG/CMTPROVIDER/](https://www.hnf-cure.org/cmtprovider/)

FOR PATIENTS: [HTTPS://WWW.HNF-CURE.ORG/PROVIDER-DIRECTORY/](https://www.hnf-cure.org/provider-directory/)



Meet Dr. Nivedita Jerath

University of Florida Center of Excellence



Q: Tell us about yourself?

A: I love my patients and treat them like my own family. I grew up in Augusta, Georgia, played tennis at a national level and was blessed to be able to join Harvard University. I loved to help others, so I focused my energies on a medical career like my mom who is also a neurologist. I joined the Mayo Clinic College of Medicine for medical school, and then went back to Harvard University for my neurology residency at Mass General/Brigham and Women's hospital. I finally decided to join my husband in Iowa City after 13 years of long distance, where I did my fellowship and received a Muscular Dystrophy Association clinical training grant to study driving ability in those with CMT1A. I wanted to be close to home, so I moved from Iowa City to the University of Florida in Gainesville, FL.

Q: Why did you choose CMT as your focus of study and research?

A: I was always interested in peripheral neuropathy and was blessed to team up with a superb mentor, Dr. Michael Shy at the University of Iowa. I learned so much and realized that despite all my previous training, the first time I truly learned about CMT was in Iowa City, where I realized how rare and under-recognized CMT was, which is why I have chosen to study it.

Q: Why is CMT your passion?

A: For many reasons! I love finding new genes and mutations that cause peripheral neuropathies. Genetics is a new field of neurology, in which "idiopathic neuropathies" can now be explained by genetic mutations. Finding these genes will help us formulate treatments in the future. The scientific correlation between a genetic mutation and its clinical manifestations is fascinating to me. There is a lot to achieve in scientific knowledge and treatments in those with CMT. Also, CMT involves the most wonderful, kind and intelligent people I have ever met. I enjoy helping and treating patients and their families who have CMT.

Q: Tell us what patients will experience when they come to the HNF Center of Excellence (COE) at the University of Florida?

A: Patients will come to the UF and have a world class experience with an amazing facility, in-depth physician evaluation and opportunities to get other therapies that they need. They will also be involved in research studies including ones which will enhance neuromuscular physiology knowledge and driving studies.

Q: What are the benefits of a patient being evaluated at a HNF Centers of Excellence?

A: The benefits are many including a clinical research evaluation, evaluation for pain, evaluation for potential enrollment in a treatment trial, latest therapeutic advice, physical therapy evaluation, occupational therapy evaluation, genetic testing, podiatry consult, orthopedic surgery, and orthotics evaluation.

Q: What other doctors will patients be able to see when they visit the Centers of Excellence?

A: Pain physician, podiatrist, orthopedic surgeon, social worker, physical therapist, occupational therapist, research evaluation, and orthotist as needed.

Q: What kind of success stories have you seen so far with CMT patients?

A: The most important success stories involve patients getting a diagnosis that they have CMT and families finding out what genetic mutation is in their family. This is important for their kids and grandkids. Many patients are relieved to get a diagnosis—they knew something was wrong with them but didn't know what. It takes some experience and skill to recognize that a patient has a hereditary neuropathy.

On follow up visits, patients are excited to show off their ankle-foot orthotics or that a slight adjustment in their pain medication, stretching, or exercise regimen helped them. The diagnosis and visit give many patients the peace of mind that they've needed after years of searching or waiting for a diagnosis or a genetic cause. Some patients who are dealing with pain are overjoyed after visiting our pain specialist to help with the pain management. Other patients are excited to get involved with our research studies.

Q: How can patients make an appointment at the center?

A: Patients can call the access center at 352-294-5000 and specifically request for a visit with Dr. Jerath in the CMT clinic; no referral is necessary for an appointment with Dr. Jerath and multiple family members are welcome. Patients can also email Dr. Jerath at njerath@ufl.edu if they need to communicate with her for any reason.

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 is to ensure care results in positive outcomes for each individual patient's clinical experience. We are honored to partner with these premier Centers and their leading experts to improve the future for people with inherited neuropathies.

CALIFORNIA

Cedars-Sinai Medical Center

127 S. San Vicente Blvd.
Advanced Health Sciences Pavilion,
A6600
Los Angeles, CA 90048
Contact: Dana Fine
Phone: 310-423-8497
Email: Dana.Fine@cshs.org

Stanford Neuroscience Health Center Neuromuscular Clinic

213 Quarry Road, 1st Floor
Palo Alto, CA 94304
Phone: 650-723-6469.
Contact: Jennifer Fisher
Email: jnfisher@stanford.edu

CONNECTICUT

Hospital for Special Care

Charles H. Kaman Neuromuscular Center
2150 Corbin Avenue
New Britain, CT 06053
Contact: Boguslawa Koczon-Jaremko
Phone: 860-612-6356
Email: Bkoczon-Jaremko@hfsc.org

FLORIDA

University of Florida Health

2004 Mowry Road
PO Box 100332
Gainesville, FL 32610
Contact: Tracie Kurtz, RN, CCRP
Phone: 352-273-8517
Email: tlkurtz@ufl.edu

University of Miami

Professional Arts Center (PAC)
1150 NW 14th Street, 6th Floor
Miami, FL 33136
Contact: Meri Jaime (for appointments)
Phone: 305-243-7400
Email: MJaime@med.miami.edu

KANSAS

University of Kansas Medical Center

Landon Center on Aging
3599 Rainbow Boulevard, MS 2012
Kansas City, KS 66160
Contact: Nicole Jenci
Phone: 913-945-9934
Email: njenci@kumc.edu

MASSACHUSETTS

Brigham and Women's Hospital

Department of Neurology
75 Francis St, Tower 5th Floor
Boston, MA 02116
Contact: Kristen Roe
Phone: 617-525-6763
Email: kroec@partners.org

MICHIGAN

University Of Michigan

Pediatric Rehabilitation Center
2205 Commonwealth Blvd.
Ann Arbor MI 48105
Contact: Keianna Banbury
Phone: 734-763-2554
Email: kbanbury@med.umich.edu

MINNESOTA

University of Minnesota Health

Neurology Clinic
14500 99th Avenue N
Maple Grove, MN 55369
For Research Studies:
Phone: 612-624-7745
E-Mail: CNRU@umn.edu
For Clinic Appointments:
Phone: 763-898-1080

MISSOURI

St. Louis University Medical Center

Department of Neurology
1438 South Grand Boulevard
St. Louis, MO 63104
Contact: Susan Eller
Phone: 314-977-4867
Email: ellersc@slu.edu

NEW JERSEY

Hackensack University Medical Center

Neuroscience Institute, Department
of Neurology
360 Essex Street, #203
Hackensack, NJ 07601
Contact: Annerys Santos
Phone: 551-996-8100
Email:
Annerys.Santos@HackensackMeridian.org

NEW YORK

Columbia University

Department of Neurology
710 West 168th Street
New York, NY 10032
Contact:
For clinical appointments: Allan Paras
Phone: 212-305-0405
For research studies:
Phone: 212-305-6035
Email: ap3476@cumc.columbia.edu

WASHINGTON

St. Luke's Rehabilitation Institute

715 South Cowley Street, Suite 210
Spokane, WA 99202
Contact: Ann Cooper
Phone: 509-939-8079
Email: coopera@st-lukes.org

The CMT Community Needs You!

Start the New Year by getting involved and helping HNF conquer CMT. There are many ways you can get involved.

TD Bank Five Boro Bike Tour

May 6, 2018

Join the HNF Team and help raise funds for CMT research! Spots are available for patients, caregivers, family members, or friends. A great way to see New York and raise funds for CMT!

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

TCS New York City Marathon

November 4, 2018

Join Team CMT on Sunday, November 4 as we run the iconic TCS New York City Marathon. Help us pound the pavement by supporting our runners and cheer us along the race course or donate to our fundraising pages.

[HTTPS://WWW.HNF-CURE.ORG/TCS-NEW-YORK-CITY-MARATHON/](https://www.hnf-cure.org/tcs-new-york-city-marathon/)

Externally-Led Patient Focused Drug Development CMT Meeting

September 28, 2018

Let your voice be heard! Attend this groundbreaking meeting that will provide the FDA with perspectives from patients on symptoms and how CMT impacts them in their daily life.

REGISTER TODAY:

[WWW.CMT-PFDD.ORG](http://www.cmt-pfdd.org)

Research

Please donate to one or more of our research initiatives today!

[HTTPS://WWW.HNF-CURE.ORG/DONATE-CMT-RESEARCH/](https://www.hnf-cure.org/donate-cmt-research/)

Team CMT

Join the movement to make a difference for those living with Charcot-Marie-Tooth. Team CMT is a grassroots community fundraising program with 213 members in 39 states and 12 countries.

JOIN TODAY:

[HTTPS://WWW.HNF-CURE.ORG/TEAM-CMT/](https://www.hnf-cure.org/team-cmt/)

Birthday for a Cause

Have a birthday coming up? Ask your family and friends for the ultimate gift: treatments for CMT. Make a fundraising page and have a customized link to send out for your birthday.

[HTTPS://HNF.DONORPAGES.COM/BIRTHDAY/](https://hnf.donorpages.com/birthday/)

Legacy Planning

Give the gift of a lifetime. You can support the Hereditary Neuropathy Foundation in many ways, with gifts that do not require an immediate change to your current spending or savings. By considering a planned gift through your estate, life insurance, will or retirement plans, you can achieve your philanthropic goals and ensure that our mission will continue.

[HTTPS://WWW.HNF-CURE.ORG/LEGACY-PLANNING/](https://www.hnf-cure.org/legacy-planning/)

“Em-bracing Fashion”

with Lainie Ishbia

Tell us about yourself.

I'm a 47-year-old mom, wife, blogger, social worker, empowerment speaker/educator and fashionista. Besides hanging with my family, I love entertaining, traveling, attending my monthly book group (more for the wine than the books) and thrift store shopping.

Why did you start Trend-ABLE?

I inherited CMT from my mom and, unfortunately, my daughter Zoe, inherited CMT from me. In my professional life as a therapist and girl-empowerment-speaker, I focused on helping teenage girls and women with their self-esteem related issues. There is a definite connection between self-confidence and taking pride in one's appearance. When people felt positive about what they were wearing and how they looked, they also felt better about themselves on the inside where it matters the most.

As a teenager, I had a very poor self-image. I felt fat and ugly and hid my body inside of baggy clothing. Having Charcot-Marie-Tooth, which challenged my mobility, gave me an excuse to get out of gym class and ultimately not challenge myself physically. I let my disability limit me and did not take pride in how I looked because I thought it was hopeless. This led to depression and ultimately a cry for help at age 16.

I eventually figured it all out and grew up. I stopped letting my disability be an excuse for not exercising and began taking care of myself. I ate better, lost weight, started weight training, learned how to apply makeup and began dressing in clothing that made me feel good and highlighted the parts of my body I didn't mind. Even though my physical problems were slowly getting worse, I was more confident than ever.

Having a physical disability that isn't obvious to others definitely has its own set of challenges. I don't wear my leg braces on the outside of my clothing or focus on the fact that my hands don't work so well. I just live my life but it's often a struggle both internally and with things like finding cute shoes that fit or putting on necklaces. Since my disability isn't obvious, I sometimes get self-conscious having to balance at parties and feeling confident. Dating was an issue for me after my divorce.



I saw a ton of support groups for my disease but none of them were focused on the issues I really cared about and there wasn't a group focusing specifically on women with invisible physical disabilities. Trend-ABLE is a place for Perfectly Imperfect Women who want to look and feel their best.

How has launching the website changed your life in a short period of time?

I launched my Trend-ABLE website less than two weeks ago, and at this point over 2000 people have visited the site. Both the Facebook and Instagram pages now have followers from all over the world. I get emails from people and organizations daily with either awesome feedback and/or questions. It feels amazing to have created something that was clearly needed.

What are your future goals for Trend-ABLE?

Right now, I am focused on growing the Perfectly Imperfect community. As Trend-ABLE grows, so will my understanding of my tribe's wants and unmet needs. So, stay tuned!

What is your top fashion tip for individuals with CMT?

Be creative and don't give up. Sometimes what seems impossible just needs to be modified. For example, a simple thing like sewing up the buttons on a stretchy shirt and making it a pullover or putting magnetic closures on your favorite jewelry. We call these "Trend-ABLE TWEAKS" on the website.

What do you want readers to take away from your blog?

I want readers to feel like their time was well spent. I want them to feel like, "She gets it" and "I'm going to use that tip/idea". We have too many social media time-wasters that leave us feeling bad about ourselves. I want Trend-ABLE to empower women with invisible physical disabilities and challenges.

VISIT TREND-ABLE: [HTTP://TREND-ABLE.COM/](http://trend-able.com/)

Breathing Problems?

What Do They Mean, Breathing Problems?

BY MICHELLE BROSE



Though Charcot-Marie-Tooth is rarely fatal, respiratory complications are one of the ways it's possible to die from CMT. Breathing requires the coordinated movement of several muscle groups; the diaphragm and abdominal muscles, the intercostal muscles that expand your rib cage, and even the bulbar muscles in your neck. It's possible for Charcot-Marie-Tooth to cause damage to the nerves that power these muscles, resulting in weakness, atrophy, and respiratory insufficiency. But the symptoms of respiratory problems in people with neuropathy aren't well known, even among doctors. It's usually more subtle than simply feeling short of breath, so people often attribute their symptoms to other causes.

Hypercapnia

The first sign of breathing problems in CMT usually isn't shortness of breath; it's hypercapnia, the retention of carbon dioxide. The symptoms generally begin to be noticeable at night, because your weakened diaphragm is being pressed on by the contents of your abdomen and your chest can't expand fully while lying flat. When you're breathing shallowly at night, the carbon dioxide in your lungs isn't being exhaled completely, and it begins to build up in your blood. Symptoms include headaches, fatigue, difficulty concentrating or staying awake during the day, and restless sleep. You may have anxiety when lying flat, nightmares, or unusually vivid dreams from your REM sleep being interrupted.

During the day, you might have difficulty making yourself heard when speaking, or find that your sentences are trailing off because you have to take a breath in the middle of them. You may also catch yourself using your shoulder and neck muscles to help you get a breath, or notice that your diaphragm is retracting with the effort to breathe.

If the carbon dioxide buildup isn't stopped, you may experience an altered mental state. You may become confused, and have others tell you you're not making sense when you speak. You may "black out"; your brain temporarily losing the ability to form short-term memories. You might even lose consciousness. In a serious respiratory crisis like this, emergency room doctors may decide to perform a tracheostomy and put you on a ventilator.

If you're experiencing any of the above symptoms, you can avoid a crisis by using a BiPAP machine with a facemask at night.

It will assist your natural breathing by helping you take deeper breaths while you sleep, which will exhale the excess carbon dioxide. You'll wake up feeling refreshed, with more energy to breathe deeply during the day.

People with more severe forms of CMT may eventually find that they need ventilatory support in the day-time as well. That can also be provided noninvasively, with either a facemask or a mouthpiece, for people who wish to avoid a tracheostomy tube.

Weak cough

When the respiratory muscles are too weak to provide an effective cough, mucus can build up in the lungs, trapping bacteria and preventing oxygen from getting in. To prevent pneumonia, you could use an "assisted coughing" method like having somebody push up sharply on your diaphragm while you cough. There are also postural drainage techniques that can make it easier to dislodge and cough up mucus, by raising your hips above your head and shoulders when you're lying down. The most effective way of clearing your lungs is a noninvasive machine called the Cough Assist; it inflates your lungs via facemask or trach tube, then pulls it and the mucus out again.

What's wrong with getting extra oxygen?

When you get supplemental oxygen, the higher levels of O₂ in your blood trick the respiratory center in your brain into thinking that you're breathing more deeply than you really are. Your brain decides it's safe to take it easy and put less effort into breathing. Extra carbon dioxide will quickly build up to

dangerous levels, eventually overwhelming the oxygen being carried by your blood. In extreme cases, you could suffocate, and require emergency intubation and invasive ventilation. Dr. John Bach, who literally wrote the book on pulmonary rehabilitation, often says that “Giving oxygen [to patients with neuromuscular diseases] is like applying a Band-Aid to cancer.”

What can I do to prevent respiratory complications?

It’s important to be proactive when it comes to your breathing. If you delay getting care for breathing problems, you may put yourself at risk for a respiratory crisis or pneumonia. The first thing to do is get a baseline pulmonary function test from a pulmonologist who’s been trained in how to manage patients with neuromuscular diseases. Not all of them understand our

unique needs, so it’s best to get a recommendation from an HNF Center of Excellence (<https://www.hnf-cure.org/centersofexcellence/>). Follow up on this regularly (about once a year) to check for any changes or decline.

Part of the test is checking your carbon dioxide levels. This can be done one of two ways; either by the traditional arterial blood gases (ABG) test, or by measuring the amount you exhale with a capnometer. Opinions vary as to which method is more accurate, but if you have a fear of needles, you can seek out a doctor who will use a capnometer instead.

Most importantly, always listen to your body. Don’t delay bringing up any concerns about your breathing with your doctor in hopes they’ll go away. Once your respiratory function has been addressed, you’ll always breathe easier.

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

Everything seems okay until your...

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

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

CMT CAREERS

Melissa Amalfitano (Pastry Chef)

CMT, No Piece of Cake



Sometimes physical challenges can dictate job and career paths. When our bodies are limited, fear can weave itself between our passions and confidence. It takes a strong and bold mindset to move towards something we already know will be extra difficult.

Melissa Amalfitano did exactly that when she made the decision to attend the Culinary Academy of Long Island for Pastry Arts 10 years ago. Diagnosed at only two years old, Melissa was well aware of the effects CMT had on daily living. Paralysis of her feet made standing in leg braces for long period incredibly painful. Grabbing and holding objects with atrophied hands took an extra dose of energy and patience. Despite concern and opposition from others, Melissa started classes and graduated nine months later at the top of her class.

Let's hear from Melissa:

It started when cake-challenge shows became popular. I loved watching people creating these incredible pieces of edible art. I became inspired and began creating my own cakes for family and friends. I would sit for hours molding all sorts of different candies to use as decorations. It felt like I was a kid playing with Playdoh again. Soon, several people were telling me that I should consider making it into a career for myself.

Time is always against me. Whenever I try to speed up, my hands just go numb and don't work well. I'll never forget this woman I trained for in a bake shop yelling at me, "What's with your hands? You can't do this if there's something wrong with your hands...you are too slow!" I left that day in tears and never returned. I questioned myself the whole drive home. "Maybe she's right...I'm a fool to have gone back to school to make a career of something using my hands".

But that feeling of helplessness soon turned to determination. It led me back to my first internship where I was accepted and where my disability didn't matter. My two mentors were very understanding and previous TV cake stars themselves. They gave me all time I needed, and that's when my skills and confidence grew. **That's when I watched my ability begin to soar.** I'll always be grateful to them for seeing so much more in me.

It takes plenty of patience to create edible art; having a disability on top of that requires even more for my body to adapt. **This type of work is extremely physical.** You're spending hours on your feet, and hunched in a chair, decorating. I remember at the end of some weeks, I could barely walk without pain. My hands would be so overworked, that I struggled to tie my shoes or hold a fork.

I've since adapted to taking more breaks. As far as my hands go, I hold everything differently...from knives to pastry bags. I can't explain how exactly, but I always find a way to adjust how I work; whether it's piping a certain design on a cookie or holding a knife to sculpt a cake. **My mind and body just know what to do.**

I'm constantly challenging myself to create different designs and strive to make it better every time. Each time I hear myself thinking, "There's no way it's going to come out that way, how am I going to pull it off?" But I do, and each time it's better than I imagined. My customers' incredible reactions are what makes it all worthwhile. **When they see my creations, no one is seeing a woman with a disability. They are only seeing talent.** I'm going to continue to challenge myself and this disease. It has limited me to certain areas of my life, but I refuse to let it take away from this one.

I'm am blessed to always have the support of my husband, parents, family, and friends who continue to push and love me.

Meet the 2018 TD BANK FIVE BORO BIKE TOUR TEAM CMT

BY COURTNEY HOLLETT, EXECUTIVE DIRECTOR

The Hereditary Neuropathy Foundation (HNF) is thrilled to have been chosen for the eighth year in a row as a Charity Partner for the 2018 TD Bank Five Boro Bike Tour. This means more participants riding to increase awareness of Charcot-Marie-Tooth (CMT), and thousands more dollars raised to fund research for treatments and a cure!

The ride takes place on Sunday May 6, 2018 and Team CMT riders will cover 42 miles through the five NY boroughs. The ride starts just north of Battery Park, up through Manhattan, Central Park, around a brief loop in the Bronx and down to the Queensboro Bridge. After a ride over the Pulaski bridge passing through Brooklyn, participants will enjoy an incredible view from the lower deck of the Verrazano Bridge, a entire one mile uphill ride with a 1 mile downhill ride of relief before entering the X Park to celebrate another amazing year!

Here's a snapshot of the inspiring Team CMT riders and why they are participating in this year's TD Bank Five Boro Bike Tour:

Matt & Donna Downing

A husband and wife team that works tirelessly with HNF to raise funds for HNF's Therapeutic Research In Accelerated Discovery (TRIAD) program that supports CMT research. Matt also is HNF's Board of Director Secretary and works behind the scenes. Matt has biked all eight years in the TD Five Boro Bike Tour.

[HTTPS://HNF.DONORPAGES.COM/BIKENEWYORK2018/DOWNING/](https://hnf.donorpages.com/bikenyork2018/downing/)

Debi Houliares

Debi is an HNF board member and is riding in honor of her son Zach who has CMT6, an extremely debilitating form that also causes blindness. Debi is passionate about raising funds to support gene therapy approaches to cure CMT.

[HTTPS://HNF.DONORPAGES.COM/BIKENEWYORK2018/DEBORAHHOULIARES](https://hnf.donorpages.com/bikenyork2018/deborahhouliares/)

Allison Moore

Allison is the CEO/Founder of HNF and has participated in this event year after year. She has dedicated her life to those with CMT and has been critical in collaborating with patients, researchers, industry and all stakeholders in accelerating therapies that are now in clinical trials.

[HTTPS://HNF.DONORPAGES.COM/BIKENEWYORK2018/ALLISONMOORE/](https://hnf.donorpages.com/bikenyork2018/allisonmoore/)

Tess Quadrozzi

Tess is a dedicated year after year rider in the TD Bank Five Boro Bike Tour in honor of her friend Allison Moore and we are grateful for her dedication. Tess is an avid biker and Allison says,

no one seems to love this ride more than her! She often will take Allison along on her long rides leading up to the big day so she is as prepared to take on the full 42 mile ride.

[HTTPS://HNF.DONORPAGES.COM/BIKENEWYORK2018/TESSQUADROZZI/](https://hnf.donorpages.com/bikenyork2018/teessquadrozzi/)

Patrick & Joe Reilly

This father son team are riding for their daughter/sister Dakota who has CMT1A and had so much fun last year that they decided to join the team again in 2018. Dakota and her mother Kerin tireless support HNF and are pivotal in shedding light on how difficult it is to live with CMT and have been in the spotlight - national news.

[HTTPS://HNF.DONORPAGES.COM/BIKENEWYORK2018/JOEANDPATRICK/](https://hnf.donorpages.com/bikenyork2018/joeandpatrick/)
VIDEO CLIP: [WWW.HNF-CURE.ORG/PHARNEXT-PLEODRUG-CMT1A/](http://www.hnf-cure.org/pharnext-pleodrug-cmt1a/)

Dr. Florian Thomas

Our special friend and caring neurologist who is always there to support HNF, and its members (especially those patients and families) takes on NYC with his lovely wife, Sarah Griesbach and will ride in tandem to support CMT research!

[HTTPS://HNF.DONORPAGES.COM/BIKENEWYORK2018/FLORIANTHOMAS/](https://hnf.donorpages.com/bikenyork2018/florianthomas/)

Dave Washabaugh

Dave is an avid cyclist and will ride for the second year as a charity rider. He and some of his family members have CMT, so he has dedicated this race to raise funds for CMT. Dave has been an important contributor to our patient centered research initiatives.

[HTTPS://HNF.DONORPAGES.COM/BIKENEWYORK2018/DAVEWASHABAUGH2017/](https://hnf.donorpages.com/bikenyork2018/davewashabaugh2017/)

Jeffrey Wilson

Jeffrey joined Team CMT to ride in the TD Bank Five Boro Bike Tour. He is riding to raise funds for research and to help people with CMT and their families. Jeff's son has CMT. This will be Jeff's third year in a row participating in this ride. In fact, he rode in the very first Spin-For-Cure and we are grateful to him and the rest of our riders for their selflessness in supporting our community.

Rob Walling

Rob is an actuary from Illinois who loves road cycling. He and two of his children, including a daughter at NYU, have CMT-1A. Rob is actively involved in CMT genetic research at the University of Iowa and with Pharnext clinical trial and looks forward to raising funds and awareness through the ride.

[HTTPS://HNF.DONORPAGES.COM/BIKENEWYORK2018/ROBWALLING/](https://hnf.donorpages.com/bikenyork2018/robwalling/)



ANNUAL PATIENT-CENTERED SUMMIT

Utilizes Voice Activation Technology to Capture Pain in Patient Reported Outcomes in CMT/HNPP

BY ALLISON MOORE FOUNDER & CEO/PRINCIPAL INVESTIGATOR, HEREDITARY NEUROPATHY FOUNDATION

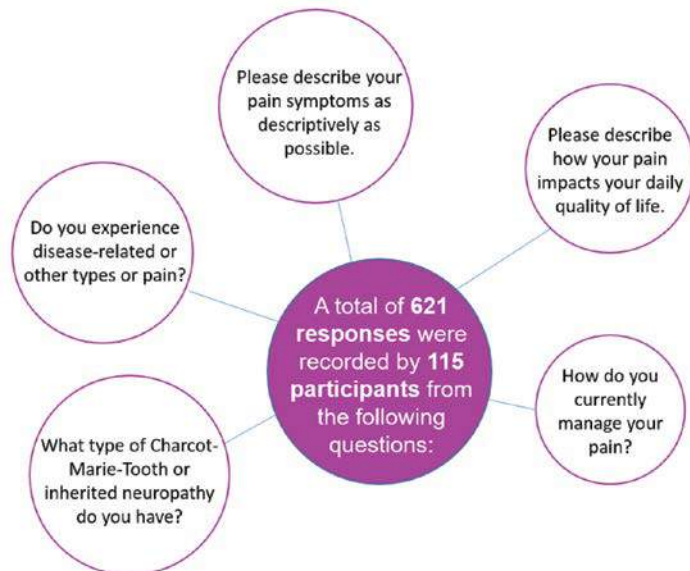
On November 3, 2017, nearly 100 participants gathered at the Samberg Conference Center on the Massachusetts Institute of Technology (MIT) campus in Cambridge, MA for the Hereditary Neuropathy Foundation (HNF) Patient-Centered Charcot-Marie-Tooth (CMT) / Hereditary Neuropathy Pressure Palsies (HNPP) Pain Summit. The meeting brought together people with hereditary neuropathies and their family members, caregivers, clinicians, researchers, funding agencies, payors, leading pain experts and pharma industry to provide an in-depth look at chronic pain within the CMT/HNPP community, including its impact on quality of life. Funded in part by a Eugene Washington PCORI Engagement Award and industry partners; Pharnext Pharma, Acceleron Pharma, Flex Pharma and a grant from Pfizer, this one day conference offered expert sessions as well as breakout sessions, primarily

patient-led, with a focus on patient engagement methods: all with an emphasis on identifying gaps that are hindering patient care in neuropathic and musculoskeletal pain, diagnosis and identifying biomarkers and outcome measures to support therapy development.

The prevalence of pain in this patient community became apparent during HNF's 2016 inaugural Patient-Centered CMT Summit. To validate this issue, HNF analyzed data from its Global Registry for Inherited Neuropathies (GRIN), to determine which co-morbidities were most prevalent and most important to patients. One question asked the one word they would use to describe their disease: Pain was the Number One response. As a result, HNF decided that the 2017 Summit needed to focus on assessing and addressing the gaps in pain management for the hereditary neuropathy patient community.

To further our knowledge of the patients experience on the impact pain is having on quality of life, and to inform the stakeholder community at the Summit, we captured the CMT patient's experience with pain in their own words. HNF partnered with the innovative, voice-powered survey platform, True Reply — www.truereply.com — to record patient responses to a 5-question survey in their own voice. The study was run over a 30-day period prior to the Summit.

A total of **621** responses were recorded by **115** participants from the following questions:



The depth and variety of the patient responses to these questions were revelatory, but not surprising, to HNF Founder and CEO, Allison Moore:

“Hearing about our patients’ experiences with pain in their own words was both enlightening and heartbreaking at the same time. Our patients are hurting badly in so many ways, and they need guidance and protocols from their healthcare providers to help manage their pain so they can go about their daily lives as pain-free as possible.”

KEY TAKEAWAYS FROM THE STUDY INCLUDED:



90%

of patients indicated that their pain has a moderate to severe impact on their quality of life.



76%

of patients are managing their pain on their own, with a combination of over-the-counter medications and alternative therapies.

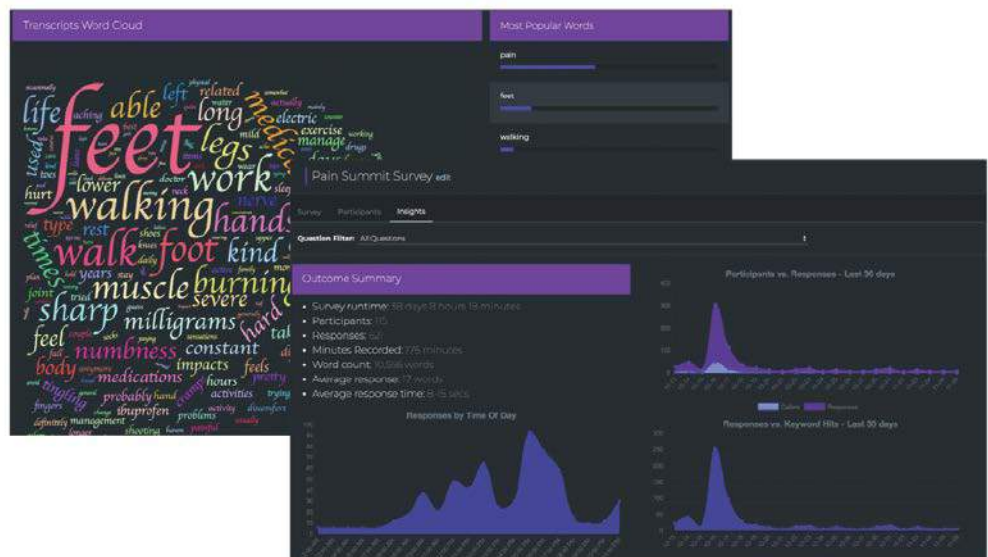


63%

of patients described their pain as numbness, sharp, burning or stabbing.

Jose Cotto, Founder and CEO of True Reply, helped HNF analyze and quantify the results. Said Jose:

“The ability of True Reply to quantitatively analyze patient responses in real-time while also giving researchers and clinicians access to qualitative data such as patient voice tone, cadence and stress levels is a real game changer for Patient Reported Outcome (PRO) studies.”



HNF is looking forward to integrating True Reply technology into future CMT PRO studies leading up to HNF’s 3rd Annual Patient-Centered CMT Summit — www.cmtsummit.org — on September 29, 2018.

In addition, as part of the FDA’s externally-led Patient-Focused Drug Development (PFDD) initiative, HNF will be hosting a PFDD meeting in Washington, D.C. on September 28th, the day before the Summit. FDA’s PFDD initiative aims to more systematically obtain the patient’s perspective on the burden of specific diseases and current treatments. This meeting will inform the FDA, drug developers and other key stakeholders, what is most important to patients and how patients view the benefits and risks of treatments for CMT.

“The voice of the CMT patient can no longer be ignored when it comes to the protocols, treatments and therapeutics that are being developed to treat this disease. We are looking forward to using technologies like True Reply to help us tell our patients’ stories in their own words so we can address their immediate quality of life issues while waiting for desperately needed therapeutics to move through the pipeline and be approved for commercial use.”

– Allison Moore

VISIT: [HTTPS://TRUEREPLY.COM/HEALTHCARE](https://truereply.com/healthcare)

Events

5/6/18

6/2/18

9/28/18

9/29/18

UPCOMING 2018 EVENTS

Save the date!

**TD Bank Five Boro
Bike Tour**
New York, NY

**An Evening for
Charity**
BH Kitchen & Bar
New York, NY

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

**Patient-Centered
CMT Summit**
Washington, DC

HNF Chosen to be 2018 TCS New York City Marathon Charity Partner!

BY COURTNEY HOLLETT, EXECUTIVE DIRECTOR

The Hereditary Neuropathy Foundation is ecstatic to be included among many prestigious charities as a charity partner for the TCS New York City Marathon on November 4, 2018.

Our founder/CEO Allison Moore has been holding out hope for years to have HNF accepted as a charity partner. While training in 1995-96 for the Marathon, she was diagnosed with a rare cancer, Synovial Sarcoma. She was given the chemotherapeutic agent vincristine, which is one of the most toxic drugs on the neurotoxic drug list for those with CMT. Allison soon experienced the symptoms of CMT after receiving vincristine. Her dreams of running the marathon were shattered. Allison's dream, however, will come true as a participant in this year's Marathon. Although she cannot run it, she has committed to speed walk the race. This is no easy feat!

The course is a 26.2-mile race through The Big Apple. The first NYC Marathon took place in 1970 and was held in Central Park. Out of the 127 participants, only 55 men finished and the sole female runner dropped out. Fast forward to 2017: over 50,773 participants completed the race and the world witnessed a historic win by Shalane Flanagan, the first American woman champion in 40 years!

The other Team CMT members who will join Allison as they all will dedicate the next 10 months to training, increasing awareness and raising funds for CMT Research.

TO DONATE



Pictured (l to r) Joy Kaye, Allison Moore, Kerin Reilly, Chris Wodke

We are honored that these runners will pound the pavement in this iconic race. Please consider making a donation to these amazing women who are 0.5 % of the population that will conquer this historic race!

HNF will be setting up cheering spots along the race course. We hope you can join us. You can email courtney@hnf-cure.org for more information.

— Follow Allison —

as she blogs about her training for the TCS New York City Marathon. She will share with the CMT Community her triumphs as she conquers this amazing feat!

<https://www.hnf-cure.org/allison-moore-tcs-new-york-city-marathon/>

It has been almost three weeks since I have started training for the marathon, a feat that I am determined to complete. So far it's been interesting as I navigate all the equipment at the new gym I recently joined and overcoming many challenges as I build up to a 26.1 mile speed walk.



**HEREDITARY
NEUROPATHY
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 Hereditary Neuropathy Foundation

 @CMTNeuropathy



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